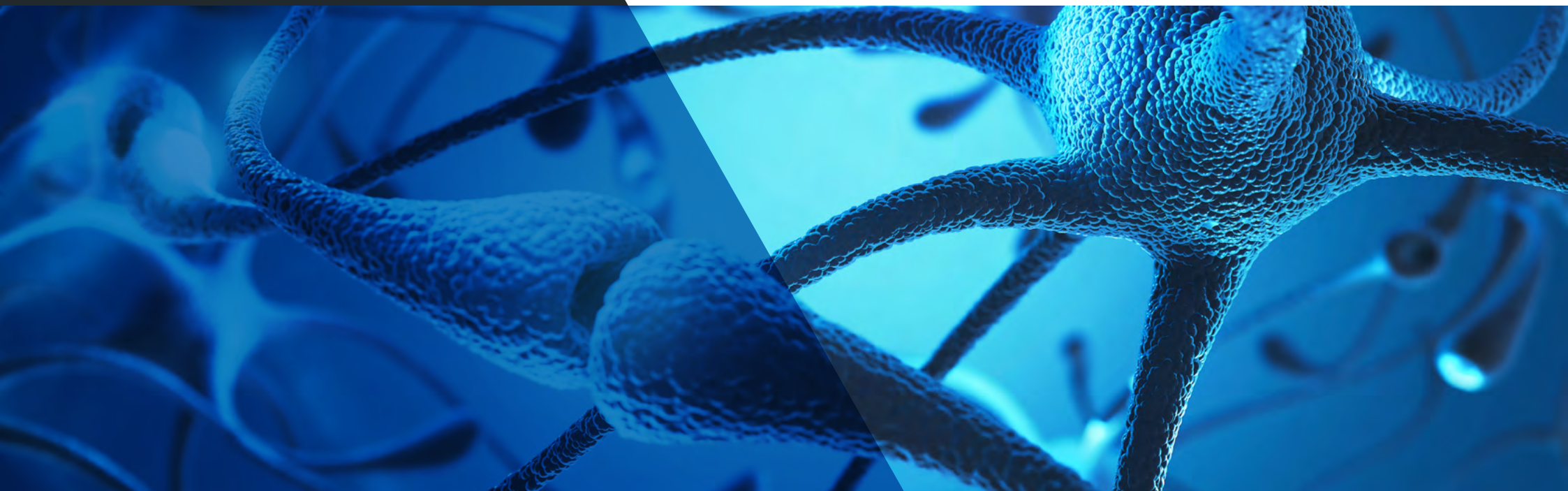


**SJD**

Sant Joan de Déu  
Institut de Recerca

ANNUAL SCIENTIFIC  
**REPORT**  
**2019**



science · innovation · commitment

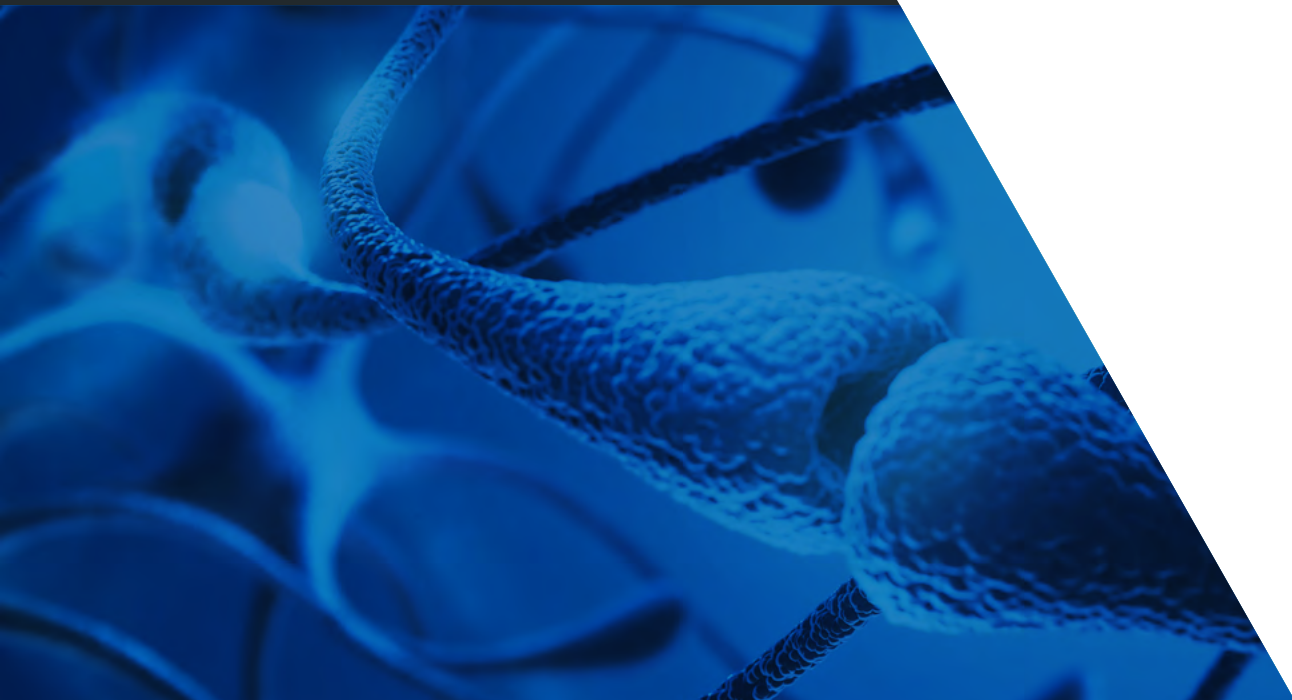
## Index

|                                                     |           |                                                                                       |           |                                                                             |           |                                                              |           |
|-----------------------------------------------------|-----------|---------------------------------------------------------------------------------------|-----------|-----------------------------------------------------------------------------|-----------|--------------------------------------------------------------|-----------|
| <b>1. FOREWORD</b>                                  | <b>3</b>  | <b>7. NEUROSCIENCE</b>                                                                | <b>26</b> | <b>8. ADAPTIVE RESPONSE, METABOLISM AND IMMUNOBIOLOGY</b>                   | <b>44</b> | <b>10. HEALTH TECHNOLOGIES AND INNOVATION</b>                | <b>64</b> |
| Director's foreword                                 | 4         | Metabolic and mitochondrial medicine                                                  | 28        | Metabolic endocrinology                                                     | 46        | Medical technologies:                                        |           |
| Governance and structures                           | 5         | Applied research in neuromuscular diseases                                            | 29        | Metabolic diseases of paediatric origin                                     | 47        | Bioinformatics and biomedical signals                        | 66        |
| Global numbers 2019                                 | 6         | Pediatric neurometabolism: neural communication mechanisms and personalized therapies | 30        | Molecular biology and gene regulation of adipose tissue and its pathologies | 48        | Medical technologies:                                        |           |
| Global numbers 2015-2019                            | 7         | Neurogenetics and Molecular Medicine                                                  | 31        | Neonatology research group                                                  | 49        | Biomaterials and Tissue Engineering                          | 67        |
| <b>2. STAFF</b>                                     | <b>8</b>  | Neonatal brain                                                                        | 32        | Pharmacological targets in inflammation and metabolic diseases              | 50        | Medical technologies:                                        |           |
| Research staff                                      | 9         | Neuropsychology                                                                       | 33        | Study of diseases due to immune dysfunction in pediatrics (GEMDIP)          | 51        | Analysis for Rehabilitation                                  | 68        |
| HRS4R award                                         | 9         | Cognitive neuroscience                                                                | 34        | Infectious diseases and microbiome                                          | 52        | Medical technologies:                                        |           |
| <b>3. PROJECTS AND CLINICAL TRIALS</b>              | <b>10</b> | Attention, Perception and Language Acquisition (APAL)                                 | 35        | Gut microbiota                                                              | 53        | Instrumentation and eHealth                                  | 69        |
| New projects                                        | 11        | Human molecular genetics I                                                            | 36        | Environment and complications in pregnancy                                  | 54        | Medical technologies:                                        |           |
| Active projects                                     | 11        | Human molecular genetics II                                                           | 37        | Cardiovascular diseases in development                                      | 55        | Graphic computing: serious games and gamification            | 69        |
| Active clinical trials and observational studies    | 12        | Etiopathogenesis and treatment of severe mental disorders (MERITT)                    | 38        | Immune and respiratory dysfunction in the critical child                    | 56        | Medical technologies:                                        |           |
| New clinical trials and observational studies       | 12        | Research in child and adolescent mental health                                        | 39        | <b>9. PAEDIATRIC CANCER</b>                                                 | <b>57</b> | Robotics and vision                                          | 70        |
| <b>4. SCIENTIFIC PRODUCTION AND RESULTS</b>         | <b>13</b> | Epidemiology of mental disorders and aging                                            | 40        | Translational Genomics                                                      | 59        | Medical technologies:                                        |           |
| Scientific publications                             | 14        | Health Technology Assessment in Primary Care and Mental Health (PRISMA)               | 41        | Sarcomas and Histiocytosis                                                  | 60        | Medical imaging                                              | 70        |
| Highest impact factor journals                      | 14        | Psychological research group on fibromyalgia and chronic pain (AGORA GROUP)           | 42        | Therapy Development in Pediatric Oncology                                   | 61        | Medical technologies:                                        |           |
| Patents and utility models                          | 18        | Cerebral Imprints of Gender, Development and Pathology                                | 43        | Paediatric leukaemia and haematopoiesis                                     | 62        | Biomechanical                                                | 71        |
| Thesis                                              | 19        |                                                                                       |           | Molecular pharmacology and experimental therapies                           | 63        | Patients Engagement in Research                              | 72        |
| <b>5. CORE FACILITIES</b>                           | <b>21</b> |                                                                                       |           |                                                                             |           | Innovation in Health Technologies                            | 73        |
| <b>6. TRAINING, COMMUNICATION AND PARTICIPATION</b> | <b>23</b> |                                                                                       |           |                                                                             |           | Diagnostic and Therapeutic Image                             | 74        |
| Seminars                                            | 24        |                                                                                       |           |                                                                             |           | <b>11. APPLIED CLINICAL RESEARCH</b>                         | <b>75</b> |
| Training programme                                  | 25        |                                                                                       |           |                                                                             |           | Influence of the environment on child and adolescent welfare | 77        |
| Communication and public engagement                 | 25        |                                                                                       |           |                                                                             |           | Paediatric ophthalmology                                     | 78        |
|                                                     |           |                                                                                       |           |                                                                             |           | Hospital Dentistry and Periodontal Medicine                  | 78        |
|                                                     |           |                                                                                       |           |                                                                             |           | Research in nursing                                          | 79        |
|                                                     |           |                                                                                       |           |                                                                             |           | <b>ANNEX 1</b>                                               | <b>80</b> |
|                                                     |           |                                                                                       |           |                                                                             |           | Active clinical trials and observational studies during 2019 | 81        |



ANNUAL SCIENTIFIC  
**REPORT**  
**2019**

**SJD**  
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Institut de Recerca



**1**

## **FOREWORD**

Director's foreword  
Governance and structures  
Global numbers 2019  
Global numbers 2019-2015

## Director's foreword

Once again, it is a great pleasure to present the work carried out at the Institut de Recerca Sant Joan de Déu (IRSJD). Within the framework of the IRSJD Strategic Plan, our scientific activity in 2019 has been structured around four main research programmes: Neurosciences; Adaptive Response, Metabolism and Immunobiology; Paediatric Cancer, and Health and Innovation Technologies. A fifth area has also been integrated, Associated Clinical Groups, consisting of research groups with a stronger patient care and clinical research profile. This annual report represents the joint effort of our team here at the Institute in carrying out high quality translational research.

Briefly summarising some of the other activities undertaken within the framework of the Strategic Plan, I should highlight, firstly, our new Research and Evaluation Career Plan created with the joint efforts of researchers and Scientific Management to establish and define the typology, classification

and evaluation of research personnel. Secondly, I should mention the efforts dedicated to creating shared spaces for scientific reflection and to foster further research and collaboration proposals. For example, we held a 1-day conference on innovation, ("Innovation Unconference" at the Research Centre for Biomedical Engineering (CREB) of Universitat Politècnica de Catalunya) and another on neuroscience ("The multi OMICS Day" at Hospital Sant Joan de Déu Barcelona). Finally, I must also expressly welcome on-board Dr Marisol Montolio (Scientific Director of the Duchenne Parent Project Spain) as a member of our external Scientific Committee at IRSJD, conferring on patients the voice they deserve.

The Institute's indicators for 2019 have remained stable, with improvements in some aspects, more competitive projects, more publications in prestigious journals and a considerable increase in the number of clinical trials. We must, therefore, congratulate

all those investigators who have made this possible. To mention just a few, I would like to highlight some of the especially relevant accomplishments of the Paediatric Cancer Programme: coordination of a new European project with strong ties with Latin America (M. Camós), coordination of three projects funded by *La Marató TV3* (J. Mora, C. Lavarino and A. Montero-Carcaboso) and participation in another (S. Mateo), and an excellent paper led by A. Montero-Carcaboso published in *Science Translational Medicine*. We also coordinated another European project in the area of Mental Health (JM. Haro), and participated in another ten projects, reinforcing the international collaboration of other researchers in other programmes. Furthermore, we have been awarded over twenty new projects under the National Plan for Scientific and Technical Research and Innovation which allow continuity of consolidated lines of research as well as opening of new ones. Our investigators

have published in the *Lancet Diabetes Endocrinology* (L. Ibáñez), *Neurology Neuroimmunology & Neuroinflammation* (L. Alsina), *British Journal of Pharmacology* (M. Vázquez), *Lancet Neurology* (J. Hoenicka), *Annals of Neurology* (M. Serrano), *Journal of the American Academy of Child & Adolescent Psychiatry* (E. Camprodon-Rosanas), *Journal of Clinical Medicine* (R. Artuch), *Bioinformatics* (A. Perera) and *PLoS Computational Biology* (S. Picart). This is also evidence of rigorous efforts, scientific collaboration and translation of knowledge into clinical practice.

Finally, and having thanked you all again for this year's achievements, I would like to remind you that only by carrying out quality research in any of our specific areas can we consolidate our Research Institute as an international centre of excellence in Paediatric Medicine.

**Francesc Palau Martínez**  
Director,  
*Institut de Recerca Sant Joan de Déu*

## Governance and structures

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Scientific Manager, Institut de Recerca  
Sant Joan de Déu

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Investigaciones Oncológicas (CNIO)

**Jordi Camí i Morell**  
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**Marisa Montolio**  
Scientific director. Duchenne Parent  
Project, Spain

**Michael B. King**  
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of Psychiatry

**Carlos Rodríguez-Galindo**  
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Children's Research

**Maithe Tauber**  
Hospital Paediatric endocrinologist.  
Institut des Sciences du Cerveau,  
de la Cognition et du Comportement  
de Toulouse (ISC3T)

### RESEARCH COMMISSION

**Rafael Artuch Iriberry**  
President

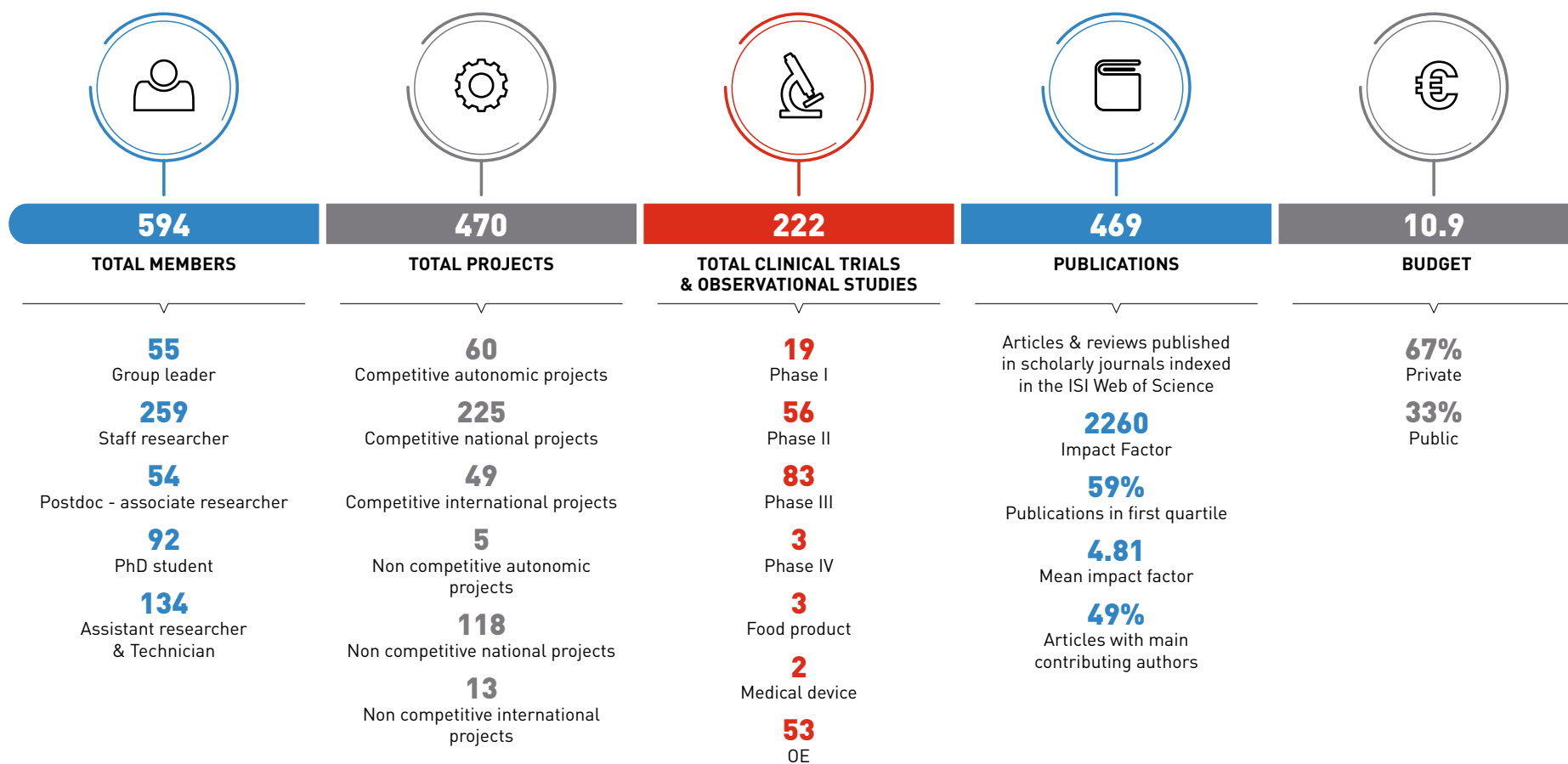
#### Vocals

Ester Camprodón Rosanas  
Alícia Casals Gelpí  
Carles Escera Micó  
Claudia Fortuny Guash  
Àngels García Cazorla  
Juan José García García  
Maria Dolores Gómez Roig  
Lourdes Ibáñez Toda  
Josep Jiménez Chillarón  
Cinzia Lavarino  
Irene Martínez de Albeniz Margalef  
Àngel Montero Carcaboso  
Carmen Muñoz Almagro  
Susana Ochoa Güerre  
Beatriz Olaya Guzmán  
Francesc Palau Martínez  
Marçal Pastor Anglada

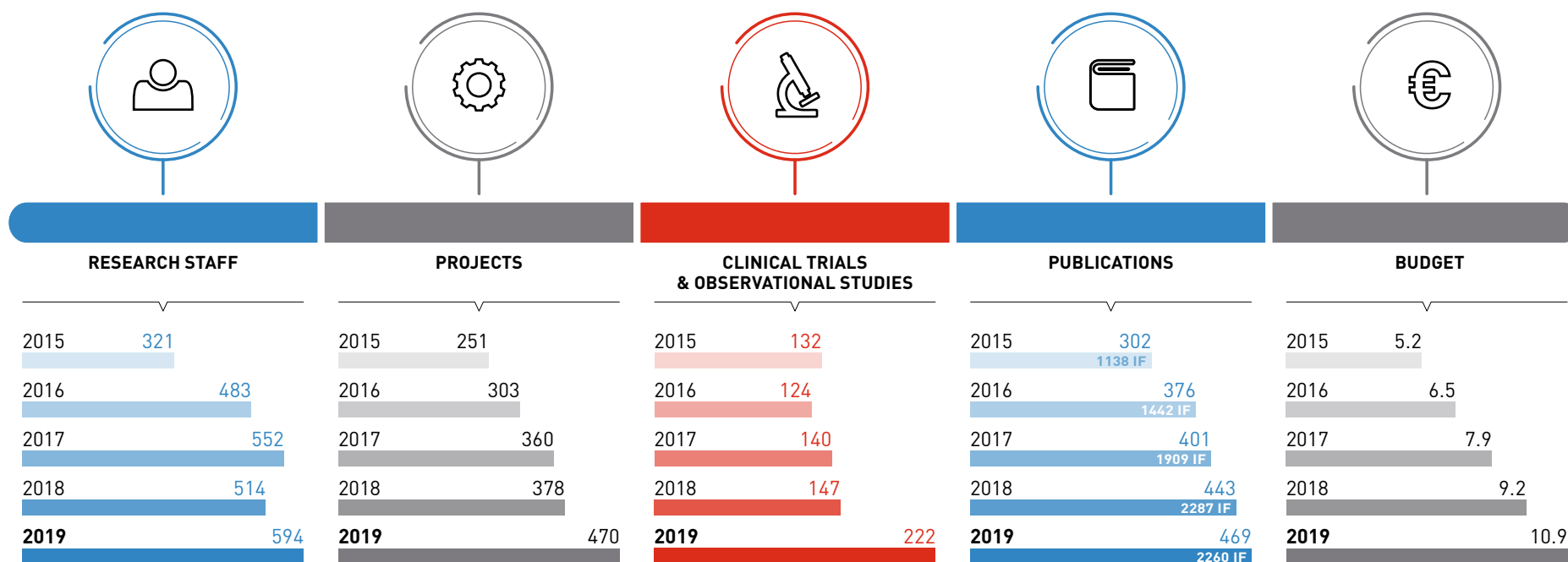
**Júlia Ribot Ballabriga**  
Secretary



## Global numbers 2019



## Global numbers 2015-2019





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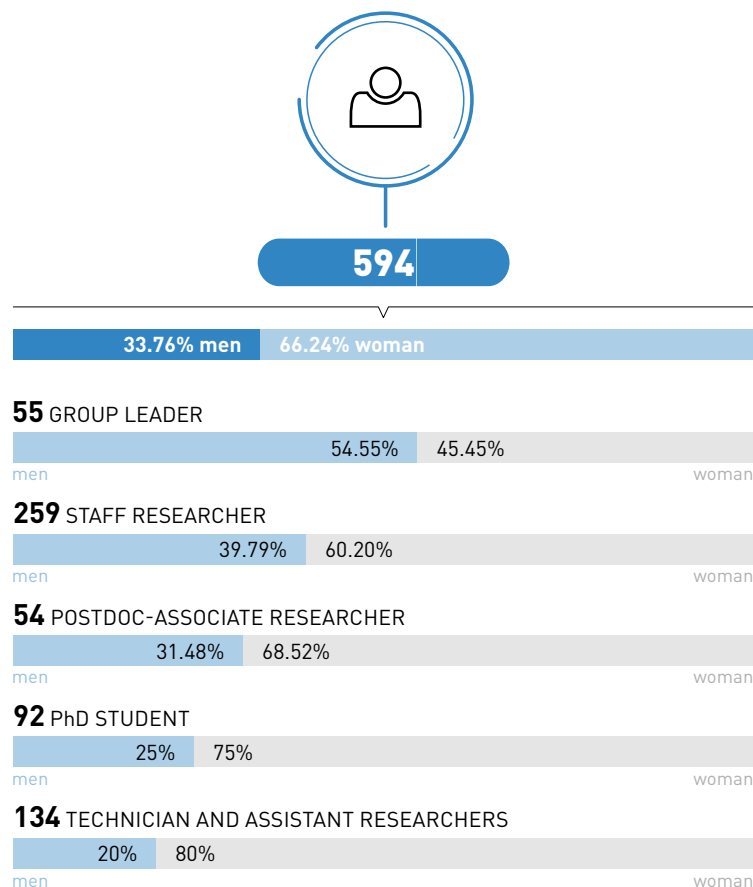
## **2** **STAFF**

Research staff  
HRS4R award





## Research staff



## HRS4R award

**HRS4R**  
Human Resources Strategy  
for Researchers

WEB  
<http://irsjd.org/ca/recerca/hrs4r-hr-excellence-research/>

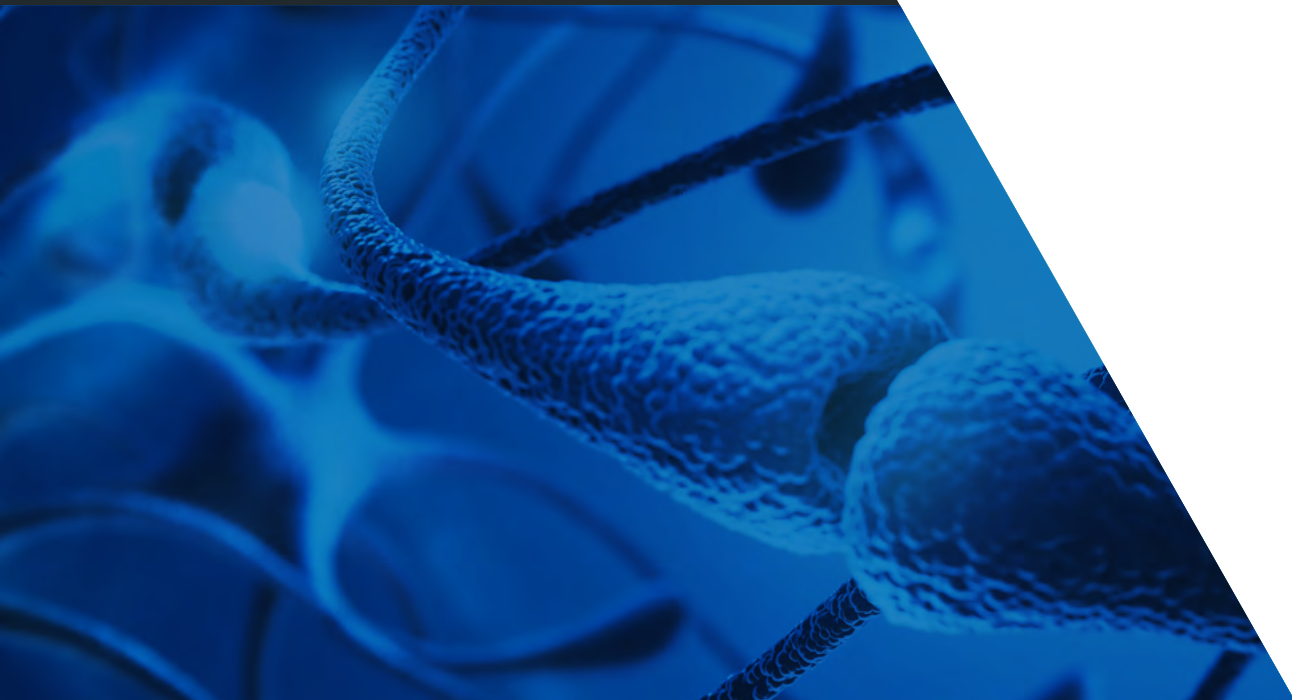
Fundació de Recerca Sant Joan de Déu is awarded with the logo "HR Excellence in research" since 2018. Thus, it has acquired a commitment to implement the European Charter for Researchers and the Code of Conduct for the Recruitment of Researchers (Charter & Code) in their policies and practices.

In 2019, IRSJD is has implemented: Strength awareness, commitment and implementation of the Charter & Code criteria, Design and implement the FSJD's Researchers' Career Plan, Design and implement the FSJD's Career Evaluation Process, Design and implement the FSJD's Recruitment Procedure, based upon OTM-R criteria, Update the FSJD's Research Committee attributions, Define the FSJD's Tech Transfer Policy, Design and implement the FSJD's Gender Plan, Write the FSJD's Welcome Manual, Implementation of occupational risk prevention measures.



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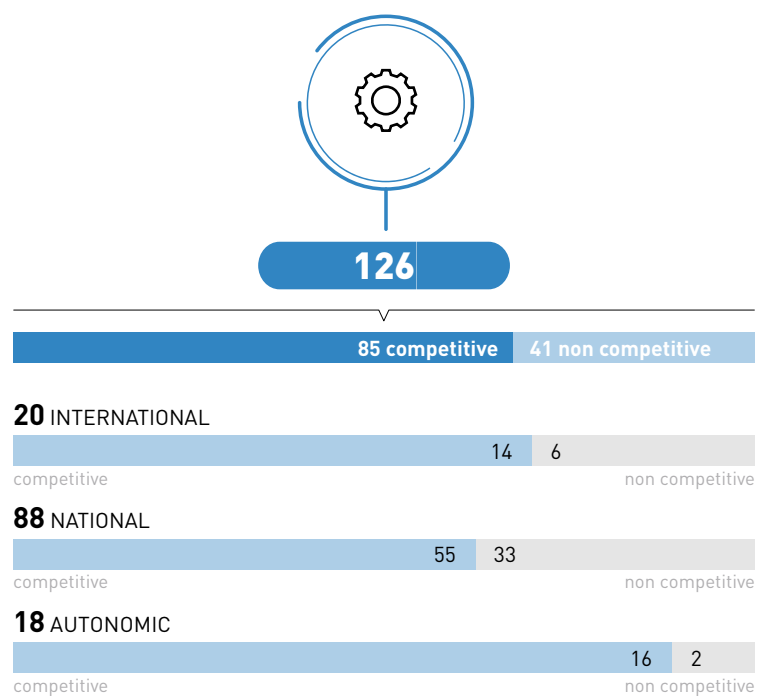


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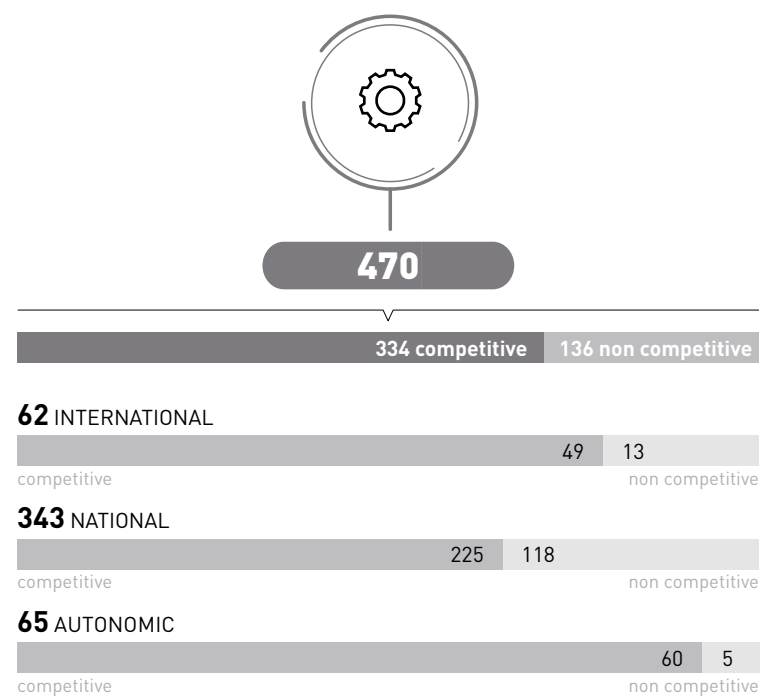
## **PROJECTS AND CLINICAL TRIALS**

New projects  
Active projects  
Active clinical trials and observational studies  
New clinical trials and observational studies

## New projects



## Active projects



## Active clinical trials and observational studies

### CLINICAL TRIALS by type

Phase I: 19

Phase II: 56

Phase III: 83

Phase IV: 3

Food product: 3

Medical Device: 2

## 222

Clinical trials and EPAS by research area

|           |                                                                               |
|-----------|-------------------------------------------------------------------------------|
| <b>48</b> | <b>Oncology</b> (41 Clinical trials 7 OE)                                     |
| <b>46</b> | <b>Paediatrics</b> (36 Clinical trials 10 OE)                                 |
| <b>43</b> | <b>Neurology</b> (36 Clinical trials 7 OE)                                    |
| <b>14</b> | <b>Endocrinology</b> (5 Clinical trials 9 OE)                                 |
| <b>13</b> | <b>Clinical haematology</b> (8 Clinical trials 5 OE)                          |
| <b>10</b> | <b>Dermatology</b> (8 Clinical trials 2 OE)                                   |
| <b>8</b>  | <b>Allergy and clinical immunology</b><br>(4 Clinical trials 4 OE)            |
| <b>6</b>  | <b>Neonatology</b> (6 Clinical trials 0 OE)                                   |
| <b>6</b>  | <b>Nephrology</b> (3 Clinical trials 3 OE)                                    |
| <b>5</b>  | <b>Gastroenterology, hepatology and nutrition</b><br>(3 Clinical trials 2 OE) |
| <b>5</b>  | <b>Gynaecology and obstetrics</b><br>(2 Clinical trials 3 OE)                 |
| <b>3</b>  | <b>Anaesthesia</b> (0 Clinical trials 3 OE)                                   |
| <b>3</b>  | <b>UCI</b> (3 Clinical trials 0 OE)                                           |
| <b>3</b>  | <b>Ophthalmology</b> (3 Clinical trials 0 OE)                                 |
| <b>3</b>  | <b>Orthopaedics and traumatology</b><br>(2 Clinical trials 1 OE)              |
| <b>2</b>  | <b>Psychiatry and psychology</b><br>(2 Clinical trials 0 OE)                  |
| <b>2</b>  | <b>Rehabilitation and physical medicine</b><br>(2 Clinical trials 0 OE)       |
| <b>1</b>  | <b>Cardiology</b> (1 Clinical trials 0 OE)                                    |
| <b>1</b>  | <b>Surgery</b> (1 Clinical trials 0 OE)                                       |

## New clinical trials and observational studies

### CLINICAL TRIALS by type

Phase I: 8

Phase II: 14

Phase III: 23

Phase IV: 1

Medical Device: 1

## 53

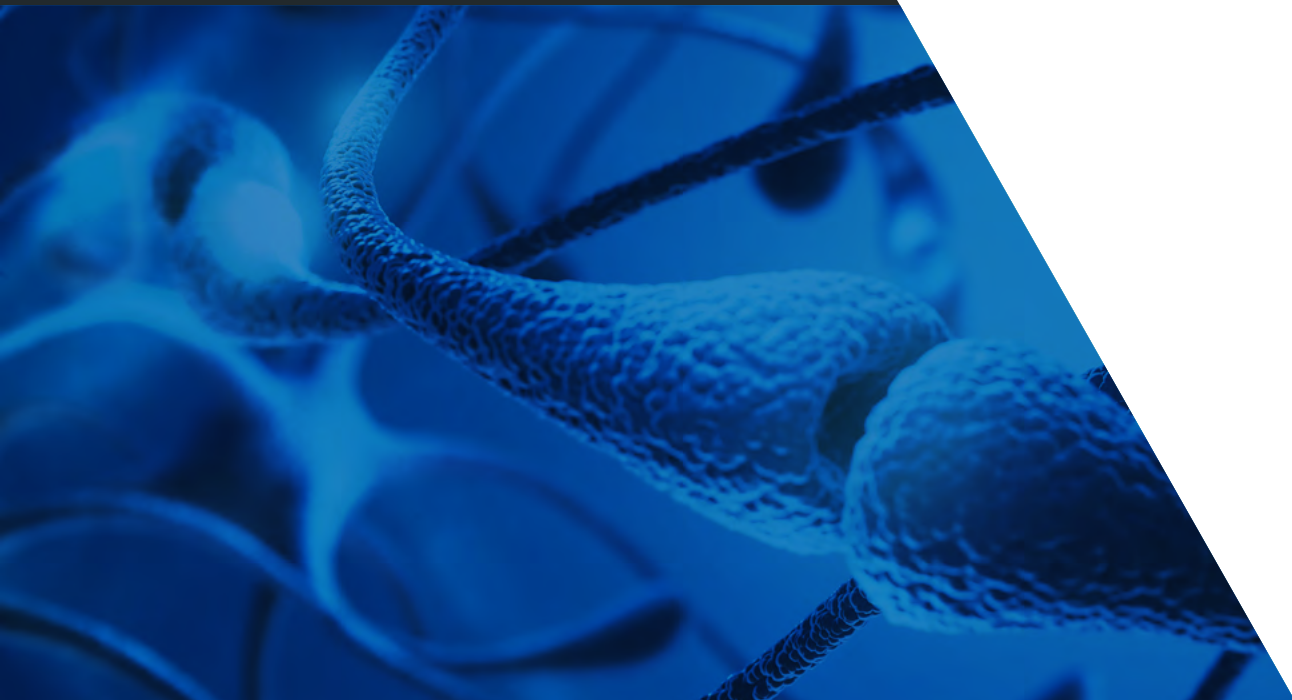
Clinical trials and EPAS by research area

|           |                                                                               |
|-----------|-------------------------------------------------------------------------------|
| <b>13</b> | <b>Neurology</b> (12 Clinical trials 1 OE)                                    |
| <b>11</b> | <b>Oncology</b> (8 Clinical trials 3 OE)                                      |
| <b>8</b>  | <b>Paediatrics</b> (8 Clinical trials 0 OE)                                   |
| <b>5</b>  | <b>Dermatology</b> (4 Clinical trials 1 OE)                                   |
| <b>2</b>  | <b>Allergy and clinical immunology</b><br>(2 Clinical trials 0 OE)            |
| <b>2</b>  | <b>Clinical haematology</b> (1 Clinical trials 1 OE)                          |
| <b>2</b>  | <b>Endocrinology</b> (2 Clinical trials 0 OE)                                 |
| <b>2</b>  | <b>Ophthalmology</b> (2 Clinical trials 0 OE)                                 |
| <b>2</b>  | <b>UCI</b> (2 Clinical trials 0 OE)                                           |
| <b>1</b>  | <b>Cardiology</b> (1 Clinical trials 0 OE)                                    |
| <b>1</b>  | <b>Gastroenterology, hepatology and nutrition</b><br>(1 Clinical trials 0 OE) |
| <b>1</b>  | <b>Nephrology</b> (1 Clinical trials 0 OE)                                    |
| <b>1</b>  | <b>Neonatology</b> (1 Clinical trials 0 OE)                                   |
| <b>1</b>  | <b>Orthopaedics and traumatology</b><br>(1 Clinical trials 0 OE)              |
| <b>1</b>  | <b>Rehabilitation and physical medicine</b><br>(1 Clinical trials 0 OE)       |



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# 4 SCIENTIFIC PRODUCTION AND RESULTS

Scientific publications  
Highest impact factor journals  
Patents and utility models  
Thesis

## Scientific publications

|                      | Output     | Total IF    | Mean IF    | % Q1          | % D1          |
|----------------------|------------|-------------|------------|---------------|---------------|
| Article              | 468        | 2259        | 4.8        | 59.40%        | 22.40%        |
| Editorial material   | 6          | 17          | 2.8        | 33.33%        | 16.67%        |
| Letter               | 18         | 102         | 5.7        | 61.11%        | 38.89%        |
| Review               | 1          | 2           | 2.1        | 0.00%         | 0.00%         |
| <b>Total general</b> | <b>493</b> | <b>2380</b> | <b>4.8</b> | <b>59.03%</b> | <b>22.92%</b> |

## Highest impact factor journals

### Original articles in first decil journals

Number of original articles in first decil journals · 37

| Journal              | IF | Total articles |
|----------------------|----|----------------|
| NAT COMMUN           | 4  | 11,878         |
| JAMA Pediatr         | 3  | 12,004         |
| Nat Genet            | 3  | 25,455         |
| NAT MED              | 3  | 30,641         |
| AGEING RES REV       | 2  | 10,390         |
| Ann Rheum Dis        | 2  | 14,299         |
| Lancet Psychiatry    | 2  | 18,329         |
| Acta Neuropathol     | 1  | 18,174         |
| Blood                | 1  | 16,601         |
| CIRC RES             | 1  | 15,862         |
| EMBO MOL MED         | 1  | 10,624         |
| J ALLERGY CLIN IMMUN | 1  | 14,110         |
| J Am Coll Cardiol    | 1  | 18,639         |

| Journal              | IF | Total articles |
|----------------------|----|----------------|
| JAMA PSYCHIAT        | 1  | 15,916         |
| LANCET DIABETES ENDO | 1  | 24,540         |
| LANCET HIV           | 1  | 14,753         |
| Lancet Oncol         | 1  | 35,386         |
| LANCET PUBLIC HEALTH | 1  | 11,600         |
| Mol Psychiatr        | 1  | 11,973         |
| NAT HUM BEHAV        | 1  | 10,575         |
| Nature               | 1  | 43,070         |
| PROG RETIN EYE RES   | 1  | 11,768         |
| SCI TRANSL MED       | 1  | 17,200         |
| THORAX               | 1  | 10,307         |
| World Psychiatry     | 1  | 34,024         |



## Originals articles with IRSJD main contribution • 32

| Original article                                                                                                                                                                                                                                                | Journal              | IF    | Program                                           |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------|-------|---------------------------------------------------|
| Barba A, Díez Escudero A, Español M, Bonany M, Sadowska JM, Guillem J, Öhman-Mägi C, Persson C, Manzanares MC, Franch J and Ginebra MP. Impact of Biomimicry in the Design of Osteoinductive Bone Substitutes: Nanoscale Matters.. ACS Appl Mater Interfaces 20 | ACS APPL MATER INTER | 8,456 | Health Technologies and Innovation                |
| Sadowska JM, Guillem J and Ginebra MP. The Influence of Physicochemical Properties of Biomimetic Hydroxyapatite on the In Vitro Behavior of Endothelial Progenitor Cells and Their Interaction with Mesenchymal Stem Cells.. ADV HEALTHC MATER 2019. 8(2): e180 | ADV HEALTHC MATER    | 6,270 | Health Technologies and Innovation                |
| Martinez-Monseny T, Bolasell M, Callejón L, Cuadras-Palleja D, Freniche V, Itzep DC, Gassiot S, Arango P, Casas-Alba D, de la Morena E, Corral J, Montero-Sanchez R, Pérez-Cerdá C, Pérez-Dueñas B, Artuch-Iriberry R, Jaeken J, Serrano M and and the CDG Span | Ann Neurol           | 9,496 | Neuroscience                                      |
| Kanaan-Izquierdo S, Ziyatdinov A, Burgueño MA and Perera A. Multiview: a software package for multiview pattern recognition methods.. Bioinformatics 2019. 35(16): 2877 - 2879. FI: 4,531(Q1)                                                                   | Bioinformatics       | 4,531 | Health Technologies and Innovation                |
| Zarei M, Pujol E, Quesada T, Villarroya F, Barroso E, Vázquez S, Pizarro J, Palomer FX and Vazquez M. Oral administration of a new HRI activator as a new strategy to improve high-fat-diet-induced glucose intolerance, hepatic steatosis, and hypertriglyceri | BRIT J PHARMACOL     | 6,583 | Adaptative response, metabolism and immunobiology |
| Birules J, Bosch L, Brieke R, Pons F and Lewkowicz DJ. Inside bilingualism: Language background modulates selective attention to a talker's mouth.. Dev Sci 2019. 22(3): e12755. FI: 4,096(Q1)                                                                  | DEVELOPMENTAL SCI    | 4,096 | Neuroscience                                      |
| Cairo M, Campderros L, Gavalda A, Cereijo R, Delgado-Anglés A, Quesada T, Giralt M, Villarroya J and Villarroya F. Parkin controls brown adipose tissue plasticity in response to adaptive thermogenesis.. Embo Rep 2019. 20(5). FI: 8,383(Q1)                  | EMBO REP             | 8,383 | Adaptative response, metabolism and immunobiology |
| Stickley A, Hans Oh, Sumiyoshi T, McKee M and Koyanagi A. Injury and depression among 212 039 individuals in 40 low- and middle-income countries.. Epidemiol Psychiatr Sci 2019. 29: 1 - 10. FI: 6,402(Q1)                                                      | EPIDEMIOL PSYCH SCI  | 6,402 | Neuroscience                                      |
| Ribas-Prats T, Almeida-Toledano L, Costa-Faidella J, Plana M, Corral MJ, Gómez-Roig MD and Escera C. The frequency-following response (FFR) to speech stimuli: A normative dataset in healthy newborns.. Hear Res 2019. 371: 28 - 39. FI: 2,952(Q1)             | HEARING RES          | 2,952 | Adaptative response, metabolism and immunobiology |
| Ashdown-Franks G, Vancampfort D, Firth J, Smith L, Sabiston CM, Stubbs B and Koyanagi A. Association of leisure-time sedentary behavior with fast food and carbonated soft drink consumption among 133,555 adolescents aged 12-15 years in 44 low- and middle-i | INT J BEHAV NUTR PHY | 6,037 | Neuroscience                                      |
| Sánchez-Niubó A, Egea-Cortés L, Olaya B, Caballero FF, Ayuso-Mateos JL, Prina M, Bobak M, Arndt H, Tobiasz-Adamczyk B, Pajak A, Leonardi M, Koupil I, Panagiotakos D, Tamosiunas A, Scherbov S, Sanderson W, Koskinen S, Chatterji S and Haro JM. Cohort Profil | Int J Epidemiol      | 7,339 | Neuroscience                                      |

| Original article                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | Journal              | IF     | Program                                           |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------|--------|---------------------------------------------------|
| Camprodon-Rosanas E, Pujol J, Martínez-Vilavella G, Blanco-Hinojo L, Medrano-Martorell S, Batlle S, Forns J, Ribas-Fitó N, Dolz M and Sunyer J. Brain Structure and Function in School-Aged Children With Sluggish Cognitive Tempo Symptoms.. J Am Acad Child A                                                                                                                                                                                                                                                                                                     | J AM ACAD CHILD PSY  | 6,391  | Neuroscience                                      |
| Koyanagi A, Hans Oh, Carvalho AF, Smith L, Haro JM, Vancampfort D, Stubbs B and DeVylder JE. Bullying Victimization and Suicide Attempt Among Adolescents Aged 12-15 Years From 48 Countries.. J Am Acad Child Adolesc Psychiatry 2019. 58(9): 907-918.e4. FI:                                                                                                                                                                                                                                                                                                      | J AM ACAD CHILD PSY  | 6,391  | Neuroscience                                      |
| Adrián Pérez Aranda, Francesco D'Amico, Feliu-Soler A, McCracken LM, Peñarrubia-María MT, Andrés Rodríguez L, Angarita-Osorio N, Martin Knapp, García-Campayo J and Luciano JV. Cost-Utility of Mindfulness-Based Stress Reduction for Fibromyalgia versus a Mu                                                                                                                                                                                                                                                                                                     | J CLIN MED           | 5,688  | Neuroscience                                      |
| Jou-Munoz C, Ortigoza-Escobar JD, O'Callaghan-Gordo M, Nascimento-Osorio A, Darling A, Pias-Peleteiro L, Pérez-Dueñas B, Pineda M, Codina-Bergadà A, Arjona-Fernandez C, Armstrong-Moron J, Palau F, Ribes A, Gort L, Tort F, Navas P, Ruiz-Pesini E, Emperador S, Lopez-Gallardo E, Bayona-Bafaluy P, Montero-Sanchez R, Jimenez-Mallebrera C, Garcia-Cazorla A, Montoya C, Yubero-Siles D and Artuch-Iriberry R. Muscle Involvement in a Large Cohort of Pediatric Patients with Genetic Diagnosis of Mitochondrial Disease. J Clin Med 2019. 8(1). FI: 5,688(Q1) | J CLIN MED           | 5,688  | Neuroscience                                      |
| Solé-Ribalta A, Rodríguez-Fanjul X, Juan Manuel Carretero Bellón, Pascual-Sala C, Martorell-Sampol L, Bobillo-Perez S and Morillo-Palomo A. Neonatal Marfan Syndrome: A Rare, Severe, and Life-Threatening Genetic Disease.. J Pediatr 2019. 211: 221-221.e2.                                                                                                                                                                                                                                                                                                       | J PEDIATR-US         | 3,739  | Adaptative response, metabolism and immunobiology |
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| Pons-Odena M, Medina A and Modesto-Alapont V. Treachery on the Rocks in the Pediatric Intensive Care Unit.. JAMA Pediatr 2019. 173(7): 699 - 699. FI: 12,004(Q1)                                                                                                                                                                                                                                                                                                                                                                                                    | JAMA Pediatr         | 12,004 | Adaptative response, metabolism and immunobiology |
| Ibañez-Toda L and de Zegher F. Exploring the use of metformin in pregnant women with polycystic ovary syndrome: new evidence, new wisdom. Lancet Diabetes Endocrinol 2019. 7(4): 242 - 243. FI: 24,540(Q1)                                                                                                                                                                                                                                                                                                                                                          | LANCET DIABETES ENDO | 24,540 | Adaptative response, metabolism and immunobiology |
| Botteri G, Salvadó L, Gumà A, Hamilton DL, Meakin PJ, Montagut G, Ashford MLJ, Ceperuelo-Mallafre V, Fernández-Veledo S, Vendrell J, Calderón-Dominguez M, Serra D, Herrero L, Pizarro J, Barroso E, Palomer FX and Vazquez M. Corrigendum to "The BACE1 produc                                                                                                                                                                                                                                                                                                     | Metabolism           | 6,513  | Adaptative response, metabolism and immunobiology |
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| Armangue-Salvador T, Baucells BJ, Vlasea A, Petit-Pedrol M, Esteve-Sole A, Deyà-Martinez A, Ruiz-García R, Juan M, Pérez de Diego R, Dalmau J and Alsina L. Toll-like receptor 3 deficiency in autoimmune encephalitis post-herpes simplex encephalitis.. Neuro                                                                                                                                                                                                                                                                                                     | NEUROL-NEUROIMMUNOL  | 7,353  | Adaptative response, metabolism and immunobiology |

| Original article                                                                                                                                                                                                                                                                                                        | Journal             | IF     | Program                                           |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------|--------|---------------------------------------------------|
| Carrera-García L, Natera-de Benito D, Lleixà C, Ortez-Gonzalez CI, Colomer J, Nascimento-Osorio A, Saiz A, Dalmau J, Querol L and Armangue-Salvador T. Chronic inflammatory demyelinating polyneuropathy associated with contactin-1 antibodies in a child.. Neurol Neuroimmunol Neuroinflamm 2019. 6(5). FI: 7,353(Q1) | NEUROL-NEUROIMMUNOL | 7,353  | Neuroscience                                      |
| Adrián Pérez Aranda, Andrés Rodríguez L, Feliu-Soler A, Nuñez C, Stephan-Otto C, Pastor-Mira MA, López-Roig S, Peñacoba C, Calandre EP, Slim M, Salgueiro M, Feixas G and Luciano JV. Clustering a large Spanish sample of patients with fibromyalgia using the                                                         | Pain                | 6,029  | Neuroscience                                      |
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| de Sousa-Malpique RM, Sánchez-Infantes D, García-Beltran C, Taxerås SD, López-Bermejo A, de Zegher F and Ibañez-Toda L. Towards a circulating marker of hepato-visceral fat excess: S100A4 in adolescent girls with polycystic ovary syndrome Evidence from ran                                                         | Pediatr Obes        | 3,713  | Adaptative response, metabolism and immunobiology |
| de Zegher F, de Sousa-Malpique RM, García-Beltran C and Ibañez-Toda L. Towards a simple marker of hepato-visceral adiposity and insulin resistance: The Z-score change from weight-at-birth to BMI-in-childhood.. Pediatr Obes 2019. 14(10): e12533. FI: 3,713{                                                         | Pediatr Obes        | 3,713  | Adaptative response, metabolism and immunobiology |
| Sebastiani G, García-Beltran C, Pie S, Guerra A, López-Bermejo A, Sanchez-de-Toledo J, de Zegher F, Rosés F and Ibañez-Toda L. The sequence of prenatal growth restraint and postnatal catch-up growth: normal heart but thicker intima-media and more pre-peri                                                         | Pediatr Obes        | 3,713  | Adaptative response, metabolism and immunobiology |
| Picart S, Barrett SJ, Willé DR, Perera A, Gutteridge A and Dessailly BH. Benchmarking network propagation methods for disease gene identification.. PLOS COMPUT BIOL 2019. 15(9): e1007276. FI: 3,955(Q1)                                                                                                               | PLoS Comput Biol    | 3,955  | Health Technologies and Innovation                |
| Jacob L, Haro JM and Koyanagi A. Association between intelligence quotient and violence perpetration in the English general population.. Psychol Med 2019. 49(8): 1316 - 1323. FI: 5,641(Q1)                                                                                                                            | Psychol Med         | 5,641  | Neuroscience                                      |
| Nuñez C, Stephan-Otto C, Usall J, Bioque M, Lobo A, González-Pinto A, Pina-Camacho L, Vieta E, Castro-Fornieles J, Rodríguez-Jimenez R, Butjosa Molines A, Janssen J, Bibiana Cabrera Llorca, Parellada M, Bernardo M and PEPs group. Neutrophil Count Is Assoc                                                         | SCHIZOPHRENIA BULL  | 7,289  | Neuroscience                                      |
| Pascual-Pastó G, Bazan-Peregrino M, Gene-Olaciregui N, Restrepo-Perdomo CA, Mato-Berciano A, Ottaviani D, Weber K, Correa MG, Paco-Mercader S, Vilà-Ubach M, Cuadrado-Vilanova M, Castillo-Ecija H, Botteri G, García-Gerique L, Moreno-Gilabert H, Gimenez-Ale                                                         | SCI TRANSL MED      | 17,200 | Paediatric cancer                                 |

## Patents and utility models

### Patents and license agreements

#### USE OF VIRAL VECTORS IN THE TREATMENT OF RETINOBLASTOMA

Date of grant: 28/02/2019  
Country of protection: US  
Grant number: US2019/0350992  
Applicants: VCN Biosciences SL; SJD Barcelona Children's Hospital  
Inventors: Manuel Maria Cascallo Piqueras; Miriam Bazan Peregrino; Angel Montero Carcaboso; Guillermo Luis Chantada

#### TREATMENT OF HEPATIC STEATOSIS RELATED OLIGO-OVULATION

Date of grant: 21/11/2019  
Country of protection: US  
Grant number: US2019/0060328  
Applicants: Katholieke Universiteit Leuven; SJD Barcelona Children's Hospital  
Inventors: Lourdes Ibáñez; Francis De Zegher

#### EQUIPMENT WITH BIOFEEDBACK FOR TRAINING THE MUSCLES OF THE PELVIC FLOOR

Date of grant: 08/01/2019  
Country of protection: China  
Grant number: CN104703537A  
Applicants: Universitat Politècnica de Catalunya, Hospital Clínic de Barcelona  
Inventors: Montserrat Espuna Pons, Miguel Angel Mañanas Villanueva,

Amelia Perez Gonzalez, Juan Jose Ramos Castro

#### PORTABLE DEVICE AND METHOD FOR MEASURING ELECTROMYOGRAPHIC SIGNALS OF AN USER

Reference: EP3324843A1  
Applicants: Universitat Politècnica de Catalunya  
Inventors: Miguel Angel Mañanas Villanueva, Mónica Rojas Martinez  
Information about: Intention to grant announced (18/09/2019)

#### EQUIPMENT WITH BIOFEEDBACK FOR TRAINING THE MUSCLES OF THE PELVIC FLOOR

Reference: US2019/0060328  
Applicants: Universitat Politècnica de Catalunya, Hospital Clínic de Barcelona  
Inventors: Montserrat Espuna Pons, Miguel Angel Mañanas Villanueva, Amelia Perez Gonzalez, Juan Jose Ramos Castro  
Information about: Information on status: patent application and granting procedure in general (21/11/2019)

### Portfolio of spin-offs

#### BCN Innova

2008 · <http://www.bcninnova.com/>  
**Dr. Joan Prat (IRSJD)**

BCN Innova, a spin-off company of Sant Joan de Déu-Barcelona Children's Hospital, has developed a device (GazeLab) that substantially improves the diagnosis and treatment of strabismus, reducing the number of second operations that are now performed to achieve the expected results.

GazeLab offers two main benefits:

1. It allows objective exploration on a revolutionary level of precision, with much better results than the subjective-measurement model that has prevailed until now.
2. It provides a 50% reduction in the number of second operations, obviating those which are surgically unnecessary.

#### CEBIOTEX

2012 · <http://www.cebiotex.com/>  
**Dr. Joan Bertran Llavinà (CEO)**  
**Angel Montero (IRSJD)**

Cebiotex has developed and characterised at preclinical level, in vitro and in vivo, nonwoven nanofiber membranes for the local release of active ingredients in the local treatment of the surgical site after tumour excision surgery. The purpose of the local release of drugs is to achieve high concentrations of active ingredients in the area where the release device is placed, avoiding systemic toxicity and increasing the effectiveness of the drug against

possible tumoral remnants. The activity of Cebiotex is based on the interaction of researchers from two Catalan institutions: the Polytechnic University of Catalonia (UPC) and Sant Joan de Déu-Barcelona Children's Hospital (HSJD).

#### OMADRE

2019 · <https://www.omadre.com/>  
**Dr. Jaume Perez Payarols (IRSJD)**

The company Omadre was created to provide hospitals with the tools required for the efficient management of mother's milk, increasing the safety of existing processes in this way.

#### DBGEN Ocular Genomics

2018 · <https://dbgen.com/>  
**Co-funders Roser González-Duarte and Gemma Marfany**

DBGen is a company focused on the genetic diagnosis of hereditary eye disorders, providing the highest level of quality, diagnostic effectiveness and close contact with patients, ophthalmologists, and enterprises in need of its services. At DBGGen, we offer high-quality, reliable and accessible genetic diagnosis, allowing the clinical diagnosis to be assured and guidance to be provided to the patient with respect to emerging therapies for genetic disorders of the eyes. Genetic diagnosis is indispensable to identify family members possessing a genetic defect, permitting the most suitable advice to be given in each case.

## Thesis

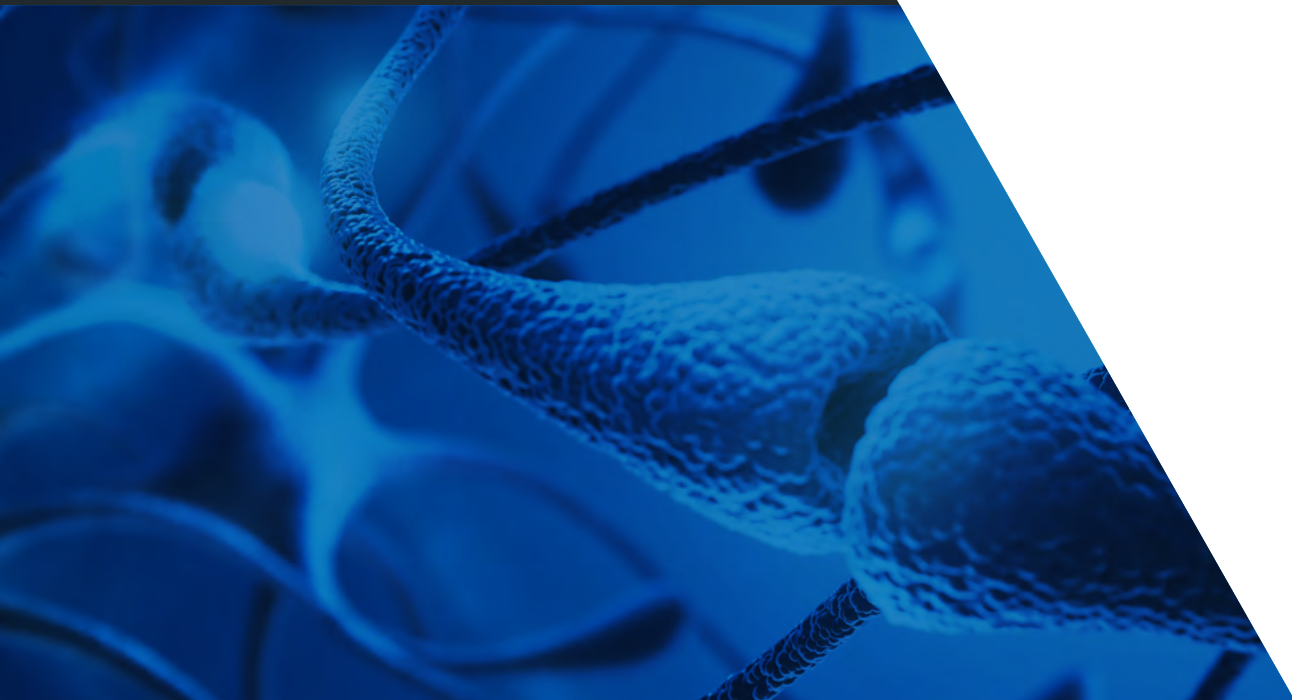
| Thesis Author                   | Title                                                                                                                                                                                   | Thesis directors                                                           | Date    |
|---------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------|---------|
| Natàlia Gorina-Careta           | Contribution of the subcortical auditory pathway to the perception and processing of sounds                                                                                             | Carles Escera                                                              | January |
| A. Barrios-Luque                | ECG como herramienta para diagnóstico para la enfermedad de Kawasaki                                                                                                                    | Georgia Sarquella Brugada                                                  | March   |
| Elvira Reche Camba              | Evaluación de la regulación emocional cognitiva en fibromialgia mediante el instrumento Cognitive Emotion Regulation Questionnaire (CERQ)                                               | Xavier Borràs Hernández, Albert Feliu Soler and Juan Vicente Luciano Devis | April   |
| Jonatan Ottino González         | Overweight, allostatic load and neuroimaging                                                                                                                                            | Maria Àngels Jurado Luque and Maite Garolera Freixa                        | April   |
| Didac Casas Alba                | Infeccions per Enterovirus i Paraechovirus que produeixen patologies neurològiques i sistèmiques en població pediàtrica. Vigilància epidemiològica i estudi de biomarcadors de gravetat | Carmen Muñoz Almagro<br>Cristian Launes Montaña (director)                 | April   |
| C. Iglesias-Gomez               | Implementación de un Modelo de evaluación de conocimiento en Cardiología Pediátrica                                                                                                     | Georgia Sarquella Brugada                                                  | June    |
| M. Castany                      | Use of molecular point of care (MPOC) systems for accurate and rapid diagnosis of gastrointestinal pathogens: impact of diagnosis yield, patient management and laboratory workflow     | Carmen Muñoz Almagro                                                       | June    |
| Toral M                         | Análisis de la influencia del consumo de tóxicos (alcohol, tabaco y drogas de abuso) durante el embarazo sobre la prematuridad                                                          | Maria Dolores Gómez-Roig                                                   | June    |
| Antonio Martínez Monseny        | Caracterización de pacientes pmm2-cdg: espectro fenotípico y correlación molecular                                                                                                      | Mercedes Serrano Gimare and Alfredo García-Alix Pérez                      | June    |
| Maria Iglesias González         | Effectiveness and feasibility of watchful waiting for the management of major depression in primary care                                                                                | Josep Maria Haro (tutor)<br>Antoni Serrano and Maria Rubio (co-directors)  | June    |
| Susanna Gassiot                 | Mieloma Múltiple. Canvis en el maneig clínic associats a la incorporació de nous fàrmacs.                                                                                               | Albert Oriol                                                               | June    |
| Laura Camperros Traver          | GDF15, un nou factor secretable regulador del metabolisme                                                                                                                               | Francesc Villaroya i Gombau and Joan Villaroya i Terrade                   | July    |
| Francisco López-Caballero       | Evoked and induced activity in the auditory nervous system: deviance detection and brainwave entrainment                                                                                | Carles Escera                                                              | July    |
| Cristina Duran Fernández-Feijóo | Neuroprotection with hypothermia and allopurinol in hypoxic-ischemic damage. Role of the antioxidant effect                                                                             | Marta Camprubí                                                             | July    |
| David Cornet Bartolomé          | Molecular determinants of human oocyte quality in assisted reproduction.                                                                                                                | Daniel Grinberg Vaisman and Rita Vassena                                   | July    |

| Thesis Author              | Title                                                                                                                                                                                           | Thesis directors                                                                                             | Date      |
|----------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------|-----------|
| Pedro Brotons de los Reyes | Infección respiratoria aguda en el paciente pediátrico: valor de la cuantificación de la carga bacteriana y de la detección viral en muestra nasofaríngea mediante pruebas rápidas moleculares. | Carmen Muñoz Almagro,<br>Iolanda Jordán García (director)                                                    | July      |
| Albert Peiret Giménez      | Modelling and simulation of multibody systems with unilateral contact                                                                                                                           | József Kövecses and<br>Josep M. Font Llagunes                                                                | August    |
| Alejandro Bosch            | Confort y desconfort en el paciente crítico pediátrico, adaptación transcultural y validación de la escala comfort behavior scale                                                               | Anna Falcó and Iolanda Jordan                                                                                | September |
| Nagore Gene Olaciregui     | New therapeutic approaches in diffuse intrinsic pontine glioma (DIPG)                                                                                                                           | Angel Montero Carcaboso (director)<br>Teresa Ribalta Farrés (tutor)                                          | September |
| Mónica Sánchez Aulet       | Biomarcadores y Trastorno Bipolar                                                                                                                                                               | Maria Paz García-Portilla and<br>Belen Arranz, Eduard Vieta (tutor)                                          | September |
| Aida Mata Ventosa          | Implicacions fisiopatològiques de l'alteració del transportador concentratiu de nucleòsids hCNT1.                                                                                               | Sandra Pérez Torras and<br>Marçal Pastor Anglada                                                             | November  |
| Neus Roca Ayats            | Identification and functional characterization of genetic loci involved in osteoporosis and atypical femoral fracture                                                                           | Daniel Grinberg Vaisman and<br>Susanna Balcells Comas                                                        | November  |
| Alba Tristan Noguero       | Deficiencia de la Tirosina Hidroxilasa: Estudios en pacientes y en un modelo celular                                                                                                            | M <sup>a</sup> Angels García Cazorla (tutor),<br>Antonella Consiglio and<br>Alcántara-Horrillo S (directors) | November  |
| Vittoria Spinosa           | Motor-auditory interaction: a window towards associative and predictive processing                                                                                                              | Iria SanMiguel and<br>Carles Escera                                                                          | December  |
| Nuria Elvira Sanz Marcos   | Estudio longitudinal de la composición corporal y del perfil endocrino-metabólico en niños sanos: influencia de la composición de ácidos grasos poliinsaturados maternos durante la gestación   | Lourdes Ibañez Toda                                                                                          | December  |
| Maite Miranda-García       | Efectividad de la acupuntura y moxibustión en la versión espontánea del feto en presentación de nalgas                                                                                          | Maria Dolores Gómez- Roig                                                                                    | December  |
| Elisabeth Figuerola Bou    | Polycomb and KDM6A Roles in the Epigenetic Dynamics of Ewing Sarcoma Tumorigenesis                                                                                                              | Jaume Mora Graupera and<br>Sara Sánchez Molina                                                               | December  |



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**5**  
**CORE FACILITIES**



The Institut de Recerca Sant Joan de Déu makes available to its researchers a set of fundamental services and infrastructures for the performance of their projects. In order to facilitate high-level research, the IRJSD offers its professionals technical, scientific and support services as well as the most suitable infrastructures required to carry out their work:

### **Scientific core facilities**

- Confocal Microscopy Unit
- Paediatric Biobank for research
- Neurological tissues Biobank for research
- Sequencing Unit
- Methodology and statistics
- Animal housing facility
- Neuroimaging
- Neuropsychological evaluations
- Library
- Clinical Trials Unit
- Biological Sample Processing and Molecular Analysis
- Unit of Design, Analysis and Interpretation of Omic data (DAI-Omics)

### **Management support services**

- Administration and corporate finance
- Human Area
- Clinical Research Ethics Committee
- Research management and Promotion
- Management of knowledge and communication

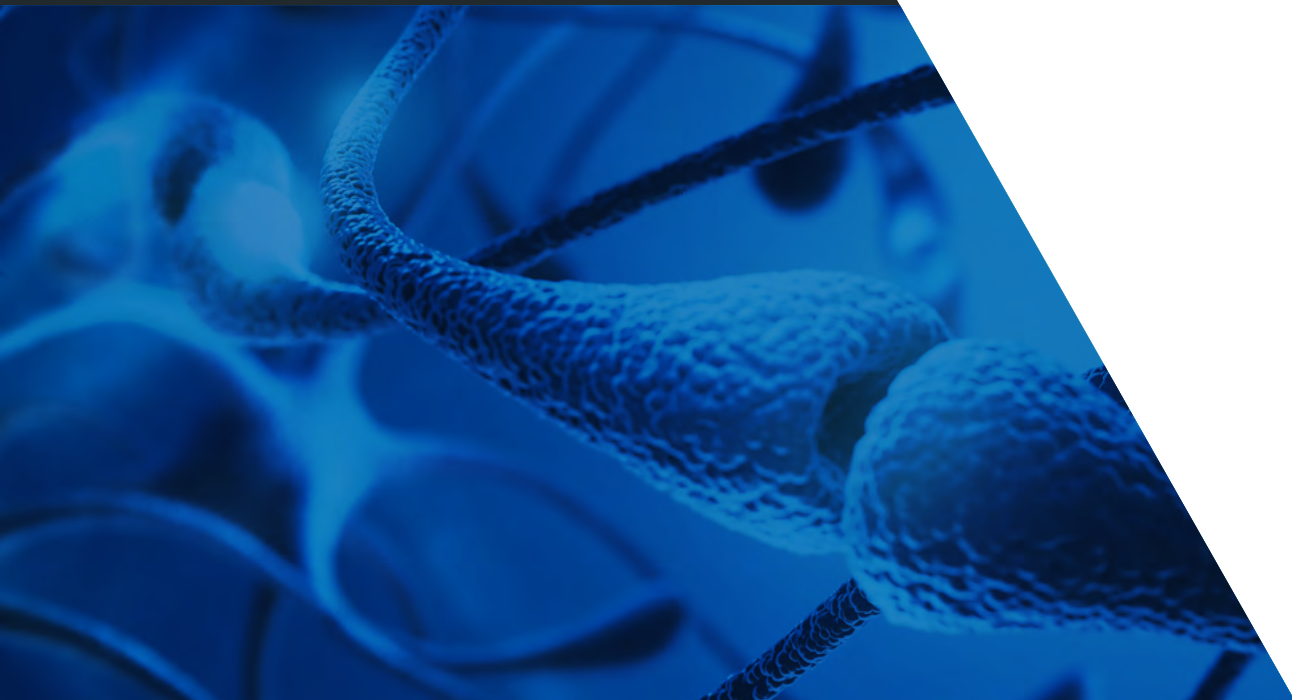
### **Other scientific and technical services**

- UBNeuro Labs and facilities
- IBUB Labs and facilities
- CREB Labs and facilities



**SJD**

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**6**

## **TRAINING, COMMUNICATION AND PARTICIPATION**

Seminars  
Training programme  
Communication and public engagement

## Seminars

**20** SEMINARS IN 2019

**467** ATTENDEES IN 2019

### Relation of research seminars and IRSJD day of debates

15 JANUARY

**Omics: theory and practice of the various methods**

Raúl Cabrera Rubio, PhD

29 JANUARY

**Keys to the preparation of competitive research projects**

Rafa Artuch, PhD and  
Angel Montero, PhD

12 FEBRUARY

**Treatment of early intrauterine growth restriction with low-molecular-weight heparin**

Eduard Mazarico Gallego, PhD

26 FEBRUARY

**Role of transporters in disease**

Sandra Pérez Torra, PhD

13 MARCH

**Collaborative intelligence and data model in the Share4Rare project**

Alexandre Perera i Lluna, PhD

26 MARCH

**Neuroimaging biomarkers in mental and neurological pathology**

Christian Stephan-Otto, PhD

5 APRIL

**HORIZON EUROPE EU Research and Innovation Framework Programme for 2021-2027**

Marina Martínez-García, PhD

30 APRIL

**Cancer virotherapy: The paradigms they are a-changing**

Juan Fueyo-Margareto, PhD

13 MAY

**New Infrastructures seminar**

Giuseppe Battaglia, PhD and  
Jaume Comas, PhD

14 MAY

**The air we breathe. New looks at the unknown etiologies of paediatric disorders**

Xavier Rodó, PhD

16 MAY

**II Day of Debate: IRSJD 2019 - Innovation Unconference**

28 MAY

**Cellular models and therapeutic approaches for Sanfilippo syndrome type C**

Noelia Benetó Gandia

11 JUNE

**Inhibition of phosphatidylinositol 3-kinase  $\alpha$  (PI3K $\alpha$ ) prevents heterotopic ossification**

Francesc Ventura, PhD

25 JUNE

**Functional study of GDAP1 by computational analysis of confocal microscopy images**

Lara Cantarero Abad, PhD

2 JULY

**Type 1 Diabetes and the Developing Brain**

Tandy Aye, PhD

16 JULY

**CNS penetrating polymersomes for glioma targeting**

Giuseppe Battaglia PhD

13 SEPTEMBER

**Generation of cellular and murine models of phenylketonuria and propionic acidemia by CRISPR/Cas9**

Aristides López Márquez, PhD

17 SEPTEMBER

**New therapeutic alternatives for diffuse intrinsic pontine glioma (DIPG)**

Nagore Gené Olaciregui

6 NOVEMBER

**III Day of Debate: IRSJD 2019. Neuroscience area · The multi OMICS day**

12 NOVEMBER

**Polycomb and KDM6A Roles in the Epigenetic Dynamics of Ewing Sarcoma Tumorigenesis**

Elisabeth Figuerola Bou

26 NOVEMBER

**Challenges in neuropsychological assessment and intervention in cerebral palsy**

Roser Pueyo, PhD, Montse Blasco and María García-Galant

10 DECEMBER

**Animal models for the study of psychiatric disturbances**

Noèlia Fernández-Castillo, PhD

## Training programme

**16** COURSES IN 2019  
**155** ATTENDEES IN 2019

To promote the development of the different competence dimensions, four types of training modalities are established:

- Knowledge and techniques
- Soft Skills
- Organization and Governance
- Institutional training (Values)

### Courses

- Digital skills: Information search (online)
- Patients involvement: Introductory session
- Specialization session: Biobank
- Specialization session: Confocal Microscopy
- ICTs and health: Digital Health
- Patients involvement (online)
- Clinical research with Medical Devices
- Specialization session: Laboratory Services
- Sources of information, bibliographic research and scientific impact
- Introduction to the methodology
- Introduction to statistics
- Intellectual property: Patents and licenses
- Fundraising
- Systematic reviews and meta-analysis
- Introductory course to R-Software
- RGPD in Research

## Communication and public engagement

### Activities

**13** Activities organized  
**1** Secondary school students  
**190** Participants  
**3** Researchers  
**12** General public  
**749** Assistants

### Participation in

- La Marató de TV3
- Europeans Researcher's Night
- Barcelona "Festa de la Ciència"
- National Xeroderma Congress
- International Gliomatosis Congress
- RareHack

### Internet and Social media

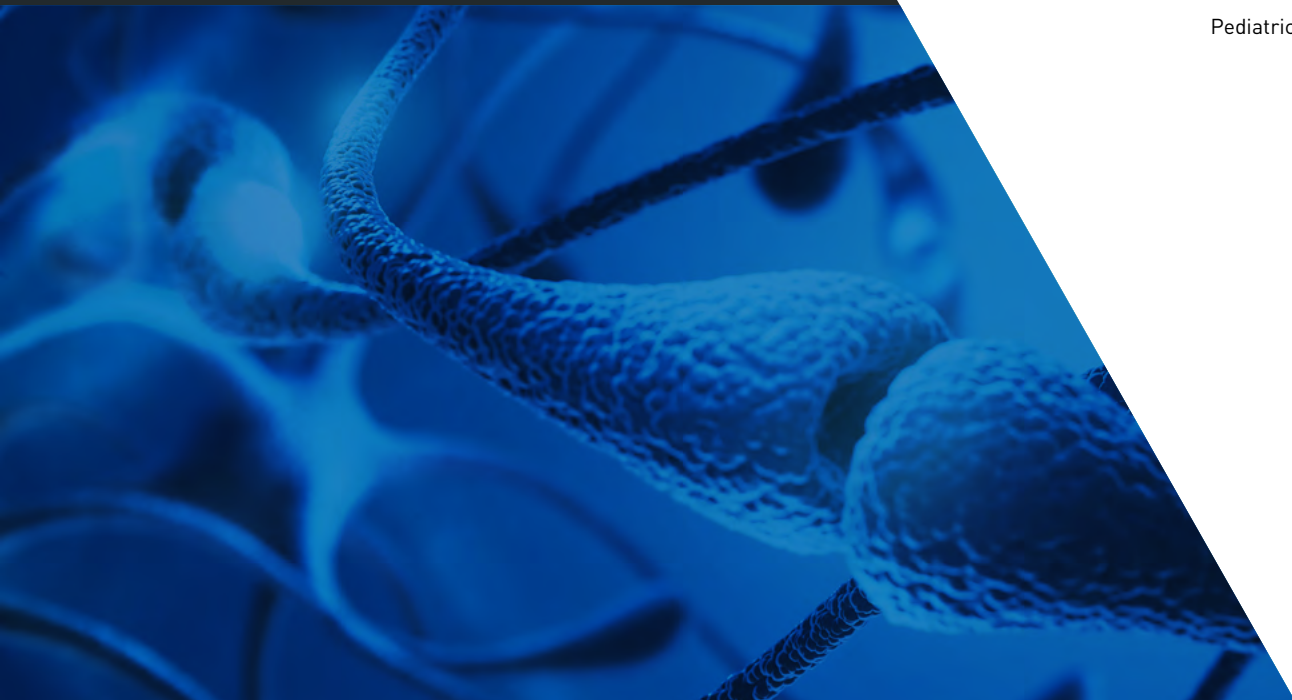
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**223.376** number of visits to pages



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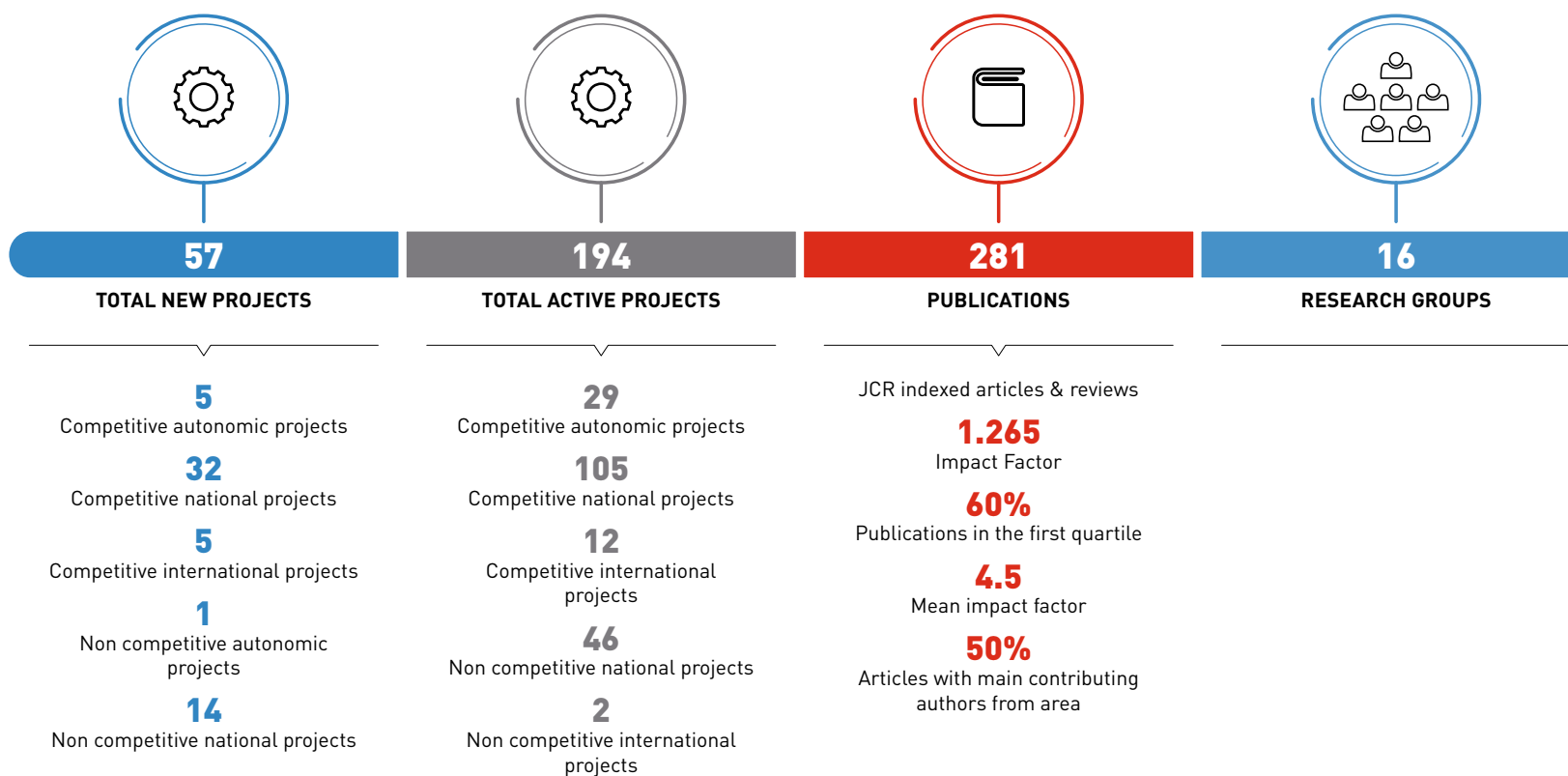
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# 7

## NEUROSCIENCE

Metabolic and mitochondrial medicine  
Applied research in neuromuscular diseases  
Pediatric neurometabolism: neural communication mechanisms and personalized therapies  
Neurogenetics and Molecular Medicine  
Neonatal brain  
Neuropsychology  
Cognitive neuroscience  
Attention, Perception and Language Acquisition (APAL)  
Human molecular genetics I  
Human molecular genetics II  
Etiopathogenesis and treatment of severe mental disorders (MERITT)  
Research in child and adolescent mental health  
Epidemiology of mental disorders and aging  
Health technology assessment in primary care and mental health (PRISMA)  
Psychological research group on fibromyalgia and chronic pain (AGORA GROUP)  
Cerebral imprints of gender, development and pathology



**METABOLIC AND MITOCHONDRIAL MEDICINE**

Rafael Artuch Iriberry, MD, PhD

We have maintained a good level of scientific output, notably including the description of two new genes associated with human pathology for the first time (ADCK2 and collectrin), and the description of a new treatment option (AZATAX) for congenital disorders of glycosylation. We are in a process of technological renewal, which has led us for the first time to carry out a project on health technologies for the development of a biosensor to monitor the ammonium value in point-of-care and home formats. We have also developed new procedures for the metabolomic phenotyping of cellular and animal models.

**Members****STAFF RESEARCHER**

Jaume Campistol Plana, Didac Casas Alba, Rosa Gassió Subirachs, Laura Marti Sanchez, Silvia Maria Meavilla Olivas, Aida Ormazabal Herrero, Mercedes Serrano Gimaré, Cristina Sierra March, Roser Urreiziti Frexedas, Délia Yubero Siles

**POSTDOC - ASSOCIATE RESEARCHER**

Mercedes Casado Rio, Juan Daría Ortigoza Escobar, Raquel Montero Sánchez

**PHD STUDENT**

Abraham José Paredes Fuentes

**TECHNICIAN**

Yahel Calzadilla Borrás, Judit Diaz Blanco, Aroa Fernández de Miguel, Juan Moreno García, Tania Parda Chofle

**Selected projects**

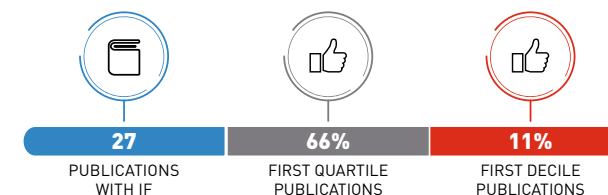
- FIS 2017\_Atrofia cerebelosa en la infancia: desarrollo de herramientas clínico-radiológicas para un fenotipado de precisión, algoritmo diagnóstico integral e identificación de nuevos genes. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI17/00101. PI: Mercedes Serrano Gimaré. 2018 - 2020.
- Biosensores para la monitorización de Enfermedades Metabólicas Hereditarias. Desarrollo y evaluación de prototipos funcionales para validación conceptual. Instituto de Salud Carlos III (ISCIII). DTS. DTS18/00075. PI: Rafael Artuch Iriberry. 2019-2020.
- Diagnóstico molecular de los defectos de la fosforización oxidativa mitocondrial: patogénesis de las deficiencias de CoQ10. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI17/00109. PI: Rafael Artuch Iriberry. 2018-2020.

**Selected publications**

- Jou-Munoz C, Ortigoza-Escobar JD, O'Callaghan-Gordo M, Nascimento-Osorio A, Darling A, Pias-Peleiteiro L, et al. Muscle Involvement in a Large Cohort of Pediatric Patients with Genetic Diagnosis of Mitochondrial Disease. J Clin Med. 2019; 8(1). FI: 5,688(Q1)
- Martinez-Monseny T, Cuadras-Palleja D, Bolasell-Girgas M, Muchart-Lopez J, Arjona-Fernandez C, Borregan M, et al. From gestalt to gene: early predictive dysmorphic features of PMM2-CDG. J Med Genet. 2019; 56(4): 236 - 245. FI: 5,899(Q1)
- Martinez-Monseny T, Bolasell-Girgas M, Callejón L, Cuadras-Palleja D, Freniche V, Itzep DC, et al. AZATAX: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2-CDG). Ann Neurol. 2019; 85(5): 740 - 751. FI: 9,496(Q1)
- Montero-Sanchez R, Yubero-Siles D, Salgado MC, González MJ, Campistol-Plana J, O'Callaghan-Gordo M, et al. Plasma coenzyme Q 10 status is impaired in selected genetic conditions. Sci Rep. 2019; 9(1): 793 - 793. FI: 4,011(Q1)

**Networks**

- CIBERER, Centro de investigación biomédica en red para enfermedades raras: Año inicio 2007
- METABERN: Red europea de referencia para enfermedades metabólicas hereditarias. Año de inicio 2018
- ERNDIM: Asesor científico para el European quality control schemes for metabolic diseases





**APPLIED RESEARCH IN  
NEUROMUSCULAR DISEASES**

Cecilia Jiménez Mallebrera, PhD

**24**PUBLICATIONS  
WITH IF**46%**FIRST QUARTILE  
PUBLICATIONS**25%**FIRST DECILE  
PUBLICATIONS

2019 was a turning point for our group since we have consolidated a Gene Therapy program which started with a project to correct mutations in collagen VI employing CRISPR/Cas9 technology with the support of public (CIBERER) and private funds (F. Noelia). To foster gene therapies we actively participate in an EU COST Action together with groups from over 27 countries. Clinical trials and new treatments for neuromuscular diseases are exploding and our group is part of this effort, participating in 15 clinical trials and delivering groundbreaking treatments such as Spinraza® for Spinal Muscular Atrophy (SMA). To optimize this therapy and based on our expertise in biomarker discovery we are investigating extracellular vesicles and long non-coding RNAs in SMA. We aim to provide 360-degrees answers to neuromuscular diseases, an example of this is our psychological support program in schools for children with muscular dystrophies and the investigation of innovative motor function monitoring devices using artificial intelligence. We celebrated the 5th open day for patients and families with hundreds of attendees where the Share4Rare platform was presented. The 7th edition of the Neuromuscular Diseases course that we offer every two-years focused on the latest therapeutic and scientific advancements.

**Members****STAFF RESEARCHER**

Julita Medina, Andrés Nascimento, Carlos Ignacio Ortez Gonzalez, Irene Beatriz Zschaecck Luzardo

**POSTDOC - ASSOCIATE RESEARCHER**

Nafiel Natera de Benito, Selena Trifunov, Arístides López Márquez

**PHD STUDENT**

Cristina Jou Muñoz

**ASSISTANT RESEARCHER**

Maria del Carmen Badosa Gallego, Laura Carrera García, Anna Codina, Jessica Exposito Escudero, Raquel Gutiérrez, Sonia Segovia Simón

**TECHNICIAN**

Arístides López Márquez

**Selected projects**

- Biomarcadores precoces pronósticos y de respuesta a tratamiento en la Atrofia Muscular Espinal. Asociación galiciAME. PRIMERA CONVOCATORIA PARA FOMENTAR LA INVESTIGACIÓN DE LA AME. V. PI: Cecilia Jiménez Mallebrera. 2019 - 2020.
- 6MWT+: The Six Minutes Walking Test PLUS System. EIT Health e.V. EIT- Innovations by Ideas (BP 2019). PI: Julita Medina. 2019 - 2020.
- Nueva aproximación terapéutica para las miopatías de Ulrich y Bethlem basada en la edición de mutaciones dominantes del colágeno VI en fibroblastos de pacientes. OSSJD - Obra Social Sant Joan de Déu. Somos Uno 2018. PI: Cecilia Jiménez Mallebrera. 2018 - 2021.
- Aplicación de la proteómica y biología de sistemas al estudio del matrisoma y miRNAs en las Distrofias Musculares Congénitas. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI16/00579. PI: Cecilia Jiménez Mallebrera. 2017 - 2020.

**Selected publications**

- Berciano J, Natera-de Benito D. Description of Restrictively Defined Acute Flaccid Myelitis. JAMA Pediatr; 2019;173(7): 701 - 702. FI: 12,004(Q1)
- Carrera-García L, Natera-de Benito D, Lleixà C, Ortez-Gonzalez CI, Colomer J, Nascimento-Osorio A, et al. Chronic inflammatory demyelinating polyneuropathy associated with contactin-1 antibodies in a child. Neurol Neuroimmunol Neuroinflamm. 2019; 6(5). FI: 7,353(Q1)
- Domínguez-González C, Madruga-Garrido M, Mavillard F, Garone C, Aguirre-Rodríguez FJ, Donati MA, et al. Deoxynucleoside Therapy for Thymidine Kinase 2-Deficient Myopathy. Ann Neurol. 2019; 86(2): 293 - 303. FI: 9,496(Q1)
- Gonorazky HD, Naumenko S, Ramani AK, Nelakuditi V, Mashouri P, Wang P, et al. Expanding the Boundaries of RNA Sequencing as a Diagnostic Tool for Rare Mendelian Disease. Am J Hum Genet. 2019; 104(3): 466 - 483. FI: 9,924(Q1)
- Bazaga A, Roldan M, Badosa-Gallego MC, Jimenez-Mallebrera C, Porta JM. A Convolutional Neural Network for the Automatic

Diagnosis of Collagen VI-related Muscular Dystrophies. Appl Soft Comput. 2019; 85: 150772. FI: 5,472(Q1)

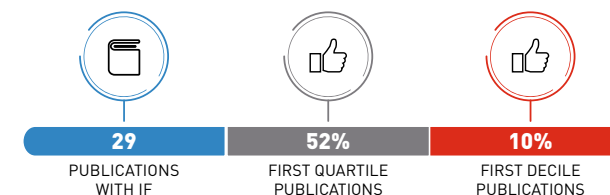
- Milev MP, Stanga D, Schänzer A, Nascimento-Osorio A, Saint-Dic D, Ortez-Gonzalez CI, et al. Characterization of three TRAPPC11 variants suggests a critical role for the extreme carboxy terminus of the protein. Sci Rep. 2019; 9(1): 14036 - 14036. FI: 3,998(Q1)

**Networks**

- Neuromuscular European Reference Network (EURO-ERN): miembros del Management Committee and Education Board
- Treat-NMD
- CSUR
- CIBERER (U703)
- RETICS-Plataforma Nacional de Biobancos, SGR-AGAUR Generalitat de Catalunya
- Sociedad Española de Neurología
- Sociedad Europea de Neuropediatría
- Sociedad Catalana de Neurologia
- World Muscle Society
- European Society for Gene and Cell Therapy

## PEDIATRIC NEUROMETABOLISM: NEURAL COMMUNICATION MECHANISMS AND PERSONALIZED THERAPIES

M<sup>a</sup> Angels García Cazorla, MD, PhD



In 2019, our team began to develop a platform of individualised therapies for disorders altering neuronal communication, mainly including synaptopathies, by using such tools as metabolic and neuronal plasticity modulation. Likewise, in collaboration with international networks, we are defining new neurometabolic disorders, new pathophysiological classifications, and patient registers.

### Members

#### STAFF RESEARCHER

Javier Aparicio, Jordi Muchart López

#### POSTDOC - ASSOCIATE RESEARCHER

Judith Armstrong, Roser Colomé, Alejandra Darling, Alfonso Luis de Oyarzabal Sanz, Carme Fons Estupiña, Mar O'Callaghan

#### PHD STUDENT

Ines Fernanda Medina Rivera, Natalia Alexandra Julia Palacios, Alba Tristan Noguero

#### ASSISTANT RESEARCHER

Uliana Musokhranova

#### TECHNICIAN

Cristina Grau

### Selected projects

- Caracterización clínica y molecular del síndrome de Rett: elucidar los casos no resueltos. Instituto de Salud Carlos III (ISCIII). FIS. PI15/01159. PI: Judith Silvia Armstrong Morón. 2016 -. 2019;
- Aproximaciones terapéuticas en un modelo celular de síndrome de Rett. OSSJD - Obra Social Sant Joan de Déu. Mi Princesa Rett. PI: M<sup>a</sup>Angels García Cazorla. 2014 - 2020.
- Medicina personalizada en los defectos genéticos de la neurotransmisión en pediatría. Nuevo enfoque basado en el metabolismo de la sinapsis neuronal. Instituto de Salud Carlos III (ISCIII). PI. PI18/00111. PI: M<sup>a</sup>Angels García Cazorla. 2019 - 2021.
- Therapeutic correction of tyrosine hydroxylase deficiency (THD): a neuropediatric disorder of dopamine synthesis and a model of parkinsonism". Hospital Sant Joan de Déu - Esplugues HSJD. Convocatòria de Beques de Recerca IRSJD - Carmen de Torres 2018. BR201802. PI: M<sup>a</sup>Angels García Cazorla. 2019 - 2022.

### Selected publications

- Castells AA, Gualdi D, Balada R, Tristan-Noguero A, Cortés-Saladelafont E, Ramos F, et al. Discovery of Biomarker Panels for Neural Dysfunction in Inborn Errors of Amino Acid Metabolism. Sci Rep. 2019; 9(1): 9128 - 9128. FI: 4,011(Q1)
- Pillai NR, Yubero-Siles D, Shayota BJ, De Oyarzabal-Sanz AL, Ghosh R, Sun Q, et al. Loss of CLTRN function produces a neuropsychiatric disorder and a biochemical phenotype that mimics Hartnup disease. Am J Med Genet A. 2019; 179(12): 2459 - 2468. FI: 2,197(Q3)
- Saudubray JM, Mochel F, Lamari F and Garcia-Cazorla A. Proposal for a simplified classification of IMD based on a pathophysiological approach: A practical guide for clinicians. J Inher Metab Dis. 2019; 42(4): 706 - 727. FI: 4,287(Q1)
- Soto D, Olivella M, Grau C, Armstrong-Moron J, Alcon C, Gasull X, et al. l-Serine dietary supplementation is associated with clinical improvement of loss-of-function GRIN2B -related pediatric encephalopathy. Sci Signal. 2019; 12(586). FI: 6,565(Q1)

### Networks

- 2007 - CIBERER
- 2017 - European Reference Network for Hereditary Metabolic Diseases (METABERN)
- 2014 - Connecting the growing brain
- 2015 - INTD - International Working Group on Neurotransmitter Related Disorders

**NEUROGENETICS  
AND MOLECULAR MEDICINE**Janet Hoenicka, MD, PhD  
Francesc Palau Martínez, MD, PhD**24**PUBLICATIONS  
WITH IF**66%**FIRST QUARTILE  
PUBLICATIONS**29%**FIRST DECILE  
PUBLICATIONS

The NeuroGene research group studies neurological and psychiatric disorders of genetic origin with the aim to improve their etiologic diagnosis and to increase our knowledge of their pathophysiology and pharmacological screening. NeuroGene carries out its scientific and translational work in the field of experimental biomedicine, within the framework of the Paediatric Institute for Rare Diseases (IPER). The scientific milestones which we achieved and the activities which we carried out in 2019 included: (1) the demonstration of neuroinflammation in axonal forms of Charcot-Marie-Tooth disease caused by the *GDAP1* gene (Fernandez-Lizarbe S et al, *Exp Neurol* 2019); (2) the study of the genetic architecture of Parkinson's disease in Spain (Bandrés-Ciga S et al, *Mov Disord* 2019); (3) the implementation of the "Translational Diagnosis" programme (Pijuan J et al., 2020 under revision) and the "Precision Medicine in Rare Diseases" programme (Palau F, *SEBBM Revista* 2020). Our training and dissemination activities included the coordination of the "Advances in Genetic Medicine, Genomic Medicine and Rare Diseases" course, and F. Palau's responsibility as editor-in-chief of *Orphanet Journal of Rare Diseases*.

**Members****STAFF RESEARCHER**Guerau Fernandez Isern,  
Joan Maynou Fernández,  
Mónica Roldan Molina**POSTDOC - ASSOCIATE RESEARCHER**

Lara Cantarero Abad, Esther Cuatrecasas Capdevila, Laura Dominguez Berzosa, Cristina Hernando Davalillo, Antonio F Martinez Monseny, Loreto Martorell Sampol, Jordi Pijuan Marquilles, Maria Rodriguez Sanz

**PHD STUDENT**

Elena Juárez Escoto

**ASSISTANT RESEARCHER**Adrian Alcalá Sanmartin,  
Arola Altimir Esquís,  
Alba Pascual Rodríguez**TECHNICIAN**

Daniel Castillo Mestres, Patricia Fernández Pareja, Jordi Genovès Escarré, Sara Manresa, Paola Pacheco Fernández, Patricia Peral García, Lluís Ramínez, Carlota Ros Bonaventura, Johanna Troya Balseca, Teresa Zabala Vidal

**Selected projects**

- El Paisaje de la Biología Axonal y las Membranas Asociadas a Mitocondrias en las Enfermedades Neurogenéticas. Ministerio de Economía Y Competitividad (MINECO). Retos colaboración 2015. SAF2015-66625-R. PI: Francesc Palau Martínez. 2016-2019.
- El paisaje entre el fenotipo y el genotipo de enfermedades neurológicas del desarrollo: Validación de un modelo de biología funcional clínica. Fundación Isabel Gemio Inv. De Distrofias Musculares y Enfermedades Raras. Fundación Isabel Gemio. PI: Francesc Palau Martínez. 2017 - 2020.
- El modelo de genómica clínica - genómica funcional en la innovación diagnóstica de las enfermedades neurogenéticas. Hospital Sant Joan de Déu - Esplugues HSJD. Beques de Recerca. BR201701. PI: Janet Hoenicka. 2018 - 2021

**Selected publications**

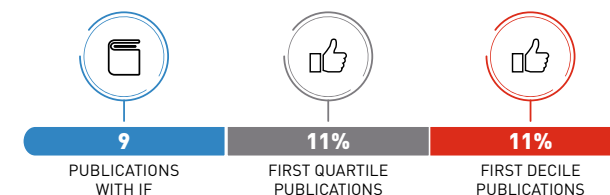
- Bandrés-Ciga S, Ahmed S, Sabir MS, Blauwendraat C, Adarmes-Gómez AD, Bernal-Bernal I, et al. The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. *Mov Disord*. 2019; 34(12): 1851 - 1863. FI: 8,222(Q1)
- Fernandez-Lizarbe S, Civera-Tregon A, Cantarero-Abad L, Herrer I, Juárez P, Hoenicka J and Palau-Martínez F. Neuroinflammation in the pathogenesis of axonal Charcot-Marie-Tooth disease caused by lack of *GDAP1*. *Exp Neurol*. 2019; 320: 113004 - 113004. FI: 4,562(Q1)
- Martinez-Monseny T, Cuadras-Palleja D, Bolasell-Girgas M, Muchart-Lopez J, Arjona-Fernandez C, Borregan M, et al. From gestalt to gene: early predictive dysmorphic features of *PMM2-CDG*. *J Med Genet*. 2019; 56(4): 236 - 245. FI: 5,899(Q1)
- Mollá B, Muñoz-Lasso DC, Calap P, Fernández-Vilata A, de la Iglesia-Vaya M, Pallardó FV, et al. Phosphodiesterase Inhibitors Revert Axonal Dystrophy in Friedreich's Ataxia Mouse Model. *Neurotherapeutics*. 2019; 16(2): 432 - 449. FI: 5,552(Q1)

**Networks**

- Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER), desde 2006
- Centro de Investigación Biomédica en Red de Salud Mental (CIBERSAM), desde 2007
- Orphanet, desde 2010. Coordinador de Orphanet-España, CIBERER
- ERN on Intellectual Disability and Congenital Anomalies - ITHACA (Unidad Afiliada)

## NEONATAL BRAIN

Alfredo Garcia-Alix Pérez, MD, PhD



### Members

#### STAFF RESEARCHER

Thais Agut Quijano, Ana Alarcón Allen, Nuria Carreras Blesa, Jonathan Francisco Olival Tremaria, Carla Skilham, Nuria Herranz Rubia

### Selected projects

- Elaboración de un atlas morfométrico normalizado de la maduración cerebral del recién nacido prematuro y valoración del impacto de la patología cerebral en el proceso madurativo. Instituto de Salud Carlos III (ISCIII). PI: PI18/00110. PI: Thais Agut Quijano. 2019 - 2021.
- Contratos Río Hortega. Instituto de Salud Carlos III (ISCIII). Río Hortega. CM18/00258. PI: Alfredo Garcia-Alix. 2019 - 2020.
- Rare Coding Genetic Variants Associated with neonatal arterial ischemic stroke. Fundació La Marató de TV3. Ictus i lesions medul·lars/cerebrals traumàtiques. 201714.10. PI: Alfredo Garcia-Alix. 2018 - 2021.

### Selected publications

- Batista Muñoz A, Hadley S, Iriando-Sanz M, Agut-Quijano T and Camprubí-Camprubí M. Role of beta-2-microglobulin as a biomarker in very preterm and extremely preterm infants with CNS inflammation. PLoS One. 2019; 14(5): e0216498. FI: 2,776(Q2)
- Echeverría-Palacio CM, Agut-Quijano T, Arnaez-Solis J, Valls-Lafon A, Reyne M and Garcia-Alix A. Neuron-Specific Enolase in Cerebrospinal Fluid Predicts Brain Injury After Sudden Unexpected Postnatal Collapse. Pediatr Neurol. 2019; 101: 71 - 77. FI: 2,326(Q2)
- Olivé G, Agut-Quijano T, Echeverría-Palacio CM, Arca-Diaz G and Garcia-Alix A. Usefulness of Cranial Ultrasound for Detecting Neonatal Middle Cerebral Artery Stroke. ULTRASOUND MED BIOL. 2019; 45(3): 885 - 890. FI: 2,205(Q2)

- Núñez C, Arca G, Agut T, Stephan-Otto C, García-Alix A, Precise neonatal arterial ischemic stroke classification with a three-dimensional map of the arterial territories of the neonatal brain. Pediatric Research. 2019:1-6.
- Arnaez-Solis J, Herranz Rubia N, Garcia-Alix A, ESP-EHI Group. Perinatal hypoxic-ischaemic encephalopathy: a national survey of end-of-life decisions and palliative care. BMJ Support Palliat Care. 2019. FI: (Q)

## NEUROPSYCHOLOGY

Roser Pueyo Benito, PhD

In 2019, the Neuropsychology group published 6 papers in international journals, four of which were 1st quartile journals and one was a 2nd quartile journal. Six research projects obtained in competitive calls are being directed. Our group also participated in a European project on transference and one of our members defended an international doctoral thesis. Likewise, our group contributed to the development of the European Dyskinetic Cerebral Palsy Network (EDCPN), and another of our members obtained the ICREA-Acadèmia Prize.

### Members

#### STAFF RESEARCHER

Júlia Ballester Plané, Xavier Caldú Ferrús, María Ángeles Jurado, Olga Laporta Hoyos, Maria Mataró Serrat, Júlia Miralbell Blanch

#### POSTDOC - ASSOCIATE RESEARCHER

Mar Ariza Gonzalez, Cristina Sánchez Castañeda

#### PHD STUDENT

Montse Blasco Sierra, Adria Bermudo Ballaquet, Alba Castells Sánchez, María García Galant, Nomeí Lamonja Vicente, Sandra Luís Ruiz, Jonatan Ottino González, Xavier Prats Soteras, Francesca Roig Coll

### Selected projects

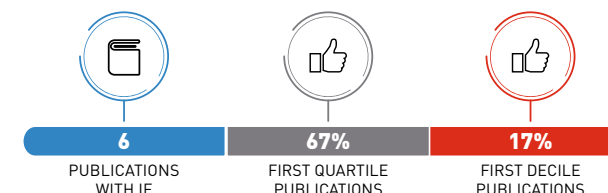
- Executive function training in childhood obesity: food choice, quality of life and brain connectivity (TOUCH). Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 201616-10. PI: María Ángeles Jurado Luque. 2017-2020.
- Entrenamiento ejecutivo en parálisis cerebral: participación, calidad de vida y conectividad cerebral. Ministerio de Economía y Competitividad. Convocatoria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Projectes I+D+I. PSI2016-75979-R. PI: Roser Pueyo Benito. 2016-2020.
- Ajut per incentivar i consolidar la recerca d'excel·lència ja existent a les universitats públiques de Catalunya. Programa ICREA Academia 2018. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). Convocatòria del Programa Icrea Acadèmia. PI: Maria Mataró Serrat. 2019-2023.

### Selected publications

- de Lange SC, Scholtens LH, Alzheimers Disease Neuroimaging Initiative, van den Berg LH, Boks MP, Bozzali M, et al. Shared vulnerability for connectome alterations across psychiatric and neurological brain disorders. Nat Hum Behav. 2019; 3(9): 988 - 998. FI: 10,575(Q1)
- Laporta O, Ballester J, Leiva D, Ribas T, Miralbell J, Torroja-Nualart C, et al. Executive function and general intellectual functioning in dyskinetic cerebral palsy: Comparison with spastic cerebral palsy and typically developing controls. Eur J Paediatr Neurol. 2019; 23(4): 546 - 559. FI: 2,496(Q1)
- Caldú X, Ottino-González J, Sánchez-Garre C, Hernan I, Tor E, Sender-Palacios MJ, et al. Effect of the catechol-O-methyltransferase Val 158 Met polymorphism on theory of mind in obesity. Eur Eat Disord Rev. 2019; 27(4): 401 - 409. FI: 3,154(Q1)
- Ottino-González, J, Jurado MA, García-García I, Caldú X, Prats-Soteras X, Tor E, et al. Allostatic load and executive functions in overweight adults. Psychoneuroendocrinology. 2019; 106:165-170. FI: 4,013(Q1)

### Networks

- European dyskinetic cerebral palsy network (EDCPN) des del 2019; Roser Pueyo



**COGNITIVE NEUROSCIENCE**

Carles Escera Micó, PhD

2019 has been quite active and successful for the Cognitive Neuroscience Research Group. We published the first paper (Ribas-Prats et al., 2019, Hear. Res.) in collaboration with the Foetal Environment and Obstetrical Complication Research Group, establishing normative values for the use of the frequency-following response (FFR) as a biomarker for neurocognitive development. Likewise, we secured competitive funding from the Knowledge Generation Programme of the Spanish Ministry of Science for this project. Using the FFR, we described subcortical deficits in the suppression of repeating auditory information in autism as a potential mechanism for hypersensitivities in this disorder (Font-Alaminos et al., 2019, Biol. Psychol.). Furthermore, we continued our research on the neural mechanism of auditory cognition, highlighting the role of regularity encoding in human neuroimaging experiments (Cacciaglia et al., 2019, Neuroimage) and animal models (Malmierca et al., 2019, Neuroimage). Three PhD theses (International Diploma) have been defended, two of them dealing with the mechanisms generating the FFR. Lastly, we started our ERC-Advance Grant

research, investigating the role of the soundscape (the acoustic signature of a particular location, with a focus on rock art sites) in driving auditory-induced emotions and mental states.

**Members****STAFF RESEARCHER**

Imma Clemente Lapena, Maria José Corral López, Jordi Costa Faidella, Maribel Nuñez Peña, Iria San Miguel Insúa, Hans Supèr, Marc Via García

**POSTDOC - ASSOCIATE RESEARCHER**

Natàlia Gorina

**PHD STUDENT**

Sonia Arenillas Alcón, Alejandro Lerer, Marta Puertollano Rodríguez, Teresa Ribas Prats

**Selected projects**

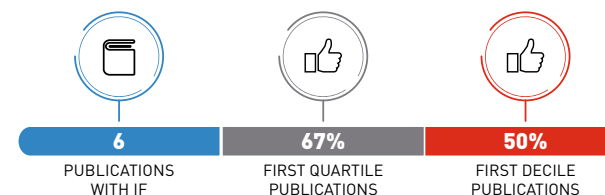
- The sound of special places: exploring rock art soundscapes and the sacred (role: Principal Investigator of Psychophysics (RL2) and Cognitive Neuroscience (RL3) work packages), 2018-2023. Budget: 316,000 €. European Research Council. ERC Advanced Grants. 787842. PI: Carles Escera Micó. 2018 - 2023
- Papel de los genes y de factores epigenéticos en la modulación de potenciales cerebrales asociados al procesamiento del lenguaje y de la música. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Modalitat Projectes d'R+D de Generació del Coneixement. PGC2018-094765-B-I00. PI: Marc Vila García. 2019 - 2021.
- Ajut per incentivar i consolidar la recerca d'excel·lència ja existent a les universitats públiques de Catalunya. Programa ICREA Acadèmia 2015. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). Convocatòria del Programa Icrea Acadèmia. PI: Carles Escera Micó. 2016 - 2020.

- Caracterización de la respuesta de seguimiento de frecuencia (RSF) en recién nacidos como posible biomarcador de desarrollo neurocognitivo. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Modalitat Projectes d'R+D de Generació del Coneixement. PGC2018-094765-B-I00. PI: Carles Escera Micó. 2019 - 2021.

**Selected publications**

- Cacciaglia R, Costa-Faidella J, Zarnowiec K, Grimm S, Escera C. Auditory predictions shape the neural responses to stimulus repetition and sensory change. Neuroimage. 2019; 186: 200 - 210. FI: 5,812(Q1)
- Font-Alaminos M, Ribas-Prats T, Gorina N, Escera C. Emergence of prediction error along the human auditory hierarchy. Hear Res. 107954 - 107954. FI: (Q2)
- Malmierca MS, Niño-Aguillón BE, Nieto-Diego J, Porteros Á, Pérez-González D, Escera C. Pattern-sensitive neurons reveal encoding

- of complex auditory regularities in the rat inferior colliculus. Neuroimage 2019; 184: 889 - 900. FI: 5,812(Q1)
- Ribas-Prats T, Almeida-Toledano L, Costa-Faidella J, Plana M, Corral MJ, Gómez-Roig MD and Escera C. The frequency-following response (FFR) to speech stimuli: A normative dataset in healthy newborns. Hear Res. 2019; 371: 28 - 39. FI: 2,952(Q1)



## ATTENTION, PERCEPTION AND LANGUAGE ACQUISITION (APAL)

Laura Bosch Galceran, PhD

Our main research focus was on the assessment of different factors (linguistic environment, prematurity, and sensory and auditory integration) in the audiovisual processing of speech in the prelexical stage. Through the use of eye-tracking technology, we observed how the infant's attention is modulated by maturational and experiential factors in various tasks (speech perception and tracking of the adult's gaze), which are significant for learning in interactive contexts. We collaborated with Neonatology (N. Carreras, preterm infant population), and with ORL (M.A. Claveria) and Neuroradiology (M. Rebollo) on the project on biomarkers that impact the language learning process in pre-implantation deaf patients. Likewise, we completed a follow-up study of children born small for gestational age who were assessed at age 7 years in order to identify markers of school-age learning difficulties risk (Dr L. Ibáñez's team, Endocrinology).

### Members

#### STAFF RESEARCHER

Ferran Pons Gimeno, Marta Ramon Casas

#### POSTDOC - ASSOCIATE RESEARCHER

Clément François

#### PHD STUDENT

Joan Birulés Muntané

#### ASSISTANT RESEARCHER

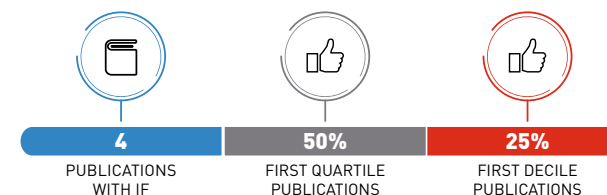
Jessica Sánchez-Galán Frauca

### Selected projects

- Pròrroga del conveni de col·laboració per una recerca sobre cognició i percepció de llengües en nens. Fundació Privada Betània-Patmos. PI: Ferran Pons Gimeno. 2019-2023.
- Variabilidad en el procesamiento audiovisual del habla en la infancia: rol de la experiencia lingüística, maduración neurológica y capacidad sensorial. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Modalitat Projectes d'R+D de Generació del Coneixement. PGC2018-097487-B-I00. PI: Ferran Pons Gimeno and Laura Bosch Galceran. 2019-2021.

### Selected publications

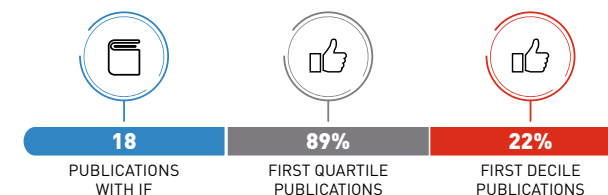
- Pons F, Bosch L, Lewkowicz DJ. Twelve-month-old infants' attention to the eyes of a talking face is associated with communication and social skills. *Infant Behav Dev.* 2019; 54: 80 - 84. FI: 1,353(Q4)
- Birules J, Bosch L, Brieke R, Pons F and Lewkowicz DJ. Inside bilingualism: Language background modulates selective attention to a talker's mouth. *Dev Sci.* 2019; 22(3): e12755. FI: 4,096(Q1)





**HUMAN MOLECULAR GENETICS I**

Daniel Raúl Grinberg Vaisman, PhD



In 2019 we described new genes and mutations responsible for intellectual disability in patients originally diagnosed as C syndrome or Bohring-Opitz (PIGT, DPH1, ASXL1, TRAF7), and in other patients with rare diseases such as familial juvenile idiopathic arthritis (LACC1), intellectual disability (SETD1B) or microcephaly (TUBB5). We analysed the C7orf76 locus in relation to osteoporosis. We also took part in multi-centre studies on mutations in RASopathies, stroke and its recovery, and psychiatric disturbances, identifying for the first time genes causing predisposition to ADHD. We also conducted studies at genomic level on the genetics of autism, of cocaine dependence, and of aggressiveness, and on the epigenetics of ADHD. In the field of lysosomal disorders, we generated iPS cells for Sanfilippo syndrome type C by the CRISPR/Cas9 technique and we differentiated them into neurons. Likewise, we participated in the development of IT tools for exome analysis. Lastly, we launched a project aimed to generate and to study neuronal models based on iPS cells derived from patients with Williams-Beuren syndrome or 7q11.23 microduplication syndrome.

**Members****STAFF RESEARCHER**

Susana Balcells Comas, Bru Cormand Rifà, Roser Crorominas Castiñeira, Raquel Rabionet Janssen

**POSTDOC - ASSOCIATE RESEARCHER**

Judit Cabana Domínguez, Noelia Fernández Castillo, Bárbara Torrico Avilés, Roser Urreiziti Frexedas

**PHD STUDENT**

Ester Antón Galindo, Noelia Benetó Gandia, Laura Castilla Vallmanya, Nuria Martínez Gil, Laura Pineda Cirera, Neus Roca Ayats, Anu Shivalikanjli

**TECHNICIAN**

Mónica Cozar Morillo

**Selected projects**

- Sinergias en el estudio genético y la búsqueda de terapias para el síndrome de Opitz C, la fractura femoral atípica, la osteoporosis y enfermedades lisosomales. Ministerio de Economía y Competitividad. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Projectes I+D+I. SAF2016-75948-R. PI: Susana Balcells Comas. 2016 - 2019.
- Effects of Nutrition and Lifestyle on Impulsive, Compulsive, and Externalizing behaviors (Eat2beNICE). Unió Europea. HORIZON 2020 (2014-2020). P2. SC2: SUSTAINABLE FOOD SECURITY - RESILIENT AND RESOURCE-EFFICIENT VALUE CHAINS (H2020-SFS-2016-2017). 728018. PI: Bru Cormand Rifà. 2017 - 2022.
- GENIUS: GENetic Influences on functional outcome after Stroke. Fundació La Marató de TV3. Ictus i lesions medul·lars/cerebrals traumàtiques. Reg. 70/307 Proj. 201726. PI: Raquel Rabionat Janssen. 2017 - 2019.

**Selected publications**

- Mason S, Castilla-Vallmanya L, James C, Andrews PI, Balcells S, Grinberg-Vaisman DR, et al. Case report of a child bearing a novel deleterious splicing variant in PIGT. Medicine (Baltimore). 2019; 98(8): e14524. FI: 1,870 (Q2)
- Roca N, Martinez N, Cozar M, Gerousi M, Garcia-Giralt N, Ovejero D, et al. Functional characterization of the C7ORF76 genomic region, a prominent GWAS signal for osteoporosis in 7q21.3. Bone. 2019; 123: 39 - 47. FI: 4,360 (Q1)
- Pineda L, Shivalikanjli A, Cabana J, Demontis D, Rajagopal VM, Børglum AD, et al. Exploring genetic variation that influences brain methylation in attention-deficit/hyperactivity disorder. Transl Psychiatry. 2019; 9(1): 242 - 242. FI: 5,182 (Q1)
- Beneto N, Cozar M, García-Morant M, Creus-Bachiller E, Vilageliu L, Grinberg-Vaisman DR and Canals I. Generation of two compound heterozygous HGSNAT-mutated lines from healthy induced pluripotent stem cells using CRISPR/Cas9 to model Sanfilippo C syndrome. Stem Cell Res. 2019; 41: 101616 - 101616. FI: 3,929 (Q1)

**Networks**

- CIBERER, 2007
- GEFOS-GENOMOS, 2002
- GEMSTONE (COST ACTION), 2019
- DARTER (COST ACTION), 2018
- IMpACT, 2007
- PGC, 2009
- IHGC, 2007



**HUMAN MOLECULAR GENETICS II**

Gemma Marfany Nadal, PhD

This year our group consolidated its activity in rare disease research, ranging from genetic diagnosis to the creation of models, the analysis of gene networks and a possible therapy. We bolstered our line of genetic diagnosis of hereditary eye disorders by founding and strengthening a spin-off, DBGen Ocular Genomics. Likewise, we established collaborations addressed to advancing in the research on and treatment of rare diseases, with groups at the University of Oxford (specialists in nanoparticles for nanomedicine), at the CIPF in Valencia (specialists in generating iPSCs and in differentiation into retina), and at the University of Barcelona (UB) (bioinformatics groups, for the analysis of gene networks and complex systems).

**Members****POSTDOC - ASSOCIATE RESEARCHER**

Serena Mirra

**PHD STUDENT**

Izarbe Aïsa Marin, Elena Borrego Domènech, Vasileios Toulis

**Selected projects**

- Modelos animales para las distrofias de retina: estrategias de edición genómica, fenotipado y nanoterapia. Ministerio de Economía y Competitividad. SAF2016-80937-R. Proyectos de I+D+I 2016, programa estatal de investigación, desarrollo e innovación orientada a los retos de la sociedad. PI: Gemma Marfany Nadal (coPI: Roser Gonzalez Duarte). 2016-2020.
- Enfermedades Raras. Ministerio de Sanidad y Consumo. Convocatòria d'ajuts destinats a finançar estructures estables d'investigació cooperativa, a l'àrea de biomedicina i ciències de la salut, en el marc del Programa Ingenio 2010, Programa Consolider, accions CIBER. CB06/07/0041. PI: Gemma Marfany Nadal (coPI: Roser Gonzalez Duarte). 2018-2020.
- Contractes KnowHow. DBGen Ocular Genomics, S.L. Fundació spin-off de diagnòstic genètic de malalties hereditàries de la visió. 309710. PI: Gemma Marfany Nadal (coPI: Roser Gonzalez Duarte). 2018 - 2023.

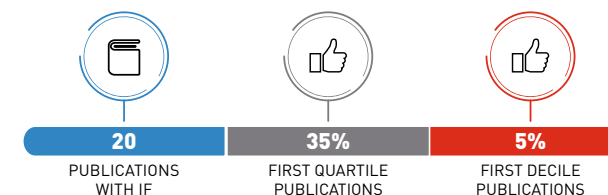
**Selected publications**

- Trigueros S, Domènech EB, Toulis V and Marfany G. In Vitro Gene Delivery in Retinal Pigment Epithelium Cells by Plasmid DNA-Wrapped Gold Nanoparticles. Genes (Basel). 2019; 10(4). FI: 3,331(Q2)
- Bolinches-Amorós A, León M, Del Buey Furió V, Marfany G, González-Duarte R, Erceg S and Lukovic D. Generation of an iPSC line from a retinitis pigmentosa patient carrying a homozygous mutation in CERKL and a healthy sibling. Stem Cell Res. 2019; 38: 101455 - 101455. FI: 3,929(Q1)
- Arenas-Galnares R, Castillo-Lara S, Toulis V, Boloc D, González-Duarte R, Marfany G, Abril JF. RPGeNet v2.0: expanding the universe of retinal disease gene interactions network. Database, Database. 2019; 2019, baz120, doi:10.1093/database/baz120. FI: 3.68(Q1)
- Mirra S and Marfany G. Mitochondrial gymnastics in retinal cells: a resilience mechanism against oxidative stress and neurodegeneration. Adv Exp Med Biol. 2019; 1185: 513 - 5179. FI: 2,126(Q2)

**8**PUBLICATIONS  
WITH IF**37%**FIRST QUARTILE  
PUBLICATIONS

**ETIOPATHOGENESIS AND TREATMENT  
OF SEVERE MENTAL DISORDERS (MERITT)**

Susana Ochoa Güerre, MD, PhD  
Judith Usall Rodié, MD, PhD



We continued to work on various financed research projects addressed to the study of protective and risk factors in first psychotic episodes, and of metacognition and social cognition in psychosis. In the field of biomarkers, we worked on various projects in coordination with the clinical area.

During 2019, our team obtained financing for predoctoral personnel assigned to research groups. A total of 4 persons are attached to our research groups thanks to [m1] predoctoral project grants.

Our group's impact factor and our number of publications remained on levels similar to those of previous years.

**Members****STAFF RESEARCHER**

Antonio Manuel Alba Muñoz, Hilari Andres Mora, Belen Arranz Martí, Emma Casas Anguera, Marta Coromona Sadurní, Ruth Cunill Clotet, María José Escandell Morcillo, Gemma Escartin Martin, Gemma Escuder Romeva, Mar García Franco, Maria Elena Huerta Ramos, Marcia Irazábal Giménez, Francisco Martínez Zambrano, Eva Miquel Fernandez, María Trinidad Peláez Martínez, Belen Ramos JoseMaria, Elena Rubio Abadal, Luis San Molina, Sonia Vilamala Anton

**PHD STUDENT**

Lucia Beatriz Diaz Cutraro, Silvia Marcó Garcia, America Jennifer Vera Montecinos, Elia Vila Andreu

**ASSISTANT RESEARCHER**

Ana Butjosa Molines, Núria del Cacho Ortega, Marta Ferrer Quintero, Clara Serra Arumí, Marina Verdaguer Rodriguez

**COLLABORATOR RESEARCHER**

Joan Costa Escolà, Raquel López Carrilero, Alfonso Monje Hernández, Francisco Vidal Domenech, Regina Vila Badia

**Selected publications**

- Nuñez C, Stephan-Otto C, Usall J, Bioque M, Lobo A, González-Pinto A, et al. Neutrophil Count Is Associated With Reduced Gray Matter and Enlarged Ventricles in First-Episode Psychosis. *Schizophr Bull.* 2019; 45(4): 846 - 858. FI: 7,289(Q1)
- Siddi S, Ochoa S, Laroi F, Cella M, Raballo A, Saldivia S, et al. A Cross-National Investigation of Hallucination-Like Experiences in 10 Countries: The E-CLECTIC Study. *Schizophr Bull.* 2019; 45(45 Suppl 1): S43-S55. FI: 7,289(Q1)
- López-González I, Pinacho R, Vila É, Escanilla A, Ferrer I, Ramos B. Neuroinflammation in the dorsolateral prefrontal cortex in elderly chronic schizophrenia. *Eur Neuropsychopharmacol.* 2019; 29(3): 384 - 396. FI: 4,468(Q1)
- Dolz M, Tor J, De la Serna E, Pardo M, Muñoz Samons D, Rodríguez Pascual M, et al. Characterization of children and adolescents with psychosis risk syndrome: The Children and Adolescents Psychosis Risk Syndrome (CAPRIS) study. *Early Interv Psychiatry.* 2019; 13(5): 1062 - 1072. FI: 3,323(Q2)

- Vila É, Huerta-Ramos ME, Nuñez C, Usall J, Ramos B. Specificity proteins 1 and 4 in peripheral blood mononuclear cells in postmenopausal women with schizophrenia: a 24-week double-blind, randomized, parallel, placebo-controlled trial. *Eur Arch Psychiatry Clin Neurosci.* 2019; 269(8): 941 - 948. FI: 3,192(Q2)

**Networks**

- CIBERSAM
- GRup de dona l salut mental. Societat Catalana de Psiquiatria i Salut Mental

## RESEARCH IN CHILD AND ADOLESCENT MENTAL HEALTH

Montserrat Dolz Abadia, MD, PhD

The basic focus of our research group is clinical research on various psychiatric disturbances, with special emphasis on neurodevelopmental disorders. At present our efforts are centred on the study of childhood-onset psychoses (PI: Dr Dolz), attention deficit hyperactivity disorders (ADHD; PI: Dr Alda), and autism spectrum disorders (ASD; PI: Dr Mairena).

In recent years, our group has become consolidated through the performance of various projects financed by both public institutions (FIS projects) and private ones, and through collaborations with various stable research groups of the mental health biomedical research network CIBERSAM, such as Group G04 of the Hospital Clínic i Provincial de Barcelona and Group G01 of the Hospital Gregorio Marañón in Madrid. One of the highlights of 2019 was the grant of the European project SAVE, centred on the detection and treatment of trauma in children exposed to domestic violence (PI: Álvarez). In the national sphere, another highlight was the grant of a competitive project focused on the validation of a parenting programme for preschool children with autism and speech delay.

### Members

#### STAFF RESEARCHER

José Ángel Alda Díez, Mar Alvarez Segura, Ester Camprodon Rosanas, Maria Diez Juan, Fernando Lacasa Saludes, M<sup>a</sup> Ángeles Mairena García de la Torre, Josep Lluís Matalí Costa, Laia Mollà Cusí, Daniel Muñoz Samons, Marta Pardo Gallego, Eduardo Serrano Troncoso, Anna Sintés Estevez, Esther Via Virgili

#### PHD STUDENT

Ania Perez Racana, Jordina Tor Fabra

#### ASSISTANT RESEARCHER

Anna Martínez García, Sara Parrilla Carrasco, Almudena Ramírez Martín, Paula Resina Curado, Miriam Sanabra Gonzalez, Berta Sanfeliu Salvà, Carlota Saumell Andreu, Ingrid Tortadès Arumí

#### COLLABORATOR RESEARCHER

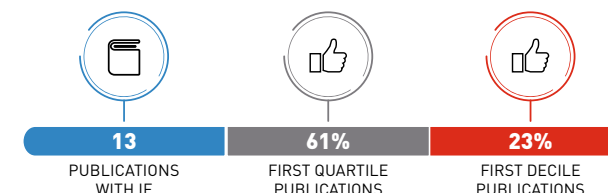
Laura Puigcerver Navarro

### Selected projects

- SAVE\_Support and treatment of traumatized children After Violence. European Commission. REC-RDAP-GBV-AG. 856677. PI: Mar Alvarez Segura. 2019 - 2021.
- First Steps: estudio piloto aleatorizado para la implementación del programa de parentalidad Incredible Years® en preescolares con autismo y retraso del lenguaje en España. Fundación Alicia Koplowitz. Ayudas a proyectos de investigación. PI: Laia Villalta Maciá. 2019 - 2021.
- Placebo-Controlled trial in Ultra-high Risk for Psychosis with Omega-3 fatty acids in Europe (PURPOSE). Stanley Medical Research Institute. EudraCT\_2015-003503-39. PI: Montserrat Dolz Abadia. 2017 - 2021.

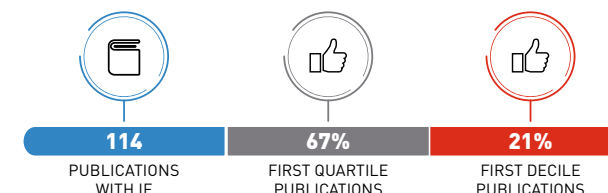
### Selected publications

- Camprodon-Rosanas E, Pujol J, Martínez-Vilavella G, Blanco-Hinojo L, Medrano-Martorell S, Batlle S, et al. Brain Structure and Function in School-Aged Children With Sluggish Cognitive Tempo Symptoms. J Am Acad Child Adolesc Psychiatry. 2019; 58(2): 256 - 266. FI: 6,391(Q1)
- Estrada-Prat X, Van Meter AR, Camprodon-Rosanas E, Batlle-Vila S, Goldstein BI, Birmaher B. Childhood factors associated with increased risk for mood episode recurrences in bipolar disorder-A systematic review. BIPOLAR DISORD. 2019; 21(6): 483 - 502. FI: 4,936(Q1)
- Dolz M, Tor J, De la Serna E, Pardo M, Muñoz Samons D, Rodríguez Pascual M, et al. Characterization of children and adolescents with psychosis risk syndrome: The Children and Adolescents Psychosis Risk Syndrome (CAPRIS) study. Early Interv Psychiatry. 2019; 13(5): 1062 - 1072. FI: 3,323(Q2)



## EPIDEMIOLOGY OF MENTAL DISORDERS AND AGING

Josep Maria Haro Abad, MD, PhD  
Ai Koyanagi, MD, PhD



### Members

#### STAFF RESEARCHER

Paula Cristobal Narvaez, Andrea De La Garza Puentes, Mireia Félez Nóbrega, Albert Sánchez Niubó, Sara Siddi, Stefano Tyrovolas

#### POSTDOC - ASSOCIATE RESEARCHER

Beatriz Olaya Guzmán

#### ASSISTANT RESEARCHER

Ivet Bayes, Joan Domènech Abella, Daniel Fernández Martínez, Aina Gabarrell Pascuet, Iago Giné Vázquez, Desireé Gutiérrez Marin, Blanca Mellor Marsa, Maria Victoria Moneta, Anna Monistrol Mula, Agnès Montoya Rimbau, Ziggi Santini, Teresa Torres Moral, Elisabeth Catharina Wilhelmina Vorstenbosch

#### MANAGEMENT

Carla Obradors Tarragó

### Selected projects

- Epidemiología de los factores asociados con el envejecimiento saludable. Ministerio de Economía Y Competitividad (MINECO). Proyectos JIN\_Jóvenes Investigadores. RTI2018-100927-J-I00. PI: Daniel Fernández Martínez. 2019 - 2022.
- ESTUDIO BIENESTAR Y SALUD POST MIGRACIÓN: DETECCIÓN NECESIDADES PSICOSOC.Y SANITARIAS. Ministerio de Trabajo, Migraciones y Seguridad Social. Convocatoria FAMI. II IN 180803 EN 162 FA 01. PI: Josep Maria Haro Abad. 2019 - 2019.
- Ayudas Juan de la Cierva - Formación. Beneficiario: Paula Cristóbal. Ministerio de Economía Y Competitividad (MINECO). Juan de la Cierva-Formación. FJCI-2017-31655. PI: Josep Maria Haro Abad. 2019 - 2020.
- SYNCHROS: SYnergies for Cohorts in Health: integrating the ROle of all Stakeholders. European Commission. H2020-SC1-HCO-09. 82884. PI: Josep Maria Haro Abad. 2019 - 2021.
- Frailty and ageing assessment: Research of global patterns and determinants. Instituto de Salud Carlos III (ISCIII). Miguel Servet I (Proyecto). CP18/00006. PI: tefanos Tyrovolas. 2019 - 2022.

### Selected publications

- Burstein R, Henry NJ, Collison ML, Marczak LB, Sligar A, Watson S, et al. Mapping 123 million neonatal, infant and child deaths between 2000 and 2017. *Nature*. 2019; 574(7778): 353 - 358. FI: 43,070(Q1)
- Vancampfort D, Firth J, Correll CU, Solmi M, Siskind D, De Hert M, et al. The impact of pharmacological and non-pharmacological interventions to improve physical health outcomes in people with schizophrenia: a meta-review of meta-analyses of randomized controlled trials. *World Psychiatry*. 2019; 18(1): 53 - 66. FI: 34,024(Q1)
- Kim JY, Son MJ, Son CY, Radua J, Eisenhut M, Gressier F, et al. Environmental risk factors and biomarkers for autism spectrum disorder: an umbrella review of the evidence. *Lancet Psychiatry*. 2019; 6(7): 590 - 600. FI: 18,329(Q1)
- Firth J, Siddiqi N, Koyanagi A, Siskind D, Rosenbaum S, Galletly Cet al. The Lancet Psychiatry Commission: a blueprint for protecting physical health in people with mental illness. *Lancet Psychiatry*. 2019; 6(8): 675 - 712. FI: 18,329(Q1)
- Degenhardt L, Bharat C, Glantz MD, Sampson NA, Al-Hamzawi A, Alonso J, et al. Association of Cohort and Individual Substance

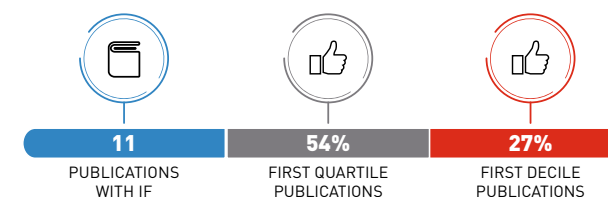
Use With Risk of Transitioning to Drug Use, Drug Use Disorder, and Remission From Disorder: Findings From the World Mental Health Surveys. *JAMA Psychiatry*. 2019; 76(7): 708 - 720. FI: 15,916(Q1)

### Networks

- CIBERSAM
- European Innovation Partnership on Active and Healthy Ageing (the Partnership)

## HEALTH TECHNOLOGY ASSESSMENT IN PRIMARY CARE AND MENTAL HEALTH (PRISMA)

Maria Rubio Valera, PhD  
Antoni Serrano Blanco, MD, PhD



The PRISMA research group has expanded its research on healthcare services with projects focused on cancer, stroke and preventive activities. Likewise, it has increased its line of initial medication non-adherence with paediatrics projects and the pilot test of the intervention, and it has kept up the collaboration with Barcelona Provincial Council for territorial diagnoses of mental health in Catalonia, while remaining active in the national research networks CIBERESP and redIAPP. In the clinical field, we lead an innovative public purchasing project for restraint-free hospitals, and we collaborated with EURECAT on the LiveIncite project for the development of an app that will minimise surgical complications. In the teaching sphere, we co-led the publication of the first Spanish-language psychiatric hospitalisation manual.

### Members

#### STAFF RESEARCHER

Luisa Baladón Higuera, Vanessa Barneda, Víctor Chavarría Romero, Laura González Suñer, Cristina López Ortiz

#### POSTDOC - ASSOCIATE RESEARCHER

Ignacio Aznar Lou, Elisabeth Parody Rua, Carlos Peña Salazar, Enric Vicens Pons

#### PHD STUDENT

Cristina Carbonell Duacastella, Maria Montserrat Gil Girbau, Carles Vilaplana Carnerero

#### ASSISTANT RESEARCHER

Claudia Costa Iracheta

#### COLLABORATOR RESEARCHER

Ana Fernández Sánchez, Carme Gallardo González, Maria Iglesias González, Ana Maria Jové Massó, Martin Knapp, Maria Antonia March Pujol, Paul McCrone, Juan Manuel Mendive Arbeloa, Maria Teresa Peñarrubia Maria, Ramón Sabes Figuera, Lluís Salvador Carulla

### Selected projects

- CIBER\_Epidemiology and Public Health. Instituto de Salud Carlos III (ISCIII). CIBER. CB16/02/00322. PI: Antonio Serrano Blanco. 2017-2020.
- Evaluación económica de una intervención compleja multiriesgo para promover conductas saludables en personas de 45-75 años atendidas en Atención Primaria: estudio Econ-EIRA3. Instituto de Salud Carlos III (ISCIII). FIS. PI15/00519. PI: Antonio Serrano Blanco. 2016-2020.
- Prevalencia e impacto de la no iniciación del tratamiento farmacológico en población pediátrica: estudio No Iniciadores-Pediátrico. Universidad Europea de Madrid S.L.U. Proyectos de Investigación en Salud. 2018/UEM51. PI: Maria Rubio Valera. 2018 - 2020.

### Selected publications

- Aznar Lou I, Carbonell Duacastella C, Rodríguez A, Mera I, Rubio-Valera M. Prevalence of Medication-Dietary Supplement Combined Use and Associated Factors. *Nutrients*. 2019; 11(10). FI: 4,171(Q1)
- Resurrección DM, Moreno-Peral P, Gómez-Herranz M, Rubio-Valera M, Pastor L, Caldas de Almeida JM, Motrico E. Factors associated with non-participation in and dropout from cardiac rehabilitation programmes: a systematic review of prospective cohort studies. *Eur J Cardiovasc Nurs*. 2019; 18(1): 38 - 47. FI: 2,497(Q1)
- Rubio-Valera M, Peñarrubia-María MT, Iglesias Gonzalez M, Knapp M, McCrone P, Roig M, et al. Cost-effectiveness of antidepressants versus active monitoring for mild-to-moderate major depressive disorder: a multisite non-randomized-controlled trial in primary care (INFAP study). *Eur J Health Econ*. 2019; 20(5): 703 - 713. FI: 2,169(Q1)
- Aznar-Lou I, Iglesias Gonzalez M, Rubio-Valera M, Peñarrubia-María MT, Mendive Arbeloa JM, Murrugarra-Centurión AG, et al. Diagnostic accuracy and treatment approach to depression in primary care: predictive factors. *Fam Pract*. 2019; 36(1): 3 - 11. FI: 1,986(Q2)

### Networks

- Consorcio de Investigación Biomédica en Red de Epidemiología y Salud Pública (CIBERESP) des de 2017.
- Red de Investigación en actividades Preventivas y de Promoción de la Salud (redIAPP). Des de 2007.

**PSYCHOLOGICAL RESEARCH GROUP  
ON FIBROMYALGIA AND CHRONIC PAIN  
(AGORA GROUP)**

Juan Vicente Luciano Devis, PhD

Dr. Albert Feliu and Dr Juan V. Luciano (PI of the AGORA group), together with Dr Xavier Borràs, directed Elvira Reche Camba's doctoral thesis, which was defended at the Autonomous University of Barcelona (UAB), obtaining the highest rating. Aside from this, one of the most important aspects to be pointed out here is that five papers of the EUDAIMON project (mindfulness in the approach to fibromyalgia) were published in 1st decile journals (PAIN, Journal of Clinical Medicine, and Brain Behavior & Immunity). The ULTREYA project ("The effects of the pilgrimage to Santiago on mindfulness, mental health and well-being") was one of the winners of the 2019 edition of the Barcelona Magic Line. Two researchers of our group (Dr Adrián Pérez and Dr Albert Feliu) undertook a three-month research stay at the London School of Economics with Prof. Martin Knapp, and another researcher (Laura Andrés) undertook a research stay at Chulalongkorn University (Thailand), also for three months, with Dr Michael Maes, a world expert on psychoneuroimmunology applied to mood disturbances.

**Members****STAFF RESEARCHER**

Antoni Rozadilla Sacanell, Lluís Torruella Barraquer

**POSTDOC - ASSOCIATE RESEARCHER**

Albert Feliu Soler

**PHD STUDENT**

Laura Andrés Rodríguez, Adrián Pérez Aranda

**ASSISTANT RESEARCHER**

Claudia Costa Iracheta

**COLLABORATOR RESEARCHER**

Jaume Aguado Carné, Xavier Borràs Hernández, Eugenia Cardeñosa Valera

**Selected projects**

- Efectos neurobiológicos de una intervención basada en mindfulness (MBSR) vs. Psico-educativa (FibroQol) en pacientes con fibromialgia (Proyecto EUDAIMON). Instituto de Salud Carlos III (ISCIII). FIS. PI15/00383. PI: Juan Vicente Luciano Devis. 2016-2019.
- Miguel Servet Tipo I (Proyecto Asociado) Cost-effectiveness of a Mindfulness-Based Stress Reduction (MBSR) treatment for fibromyalgia syndrome: A 12-month randomised, controlled trial. Instituto de Salud Carlos III (ISCIII). Miguel Servet tipo I. CP14/00087. PI: Juan Vicente Luciano Devis. 2015-2019.

**Selected publications**

- Pérez Aranda A, Feliu-Soler A, Montero-Marín J, García-Campayo J, Andrés Rodríguez L, Borràs X, et al. A randomized controlled efficacy trial of Mindfulness-Based Stress Reduction compared to an active control group and usual care for fibromyalgia: the EUDAIMON study. Pain. 2019; 160(11): 2508 - 2523. FI: 6,029(Q1)
- Pérez Aranda A, D'Amico F, Feliu-Soler A, McCracken LM,

- Peñarrubia-María MT, Andrés Rodríguez L, et al. Cost-Utility of Mindfulness-Based Stress Reduction for Fibromyalgia versus a Multicomponent Intervention and Usual Care: A 12-Month Randomized Controlled Trial (EUDAIMON Study). J Clin Med. 2019; 8(7). FI: 5,688(Q1)
- Andrés Rodríguez L, Borràs X, Feliu-Soler A, Pérez Aranda A, Rozadilla-Sacanell A, Montero-Marín J, et al. Immune-inflammatory pathways and clinical changes in fibromyalgia patients treated with Mindfulness-Based Stress Reduction (MBSR): A randomized, controlled clinical trial. Brain Behav Immun. 2019; 80: 109 - 119. FI: 6,170(Q1)
  - Pérez-Aranda A, Andrés-Rodríguez L, Feliu-Soler A, Núñez C, Stephan-Otto C, Pastor-Mira MA, et al. Clustering a large Spanish sample of patients with fibromyalgia using the FIQR: differences in clinical outcomes, economic costs, inflammatory markers and gray matter volumes. Pain. 2019; 160: 908-921. FI: 6.029 (Q1)

**11**PUBLICATIONS  
WITH IF**54%**FIRST QUARTILE  
PUBLICATIONS**27%**FIRST DECILE  
PUBLICATIONS**Networks**

- The following members of the AGORA group participate in the RedIAPP (ISCIII; RD16/0007/0012): Laura Andrés Rodríguez, Albert Feliu Soler, Juan Vicente Luciano Devis, Adrián Pérez Aranda

## CEREBRAL IMPRINTS OF GENDER, DEVELOPMENT AND PATHOLOGY

Christian Stephan-Otto Attolini, PhD

Our group has carried on with its research based on neuroimaging applied to various biomedical disciplines. For example, we reported, for the first time, that the brain of patients with a first psychotic episode shows a loss of grey matter associated with an increased blood concentration of neutrophils (an inflammation indicator). We began to explore new lines of research, one of which is addressed to analysing the early structural changes in the brain of persons at risk of developing Alzheimer's disease.

### Members

#### STAFF RESEARCHER

Christian Núñez Leánez,  
Gildas Yanin Guillaume Brebion

#### PHD STUDENT

Ma. Rosa Cambra,  
Federica Lombardini

### Selected projects

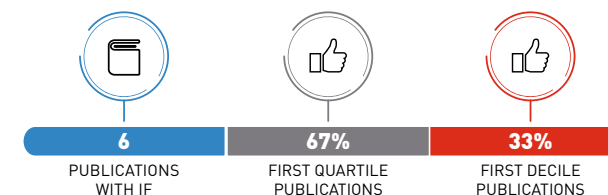
- SGR 2017\_Cerebral imprints of gender, development and pathology. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SGR 2017-2019. 2017 SGR 1297. PI: Christian Stephan-Otto Attolini. 2017 - 2020.
- Bases neurofuncionales de la anomalía del discurso interior en las alucinaciones verbales. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00047. PI: Gildas Yanin Guillaume Brebion. 2015 - 2019

### Selected publications

- Nuñez C, Stephan-Otto C, Usall J, Bioque M, Lobo A, González-Pinto A, et al. Neutrophil Count Is Associated With Reduced Gray Matter and Enlarged Ventricles in First-Episode Psychosis. Schizophr Bull. 2019; 45(4): 846 - 858. FI: 7,289(Q1)
- Núñez C, Arca G, Agut T, Stephan-Otto C, García-Alix A, Precise neonatal arterial ischemic stroke classification with a three-dimensional map of the arterial territories of the neonatal brain. Pediatric Research. 2019:1-6.
- Siddi S, Nuñez C, Senior C, Preti A, Cuevas-Esteban J, Ochoa S, et al. Depression, auditory-verbal hallucinations, and delusions in patients with schizophrenia: Different patterns of association with prefrontal gray and white matter volume. Psychiatry Res Neuroimaging. 2019; 283: 55 - 63. FI: 2,270(Q3)
- Brebion G, Stephan-Otto C, Ochoa S, Cuevas-Esteban J, Núñez Navarro A and Usall J. Clinical and non-clinical hallucinations are similarly associated with source memory errors in a visual memory task. Conscious Cogn. 2019; 76: 102823 - 102823. FI: 1,855(Q3)

### Networks

- Centro de Investigación Biomédica en Red de Salud Mental (CIBER-SAM)

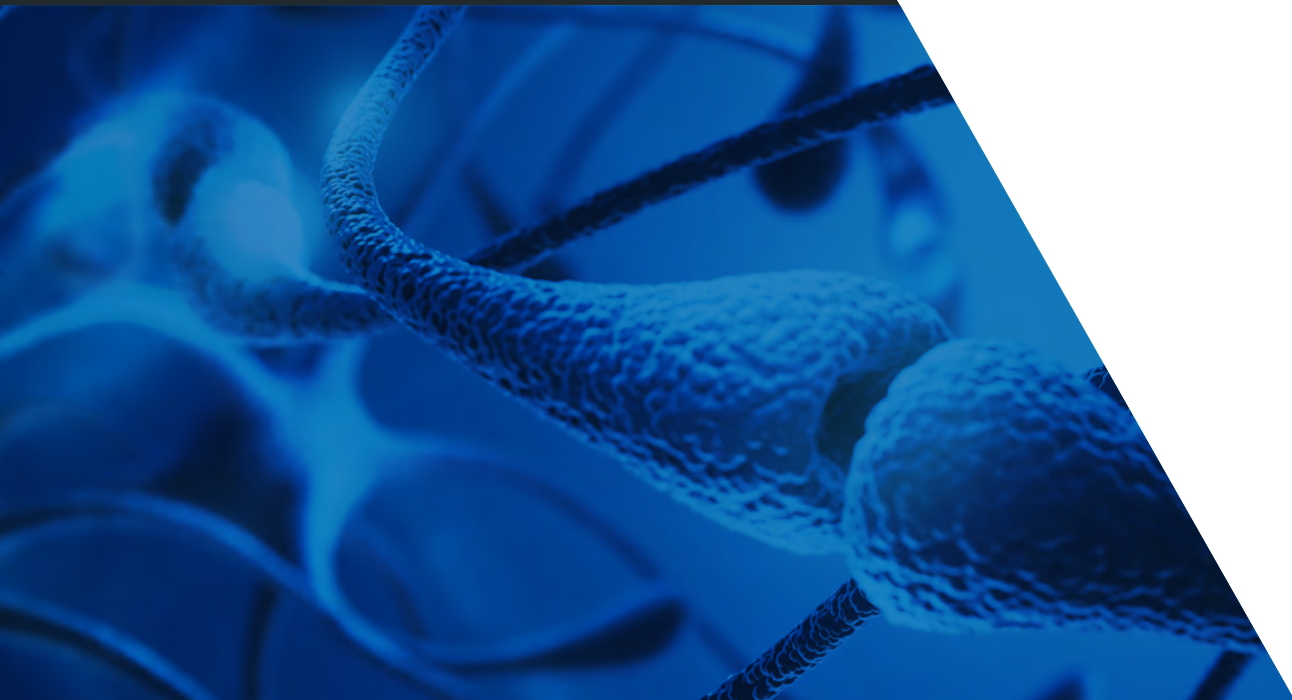






**SJD**

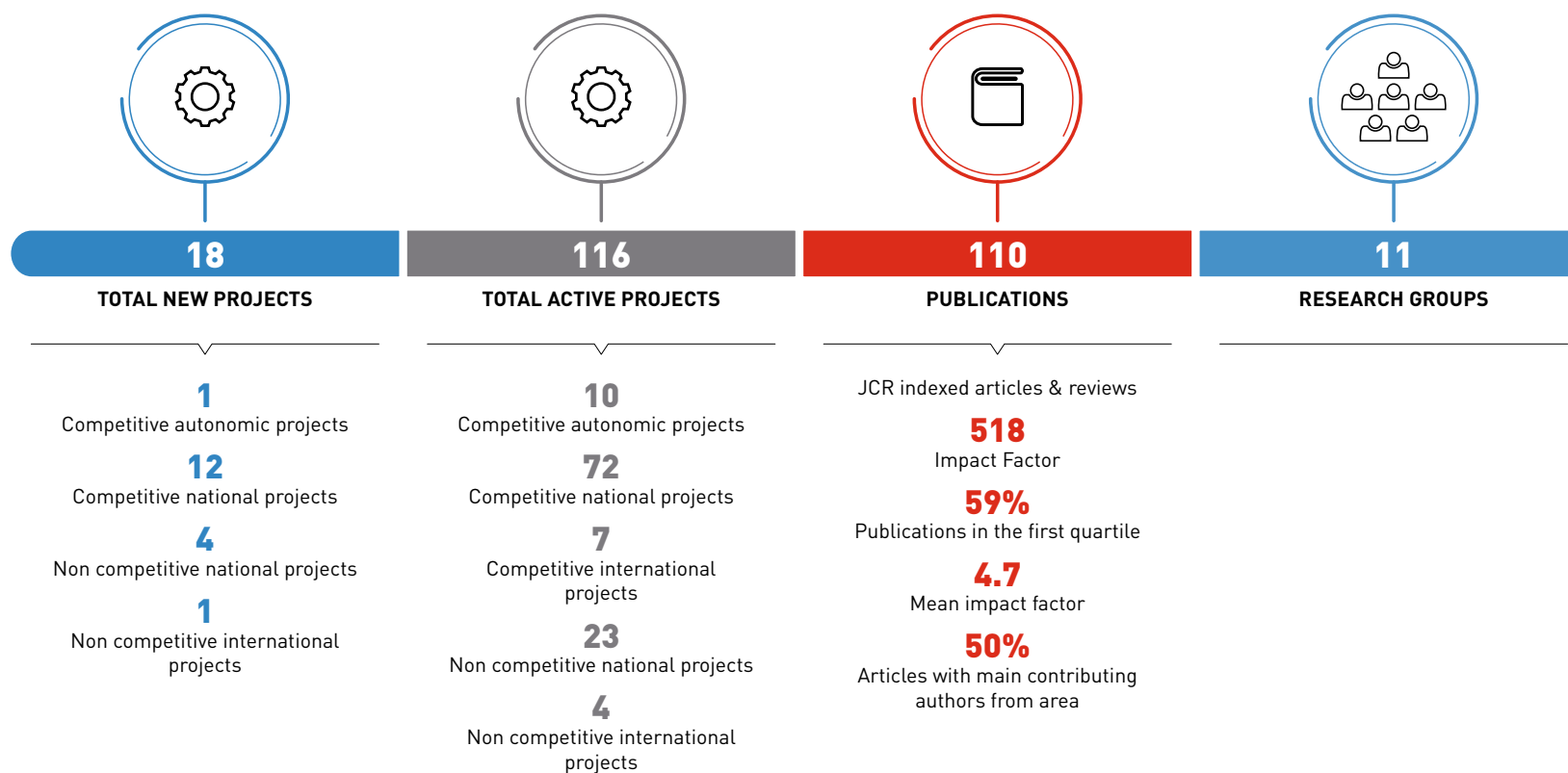
Sant Joan de Déu  
Institut de Recerca



**8**

## **ADAPTIVE RESPONSE, METABOLISM AND IMMUNOBIOLOGY**

Metabolic endocrinology  
Metabolic diseases of paediatric origin  
Molecular biology and gene regulation of adipose tissue and its pathologies  
Neonatology research group  
Pharmacological targets in inflammation and metabolic diseases  
Study of diseases due to immune dysfunction in pediatrics (GEMDIP)  
Infectious diseases and microbiome  
Gut microbiota  
Environment and complications in pregnancy  
Cardiovascular diseases in development  
Immune and respiratory dysfunction in the critical child



## METABOLIC ENDOCRINOLOGY

Lourdes Ibáñez Toda, MD, PhD

In 2019, we finished the analysis of the FIS 2015 data, which have allowed the demonstration of the effectiveness of a new pathophysiological treatment for polycystic ovary syndrome in adolescents. We also identified new prognostic markers of the treatment's effectiveness.

### Members

#### STAFF RESEARCHER

Paula Casano Sancho, Marta Díaz Silva, Silvia Xargay Torrent

#### ASSISTANT RESEARCHER

Cristina García Beltran, Esther Lizarraga Mollinedo, Rita Maria Martins de Sousa Maia Malpique, Cristina Plou Pérez

#### COLLABORATOR RESEARCHER

Judith Bassols Casadevall, Francis de Zegher, Abel López Bermejo

### Selected projects

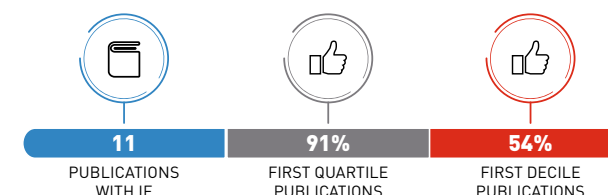
- Perfil longitudinal de exosomas circulantes y de su proteoma en niños nacidos con bajo peso y peso adecuado para la edad gestacional: Utilidad como biomarcador de síndrome metabólico. Instituto de Salud Carlos III (ISCIII). PI: PI18/00109. PI: Lourdes Ibáñez Toda. 2019-2021.
- Pioglitazona + espironolactona + metformina versus contracepción oral en adolescentes con hiperandrogenismo ovárico e hiperinsulinismo. Instituto de Salud Carlos III (ISCIII). FIS. PI15/01078. PI: Lourdes Ibáñez Toda. 2016-2019.
- Maternal diet, microbiome and infant feeding practice: roles in initializing the fetal gut microbiome and establishing the infant gut microbiome. Icahn School of Medicine at Mount Sinai. ICAHN – Microbiota. PI: Lourdes Ibáñez Toda. 2015-2019.

### Selected publications

- de Sousa-Malpique RM, Gallego-Escuredo JM, Sebastiani G, Villarroja J, López-Bermejo A, de Zegher F, et al. Brown adipose tissue in prepubertal children: associations with sex, birthweight, and metabolic profile. *Int J Obes (Lond)*. 2019; 43(2): 384 - 391. FI: 4,514(Q1)
- Ibáñez-Toda L and de Zegher F. Exploring the use of metformin in pregnant women with polycystic ovary syndrome: new evidence, new wisdom. *Lancet Diabetes Endocrinol*. 2019; 7(4): 242 - 243. FI: 24,540(Q1)
- Sebastiani G, García-Beltran C, Pie S, Guerra A, López-Bermejo A, Sanchez-de-Toledo J, et al. The sequence of prenatal growth restraint and postnatal catch-up growth: normal heart but thicker intima-media and more pre-peritoneal fat in late infancy. *Pediatr Obes*. 2019; 14(3): e12476. FI: 3,713(Q1)

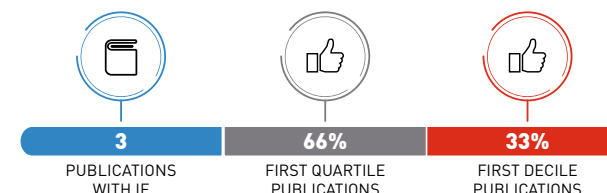
### Networks

- CIBERDEM, Instituto de Salud Carlos III, Madrid (2008), CAP DE GRUP



## METABOLIC DISEASES OF PAEDIATRIC ORIGIN

Josep Jiménez Chillarón, PhD  
Carles Lerin Martínez, PhD



### Members

#### STAFF RESEARCHER

Rubén Díaz Naderi, Marta Ramon Krauel, Montse Amat-Bou, Charlotte Juton

#### POSTDOC - ASSOCIATE RESEARCHER

Marcela Catalina Parra Vargas, Francesc Ribas Aulinas

#### COLLABORATOR RESEARCHER

Ivonne Palacios Marin, Marina Llobert

### Selected projects

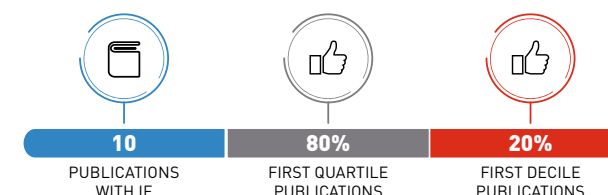
- BetAkk: Microbiota intestinal durante las primeras etapas de la vida y riesgo de obesidad y enfermedades metabólicas a largo plazo. Ministerio de Economía Y Competitividad (MINECO). Retos Investigación: Proyectos I+D+i. SAF2017-88005-R. PI: Carles Lerin Martínez. 2018-2020.
- Modifying maternal nutrition during lactation to improve offspring life-course health outcomes. Ajinomoto Co., Inc. Ajinomoto Innovation Alliance Program. Ajinomoto Innovation Alliance Program. PI: Carles Lerin Martínez. 2015-2019.
- TransEpiR: Herencia no-genómica de la obesidad mediante ARNs no codificantes de pequeño tamaño. Ministerio de Economía Y Competitividad (MINECO). Retos Investigación: Proyectos I+D+i. SAF2017-84542-R. PI: Josep Jiménez Chillarón. 2018-2020.
- Molecular determinants that link neonatal nutrition with late onset diabetes: epigenetic regulation of the circadian clock. European Foundation for the Study of Diabetes. Programme for Diabetes Research in Europe. EFSD/Lilly 2015. PI: Josep Jiménez Chillarón. 2016-2020.

### Selected publications

- Corripio R, Giménez-Palop O, Ibañez-Toda L, Ramon-Krauel M, Rivero MJ, Vela A. Controversias del tratamiento con hormona de crecimiento en pacientes con síndrome de Prader-Willi. Acta Pediatr Esp. 2019; 77(1-2): 6 - 11. FI: (Q)
- Isganaitis E, Venditti S, Matthews TJ, Lerin C, Demerath EW, Fields DA. Maternal obesity and the human milk metabolome: associations with infant body composition and postnatal weight gain. Am J Clin Nutr. 2019; 110(1): 111 - 120. FI: 6,568(Q1)
- Madsen JOB, Casteels K, Fieuws S, Kristensen K, Vanbrabant K, Ramon-Krauel M, et al. No Effect of an Automated Bolus Calculator in Pediatric Patients with Type 1 Diabetes on Multiple Daily Injections: The Expert Kids Study. Diabetes Technol Ther. 2019; 21(6): 322 - 328. FI: 4,488(Q1)
- Taxeràs SD, Galán M, Campderrós L, Piquer-Garcia, Pellitero, Martínez E, et al. Differential association between S100A4 levels and insulin resistance in prepubertal children and adult subjects with clinically severe obesity. Obes Sci Pract. 2019; 1 - 9. FI: (Q)

**MOLECULAR BIOLOGY AND  
GENE REGULATION OF ADIPOSE  
TISSUE AND ITS PATHOLOGIES**

Francesc Villarroya Gombau, PhD



In 2019, our team continued its basic and translational research on the control mechanisms of adiposity and metabolism, and their changes under pathological conditions such as obesity, diabetes, and lipodystrophies, and also in cardiovascular pathology. We identified new adipokines which are predominantly produced by brown adipose tissue, and new intracellular control factors of adipose plasticity in response to hormonal stimuli. In the translational field, we analysed the activity of the brown adipose tissue and the variations in the levels of new metabolic biomarkers in connection with pathophysiological determinants in the paediatric population.

**Members****STAFF RESEARCHER**

Marta Giralt Oms, Roser Iglesias Coll, Teresa Mampel Astals, Marion Peyrou, Anna Planavila López

**POSTDOC - ASSOCIATE RESEARCHER**

Montserrat Cairó Calzada, Rubén Cereijo Téllez, Aleix Gavalda Navarro, Joan Villarroya Terrade

**PHD STUDENT**

Laura Campderrós Traver, Moisès Castellà Giner, Alejandro Delgado Anglés, Gemma Ferrer Curriu, Samantha Morón Ros, Tania Quesada López, Celia Helena Rupérez Gonzalo

**TECHNICIAN**

Mercedes Morales Rueda, Gemma Moreno Besora, Albert Però García

**Selected projects**

- Cardioquinas y control de la función cardíaca: Nuevos actores y mecanismos, estrategias de terapia génica, y elucidación del secretoma cardíaco humano. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. RTI2018-096137-B-I00. PI: Anna Planavila López. 2019 - 2021.
- Activar el tejido adiposo pardo: una nueva estrategia para la prevención/tratamiento de la hepatocarcinogénesis potenciada por la lipotoxicidad metabólica. Asociación Española Contra el Cáncer-Catalunya (AECC). Ideas Semilla. Ayudas Fundación Científica AECC a proyectos de investigación en cáncer. PI: Francesc Villarroya Gombau. 2019 - 2021.

**Selected publications**

- Cairo M, Campderros L, Gavalda A, Cereijo R, Delgado-Anglés A, Quesada T, et al. Parkin controls brown adipose tissue plasticity in response to adaptive thermogenesis. *Embo Rep.* 2019; 20(5). FI: 8,383(Q1)
- Quesada T, Gavalda A, Moron S, Campderros L, Iglesias R, Giralt M and Villarroya F. GPR120 controls neonatal brown adipose tissue thermogenic induction. *Am J Physiol Endocrinol Metab.* 2019; 317(5): E742-E750. FI: 4,125(Q1)
- Villarroya J, Cereijo R, Gavalda-Navarro A, Peyrou M, Giralt M, Villarroya F. New insights into the secretory functions of brown adipose tissue. *J Endocrinol.* 2019; 243(2):R19-R27. FI: 4,381 (Q1)
- Malpique R, Gallego-Escuredo JM, Sebastiani G, Villarroya J, López-Bermejo A, de Zegher F, et al. Brown adipose tissue in prepubertal children: associations with sex, birthweight, and metabolic profile. *Int J Obes (Lond).* 2019; 43(2):384-391. FI: 4,515 (Q1)

**Networks**

- Red de Excelencia "Adipoplast". Ministerio de Economía y Competitividad. Desde 2017

## NEONATOLOGY RESEARCH GROUP

Martin Iriondo Sanz, MD, PhD

We analysed and published the positive results of introducing donor milk on the in-hospital health of infants born at or before 32 weeks of gestation who were admitted to our unit. We recruited very preterm (VP) mother-infant pairs and started experimental work aimed at analysing in greater depth the mechanisms by which mother's milk favourably affects a range of prematurity outcomes. We also carried out and published a detailed analysis of growth assessment methods applied to the NUTRIPREM cohort of VP infants and we continued to follow up their physical and neurological development to assess the impact of early nutritional support.

Likewise, we developed a mortality calculator for Spanish preterm infants based on data from the SEN-1,500 Network, and we defined three predictive models for neonatal death (prenatal, first 24 hours of life, and during admission).

A grant has been requested for a prospective validation project on the mortality calculator.

### Members

#### STAFF RESEARCHER

Carla Balcells Esponera, Marta Camprubi Camprubi, Beatriz Del Rey Hurtado De Mendoza, Isabel Iglesias Platas, Montserrat Izquierdo Renau, Africa Perterra Cortada

#### COLLABORATOR RESEARCHER

Marta Thió Lluch, Josep Figueras Aloy, Mattia Bosio, Albert Batista, Cristina Durán

### Selected projects

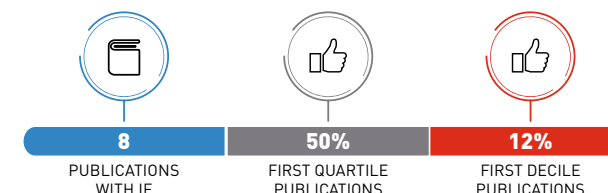
- Predicción de supervivencia sin morbilidad neonatal y sin discapacidad moderada-grave del neurodesarrollo en recién nacidos prematuros de la red española SEN1500. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI16/00748. PI: Martin Iriondo Sanz. 2017-2020.
- Leche humana: Nutrientes y factores bioactivos. Relación con el desarrollo de la microbiota intestinal, el crecimiento y la salud del recién nacido prematuro. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI17/00107. PI: Isabel Iglesias Platas. 2018-2020.
- Estrés oxidativo y mielinización en pacientes con cardiopatía congénita: nuevos horizontes terapéuticos. Asociación Española de Pediatría. Beca de Investigación AEP. Ayuda a la Investigación, clínica o básica en Pediatría. PI: Marta Camprubi Camprubi. 2019-2021.

### Selected publications

- Batista Muñoz A, Hadley S, Iriondo-Sanz M, Agut-Quijano T, Camprubi-Camprubi M. Role of beta-2-microglobulin as a biomarker in very preterm and extremely preterm infants with CNS inflammation. PLoS One. 2019; 14(5): e0216498. FI: 2,776(Q2)
- Cañizo Vázquez D, Salas García S, Izquierdo Renau M, Iglesias-Platas I. Availability of Donor Milk for Very Preterm Infants Decreased the Risk of Necrotizing Enterocolitis without Adversely Impacting Growth or Rates of Breastfeeding. Nutrients. 2019; 11(8). FI: 4,171(Q1)
- Izquierdo Renau M, Aldecoa-Bilbao V, Balcells Esponera C, Del Rey Hurtado de Mendoza B, Iriondo-Sanz M, Iglesias-Platas I. Applying Methods for Postnatal Growth Assessment in the Clinical Setting: Evaluation in a Longitudinal Cohort of Very Preterm Infants. Nutrients. 2019; 11(11). FI: 4,171(Q1)

### Networks

- Several members of the group belong to the SAMID network (RD16 / 0022/0002. IP: Óscar García Algar), since April 2018



## PHARMACOLOGICAL TARGETS IN INFLAMMATION AND METABOLIC DISEASES

Manuel Vázquez Carrera, PhD

In 2019 we demonstrated that the activation of HRI (heme-regulated eukaryotic translation initiation factor 2 $\alpha$  kinase) by a new compound which we developed may become a pharmacological target for the treatment of type 2 diabetes and other associated disorders such as non-alcoholic steatohepatitis. These new HRI activators increase FGF21 (fibroblast growth factor 21) levels and may be an alternative to the FGF21 analogues that are being studied for the treatment of non-alcoholic steatohepatitis, with the advantage of being suitable for oral administration, helping to avoid some of the adverse effects of such substances in this way.

### Members

#### STAFF RESEARCHER

F. Xavier Palomer Tarridas

#### POSTDOC - ASSOCIATE RESEARCHER

Emma Barroso Fernández, Marta Montori Grau, Lucía Peña Moreno, Javier Pizarro Delgado

#### PHD STUDENT

Davis Aguilar Recarte

### Selected projects

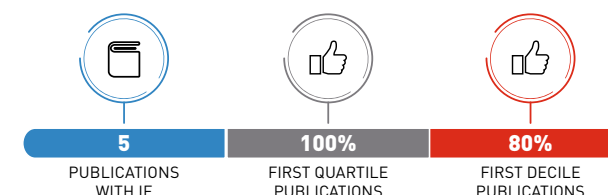
- New HRI activators for the treatment of type 2 Diabetes Mellitus. Fundació Caixa de Pensions "La Caixa". Caixa Impulse Programme 2018. CI18-00006. PI: Manuel Vazquez Carrera. 2018-2020.
- Activación de PPAR $\beta$ /d y HRI: explorando nuevos mecanismos compensatorios para el tratamiento de la inflamación, la resistencia a la insulina y la esteatohepatitis no alcohólica. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. RTI2018-093999-B-I00. PI: Manuel Vazquez Carrera. 2019 - 2021.

### Selected publications

- Busquets O, Eritja À, López BM, Ettcheto M, Manzone PR, Castro-Torres RD, et al. Role of brain c-Jun N-terminal kinase 2 in the control of the insulin receptor and its relationship with cognitive performance in a high-fat diet pre-clinical model. J NEUROCHEM. 2019; 149(2): 255 - 268. FI: 4,870(Q1)
- Zarei M, Pujol E, Quesada T, Villarroja F, Barroso E, Vázquez S, et al. Oral administration of a new HRI activator as a new strategy to improve high-fat-diet-induced glucose intolerance, hepatic steatosis, and hypertriglyceridaemia through FGF21. Br J Pharmacol. 2019; 176(13): 2292 - 2305. FI: 6,583(Q1)
- Rodríguez-Calvo R, Girona J, Rodríguez M, Samino S, Barroso E, de Gonzalo-Calvo D, et al. Fatty acid binding protein 4 (FABP4) as a potential biomarker reflecting myocardial lipid storage in type 2 diabetes. Metabolism. 2019; 96: 12 - 21. FI: 6,513(Q1)
- Márquez A, Aymerich J, Dei M, Rodríguez-Rodríguez R, Vazquez M, Pizarro J, et al. Reconfigurable multiplexed point of Care System for monitoring type 1 diabetes patients. Biosens Bioelectron. 2019; 136: 38 - 46. FI: 9,518(Q1)

### Networks

- 2007 CIBERDEM





**STUDY OF DISEASES DUE  
TO IMMUNE DYSFUNCTION  
IN PEDIATRICS (GEMDIP)**

Laia Alsina Manrique de Lara, MD, PhD  
Jordi Antón López, MD, PhD

Dr. Antón's group participated in clinical trials of key importance for the handling of autoinflammatory disorders, juvenile idiopathic arthritis, and systemic lupus erythematosus, as well as in the analysis of the safety profiles with biological drugs. It is also a national reference group in the study of such disorders as juvenile dermatomyositis, juvenile idiopathic arthritis associated uveitis, and Kawasaki disease.

The group led by Dr. Alsina centres its research on primary/ congenital and secondary immunodeficiencies, focusing on: 1) primary immunodeficiencies of the Toll-IL-1 R pathway (susceptibility to pneumococcus and herpes simplex encephalitis) and of the IL-12 /IFN- $\gamma$  pathway, which generate susceptibility to intracellular infections (mycobacteria and Leishmania species, among others); 2) primary immunodeficiencies with a predominant component of immune deregulation (inflammation, lymphoproliferative disorders, autoimmunity), with special interest in the study of the cells with a regulatory role (regulatory T and B lymphocytes) and in the use of sirolimus in their treatment; and 3) immunodeficiencies secondary to

the use of monoclonal antibodies, especially anti-TNF- $\alpha$  agents during pregnancy and B-lymphocyte depletion therapies (CART19 and rituximab, among others).

Our goal is to learn about the basis of immune dysfunction in the various pathology groups, and to identify biomarkers and new therapeutic targets.

**Members****STAFF RESEARCHER**

Rosa Bou i Torrent, Joan Calzada Hernández, Àngela Deyà Martínez, Estibaliz Iglesias Jimenez, Juan Manuel Mosquera Angarita, Mónica Piquer Gibert, Ana M<sup>a</sup> Plaza Martín, Ana Pilar García García, Judith Sánchez Manubens

**POSTDOC - ASSOCIATE RESEARCHER**

Anna Esteve Solé

**PHD STUDENT**

Yiyi Luo

**ASSISTANT RESEARCHER**

Violeta Bittermann, Clara Isabel Giménez Roca, Lucia Rodríguez Diez

**COLLABORATOR RESEARCHER**

Juan Ignacio Arostegui Gorospe, Thaïs Armangué, Manel Juan, Anna Mensa, Alexandru Vlaga

**16**PUBLICATIONS  
WITH IF**69%**FIRST QUARTILE  
PUBLICATIONS**31%**FIRST DECILE  
PUBLICATIONS**Selected projects**

- ImmunAID\_Immunome project consortium for AutoInflammatory Disorders. European Commission. H2020-SC1-PM-03-2017. 779295. PI: Jordi Antón López. 2018 - 2023.
- Estudio del efecto de la exposición intrauterina a fármacos anti-TNF- $\alpha$  en el sistema inmunitario de hijos de madres con enfermedades reumáticas inflamatorias. Instituto de Salud Carlos III (ISCIII). PI: PI18/00223. PI: Laia Alsina Manrique de Lara. 2019 - 2021.
- MDRTBNet\_Host-pathogen interactions in MultidrugResistant Tuberculosis Study of the Molecular Epidemiology, Host Immune Responses and GenomeBased Prediction and Early Identification of MDR-TB in High Tuberculosis Burden Settings. Instituto de Salud Carlos III (ISCIII). Acciones complementarias. AC16/00025. PI: Laia Alsina Manrique de Lara. 2017 - 2020.

**Selected publications**

- Armangué-Salvador T, Baucells BJ, Vlaga A, Petit-Pedrol M, Esteve-Sole A, Deyà-Martínez A, et al. Toll-like receptor 3 deficiency in autoimmune encephalitis post-herpes simplex encephalitis. *Neurol Neuroimmunol Neuroinflamm*. 2019; 6(6). FI: 7,353(Q1)
- Gattorno M, Hofer M, Federici S, Vanoni F, Bovis F, Aksentijevich I, et al. *Ann Rheum Dis*. 2019; 78(8): 1025 - 1032. FI: 14,299(Q1)
- Mensa-Vilaró A, García-Morato MB, de la Calle-Martín O, Franco-Jarava C, Martínez-Saavedra MT, González-Granado LI, et al. Unexpected relevant role of gene mosaicism in patients with primary immunodeficiency diseases. *J Allergy Clin Immunol*. 2019; 143(1): 359 - 368. FI: 14,110(Q1)
- Patterson K, Iglesias-Jimenez E, Nasrallah M, González V, Suñol M, Anton-Lopez J, et al. Anti-MOG encephalitis mimicking small vessel CNS vasculitis. *Neurol Neuroimmunol Neuroinflamm*. 2019; 6(2): e538. FI: 7,353(Q1)

**INFECTIOUS DISEASES  
AND MICROBIOME**

Carmen Muñoz Almagro, MD, PhD

**37**PUBLICATIONS  
WITH IF**43%**FIRST QUARTILE  
PUBLICATIONS**16%**FIRST DECILE  
PUBLICATIONS

In 2019 we demonstrated that the activation of HRI (heme-regulated eukaryotic translation initiation factor 2 $\alpha$  kinase) by a new compound which we developed may become a pharmacological target for the treatment of type 2 diabetes and other associated disorders such as non-alcoholic steatohepatitis. These new HRI activators increase FGF21 (fibroblast growth factor 21) levels and may be an alternative to the FGF21 analogues that are being studied for the treatment of non-alcoholic steatohepatitis, with the advantage of being suitable for oral administration, helping to avoid some of the adverse effects of such substances in this way.

**Members****STAFF RESEARCHER**

Pedro Brotons de los Reyes, Clàudia Fortuny Guasch, Vicky Fumadó, Juan José García García, Cristian Launes Montaña, Maria Goretti López Ramos, Antoni Noguera Julián

**POSTDOC - ASSOCIATE RESEARCHER**

Cristina Esteva Afonso, Mariona Fernández de Sevilla Estrach

**PHD STUDENT**

Sara Bobillo Perez, Ferran Bossacoma Busquets, Desiree Henares Bonilla, Míriam Triviño Rodríguez, Eneritz Velasco, Joan Vinent Genestar

**ASSISTANT RESEARCHER**

Susana Hernández Bou, Alba Redín Alonso, Silvia Simó Nebot

**TECHNICIAN**

Carles Cisneros Camps, Amaresh Pérez Argüello, Jesica Saucedo Verdes

**COLLABORATOR RESEARCHER**

Emilia Sánchez Ruiz

**Selected projects**

- Sol·licitud de subvencions en l'àmbit de la recerca i la innovació en salut per a la intensificació de professionals amb formació sanitària especialitzada: medicina, farmàcia, química, biologia, bioquímica, psicologia clínica, radiofísica hospitalària i infermeria, corresponents als anys 2019-2021. Departament de Salut - Generalitat de Catalunya. Intensificació facultatiu. SLT008/18/00193. PI: Antoni Noguera Julián. 2019 - 2021.
- Contratos Río Hortega. Instituto de Salud Carlos III (ISCIII). CM18/00054. PI: Carmen Muñoz Almagro. 2019 - 2020.
- Serotype and clonal distribution of streptococcus pneumoniae in Catalonia (Spain) before and after introduction of PCV13 vaccine in children. Pfizer, S.L.U. PI: Carmen Muñoz Almagro. 2019 - 2021.
- Microbiota infantil en els primers 2 anys de vida: relació amb el patró de creixement post-natal i el posterior desenvolupament de la síndrome de l'ovari poliquístic. Hospital Sant Joan de Déu - Esplugues HSJD. Convocatòria de Beques de Recerca IRSJD - Carmen de Torres 2018. BR201801. PI: Carmen Muñoz Almagro. 2019 - 2022.

- Rinovirus humano (HRV) en niños menores de 5 años, del resfriado común a la unidad de intensivos: rol de la microbiota. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. Convocatòria d'Intensificació de l'Activitat Investigadora de facultatiu. BI201901. PI: Cristian Launes Montaña. 2019 - 2020.
- Microbiota de riesgo o resiliente a la infección respiratoria aguda en niños y su utilidad en el diagnóstico diferencial entre neumonía/bronconeumonía bacteriana e infección viral respiratoria. SEIMC. Ayudas Movilidad. PI: Desiree Henares Bonilla. 2019.

**Selected publications**

- Brotons-de los Reyes P, Nogueras MM, Valls-Lafon A, Larrauri A, Dominguez A, Launes-Montana C, et al. Impact of Rapid On-demand Molecular Diagnosis of Pediatric Seasonal Influenza on Laboratory Workflow and Testing Costs: A Retrospective Study. *Pediatr Infect Dis J*. 2019; 38(6): 559 - 563. FI: 2,317(Q2)
- González-Sanz R, Casas-Alba D, Launes-Montana C, Munoz-Almagro C, Ruiz-García MM,

- Alonso M, et al. Molecular epidemiology of an enterovirus A71 outbreak associated with severe neurological disease, Spain, 2016. *Euro Surveill*. 2019; 24(7): 21 - 29. FI: 7,421(Q1)
- Hanquet G, Krizova P, Valentiner-Branth P, Ladhani SN, Nuorti JP, Lepoutre A, et al. Effect of childhood pneumococcal conjugate vaccination on invasive disease in older adults of 10 European countries: implications for adult vaccination. *Thorax*. 2019; 74(5): 473 - 482. FI: 10,307(Q1)
- Valero-Rello A, Henares-Bonilla D, Acosta L, Jane M, Jordán-García I, Godoy P and Munoz-Almagro C. Validation and Implementation of a Diagnostic Algorithm for DNA Detection of Bordetella pertussis, B. parapertussis, and B-holmesii in a Pediatric Referral Hospital in Barcelona, Spain. *J Clin Microbiol*. 2019; 57(1): UNSP e01231. FI: 4,959(Q1)

## GUT MICROBIOTA

Laura Baldomà Llavínés, PhD

Gut microbiota secreted vesicles (MVs) mediate communication with the host:

- MVs are used by gut microbiota to steadily prime the innate immune system, activating dendritic cells (DCs) in a strain-specific manner. MVs from probiotic and commensal *E. coli* strains drive complex Th responses that include a combination of Th2, Th22 and Th17. The main differences detected were in the ability to promote Th1 (protective immunity towards intracellular pathogens) and Treg (immunological tolerance) responses.
- MVs from probiotic and *E. coli* strains (phylogenetic group B) protect the barrier integrity against EPEC infection by activating compensatory mechanisms at various levels targeted to: (i) mRNA expression of the TJ proteins occludin and claudin-14, (ii) redistribution of ZO-1 and occludin from the cytoplasm to TJ structures, and (iii) maintenance of the F-actin cytoskeleton structure.

### Members

#### STAFF RESEARCHER

Josefa Badia Palacin, Rosa Giménez Claudio

#### POSTDOC - ASSOCIATE RESEARCHER

Rodrigo Vera

#### PHD STUDENT

Natalia Diaz-Garrido

### Selected projects

- Vesículas de membrana externa de microbiota y probióticos Gram-negativos como agentes modulares de la barrera intestinal y la respuesta inmunitaria. Ministerio de Economía y Competitividad. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Projectes I+D+I. AGL2016-79113-R. PI: Laura Baldomà Llavínés. 2016 - 2020.

### Selected publications

- Gonzalez-Pizarro R, Parrotta G, Vera R, Sánchez-López E, Galindo R, Kjeldsen F, et al. Ocular penetration of fluorometholone-loaded PEG-PLGA nanoparticles functionalized with cell-penetrating peptides. *Nanomedicine (Lond)*. 2019; 14(23): 3089 - 3104. FI: 4,717(Q1)
- Alvarez CS, Gimenez R, Canas MA, Vera R, Díaz-Garrido N, Badia J and Baldoma L. Extracellular vesicles and soluble factors secreted by *Escherichia coli* Nissle 1917 and ECOR63 protect against enteropathogenic *E. coli*-induced intestinal epithelial barrier dysfunction. *BMC Microbiol*. 2019; 19(1): 166 - 166. FI: 3,287(Q2)
- Diaz-Garrido N, Fabrega MJ, Vera R, Gimenez R, Badia J and Baldoma L. Membrane vesicles from the probiotic Nissle 1917 and gut resident *Escherichia coli* strains distinctly modulate human dendritic cells and subsequent T cell responses. *J Funct Foods*. 2019; 61: 103495. FI: 3,197(Q1)



3

PUBLICATIONS  
WITH IF



67%

FIRST QUARTILE  
PUBLICATIONS

## ENVIRONMENT AND COMPLICATIONS IN PREGNANCY

Maria Dolores Gómez Roig, MD, PhD

An intervention study to reduce alcohol consumption among pregnant women (EMOTIVE) was started in 2019. This project arose from the observation, made in one of our group's previous studies, that the serious problem of alcohol consumption during pregnancy is revealed by the presence of an alcohol metabolite in the hair. At the time of this writing, the recruitment phase of the study is being completed, with over 400 participants.

We also took part in the BiSC (Barcelona Life Study Cohort) project, which is aimed to provide an understanding of the effect of air pollution on placental function, foetal growth, and the cardiovascular and cerebral development of the foetus. At present our group is working on the recruitment phase and on the performance of echocardiograms and neurosonograms on 600 volunteers who live in the metropolitan area of Barcelona.

Likewise, our group is beginning a study which will involve the performance of a follow-up of the development of 100 babies (50 controls and 50 babies with growth restriction) from the 32nd week of pregnancy to the first year of life. The goal is to validate a cerebral response of the auditory

system called the "frequency-following response", which may be registered from birth and which provides information on the development and maturation of the auditory system, and on its relation to language development. For this purpose, we will perform an electroencephalogram on each baby on three occasions (0 months, 6 months and 12 months of life), a neurosonogram on each mother in the 32nd week of pregnancy, and a magnetic resonance on each baby at 0 and 6 months of life.

Lastly, our group will be participating in the ATHLETE (Advancing Tools for Human Early Lifecourse Exposome Research and Translation) study. The goal of this project is to develop advanced tools for the study of the exposome. A study will be made of a prospective exposure cohort, which will be used to quantify systematically, at both community and individual level, the effects of environmental risk factors on mental, cardiometabolic and respiratory health, as well as the health of the associated biological pathways, in the first two decades of life. In this way, acceptable and feasible exposome interventions may be implemented and the resulting evidence may be translated into prevention strategies and recommendations.

### Members

**POSTDOC - ASSOCIATE RESEARCHER**  
Eduarne Mazarico Gallego, Miriam Pérez Cruz

**PHD STUDENT**  
Marc Cahuana, Laura Almeida Toledano, Paz Ahumada, Mariona Serra, Diana Lip

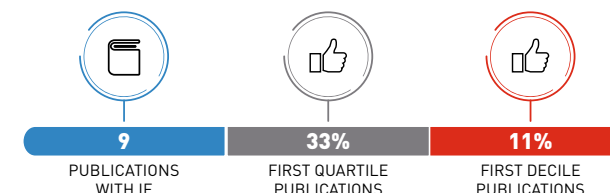
**COLLABORATOR RESEARCHER**  
Marta Muniesa, Giulia Casu  
**TECHNICIAN**  
Erica Muñoz Sanz

### Selected projects

- Traffic-related air pollution and birth weight: the roles of noise, placental function, green space, diet, physical activity, and socioeconomic status (FRONTIER). Health Effects Institute (HEI). RFA 17-1: Assessing adverse health effects of exposure to traffic-related air pollution, noise, and their interactions with socio-economic status. CR-83590201. PI: Maria Dolores Gómez Roig (PARTNER). 2018 - 2022.
- Efectividad de la entrevista motivacional repetida durante el embarazo en el abandono del consumo de alcohol. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI17/00105. PI: Maria Dolores Gómez Roig. 2018 - 2020.

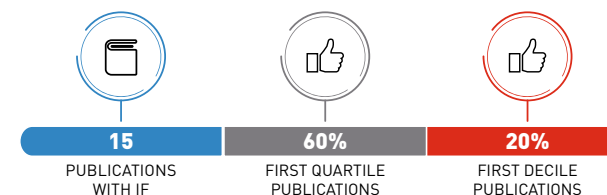
### Selected publications

- Ribas-Prats T, Almeida-Toledano L, Costa-Faidella J, Plana M, Corral MJ, Gómez-Roig MD and Escera C. The frequency-following response (FFR) to speech stimuli: A normative dataset in healthy newborns. *Hear Res.* 2019; 371: 28 - 39. FI: 2,952(Q1).
- Mazarico-Gallego E, Llorba E, Cabero L, Sanchez O, Valls-Lafon A, Martín-Ancel A, et al. Associations between neural injury markers of intrauterine growth-restricted infants and neurodevelopment at 2 years of age. *J Matern Fetal Neonatal Med.* 2019; 32(19): 3197 - 3203. FI: 1,569(Q3)
- Fernández Arias M, Mazarico-Gallego E, Gonzalez A, Muniesa M, Molinet C, Almeida-Toledano L and Gómez-Roig MD. Genetic risk assessment of thrombophilia in patients with adverse obstetric outcomes. *PLoS One.* 2019; 14(2): e0211114. FI: 2,776(Q2)



## CARDIOVASCULAR DISEASES IN DEVELOPMENT

Joan Sánchez de Toledo, MD, PhD  
Georgia Sarquella Brugada, MD, PhD



### Members

#### STAFF RESEARCHER

Esther Carro Fernandez, Sergio  
César Diaz, Isaac Moll Adrian

#### TECHNICIAN

Miriam López Abad

### Selected projects

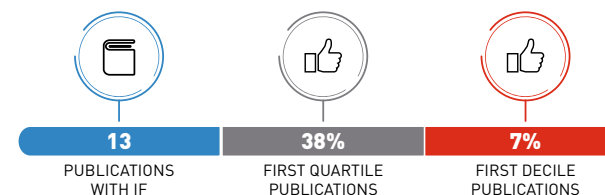
- Identification of predictors of cardiac arrhythmias and sudden death in pediatric patients affected with laminopathies. Medtronic Iberica S.A. PI: Georgia Sarquella Brugada. 2019 - 2020.
- ECG neonatal sistemático para la detección precoz de enfermedades cardíacas relacionadas con la muerte súbita del lactante. Sociedad Española de Cardiología. PI: Georgia Sarquella Brugada. 2019 - 2019.
- Utilidad de la neuromonitorización integrada como predictor del neurodesarrollo en pacientes sometidos a cirugía cardíaca en edad pediátrica: de la clínica al modelo animal pediátrico. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI17/02198. PI: Joan Sanchez de Toledo. 2018 - 2020.

### Selected publications

- Campuzano O, Fernandez-Falgueras A, Lemus X, Sarquella-Brugada G, César Diaz, Sergio, et al. Short QT Syndrome: A Comprehensive Genetic Interpretation and Clinical Translation of Rare Variants. J Clin Med. 2019; 8(7). FI: 5,688(Q1)
- Conte G, Belhassen B, Lambiase P, Ciconte G, de Asmundis C, Arbelo E, et al. Out-of-hospital cardiac arrest due to idiopathic ventricular fibrillation in patients with normal electrocardiograms: results from a multicentre long-term registry. Europace. 2019; 21(11): 1670 - 1677. FI: 5,047(Q1)
- Michowitz Y, Milman A, Andorin A, Sarquella-Brugada G, Gonzalez Corcia MC, Gourraud JB, et al. Characterization and Management of Arrhythmic Events in Young Patients With Brugada Syndrome. J Am Coll Cardiol. 2019; 73(14): 1756 - 1765. FI: 18,639(Q1)
- Milman A, Andorin A, Postema PG, Gourraud JB, Sacher F, Mabo P, et al. Ethnic differences in patients with Brugada syndrome and arrhythmic events: New insights from Survey on Arrhythmic Events in Brugada Syndrome. Heart Rhythm. 2019; 16(10): 1468 - 1474. FI: 5,225(Q1)

**IMMUNE AND RESPIRATORY  
DYSFUNCTION IN THE CRITICAL CHILD**

Francisco José Cambra Lasaosa, MD, PhD



During 2019 the researchers participated in 3 clinical trials of the industry as principal investigators and in 2 as collaborators. We have also written 7 book chapters, as well as the drafting of protocols of the Spanish Society of Pediatric Intensive Care (SECIP). For the scientific dissemination and dissemination, we participated in the Science Festival, organized by Barcelona City Council, at the end of October. We are also members of scientific societies: SECIP and ESPNIC.

This 2019 we have started our participation in the 2-year long-term international multicenter study Long Term Ventilation Kids in the context of the collaboration of the European Network ResPNIC and the PALISI network (USA-Canada). And also in the organization of the 4th International Conference on Pediatric and Neonatal Non-Invasive Ventilation.

**Members****STAFF RESEARCHER**

Mónica Balaguer Gargallo, Elisabeth Esteban Torne, Iolanda Jordán García, Martí Pons Òdena

**ASSISTANT RESEARCHER**

Carme Alexandre Galobardes, Sergio Benito Fernández, Sara Bobillo Pérez, Patricia Corniero Alonso, Aida Felipe Villalobos, Elena Fresán Ruiz, Mónica Girona Alarcón, Carmina Guitart Pardellans, Lluïsa Hernandez Platero, Nuria Millán García del Real, Susana Segura Matute, Anna Sole Ribalta, David Vila Pérez

**COLLABORATOR RESEARCHER**

Montserrat Esquerda Areste

**Selected projects**

- Contratos predoctorales de formación en investigación en salud. Beneficiari: Sara Bobillo. Instituto de Salud Carlos III (ISCIII). PFIS. FI17/00253. PI: Iolanda Jordán García. 2018 - 2022.
- Valoración del efecto neuroprotector de la dexmedetomidina en el modelo animal pediátrico de circulación extracorporea y parada circulatoria. Sociedad y Fundación Española de Cuidados Intensivos Pediátricos (SECIP). BECA FRANCISCO RUZA. PI: Sergio Benito Fernández. 2018 - 2020.
- Análisis de un algoritmo diagnóstico de neumonía grave en pacientes críticos pediátricos, mediante ecografía pulmonar y procalcitonina, como plan de mejora de la calidad asistencial. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI16/01040. PI: Iolanda Jordán García. 2017 - 2020.

**Selected publications**

- Bobillo-Perez S, Jordán-García I, Corniero-Alonso P, Balaguer-Gargallo M, Sole-Ribalta A, Esteban ME, et al. Prognostic value of biomarkers after cardiopulmonary bypass in pediatrics: The prospective PANCAP study. PLoS One. 2019; 14(6): e0215690. FI: 2,776(Q2)
- Flores-González JC, Valladares CM, Yun Castilla C, Mayordomo-Colunga J, Quesada SP, Martín Delgado CM, et al. Association of Fluid Overload With Clinical Outcomes in Critically Ill Children With Bronchiolitis: Bronchiolitis en la Unidad de Cuidados Intensivos Pediátricos (BRUCIP) Study. Pediatr Crit Care Med. 2019; 20(3): 130 - 136. FI: 2,798(Q1)
- Pons-Òdena M, Medina A, Modesto-Alapont V. Treachery on the Rocks in the Pediatric Intensive Care Unit. JAMA Pediatr. 2019; 173(7): 699 - 699. FI: 12,004(Q1)
- Valero-Rello A, Henares-Bonilla D, Acosta L, Jane M, Jordán-García I, Godoy P, Munoz-Almagro C. Validation and Implementation of a Diagnostic Algorithm for DNA Detection of Bordetella pertussis, B. parapertussis, and B-holmesii in a Pediatric Referral Hospital in

Barcelona, Spain. J Clin Microbiol. 2019; 57(1): UNSP e01231. FI: 4,959(Q1)

**Networks**

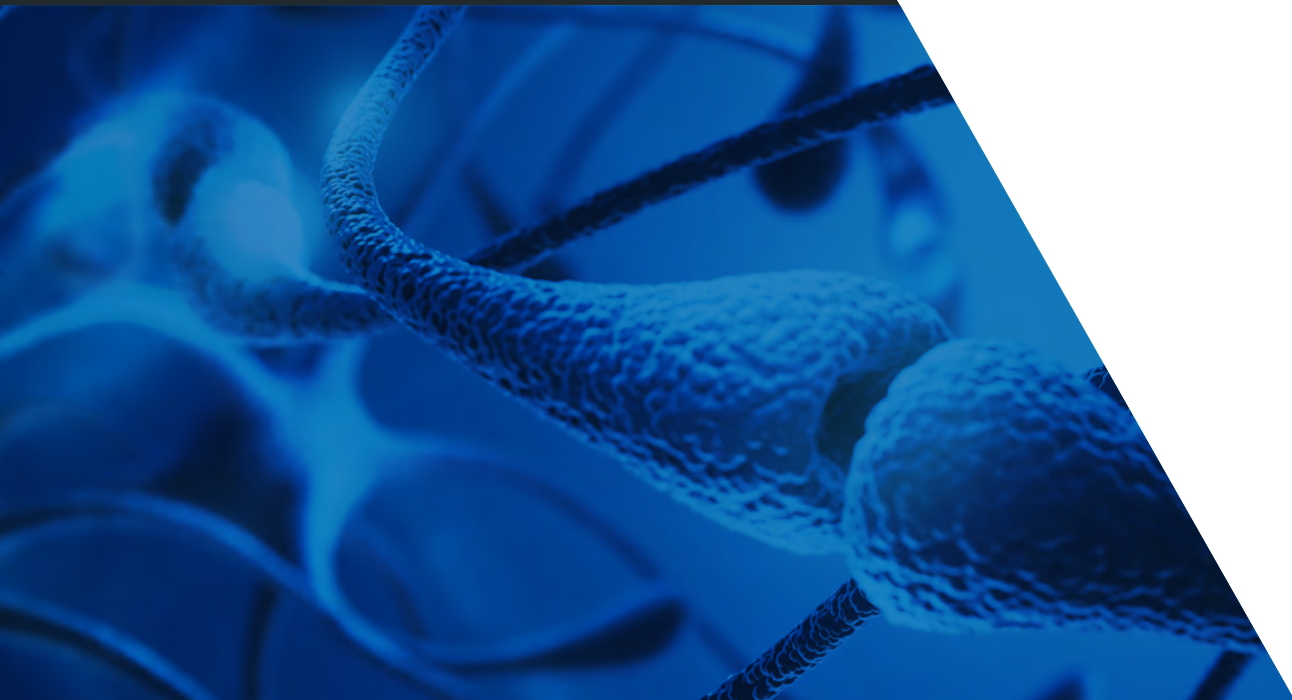
- 2017. Red de Investigación Translacional en Infectología Pediátrica (RITIP). Translational Research Network in Pediatric Infectious Diseases. IP: Pilar Calvo.





**SJD**

Sant Joan de Déu  
Institut de Recerca

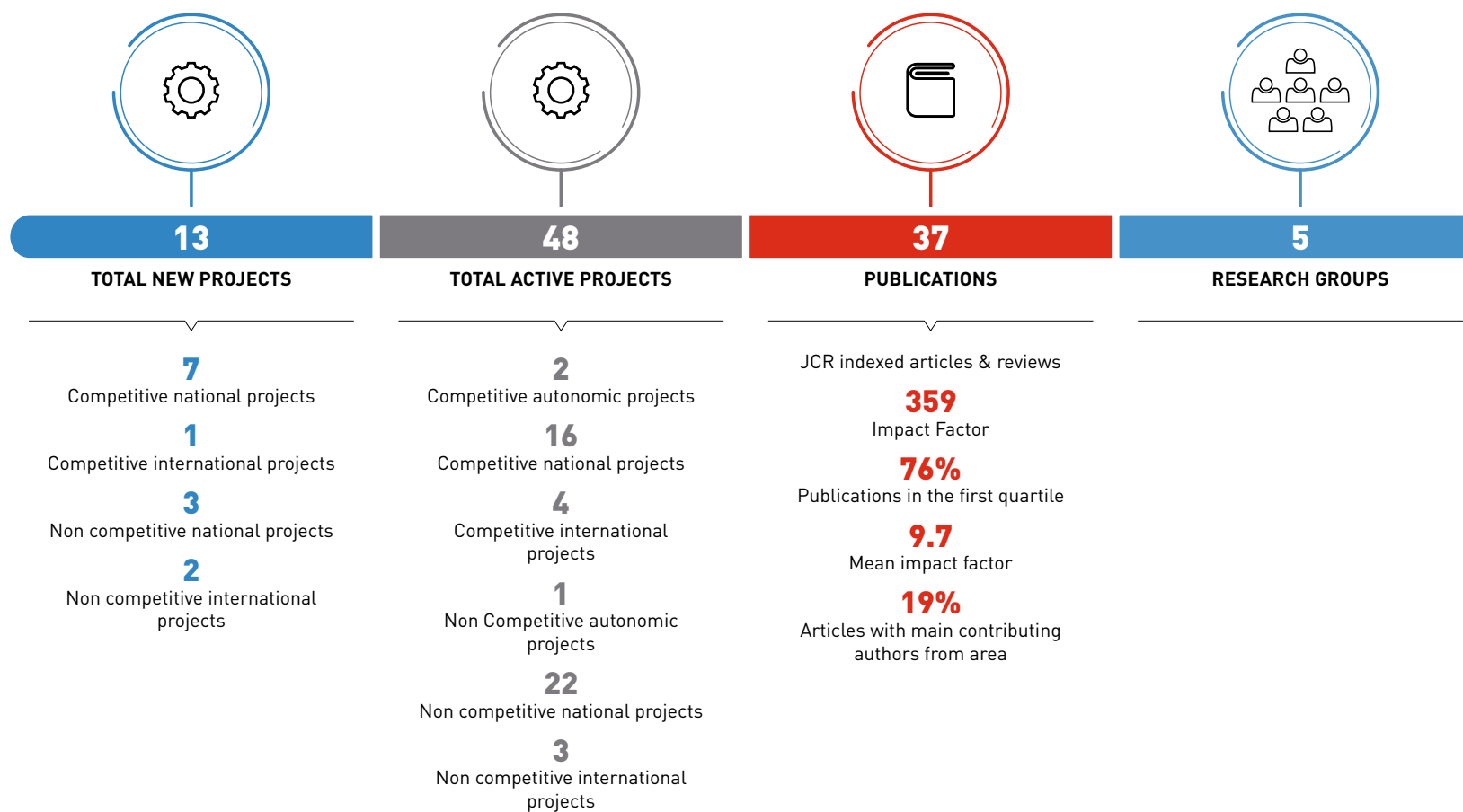


**9**

## **PAEDIATRIC CANCER**

Translational genomics  
Sarcomas and histiocytosis  
Therapy development in pediatric oncology  
Paediatric leukaemia and haematopoiesis  
Molecular pharmacology and experimental therapies





## TRANSLATIONAL GENOMICS

Cinzia Emilia Lavarino, PhD

The Translational Genomic Group has begun to develop a digital platform for the molecular classification of paediatric tumours. This digital platform will consist of a controlled-access web service that will allow the rapid and precise classification of paediatric tumours on the basis of an analysis of methylation values of epigenetic biomarkers' panels uploaded by users. The primary goal is to provide tools (classifiers and digital platform) allowing the majority of the centres to classify patients, to assign them to the respective clinical risk groups, and to quickly individualise candidate patients for targeted therapies.

### Members

#### STAFF RESEARCHER

Héctor Salvador Hernández

#### POSTDOC - ASSOCIATE RESEARCHER

Soledad Gomez Gonzalez, Marta Garcia López

#### PHD STUDENT

Laura Garcia Gerique, Alícia Garrido Garcia

#### TECHNICIAN

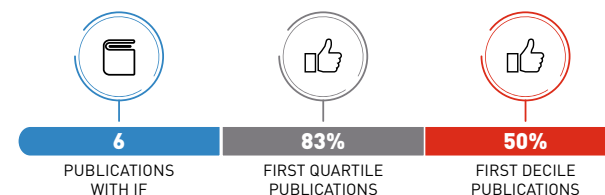
Isadora Lemos

### Selected projects

- Método para la clasificación en subgrupos moleculares de pacientes con Meduloblastoma. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI17/00353. PI: Cinzia Emilia Lavarino. 2018 - 2020.
- La epigenética en la patogénesis del neuroblastoma: nuevas perspectivas moleculares para el desarrollo de estrategias terapéuticas. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00038. PI: Cinzia Emilia Lavarino. 2015 - 2019.

### Selected publications

- Campbell K, Rossi F, Adams J, Pitsidianaki I, Barriga FM, García-Gerique L, et al. Collective cell migration and metastases induced by an epithelial-to-mesenchymal transition in *Drosophila* intestinal tumors. *Nat Commun.* 2019; 10(1): 2311 - 2311. FI: 11,878(Q1)
- D'Angelo F, Ceccarelli M, Tala Garofano L, Zhang J, Frattini V, Caruso FP, et al. The molecular landscape of glioma in patients with Neurofibromatosis 1. *Nat Med.* 2019; 25(1): 176 - 187. FI: 30,641(Q1)
- Garcia-López M, Rodriguez-Hernandez CJ, Mateo-Lozano S, Pérez-Jaume S, Gonçalves-Alves E, Lavarino C, et al. Parathyroid hormone-like hormone plays a dual role in neuroblastoma depending on PTH1R expression. *MOL ONCOL.* 2019; 13(9): 1959 - 1975. FI: 5,962(Q1)
- Pascual-Pastó G, Bazan-Peregrino M, Gene-Olaciregui N, Restrepo-Perdomo CA, Mato-Berciano A, Ottaviani D, et al. Therapeutic targeting of the RB1 pathway in retinoblastoma with the oncolytic adenovirus VCN-01. *SCI TRANSL MED.* 2019; 11(476). FI: 17,200(Q1)
- Perez-Somarriba M, Andión M, López-Pino MA, Lavarino C, Madero L and Lassaletta A. Old



drugs still work! Oral etoposide in a relapsed medulloblastoma. *Childs Nerv Syst.* 2019; 35(5): 865 - 869. FI: 1,327(Q3)

**SARCOMAS AND HISTIOCYTOSIS**

Jaume Mora Graupera, MD, PhD

As a result of the work of all the researchers, of the groups of families who give us their unconditional support, and of the translational nature of the group, all three of the projects for which applications were submitted to the year 2018 call of the telethon on cancer organised by the Catalan TV (La Marató) were awarded grants at the end of 2019. This is the first time that paediatric oncology projects have obtained financing at this event.

Lastly, the bid submitted by our centre won Barcelona's designation as the venue of the international congress of the International Society of Paediatric Oncology (SIOP) for 2022. This meeting has never been held before in Spain and our city's designation bears witness to the international recognition which our centre has achieved in the field of paediatric oncology.

**Members****STAFF RESEARCHER**

Veronica Celis Passini, Moira Garraus Oneca, Alicia Castañeda Heredia, Maite Gorostegui Obanos, Lucas Krauel Gimenez-Salinas, Maria Inmaculada Hernandez Muñoz

**POSTDOC - ASSOCIATE RESEARCHER**

Anastasia Herrero, Maria Sanchez Jimenez, Silvia Mateo Lozano, Carlos Javier Rodriguez Hernandez, Sara Sánchez Molina

**PHD STUDENT**

Elisabet Figuerola Bou

**ASSISTANT RESEARCHER**

Irene Cuervas Oliveras, Alejandro Manzanares Quintela, Jara Martin Serrano, Sara Pérez Jaume, Estela Prada Varela

**TECHNICIAN**

Soraya Morales Moreno, Óscar Muñoz Aznar

**Selected projects**

- Ipatasertib in Rhabdomyosarcoma. F. Hoffmann- La Roche Ltd. PI: Jaume Mora Graupera. 2019 - 2020.
- Modelo de terapia de precisión: tratamientos personalizados de rescate para cada paciente (en base a las anomalías genéticas del tumor y el test de fármacos en modelo animal). Fundación Inocente Inocente. Proyectos en cáncer infantil. PI: Jaume Mora Graupera. 2018 - 2019.

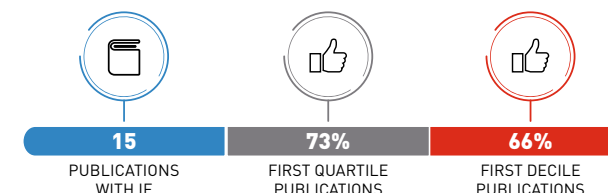
**Selected publications**

- Durham BH, Lopez Rodrigo E, Picarsic J, Abramson D, Rotemberg V, De Munck S, et al. Activating mutations in CSF1R and additional receptor tyrosine kinases in histiocytic neoplasms. Nat Med. 2019; 25(12): 1839 - 1842. FI: 30,641(Q1)
- Janin M, Ortiz-Barahona V, de Moura MC, Martínez-Cardús A, Llinàs-Arias P, Soler Vila M, et al. Epigenetic loss of RNA-methyltransferase NSUN5 in glioma targets ribosomes to drive a stress adaptive translational program. ACTA NEUROPATHOL. 2019; 138(6): 1053 - 1074. FI: 18,174(Q1)
- Pascual-Pastó G, Bazan-Peregrino M, Gene-Olaciregui N, Restrepo-Perdomo CA, Mato-Berciano A, Ottaviani D, et al. Therapeutic targeting of the RB1 pathway in retinoblastoma with the oncolytic adenovirus VCN-01. Sci Transl Med. 2019; 11(476). FI: 17,200(Q1)



## THERAPY DEVELOPMENT IN PEDIATRIC ONCOLOGY

Angel Montero Carcaboso, PhD



Our group's output for 2019 has included the publication entitled "Therapeutic targeting of the RB1 pathway in retinoblastoma with the oncolytic adenovirus VCN-01" (<http://stm.sciencemag.org/content/11/476/eaat9321>), published in the journal *Science Translational Medicine* on 23 January 2019. This study was commented on in the scientific journals *Science* and *Nature*.

### Members

#### STAFF RESEARCHER

Guillermo Chantada, Ofelia Cruz Martínez, Andrés Morales La Madrid

#### POSTDOC - ASSOCIATE RESEARCHER

Sonia Paco Mercader

#### PHD STUDENT

Helena Castillo Ecija, Maria Cuadrado Vilanova, Nagore Gene Olaciregui

#### ASSISTANT RESEARCHER

Leire Balaguer Lluna, Mercè Baulenas Farrés, Víctor Burgueño Sandoval, Francisco Javier Vicario Torre

#### TECHNICIAN

Ana Jiménez Cabaco, Mònica Vilà Ubach

### Selected projects

- Miguel Servet II. Instituto de Salud Carlos III (ISCIII). Miguel Servet II. MSII18/00009. PI: Angel Montero Carcaboso. 2019 - 2022.
- Angel Montero Carcaboso renovó su contrato con el programa Miguel Servet del Instituto de Salud Carlos III obteniendo la primera posición del ranking de su promoción Miguel Servet II de la convocatoria 2018.
- Cure4RB. Ministerio de Economía Y Competitividad (MINECO). Retos colaboración 2015. RTC-2015-4319-1. PI: Angel Montero Carcaboso. 2015 - 2019.
- Proyecto finalizado con éxito en 2019, ha permitido llevar a un ensayo clínico fase I la terapia avanzada con adenovirus VCN-01 para el tratamiento del retinoblastoma en pacientes resistentes a los tratamientos.
- Inmunoterapia del glioma difuso de protuberancia (DIPG) Instituto de Salud Carlos III (ISCIII). FIS. PI15/01161. PI: Angel Montero Carcaboso. 2016 - 2019.
- Proyecto finalizado con éxito en 2019, ha permitido llevar a un ensayo clínico fase I la terapia avanzada con vacunas de células dendríticas autólogas cargadas con lisados heterólogos de cultivos celulares de DIPG.

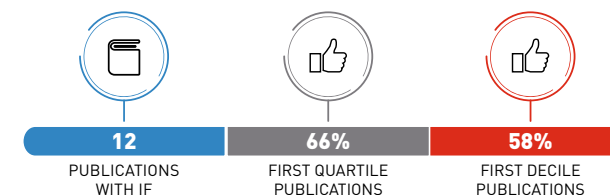
### Selected publications

- Pascual-Pastó G, Bazan-Peregrino M, Gene-Olaciregui N, Restrepo-Perdomo CA, Mato-Berciano A, Ottaviani D, et al. Therapeutic targeting of the RB1 pathway in retinoblastoma with the oncolytic adenovirus VCN-01. *SCI TRANSL MED*. 2019; 11(476). FI: 17,200(Q1). Selected as cover article and featured in *Science* and in *Nature*.
- Zaritski A, Castillo-Ecija H, Kumarasamy M, Peled E, Sverdlow Arzi R, Montero-Carcaboso A and Sosnik A. Selective Accumulation of Galactomannan Amphiphilic Nanomaterials in Pediatric Solid Tumor Xenografts Correlates with GLUT1 Gene Expression. *ACS Appl Mater Interfaces*. 2019; 11(42): 38483 - 38496. FI: 8,456(Q1)
- Carvalho D, Taylor KR, Gene-Olaciregui N, Molinari V, Clarke M, Mackay A, et al. ALK2 inhibitors display beneficial effects in preclinical models of ACVR1 mutant diffuse intrinsic pontine glioma. *Commun Biol*. 2019; 2(1): 156 - 156. FI: [Q]
- Munier FL, Beck-Popovic M, Chantada G, Cobrinik D, Kivelä TT, Lohmann D, et al. Conservative management of retinoblastoma: Challenging orthodoxy without compromising the state of metastatic grace. "Alive, with good

vision and no comorbidity". *Prog Retin Eye Res*. 2019; 73: 100764 - 100764. FI: 11,768(Q1)

**PAEDIATRIC LEUKAEMIA  
AND HAEMATOPOIESIS**

Mireia Camós Guijosa, MD, PhD



Dr. Mireia Camós coordinates the project “CLOSER. Childhood Leukemia: Overcoming distance between South America and Europe”, financed by the European Union within the Horizon 2020 programme (Grant Agreement 825749). CLOSER is a multidisciplinary collaborative international project that seeks to harmonise the handling of children with leukaemia and to reduce the inequalities in survival between Latin America and Europe. The project will be carried out between 2019 and 2023.

Within the framework of the implementation of NGS technology at our centre, in 2019 we completed a national collaborative project coordinated by Sant Joan de Déu Barcelona Children’s Hospital. The project was awarded an Unoentrecienmil Foundation grant and has dealt with the usefulness of various NGS approaches with directed panels in paediatric leukaemia. The results of the study have been presented in a paper which has been submitted to a scientific journal for its publication.

**Members****STAFF RESEARCHER**

Anna Alonso Saladrígues, Isabel Badell Serra, Ruben Buerrueco Moreno, Albert Català Temprano, Izaskun Elorza Alvarez, Susanna Gassiot Riu, Julia Marsal Ricoma, Montserrat Meseguer Meda, Susana Rives Solà, Anna Ruiz Llobet, Montserrat Torredadell Burriel, Maria Trabazo del Castillo

**POSTDOC - ASSOCIATE RESEARCHER**

Nerea Vega García

**PHD STUDENT**

Elena Esperanza Cebollada, Sandra Pont Martí

**ASSISTANT RESEARCHER**

Clara Vicente Garcés

**TECHNICIAN**

Marina Camino Estella Aguado, Soraya Morales Moreno, Cèlia Portillo Bog, Mercè Richarte Franqués, Lidia Ruiz Bocanegra, Warda Suleman

**Selected projects**

- CLOSER: Childhood Leukemia: Overcoming distance between South America and Europe Regions. European Commission. H2020-SC1-BHC-18. 825749. PI: Mireia Camós Guijosa. 2019 - 2023.
- Hacia la personalización de la profilaxis de la trombosis venosa profunda en pacientes pediátricos afectos de leucemia linfoblástica aguda: uso de test de generación de trombina. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI17/00356. PI: Ruben Berrueco Moreno. 2018 - 2020.
- Ingeniería fotónica para el diagnóstico, seguimiento y tratamiento de enfermedades oculares y sanguíneas. Ministerio de Economía, Industria y Competitividad. Proyectos de investigación. DPI2017-89414-R. PI Global: Vilaseca Ricart, Meritxell. PI-SJD: Gassiot Riu, Susanna. 2018 - 2020

**Selected publications**

- Laetsch TW, Myers GD, Baruchel A, Dietz AC, Pulsipher MA, Bittencourt H, et al. Patient-reported quality of life after tisagenlecleucel infusion in children and young adults with relapsed or refractory B-cell acute lymphoblastic leukaemia: a global, single-arm, phase 2 trial. *Lancet Oncol.* 2019; 20(12): 1710 - 1718. FI: 35,386(Q1)
- Martínez-Monseny T, Bolasell-Girgas M, Callejón L, Cuadras-Palleja D, Freniche-Rodríguez V, Itzep DC, et al. AZATAX: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2-CDG). *Ann Neurol.* 2019; 85(5): 740 - 751. FI: 9,496(Q1)
- Río P, Navarro S, Wang W, Sánchez-Domínguez R, Pujol RM, Segovia JC, et al. Successful engraftment of gene-corrected hematopoietic stem cells in non-conditioned patients with Fanconi anemia. *Nat Med.* 2019; 25(9): 1396 - 1401. FI: 30,641(Q1)
- Sánchez-Martínez D, Baroni ML, Gutierrez-Agüera F, Roca-Ho H, Blanch-Lombarte O, González-García S, et al. Fratricide-resistant CD1a-specific CAR T cells for the treatment of cortical T-cell acute

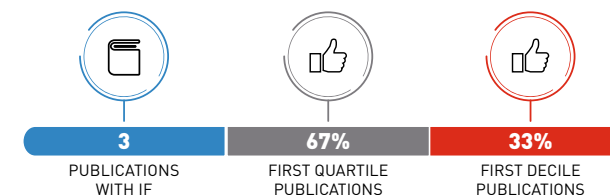
lymphoblastic leukemia. *Blood.* 2019; 133(21): 2291 - 2304. FI: 16,601(Q1)

**Networks**

- Innovative Therapies for Children with Cancer
- Grup clinic vinculat CIBER malalties rares (CIBERER), Instituto de Salud Carlos III. 2015-

**MOLECULAR PHARMACOLOGY  
AND EXPERIMENTAL THERAPIES**

Marçal Pastor-Anglada, PhD



In the course of this year, our group made two significant contributions in the field of biomedical research. From a more clinical standpoint, we identified the first case of dysfunction of a gene of the SLC28 family, encoding the pyrimidine nucleoside transporter hCNT1, which is responsible for an error of metabolism which, among others, is associated with pyrimidinuria. We also identified a new regulatory mechanism of the hENT1 and hENT2 proteins, which are responsible for the nucleoside recovery paths that are essential in both physiological and pathological situations of cell proliferation.

**Members****STAFF RESEARCHER**

Sandra Perez-Torras

**POSTDOC - ASSOCIATE RESEARCHER**

Nerea Urtasun Plans

**PHD STUDENT**

Clara Boces Pascuals, Aida Mata Ventosa, Eduard Mas Marín, Catalina Perelló Reus

**Selected projects**

- Proteínas transportadoras de nucleósidos (NT), metabolismo de nucleótidos y cáncer. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. RTI2018-094655-B-I00. PI: Marçal Pastor-Anglada. 2019 - 2021.
- Contractes de recerca/serveis. Modis Therapeutics, Inc. 310320. PI: Marçal Pastor-Anglada. 2019.
- Contracte de recerca/serveis. Cytes Biotechnologies, S.L. 310131. PI: Marçal Pastor-Anglada. 2018 - 2020.
- Farmacologia Molecular i Teràpies Experimentals (MPET). Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). Convocatòria d'ajuts per donar suport a les activitats dels grups de recerca (SGR). PI: Marçal Pastor-Anglada. 2018 - 2020.

**Selected publications**

- Perez-Torras S, Mata-Ventosa A, Drögemöller B, Tarailo-Graovac M, Meijer J, Meinsma R, et al. Deficiency of perforin and hCNT1, a novel inborn error of pyrimidine metabolism, associated with a rapidly developing lethal phenotype due to multi-organ failure. *Biochim Biophys Acta Mol Basis Dis.* 2019; 1865(6): 1182 - 1191. FI: 4,328(Q1)

**Networks**

- CIBER EHD 2006



**SJD**

Sant Joan de Déu  
Institut de Recerca

**10**

## **HEALTH TECHNOLOGIES AND INNOVATION**

Medical technologies: Bioinformatics and biomedical signals

Medical technologies: Biomaterials and tissue engineering

Medical technologies: Analysis for rehabilitation

Medical technologies: Instrumentation and eHealth

Medical technologies: Graphic computing: serious games and gamification

Medical technologies: Robotics and vision

Medical technologies: Medical imaging

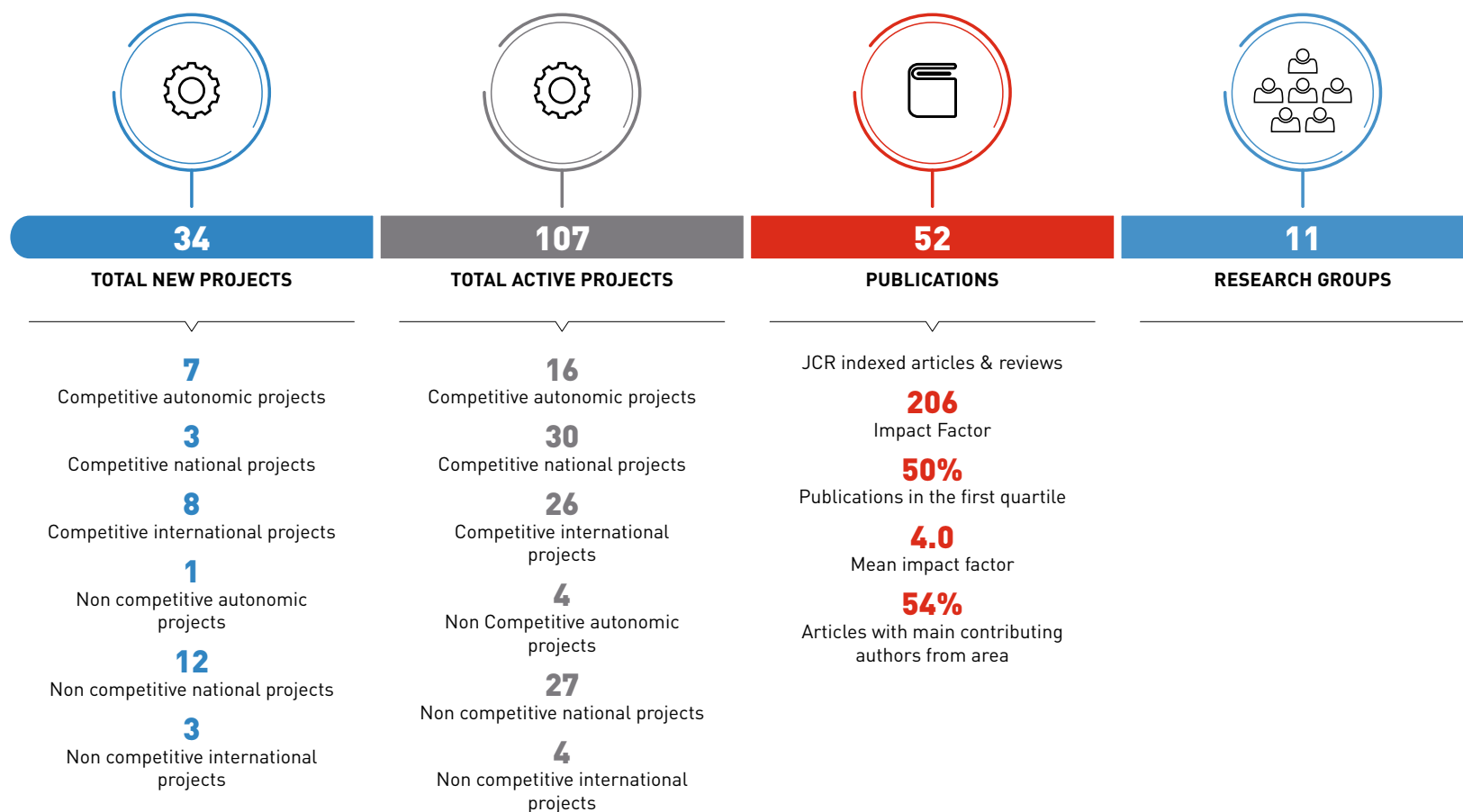
Medical technologies: Biomechanical

Patients engagement in research

Innovation in health technologies

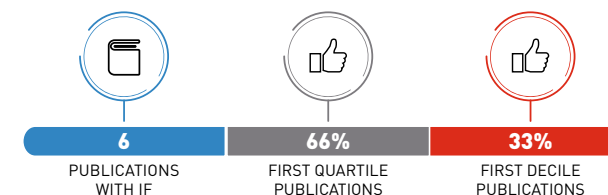
Diagnostic and therapeutic image





## MEDICAL TECHNOLOGIES: BIOINFORMATICS AND BIOMEDICAL SIGNALS

Alexandre Perera Lluna, PhD



### Members

#### STAFF RESEARCHER

Maria Barranco Altirriba, Pol Canal Noguera, Jesús Escrivá Muñoz, Jordi Fonollosa Magrinyà, Jon Garrido Aguirre, Giovana Elizabeth Gavidia Bovadilla, Pol Gil Figuerola, Pedro Gomis Román, Samir Kanaan Izquierdo, Oleksii Leonovych, Joshua Llano Viles, Ángela López del Río, Enrico Manzini, Imanol Morata Martínez, Joana Palés Huix, Flavio Palmieri, Pol Solà de los Santos, Alexander Vallmitjana Lees, Helena Vallverdú Cartié, Maria Montserrat Vallverdú Ferrer

#### PHD STUDENT

Xavier Marimon Serra, Sergi Picart Armada

#### TECHNICIAN

Maria Maqueda

### Selected projects

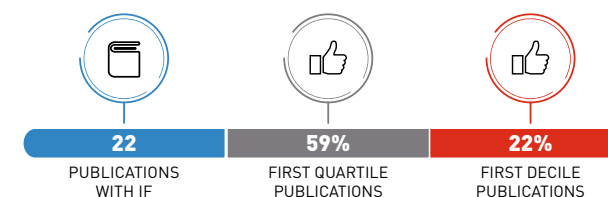
- Diacare: Desarrollo de un sistema de clusterización/segmentación de pacientes diabéticos tipo 2, según su adherencia al plan terapéutico. Pulso Ediciones SL. PI: Alexandre Perera Lluna. 2019 - 2020.
- EMPOC: Segmentación y predicción de episodios con pacientes de EMPOC. Pulso Ediciones SL. PI: Alexandre Perera Lluna. 2019 - 2020.
- AudioTec: Desarrollar un sistema de audiometría verbal con la capacidad de disponer de reconocimiento de voz automático. Devimatrix. PI: Alexandre Perera Lluna. 2019 - 2020.
- RCS: Impacte de la formació en l'Acadèmia de Ciències Mèdiques en termes d'indicadors de responsabilitat social. Acadèmia de les Ciències Mèdiques. PI: Alexandre Perera Lluna. 2019 - 2021.
- RCS: Anàlisi estadístic, bigdata i data mining. Universal Diagnostis SL. PI: Alexandre Perera Lluna. 2019 - 2019.
- Sistema basat en sensors de gas pel monitoratge i seguretat dels ancians a casa. AGAUR. PI: Jordi Fonollosa. 2019 - 2020.

### Selected publications

- Kanaan-Izquierdo S, Ziyatdinov A, Burgueño MA and Perera A. Multiview: a software package for multiview pattern recognition methods. *Bioinformatics*. 2019; 35(16): 2877 - 2879. FI: 4,531(Q1)
- Lopez-Del Rio A, Nonell-Canals A, Vidal D, Perera A. Evaluation of Cross-Validation Strategies in Sequence-Based Binding Prediction Using Deep Learning. *J Chem Inf Model*. 2019; 59(4): 1645 - 1657. FI: 3,966(Q1)
- Lupón J, Gavidia G, Ferrer Vidal-Barraquer E, de Antonio M, Perera A, López-Ayerbe J, et al. Heart Failure With Preserved Ejection Fraction Infrequently Evolves Toward a Reduced Phenotype in Long-Term Survivors. *Circ Heart Fail*. 2019; 12(3): e005652. FI: 6,526(Q1)
- Picart S, Barrett SJ, Willé DR, Perera A, Gutteridge A, Dessailly BH. Benchmarking network propagation methods for disease gene identification. *Plos Comput Biol*. 2019; 15(9): e1007276. FI: 3,955(Q1)

**MEDICAL TECHNOLOGIES:  
BIOMATERIALS AND  
TISSUE ENGINEERING**

Maria Pau Ginebra Molins, PhD



The Biomaterials, Biomechanics and Textile Engineering research group (BBT) obtained 6 European, national and regional projects with public financing in 2019. During the year, our group's members published a total of 27 papers in high-impact indexed journals, and they presented 21 papers at international conferences, as well as 18 guest lectures. During this period, a number of agreements were signed with different organisations, notably including the UPC-Klockner Chair and the UPC-AMES Chair.

**Members****STAFF RESEARCHER**

Cristina Canal Barnils, Montserrat Español Pons, José Manuel García Torres, Jose María Manero Planella, Carles Mas Moruno, Mònica Ortiz Hernández, Marta Peguerols Neyra, Miquel Punset Fusté, Alejandra Rodríguez Contreras, Daniel Rodríguez Rius, Elisa Rupérez de Gracia

**POSTDOC - ASSOCIATE RESEARCHER**

Judit Buxadera Palomero, Laura Córdoba Román, Jordi Guillem Martí, Osnat Hakimi, Cédric Labay, Irene Lodoso Torrecilla, Helena Martín Gómez, Francesco Tampieri, Juan Tornín Cavielles

**PHD STUDENT**

Mar Bonany Mariñosa, Victor Chausse Calbet, Laura del Mazo Barbara, Claudia García Mintegui, Inès Hamouda, Joanna Magdalena Konka, Miguel Mateu Sanz, Joaquim Minguella Díaz, Lluís Oliver Cervelló, Santiago Raymond Llorens, Xavier Solé Martí, Diego Torres Garrido, Èlia Vidal Girona

**TECHNICIAN**

Meritxell Molmeneu Trias, Meritxell Sánchez Hervàs

**PROJECT MANAGER**

Nelia Apariacio Bádenas

**Selected projects**

- Atmospheric Pressure pAsma meets biomaterials for bone Cancer HEaling (APACHE). European Commission. ERC Starting Grants. ERC-2016-STG 714793. PI: Cristina Canal Barnils. 2016 - 2021.
- Print4Life: "Novel bioprinting strategies for bone regeneration cancer therapies". Ministerio de Economía, Industria y Competitividad. MINECO. MAT2015-65601- R. PI: Maria Pau Ginebra Molins and Cristina Canal Barnils. 2016 - 2019.
- Duality: "Recubrimientos de biomateriales de doble acción para infecciones bacterianas e integración de tejidos". Agencia Estatal de Investigación, Ministerio de Ciencia, Innovación y Universidades. RTI2018-098075-B-C21. PI: José M. Manero and Marta Peguerols. 2019 - 2021.

**Selected publications**

- Barba A, Díez Escudero A, Español M, Bonany M, Sadowska JM, Guillem J, et al. Impact of Biomimicry in the Design of Osteoinductive Bone Substitutes: Nanoscale Matters. ACS Appl Mater Interfaces. 2019; 11(9): 8818 - 8830. FI: 8,456(Q1)
- Guillem J, Gelabert M, Heras-Parets A, Peguerols M, Ginebra MP, Manero JM. RGD Mutation of the Heparin Binding II Fragment of Fibronectin for Guiding Mesenchymal Stem Cell Behavior on Titanium Surfaces. ACS Appl Mater Interfaces. 2019; 11(4): 3666 - 3678. FI: 8,456(Q1)
- Sadowska JM, Guillem J, Ginebra MP. The Influence of Physicochemical Properties of Biomimetic Hydroxyapatite on the In Vitro Behavior of Endothelial Progenitor Cells and Their Interaction with Mesenchymal Stem Cells. Adv Healthc Mater; 2019. 8(2): e1801138. FI: 6,270(Q1)
- Sadowska JM, Wei F, Guo J, Guillem J, Lin Z, Ginebra MP, Xiao Y. The effect of biomimetic calcium deficient hydroxyapatite and sintered  $\beta$ -tricalcium phosphate on osteoimmune reaction and osteogenesis. Acta Biomater. 2019; 96: 605 - 618. FI: 6,638(Q1)

**Networks**

- Red Temática de BIOMATERIALES BACTERICIDAS PARA LA LUCHA CONTRA LA PERIIMPLANTITIS. 2019
- AMICI: Anti-Microbial Coating Innovations to prevent infectious diseases. 2015

## MEDICAL TECHNOLOGIES: ANALYSIS FOR REHABILITATION

Miquel Angel Mañana Villanueva, PhD



### Members

#### STAFF RESEARCHER

Joan Francesc Alonso Lopez, Sergio Romero Lafuente

#### POST-DOC ASSOCIATION RESEARCHER

Leidy Y. Serna Higueta, Carolina Falcone Migliorelli, Mislav Jordanic, Alejandro Bachiller Matarranz, Marjan Mansourian Gharakozlou

#### PHD STUDENT

Mehdi Shirzadi, Ana Tost Abadias

#### ASSISTANT RESEARCHER

Elena Condomines Casanoves, David Ivernon Garrido

#### TECHNICIAN

Isidre Solé Ramon

#### PROJECT MANAGER

Anna Nicolau Torra

#### COLLABORATOR RESEARCHER

Hamid R. Marateb

### Selected projects

- Multimodal analysis for assessment and rehabilitation of disabling neurological disorders. MINECO. 2017. DPI2017-83989-R. PI: Miguel Ángel Mañanas and Sergio Romero. 2018 - 2020.
- Cost effective self-management of urinary incontinence addressed to women across Europe (WOMEN-UP). Programa Europeu. H2020. H2020-PHC-2014. Ref. 643535. PI: Miguel Ángel Mañanas. 2015 - 2019.
- An online expert-based intelligent hybrid system to predict and assess motor rehabilitation outcomes in neurological disorders: NEURASSESS. Generalitat de Catalunya / European Commission. TECSPR18-1-0017. PI: Miguel Ángel Mañanas. 2019 - 2021.

### Selected publications

- Molina V, Bachiller A, de Luis R, Lubeiro A, Poza J, Hornero R, et al. Topography of activation deficits in schizophrenia during P300 task related to cognition and structural connectivity. Eur Arch Psychiatry Clin Neurosci. 2019; 269(4): 419 - 428. FI: 3,192(Q2)
- Rojas M, Alonso Lopez JF, Jordanic M, Mañanas MA and Chaler J. Analysis of Muscle Load-Sharing in Patients With Lateral Epicondylitis During Endurance Isokinetic Contractions Using Non-linear Prediction. Front Physiol. 2019; 10: 1185 - 1185. FI: 3,201(Q2)
- Salazar M, Hernández AM, Mañanas MA, Zuluaga A. Potential clinical application of surface electromyography as indicator of neuromuscular recovery during weaning tests after organophosphate poisoning. Revista Brasileira de Terapia Intensiva. 2017; 29(2): 253 - 258. FI: (Q)
- Migliorelli C, Bachiller A, Andrade AG, Alonso Lopez JF, Mañanas MA, Borja C, Giménez S, Antonijoan RM, Varga AW, Osorio RS, Romero S. Alterations in EEG connectivity in healthy young adults provide an indicator of sleep depth. Sleep. 2019; 42(6). FI: 4,5

### Networks

- 2008 Centros de Investigación Biomédica en Red en Bioingeniería, Biomateriales y Nanomedicina (CIBER-BBN)

## MEDICAL TECHNOLOGIES: INSTRUMENTATION AND eHEALTH

Xavier Rosell Ferrer, PhD



### Members

#### STAFF RESEARCHER

Ramon Bragos Barcia, Pere Riu Costa

### Selected projects

- Integrating CARTo and multi-frequency impedance system. BiosenseWebster. PI: Xavier Rosell Ferrer. 2019 - 2019.
- Electronic biopsy of in-vivo lung tissue based on electrical impedance spectroscopy. Agencia Estatal de Investigación. RTI2018-098116-B-C21. PI: Ramon Bragos

Bardia. 2019 - 2021. Grups de recerca consolidats. AGAUR. 2017 SGR 419. PI: Xavier Rosell. 2017 - 2020.

- A novel myocardial impedance mapping system for ablation of post-infarction ventricular arrhythmias in humans. Fundació La Marató de TV3. MARATO 20150830. PI: Xavier Rosell. 2016 - 2020.

### Selected publications

- Ontanilla-Clavijo G, Ampuero J, Borreguero S, Rosell Ferrer X, Romero-Gómez M. Usefulness of bioelectrical impedance analysis for monitoring patients with refractory ascites. Rev Esp Enferm Dig. 2019;111(3): 223 - 227. FI: 1,858(Q4)

## MEDICAL TECHNOLOGIES: GRAPHIC COMPUTING: SERIOUS GAMES AND GAMIFICATION

Daniela Tost Pardell, PhD

### Members

#### STAFF RESEARCHER

Dolors Ayala Vallespi, Jose Luis Eguia Gómez, Robert Joan Arinyó, Núria Pla García, Lluís Solano Albages, Marc Vigo Anglada

#### PHD STUDENT

Núria Bonet Codina, Ariel von Barnekow

### Selected projects

- BEST: Bienestar y Salud mental en el puesto de trabajo. IRSJD. 2018. C11230. PI: Dani Tost. 2019 - 2020.

**MEDICAL TECHNOLOGIES:  
ROBOTICS AND VISION**

Alicia Casals Gelpi, PhD

A new Marie Curie European Project on endoluminal robotic surgery has been started. This project is along the line of the projects underway on robotic surgery applicable in foetal and laparoscopic procedures. Specifically, this research project focusses on the processing of images and of sensory information for the intelligent control of robots. One of the assistance robots in progress, the "roller" (an intelligent walker) has been certified and transferred to an international enterprise.

**Members****STAFF RESEARCHER**

Josep Amat Girbau, Joan Aranda López, Josep Fernández Ruzafa, Manel Frigola Bourlon, Albert Hernansanz Prats, Enric Xavier Martín Rull, Antonio Benito Martínez Velasco, Luis Miguel Muñoz Morgado

**PHD STUDENT**

Marina Farahi, Maria Farahi, Martina Finocchiaro, Marien Cristina Narvaez Morata, Narcís Sayols Baixeras

**ASSISTANT RESEARCHER**

Tomás Pieras Morell, Laureano Tinoco, Alejandro Veiga

**Selected projects**

- Autonomous Intraluminal Surgery (ATLAS). H2020. H2020-ITN 2018 REF: 813782. FI: Alicia Casals. 2019 - 2023.
- Smart Autonomous RoboticAssistant Surgeon (SARAS). H2020. H2020-ICT-2017 Ref 779813. FI: Alicia Casals. 2018 - 2020.
- Agreement Regarding Purchase of R&D. FI: Antonio B. Martínez. 2018 - 2019.
- Caminadors elèctrics. TOPRO Industry. FI: Antonio B. Martínez. 2018 - 2020.

**Selected publications**

- Marbán A, Srinivasan V, Samek W, Fernandez J, Casals-Gelpi A. A recurrent convolutional neural network approach for sensorless force estimation in robotic surgery. Biomedical Signal Processing and Control. 2019; 50: 134 - 150. FI: 2,943(Q2)
- Sayols N, Hernansanz A, Parra J, Eixarch E, Gratacós E, Amat J, Casals-Gelpi A. Vision Based Robot Assistance in TTTS Fetal Surgery. Conf Proc IEEE Eng Med Biol Soc. 2019; 2019: 5855 - 5861. FI: (Q)

**Networks**

- 2019 - DIH-Hero Digital Innovation Hub
- 2017 - Cluster de Salut Mental



1

PUBLICATIONS  
WITH IF**MEDICAL TECHNOLOGIES:  
MEDICAL IMAGING**

Mercè Ginjaume Egidio, PhD

**Members****STAFF RESEARCHER**

Maria Amor Duch Guillen

**PHD STUDENT**

Marina Farahi, Maria Farahi, Martina Finocchiaro, Marien Cristina Narvaez Morata, Narcís Sayols Baixeras

**ASSISTANT RESEARCHER**

Tomás Pieras Morell, Laureano Tinoco, Alejandro Veiga

**Selected projects**

- Calibratge d'equips de mesura de radiacions ionitzants - Trescal S.L. TRESICAL, S.A. C-11045 (UPC). PI: Mercè Ginjaume. 2019 - 2019.
- Realización de una intercomparación destinada a valorar el grado de fiabilidad de los servicios de dosimetría personal externa autorizados en

España. Consejo de Seguridad Nacional. PI: Mercè Ginjaume. 2019 - 2020.

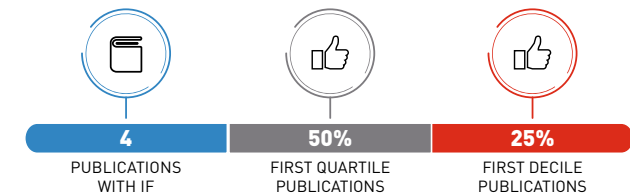
- Estimaciones de dosis ocupacionales en cristalino en instalaciones sanitarias y de investigación (EDOCI). Propuestas de vigilancia radiológica individual. Consejo de Seguridad Nacional. PI: Mercè Ginjaume. 2019 - 2022.
- Irradiació de dosímetres

Vandellós. Asociación Nuclear Ascó Vandellós. PI: Mercè Ginjaume. 2019 - 2019.

- Calibración equipos. ACPRO S.L. PI: Mercè Ginjaume. 2019 - 2019.
- Transversal Programme in Medical Image PostProcess, Training Professionals: 3D Printing. European Insitute of Innovation and Technology (EIT). PI: Mercè Ginjaume. 2019 - 2019.

## MEDICAL TECHNOLOGIES: BIOMECHANICAL

Josep Maria Font Llagunes, PhD



In 2019, the Biomechanical Engineering Lab (BIOMECH) research group of CREB-UPC has received three competitive projects (EIT Health Innovation, AGAUR Product, and MICINN National Project). Likewise, it has received financing from the Duchenne Parent Project to carry out projects on assistance robotics for paediatric patients with Duchenne. Seven papers were published in the journal JCR, and the spin-off of the ABLE Human Motion group won various prizes, including the Best Young Business Initiative Prize 2019 awarded by the management association CECOT.

### Members

#### STAFF RESEARCHER

Ana Barjau Condomines, Rosa Pàmies Vilà, Daniel Clos Costa

#### POSTDOC - ASSOCIATE RESEARCHER

Joan Lobo Prat

#### PHD STUDENT

Miriam Febrer Nafria, Mireia Claramunt Molet, Antonio Rodríguez Fernández, Jesús de Miguel Fernández

#### ASSISTANT RESEARCHER

Víctor Antonio Carmona Ortiz

### Selected projects

- Personalized robotic exoskeleton for gait rehabilitation. Fundació BBVA. PI: Josep Maria Font. 2018 - 2020.
- ABLE: exoesquelet robòtic que permet tornar a caminar de forma natural i intuïtiva després de patir una lesió medul·lar. AGAUR. 2018-PROD-00059. PI: Josep Maria Font. 2019 - 2020.
- Design of personalized robotic and neuroprosthetic wearable systems for walking assistance using a predictive simulation framework (TAILOR). Spanish Ministry of Science, Innovation and Universities (MICINN). RTI2018-097290-B-C33. PI: Josep Maria Font. 2019 - 2021.

### Selected publications

- Gomez-Perez C, Font Llagunes JM, Martori JC and Vidal Samsó J. Gait parameters in children with bilateral spastic cerebral palsy: a systematic review of randomized controlled trials. Dev Med Child Neurol. 2019; 61(7): 770 - 782. FI: 3,532(Q1)
- Pallarès-López R, Costa-Alvim F, Febrer Nafria M, Luporini L, Font Llagunes JM. Assessment of residual reduction procedures for high-speed tasks. Gait Posture. 2019; 73: 116 - 119. FI: 2,414(Q2)
- Stuhlenmiller F, Clos D, Rinderknecht S, Beckerle P, Font Llagunes JM. Impact of friction and gait parameters on the optimization of series elastic actuators for gait assistance. Mech Mach Theory. 2019; 133: 737 - 749. FI: 3,535(Q1)
- Tassani S, Font Llagunes JM, Gonzalez Ballester MA, Noailly J. Muscular tension significantly affects stability in standing posture. Gait Posture. 2019; 68: 220 - 226. FI: 2,414(Q2)

### Networks

- 2018-2020 - Red Española de Investigación en Biomecánica (DPI2017-90572-REDT, coordinator)
- 2020 - Xarxa R+D+I en Tecnologies de la Salut (AGAUR)



## PATIENTS ENGAGEMENT IN RESEARCH

Joana Claverol Torres  
Begonya Nafria Escalera

In 2019, the Patients Engagement in Research group focussed its activity on two strategic areas: the fostering of the participation of paediatric patients and their parents in the design of clinical trials, and the performance of various collaborative research projects with patients/carers affected by a rare disease. The group's mission over the course of 2019 was to provide a suitable context and an optimum methodology for patients' contribution to research, with the aim of ensuring that their collaboration will have a significant impact. The analysis of the pertinent results and the dissemination of the respective initiatives formed part of each project's activities.

### Members

#### STAFF RESEARCHER

Sara Hernández Ortega, Estibaliz Iglesias Jimenez, Nuria Noel Palacio, Francesco Schiraldi

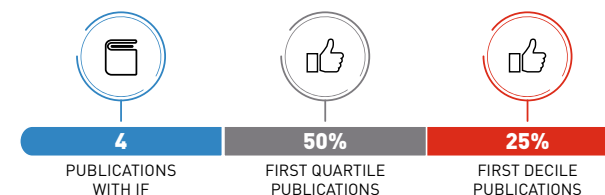
### Selected projects

- EJP RD: European Joint Programme on Rare Diseases. European Commission. H2020-SC1-BHC-04. 825575. PI: Joana Claverol Torres. 2019 - 2023.
- PARADIGM\_Patients Active in Research and Dialogues for an Improved Generation of Medicines: Advancing meaningful patient engagement in the life cycle of medicines for better health outcomes. European Commission. H2020-JTI-IMI2-2016-10-two-stage-Topic10-2nd stage. PI: Begoña Nafria Escalera. 2018 - 2020.
- SHARE4RARE: Social media platform dedicated to rare diseases, using collective intelligence for the generation of awareness and advanced knowledge on this large group of diseases. European Commission. H2020-ICT-11-2017. PI: Begoña Nafria Escalera. 2018 - 2020.
- c4c\_conect4children (Collaborative Network for European Clinical Trials For Children). European Commission. H2020-JTI-IMI2-2016-10-two-stage-Topic 4- 2nd stage. 777389. PI: Joana Claverol Torres. 2018 - 2024
- PFE00035\_ID-EPTRI (GA n° 777554). European Commission.

H2020-INFRADEV-01-2017.  
777554. PI: Joana Claverol Torres.  
2018 - 2020

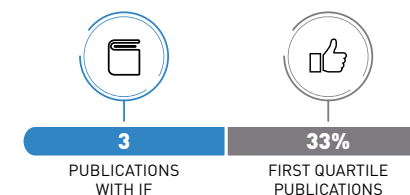
### Selected publications

- Badia X, Aguarón A, Fernández A, Gimón A, Nafria-Escalera B, Gaspar B, et al. Patient involvement in reflective multicriteria decision analysis to assist decision making in oncology. Int J Technol Assess Health Care. 2019; 35(1): 56 - 63. FI: 1,418(Q3)
- Deane K, Delbecque L, Gorbenko O, Hamoir AM, Hoos A, Nafria-Escalera B, et al. Co-creation of patient engagement quality guidance for medicines development: an international multistakeholder initiative. BMJ Innov. 2019; 5(1): 43 - 55. FI: (Q)
- Nordenmalm S, Kimland E, Ligas F, Lehmann B, Claverol J, Nafria-Escalera B, et al. Children's views on taking medicines and participating in clinical trials. Arch Dis Child. 2019; 104(9): 900 - 905. FI: 3,158(Q1)
- Tsang VWL, West L, Woods C, Koh CJ, McCune S, Mullin T, et al. Role of Patients and Parents in Pediatric Drug Development. Ther Innov Regul Sci. 2019;53(5): 601 - 608. FI: 0,901(Q4)



## INNOVATION IN HEALTH TECHNOLOGIES

Jaume Pérez Payarols, MD, PhD



### Members

#### STAFF RESEARCHER

Marta Ayats Soler, Carmen De la Gala Otero, Lucas Krauel Giménez-Salinas, José M. Quintillá Martínez, Josep Rubio Palau, Arian Tarbal Roquer, Arnau Valls Esteve

#### ASSISTANT RESEARCHER

María Garau Rolandi, Alessandra Zoccali

#### MANAGEMENT

Ariadna Ballesteros Herruzo, Paula Cañal Vega, Alex Rigol Reyes

### Selected projects

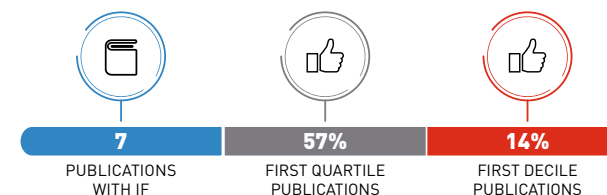
- Using indicators to influence consumer behaviour and prevent pre-disease through food and lifestyle-related interventions (STOP MetSyn). EIT Health e.V. Food4Health. 19967. PI: Arian Tarbal Roquer. 2019 - 2019.
- All@once: Training professionals and families for a healthier childhood. EIT Health e.V. EIT-CAMPUS- Citizens (BP 2019). PI: Arian Tarbal Roquer. 2019 - 2019.
- P2 - FUSE3D: Technologies per a deposició de material semifos. Departament de Salut - Generalitat de Catalunya. Technologies emergents. PI: Arnau Valls Esteve. 2019 - 2022.
- Starship\_Fellowship Network (19218). EIT Health e.V. CAMPUS, Citizens, Summer School. 19218. PI: Arnau Valls Esteve. 2019 - 2019.

### Selected publications

- Duque P, Quintillá Martínez JM, Varela JA, Garrido P, Valencia Ó, Maestre JM. Emotional response to a high-fidelity trauma simulation: An observational study. Rev Esp Anesthesiol Reanim. 2019; 66(10): 521 - 527. FI: (Q)
- Fenollosa F, Gomà JR, Buj-Corral I, Tejo-Otero A, Minguella-Canela J, Uceda R, et al. Foreseeing new multi-material FFF-Additive Manufacturing concepts meeting mimicking requirements with living tissues. Procedia Manufacturing. 2019; 41: 1063 - 1070. FI: (Q)
- Lezama-Del Valle P, Krauel L, LaQuaglia MP. Error traps and culture of safety in pediatric surgical oncology. Semin Pediatr Surg. 2019; 28(3): 164 - 171. FI: 2,462(Q1)
- Parri F, Alonso V, Albert A, Bejarano M, Vicario F, Rubio Palau J. Auricular Reconstructive Surgery Improvement to the Firmin Technique for Placing an Earring. Cleft Palate Craniofac J. 2019; 56(9): 1260 - 1262. FI: 1,471(Q3)
- Salcedo-Gil CA, Prieto Gundin A, Alberola Ferranti M, Rubio Palau J. Lymphoepithelial carcinoma of the accessory parotid gland. Br J Oral Maxillofac Surg. 2019; 57(4): 365 - 367. FI: 1,164(Q3)

## DIAGNOSTIC AND THERAPEUTIC IMAGE

Jose Luis Munuera del Cerro, MD, PhD



### Members

#### STAFF RESEARCHER

Marta Gómez Chiari, Emilio Inarejos Clemente, Marta Maristany Cucurella, Monica Rebollo Polo

### Selected projects

- EXPERT3D\_Transversal programme in medical image post-process, training professionals: 3D printing to research. EIT Health e.V. 02/04/2019. 19787. PI: Jose Luis Munuera del Cerro. 2019.
- Resonancia magnética funcional e imagen de tensor de difusión en la valoración prequirúrgica del paciente pediátrico con hipoacusia prelingual neurosensorial profunda bilateral de detección en el cribado neonatal como potenciales biomarcadores de desarrollo de lenguaje oral tras la colocación de un implante coclear. Sociedad Española de Neurorradiología (SENOR). 05/10/2017. PI: Mónica Rebollo Polo. 2017 - 2020.

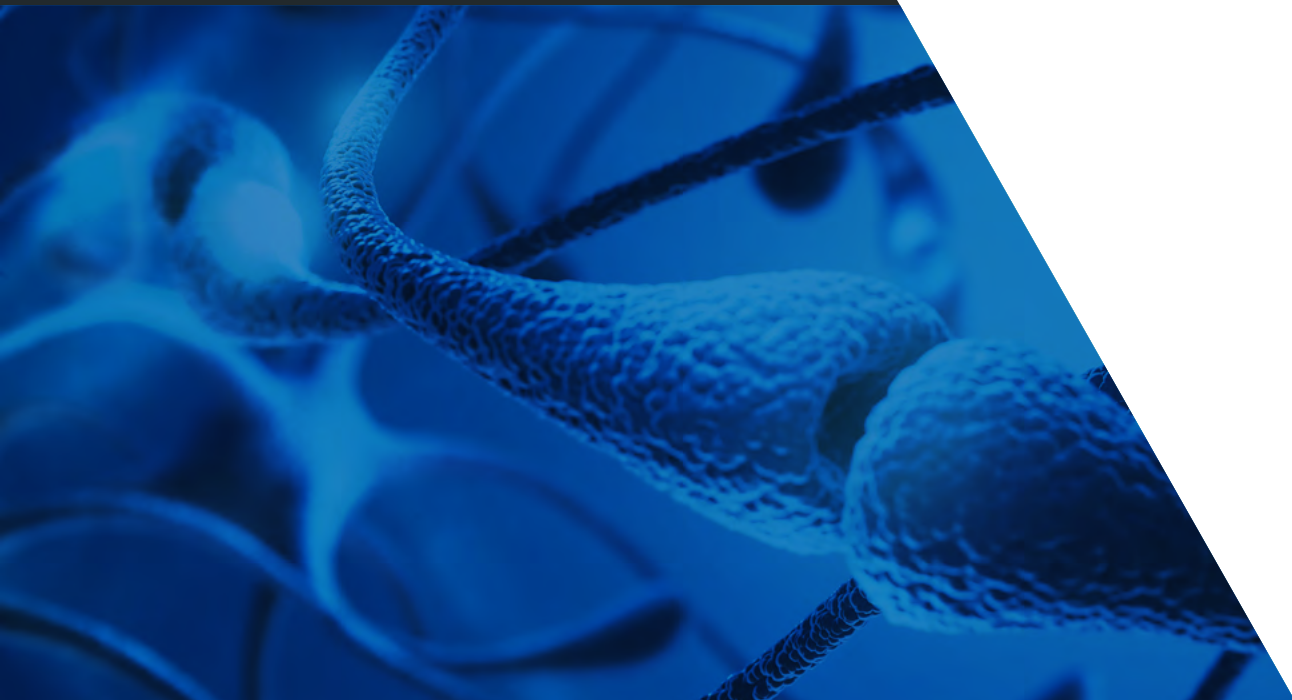
### Selected publications

- Alonso-Lana S, Moro N, McKenna PJ, Sarró S, Romaguera A, Monté GC, et al. Longitudinal brain functional changes between mania and euthymia in bipolar disorder. *Bipolar Disord.* 2019; 21(5): 449 - 457. FI: 4,936(Q1)
- Madre M, Canales-Rodríguez EJ, Fuentes-Claramonte P, Alonso-Lana S, Salgado-Pineda P, Guerrero-Pedraza A, et al. Structural abnormality in schizophrenia versus bipolar disorder: A whole brain cortical thickness, surface area, volume and gyrification analyses. *Neuroimage Clin.* 2019; 25: 102131 - 102131. FI: 3,943(Q1)
- Navallas M, Inarejos E, Iglesias-Jimenez E, Rebollo M, Anton-Lopez J, Navarro OM. Connective Tissue Disorders in Childhood: Are They All the Same?. *Radiographics.* 2019; 39(1): 229 - 250. FI: 3,923(Q1)
- Pérez-Grijalba V, Arbizu J, Romero J, Prieto E, Pesini P, Sarasa L, et al. Plasma Aβ42/40 ratio alone or combined with FDG-PET can accurately predict amyloid-PET positivity: a cross-sectional analysis from the AB255 Study. *Alzheimers Res Ther.* 2019; 11(1): 96 - 96. FI: 6,142(Q1)



**SJD**

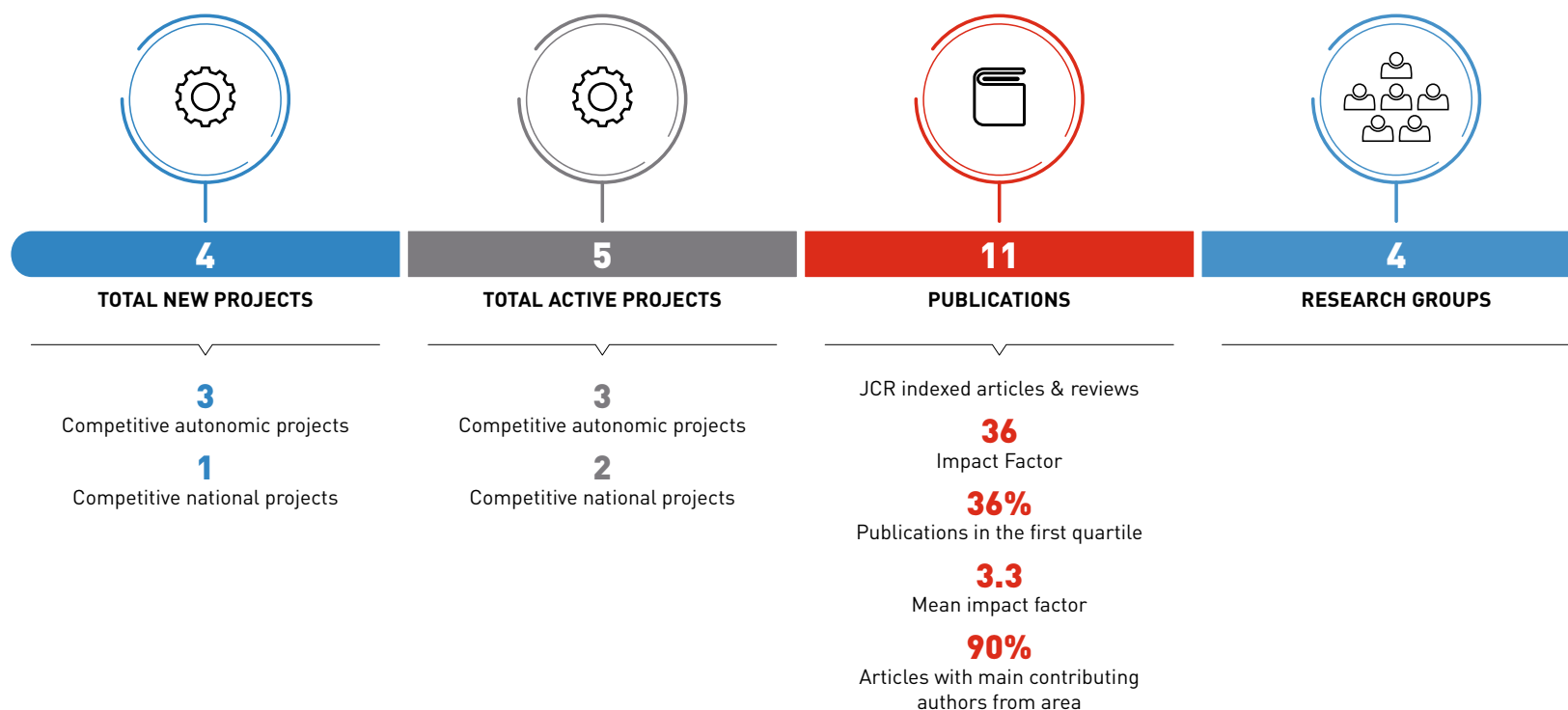
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**11**

## **APPLIED CLINICAL RESEARCH**

Influence of the environment on child and adolescent welfare  
Paediatric ophthalmology  
Hospital dentistry and periodontal medicine  
Research in nursing



## INFLUENCE OF THE ENVIRONMENT ON CHILD AND ADOLESCENT WELFARE

Carles Luaces Cubells, MD, PhD



7

PUBLICATIONS  
WITH IF



28%

FIRST QUARTILE  
PUBLICATIONS

Contribution to the development and dissemination of the protocol of action for mistreatments in childhood and adolescence in healthcare field (published by the Secretariat for Public Health, Catalan Department of Health, Barcelona, March 2019: [http://salutpublica.gencat.cat/.content/minisite/aspcat/promocio\\_salut/infancia-i-adolescencia/05maltractamentinfantjuvenil/protocol-actuacio-maltractaments-infancia.pdf](http://salutpublica.gencat.cat/.content/minisite/aspcat/promocio_salut/infancia-i-adolescencia/05maltractamentinfantjuvenil/protocol-actuacio-maltractaments-infancia.pdf))

Coordination of telemedicine sessions (1 monthly) with Spanish and Latin American centres aimed to facilitate the exchange of experiences relating to clinical situations specific to the sphere of paediatric patients.

### Members

#### STAFF RESEARCHER

Andrea Aldemira Liz, Vanessa Arias Constantí, Gemma Claret Teruel, Ana Isabel Curcoy Barcenilla, Eva Gargallo Burriel, Lidia Martínez Sánchez, David Muñoz Santanach, Sergi Navarro Vilarrubi, Cristina Parra Cotanda, Marta Simó Nebot, Victoria Trenchs Sainz de la Maza

### Selected projects

- Estudio prospectivo observacional sobre la hidratación y nutrición en pacientes pediátricos con enfermedad avanzada. Sociedad Española de cuidados paliativos pediátricos (PEDPAL). Beca Nutricia. PI: Sergi Navarro Vilarrubi. 2019-2020.

### Selected publications

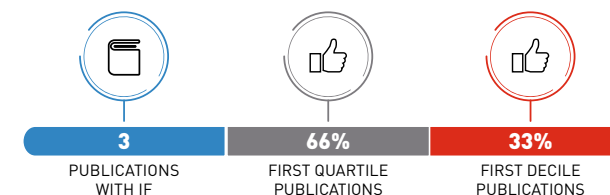
- Brotons-de los Reyes P, Nogueras MM, Valls-Lafon A, Larrauri A, Dominguez A, Launes-Montana C, Luaces-Cubells C and Munoz-Almagro C. Impact of Rapid On-demand Molecular Diagnosis of Pediatric Seasonal Influenza on Laboratory Workflow and Testing Costs: A Retrospective Study. *Pediatr Infect Dis J.* 2019; 38(6): 559 - 563. FI: 2,317(Q2)
- Cristina Parra Cotanda, Trenchs-Sainz de la Maza V, Sabater Sabaté A and Luaces-Cubells C. Perceived quality in a pediatric emergency department: what does the mother think about?. *Emergencias.* 2019; 31(2): 136 - 138. FI: 3,350(Q1)
- Faura Morros A, Cuaserna González A, Hernández-Bou S, Trenchs-Sainz de la Maza V, Camacho Díaz JA and Luaces-Cubells C. Rentabilidad diagnóstica de la ecografía renal tras la primera infección de orina en los lactantes. *An Pediatr (Barc).* 2019; 90(4): 232 - 236. FI: 1,166(Q3)
- Sarria-Guerrero JA, Luaces-Cubells C, Jiménez-Fàbrega FX, Villamor-Ordozgoiti A, Isla Pera P and Guix-Comellas EM. Pediatric televisits and telephone triage: impact on use of a hospital emergency department. *Emergencias.* 2019; 31(4): 257 - 260. FI: 3,350(Q1)

### Networks

- Research in pediatric emergency medicine (REPEM) network (The European Paediatric Emergency Medicine Research Network. Pediatric Section of the European Society for Emergency Medicine)
- Red De Investigación De La Sociedad Española De Urgencias De Pediatría / Spanish Pediatric Emergency Research Group (RISeuP/SPERG), 2012
- Red de investigación y desarrollo de la emergencia pediátrica latinoamericana (RIDEPLA)

## PAEDIATRIC OPHTHALMOLOGY

Joan Prat Bartomeu, MD, PhD



### Members

#### STAFF RESEARCHER

Ester Casas Gimeno, Jaume Català Mora, Ana Isabel Díaz Cortés, Carlos Fresno Cañada, Carmen García de Vicuña Muñoz, Enrique Jimenez Gañan, Marta Morales Ballús, M<sup>a</sup> Josep Planas Tapias, Enric Puigventós, Alicia Serra Castanera, Gemma Romeu Cerrillo, Ana Llorca Cardeñosa, Begoña Yeste Mayoral, Núria Planas Doménech, Simone Dowon Suh, Jesús Díaz Cascajosa, Elisa Carreras Bertrán

### Selected publications

- Foeldvari I, Klotsche J, Simonini G, Edelsten C, Angeles-Han ST, Bangsgaard R, et al. Proposal for a definition for response to treatment, inactive disease and damage for JIA associated uveitis based on the validation of a uveitis related JIA outcome measures from the Multinational Interdisciplinary Working Group for Uveitis in Childhood (MIWGUC). *Pediatr Rheumatol Online J*. 2019; 17(1): 66 - 66. FI: 2,673(Q1)

- Koch CR, Kara-Junior N, Santhiago MR and Morales-Ballus M. Comparison of different surgical approaches for pediatric cataracts: complications and rates of additional surgery during long-term follow-up. *Clinics (Sao Paulo)*. 2019; 74: e966. FI: 1,127(Q3)

- Pascual-Pasto G, Bazan-Peregrino M, Gene-Olaciregui N, Restrepo-Perdomo CA, Mato-Berciano A, Ottaviani D, et al. Therapeutic targeting of the RB1 pathway in retinoblastoma with the oncolytic adenovirus VCN-01. *SCI TRANSL MED*. 2019; 11(476). FI: 17,200(Q1)

## HOSPITAL DENTISTRY AND PERIODONTAL MEDICINE

Alejandro Rivera Baró, MD, PhD

### Members

#### STAFF RESEARCHER

Lluís Brunet Llobet, Jordi Moncunill Mira, Maria Lluïsa Tobella Camps, Jaume Miranda Rius, Mireia Aznar Gómez

### Selected projects

- SGR 2017\_Hospital Dentistry & Periodontal Medicine Research Group. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SGR 2017-2019. 2017 SGR 00453. PI: Lluís Brunet Llobet. 2017 - 2020.

### Selected publications

- Brunet-Garcia A, Costa-Climent MD, Pujol-Rodríguez M, Lluís Brunet Llobet and Faubel-Serra M. Sphenoid sinus mucocoele: an unusual complication of head and neck irradiation in a North African

woman. *J Clin Exp Dent*. 2019; 11(2): e208-e212. FI: (Q)

- Lluís Brunet Llobet, Lahor-Soler E, Mashala El and Miranda-Rius J. Continuous Surgical Decompression for Solitary Bone Cyst of the Jaw in a Teenage Patient. *Case Rep Dent*. 2019; 2019: 9137507 - 9137507. FI: (Q)



## RESEARCH IN NURSING

Laura Lahuerta Valls, PhD



1

PUBLICATIONS  
WITH IF

The research conducted by our group is addressed to providing the best care to our patients and their families. Our goal is to give the best possible personal, holistic and integral care based on scientific evidence, in order to empower them and to achieve a quick reintegration into their customary setting, improving their quality of life as well as the experience of their stay at our institution.

In 2019, the Nursing research group carried out the following activities:

1. We developed a care research strategy to promote and coordinate translational and interdisciplinary research in the nursing field.
2. Evidence-based practice was strengthened with the aim to provide safer and better care of the highest quality to our patients and their families.
3. We have created a clinical practice environment in which nurses can apply scientific evidence to improve the care which is provided and the results of their professional activity.
4. Wealth has been generated in the healthcare sphere through innovation and the implementation of various initiatives addressed to improving the quality of life of our patients and their families.

### Members

#### STAFF RESEARCHER

Silvia Ciprés Roig, Encarna Gómez Gamboa, Carmen Jerez Molina, Jordi Mitjà Costa, Gemma Pérez Acevedo, Concepción Agustí Mata, Dolors Navarro

### Selected projects

- Sol·licitud de subvencions per a la intensificació de professionals d'infermeria, llevadora, fisioteràpia, dietètica i nutrició, òptica i optometria, podologia i protètica dental en l'àmbit de la recerca i la innovació en salut. Departament de Salut - Generalitat de Catalunya. Intensificació Competitivo infermeria. SLT008/18/00170. PI: Encarna Gómez Gamboa. 2019 - 2020.
- Sol·licitud de subvencions per a la intensificació de professionals d'infermeria, llevadora, fisioteràpia, dietètica i nutrició, òptica i optometria, podologia i protètica dental en l'àmbit de la recerca i la innovació en salut. Departament de Salut - Generalitat de Catalunya. Intensificació infermeria. SLT008/18/00164. PI: Gemma Pérez Acevedo. 2019 - 2020.
- Eficacia y seguridad de los ácidos grasos hiperoxigenados en la microcirculación de la púrpura fulminante en pacientes pediátricos con sepsis. Fundació Infermeria i Societat. Ajudes a la Recerca en Infermeria. PR-242/17. PI: Gemma Pérez Acevedo. 2018 - 2019.

### Selected publications

- Pérez-Acevedo G, Torra-Bou JE, Marisa Manzano Canillas and Bosch-Alcaraz A. Management of purpura fulminans skin lesions in a premature neonate with sepsis: a case study. J WOUND CARE. 2019; 28(4): 198 - 203. FI: 1,948(Q3)



ANNUAL SCIENTIFIC  
**REPORT**  
**2019**

**SJD**

Sant Joan de Déu  
Institut de Recerca

**ANNEX 1**



## Active clinical trials and observational studies during 2019

### TMC278HTX2002

A Phase 2, Open-label, Single-arm, Multicenter Study to Evaluate the Pharmacokinetics, Safety, Tolerability, and Efficacy of Switching to RPV Plus Other ARVs in HIV-1-infected Children (Aged 2 to <12 years) who are Virologically Suppressed. Phase II. Promoter: Janssen - Cilag, S. A. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 19/12/2019. Commercial

### I4V-MC-JAHU

A Randomized, Double-Blind, Placebo-Controlled, Withdrawal, Safety and Efficacy Study of Oral Baricitinib in Patients from 1 Year to Less Than 18 Years Old with Systemic Juvenile Idiopathic Arthritis. Phase III. Promoter: Lilly S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 11/12/2019. Commercial

### 6MWT

Hospital Sant Joan de Déu - Esplugues HSJD. 1 (FSJD). Prod. Sanitario. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Julita Medina. Service: Rehabilitation and physical medicine. Start: 9/12/19. Non Commercial

### G040871

A phase I/II, multicenter, open label, multi arm study evaluating the safety, tolerability, pharmacokinetics, and preliminary activity of idasanutlin in combination with either chemotherapy or venetoclax in the treatment of pediatric and young adult patients with relapsed/refractory acute leukemias or solid tumors. Phase I. Promoter: F. Hoffmann- La Roche Ltd. PI: Jaume Mora Graupera. Service: Oncology. Start: 06/12/2019. Commercial

### INTERFANT-06

Guia de recomendaciones de la sehop para el tratamiento de la leucemia

linfoblástica aguda o bifenotípica del lactante menor de 1 año. No EPA. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI: José Luis Dapena Díaz. Service: Oncology. Start: 04/12/2019. Non Commercial

### NS-065/NCNP-01-301

A Phase 3 Randomized, Double-blind, Placebo-controlled, Multicenter Study to Assess the Efficacy and Safety of Viltolarsen in Ambulant Boys with Duchenne Muscular Dystrophy (DMD). Phase III. Promoter: Medpace Clinical Research, LLC. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 03/12/2019. Commercial

### I4V-MC-JAHW

Estudio sin enmascaramiento, comparativo con un tratamiento activo, en el que se evalúan la seguridad y la eficacia del tratamiento con baricitinib por vía oral en pacientes de entre 2 y menos de 18 años que presentan uveítis asociada con artritis idiopática juvenil activa o uveítis anterior crónica y anticuerpos antinucleares. Phase III. Promoter: Lilly S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 15/11/2019. Commercial

### MT-12

A one-year placebo-controlled phase III trial evaluating the efficacy and safety of the house dust mite (HDM) SLIT-tablet in children (5-11 years of age) with HDM allergic rhinitis/rhinoconjunctivitis with or without asthma. Phase III. Promoter: ALK-ABELLÓ A/S. PI: Mónica Piquer Gibert. Service: Allergy and clinical immunology. Start: 14/11/2019. Commercial

### GWEP1560

A Randomized, Double-blind, Placebo-controlled Single-ascending Dose Trial to Evaluate the Safety, Tolerability, and Pharmacokinetics of GWP42003-P in

Conjunction with Hypothermia in Neonates with Moderate or Severe Hypoxic Ischemic Encephalopathy. Phase I. Promoter: GW Research. PI: Ana Alarcon Allen. Service: Neonatology. Start: 14/11/2019. Commercial

### WTX101-301

Phase 3, Randomised, Rater-Blinded, Multi-Centre Study to Evaluate the Efficacy and Safety of WTX101 Administered for 48 Weeks Versus Standard of Care in Wilson Disease Subjects Aged 18 and Older with an Extension Phase of up to 60 Months. Phase III. Promoter: Alexion Pharmaceuticals Inc. PI: Cristina Molera Busoms. Service: Gastroenterology, hepatology and nutrition. Start: 12/11/2019. Commercial

### ZX008-1900

An Open-Label Extension Trial to Assess the Long-Term Safety of ZX008 (Fenfluramine Hydrochloride) Oral Solution as an Adjunctive Therapy for Seizures in Patients with Rare Seizure Disorders Such as Epileptic Encephalopathies Including Dravet Syndrome and Lennox- Gastaut Syndrome. Phase III. Promoter: Zogenix, Inc. PI: Alia Ramírez Camacho. Service: Neurology. Start: 07/11/2019. Commercial

### CSE6101B2201

Estudio de fase 2, multicéntrico, abierto para evaluar la dosis adecuada y la seguridad de crizanlizumab, con o sin hidroxiurea/hidroxicarbamida, en grupos secuenciales de edad descendente de pacientes pediátricos con Enfermedad de Células Falciformes con crisis vaso-oclusivas. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Anna Ruiz Llobet. Service: Clinical haematology. Start: 05/11/2019. Commercial

### GWND18064

A randomized, double-blind, placebo-controlled, trial to investigate the efficacy

and safety of cannabidiol oral solution (GWP42003-P, CBD-OS) in patients with rett syndrome. Phase III. Promoter: GW Research. PI: M<sup>a</sup>del Mar O'Callaghan Gordo. Service: Neurology. Start: 04/11/2019. Commercial

### I4V-MC-JAHX

A Phase 3 Multicenter Study to Evaluate the Long-Term Safety and Efficacy of Baricitinib in Patients from 1 Year to <18 Years of Age with Juvenile Idiopathic Arthritis (JIA). Phase III. Promoter: Lilly S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 25/10/2019. Commercial

### 20170534

Estudio de extensión abierto, multicéntrico y de un solo brazo para evaluar la seguridad y eficacia a largo plazo del tratamiento actual o anterior con denosumab en niños / adultos jóvenes con osteogénesis imperfecta. Phase III. Promoter: Amgen Inc. PI: Rosa Bou i Torrent. Service: Paediatrics. Start: 16/10/2019. Commercial

### MK-7625A-036

A Phase 1, open-label, non-comparative, multicenter clinical trial to evaluate the safety, tolerability, and pharmacokinetics of ceftolozane/tazobactam (MK-7625A) in pediatric participants with nosocomial pneumonia. Phase I. Promoter: Merck, Sharp & Dohme de España. S. A. PI: Iolanda Jordán García. Service: UCI. Start: 08/10/2019. Commercial

### 105-MS306

An Open-Label, Randomized, Multicenter, Active-Controlled, Parallel-Group Study to Evaluate the Safety, Tolerability, and Efficacy of BII017 in Pediatric Subjects Aged 10 to Less Than 18 Years for the Treatment of Relapsing Remitting Multiple Sclerosis, With Optional Open-Label

Extension. Phase III. Promoter: Biogen IDEC Research LTD. PI: Thais Armangué Salvador. Service: Neurology. Start: 10/09/2019. Commercial

#### **MT-2-02**

An exploratory, open-label, multicenter study in male pediatric patients with cerebral X-Linked adrenoleukodystrophy (CALD) to assess the effects of min 102 treatment on the progression of cerebral lesions. Phase II. Promoter: Minoryx Therapeutics S. L. PI: M<sup>a</sup>Angels García Cazorla. Service: Neurology. Start: 09/09/2019. Commercial

#### **GC-LTFU-001**

Protocolo para el seguimiento a largo plazo de pacientes tratados con linfocitos T modificados genéticamente. Phase III. Promoter: Celgene International II S. à. r. l. PI: Susana Rives Solà. Service: Oncology. Start: 23/08/2019. Commercial

#### **ARQ-092-103**

A Phase 1/2 Study of ARQ 092 in Patients with PIK3CA-related Overgrowth Spectrum and Proteus Syndrome. Phase I. Promoter: ARQULE, INC. PI: Jaume Mora Graupera. Service: Oncology. Start: 20/08/2019. Commercial

#### **UKM17\_0023**

LBL 2018 – International cooperative treatment protocol for children and adolescents with lymphoblastic lymphoma. Phase II. Promoter: Universitätsklinikum Munster. PI: Veronica Celis Passini. Service: Oncology. Start: 02/08/2019. Non Commercial

#### **M15-340**

An Open-label Multiple Dose Study to Evaluate the Pharmacokinetics, Safety, and Tolerability of Upadacitinib in Pediatric Subjects with Polyarticular

Course Juvenile Idiopathic Arthritis. Phase I. Promoter: "AbbVie Deutschland GmbH & Co. KG "AbbiVie"". PI: Jordi Antón López. Service: Paediatrics. Start: 24/07/2019. Commercial

#### **VBP15-004**

A Phase IIb Randomized, Double-blind, Parallel Group, Placebo- and Active-controlled Study with Double-Blind Extension to Assess the Efficacy and Safety of Vamorolone in Ambulant Boys with Duchenne Muscular Dystrophy (DMD). Phase II. Promoter: Reveragen Biopharma Inc. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 22/07/2019. Commercial

#### **TMC114FD2HTX1006**

Ensayo para evaluar la aceptabilidad de comprimidos ranurados recubiertos con película de darunavir/cobicistat/emtricitabina/tenofovir alafenamida (D/C/F/TAF) a dosis fija en participantes pediátricos infectados con VIH-1 de edad ≥6 a <12 años, usando comprimidos equivalentes de placebo. Phase I. Promoter: Jassen Science Ireland UC. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 19/07/2019. Commercial

#### **CC-10004-PP50-003**

A phase 3, multi-center, randomized, double-blind, placebo controlled study to assess the efficacy and safety of apremilast (CC-10004) in pediatric subjects from 6 through 17 years with moderate to severe plaque psoriasis. Phase III. Promoter: Celgene Corporation. PI: M<sup>a</sup> Asunción Vicente Villa. Service: Dermatology. Start: 12/07/2019. Commercial

#### **SRK-015-002**

Estudio de fase 2 con tratamiento activo para evaluar la eficacia y la seguridad de SRK-015 en pacientes con atrofia

muscular espinal de inicio tardío. TOPAZ. Phase II. Promoter: Scholar Rock Inc. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 11/07/2019. Commercial

#### **PK HemoA-SP / STH-OCT-2016-01**

Caracterización farmacocinética de la población con Hemofilia A en España, utilizando una aplicación médica online basada en un modelo poblacional publicado y un algoritmo Bayesiano. EPA-SP. Promoter: Fundación Española de Trombosis y Hemostasia. PI: Ruben Berrueto Moreno. Service: Clinical haematology. Start: 05/07/2019. Non Commercial

#### **BAY 86-5321/20090**

A randomized, 2-arm, open-label, phase 3 study to evaluate the efficacy, safety, and tolerability of intravitreal aflibercept versus laser photocoagulation in patients with retinopathy of prematurity (ROP). Phase III. Promoter: Bayer AG. PI: Alicia Serra Castanera. Service: Ophthalmology. Start: 05/07/2019. Commercial

#### **200075 (ZOSTER-047)**

Estudio de fase II, aleatorizado, abierto y multicéntrico para evaluar la reactogenicidad, seguridad e inmunogenicidad de la vacuna pediátrica candidata de subunidades de herpes zóster (PED-HZ/su) de GSK tras su administración intramuscular con una pauta de dos dosis a niños inmunodeprimidos receptores de trasplante renal de 1-17 años de edad. Phase II. Promoter: Glaxosmithkline, S. A. PI: Alvaro Madrid Aris. Service: Nephrology. Start: 04/07/2019. Commercial

#### **ID-085A301**

Estudio de evolución natural de pacientes pediátricos con inicio temprano de gangliosidosis GM1, gangliosidosis GM2 o enfermedad de Gaucher tipo 2

(RETRIEVE). No EPA. Promoter: Idorsia Pharmaceuticals Ltd. PI: M<sup>a</sup>del Mar O'Callaghan Gordo. Service: Neurology. Start: 02/07/2019. Commercial

#### **CQAW039B2201**

A multicenter, open-label, 8 day treatment study to assess the pharmacokinetics, safety and tolerability of fevipiprant delivered via a once daily chewable tablet in children aged 6 to <12 years with asthma. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Jaime Lozano Blasco. Service: Allergy and clinical immunology. Start: 17/06/2019. Commercial

#### **MT1621-102**

Estudio abierto en fase II del tratamiento de continuación con una combinación de nucleósidos de pirimidina en pacientes con deficiencia de timidina quinasa 2 (TK2). Phase II. Promoter: Modis Therapeutics, Inc. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 03/06/2019. Commercial

#### **CLC2696B2319**

Estudio multicéntrico, abierto, para evaluar la seguridad, tolerabilidad, farmacocinética y farmacodinámica de LCZ696, seguido de un estudio de 52 semanas, aleatorizado, doble ciego, paralelo controlado con fármaco activo para evaluar la eficacia y seguridad de LCZ696 comparado con enalapril en pacientes pediátricos con edades desde 1 mes hasta < 18 años, con insuficiencia cardíaca debida a disfunción sistólica del ventrículo izquierdo sistémico. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Joan Sanchez de Toledo. Service: Cardiology. Start: 15/05/2019. Commercial

#### **MT-2-03**

A double-blind, placebo-controlled study on the effects of min-102 on biochemical,

imaging, neurophysiological, and clinical markers in patients with Friedreich's ataxia. Phase II. Promoter: Minoryx Therapeutics BE SA. PI: Alejandra Darling. Service: Neurology. Start: 26/04/2019. Commercial

#### **DSC/14/2357/51**

Estudio abierto, de seguridad a largo plazo, de tolerabilidad y de eficacia de GIVINOSTAT en todos los pacientes con distrofia muscular duchenne que hayan sido tratados previamente en uno de los estudios de GIVINOSTAT. Phase IV. Promoter: Italfarmaco, S. A. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 24/04/2019. Commercial

#### **I4V-MC-JAHV**

A Randomized, Double-Blind, Placebo-Controlled, Withdrawal, Safety and Efficacy Study of Oral Baricitinib in Patients from 2 Years to Less Than 18 Years Old with Juvenile Idiopathic Arthritis (JIA). Phase III. Promoter: Lilly S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 24/04/2019. Commercial

#### **BST-LT-01**

Ensayo clínico fase Ib-II, prospectivo, multicéntrico, abierto y no controlado para evaluar la seguridad y eficacia inmunológica de la infusión de linfocitos T específicos frente a virus a partir del mejor donante disponible en receptores de un trasplante alogénico de progenitores hematopoyéticos. Phase II. Promoter: Banc de Sang i Teixits. PI: Julia Marsal Ricoma. Service: Oncology. Start: 11/04/2019. Non Commercial

#### **OBS15333**

Estudio longitudinal, observacional y prospectivo en pacientes pediátricos con dermatitis atópica de moderada a grave, cuya enfermedad no está controlada adecuadamente con tratamientos tópicos

prescritos o cuando dichos tratamientos no son aconsejables desde el punto de vista médico. EPA-SP. Promoter: Sanofi-Aventis S. A. PI: M<sup>o</sup> Asunción Vicente Villa. Service: Dermatology. Start: 10/04/2019. Commercial

#### **SNT-III-012-E**

A Phase III Open-Label Extension Study to Assess the Long-Term Safety and Efficacy of Idebenone in Patients with Duchenne Muscular Dystrophy (DMD) who Completed the SIDEROS Study. Phase III. Promoter: Santhera Pharmaceuticals (Switzerland) Ltd,. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 09/04/2019. Commercial

#### **B7451015**

Estudio en fase III, multicéntrico y de extensión a largo plazo para evaluar la eficacia y la seguridad de PF-04965842, en monoterapia o en combinación con medicamentos tópicos, administrado a pacientes de 12 años o más con dermatitis atópica moderada o grave. Phase III. Promoter: PFIZER INC. PI: M<sup>o</sup> Asunción Vicente Villa. Service: Dermatology. Start: 01/04/2019. Commercial

#### **B7451014**

A phase 3 randomized withdrawal, double-blind, placebo-controlled, multicenter study investigating the efficacy and safety of PF-04965842 in subjects aged 12 years and over, with moderate to severe atopic dermatitis with the option of rescue treatment in flaring subjects. Phase III. Promoter: PFIZER INC. PI: M<sup>o</sup> Asunción Vicente Villa. Service: Dermatology. Start: 18/03/2019. Commercial

#### **AZP01-CLI-003**

A Phase 2b/3 study to evaluate the safety, tolerability, and effects of Livoletide

(AZP-531), an unacylated ghrelin analog, on food-related behaviors in patients with Prader-Willi syndrome. Phase II. Promoter: Millendo Therapeutics SAS. PI: Marta Ramon Krauel. Service: Endocrinology. Start: 07/03/2019. Commercial

#### **CL1-68587-003 (UCART19-LTFU)**

Estudio de seguimiento a largo plazo (LTFU) de pacientes que han sido previamente expuestos a UCART19/ALLO-501 (células T alogénicas modificadas que expresan un receptor antigénico quimérico anti- CD19 basado en lentivirus). Phase I. Promoter: Laboratorios Servier, S. L. PI: Susana Rives Solà. Service: Oncology. Start: 26/02/2019. Commercial

#### **2PR01**

Un estudio observacional retrospectivo de pacientes con neuroblastoma de alto riesgo en España tratados con Naxitamab y el factor estimulante de colonias de granulocitos y macrófagos (GM-CSF). EPA-OD. Promoter: Y-mAbs Therapeutics A/S. PI: Jaume Mora Graupera. Service: Oncology. Start: 19/02/2019. Commercial

#### **E7389-G000-213**

A Phase 1/2 single-arm study evaluating the safety and efficacy of eribulin mesilate in combination with irinotecan in children with refractory or recurrent solid tumors. Phase II. Promoter: Eisai Limited. PI: Jaume Mora Graupera. Service: Oncology. Start: 18/02/2019. Commercial

#### **I4V-MC-JAIP**

Estudio de fase 3, multicéntrico, aleatorizado, comparativo con placebo, con enmascaramiento doble y grupos paralelos, en el que se evalúa la farmacocinética, la eficacia y la seguridad de baricitinib en niños enfermos ambulatorios con dermatitis atópica moderada o grave. Phase III. Promoter:

Lilly S. A. PI: M<sup>o</sup> Asunción Vicente Villa. Service: Dermatology. Start: 12/02/2019. Commercial

#### **I8B-MC-ITSB**

Estudio prospectivo, aleatorizado y con enmascaramiento doble, en el que se compara LY900014 con Humalog, con un grupo en el que se administra LY900014 posprandial sin enmascaramiento, en niños y adolescentes con diabetes de tipo 1: PRONTO-Peds. Phase III. Promoter: Lilly S. A. PI: Larisa Suarez Ortega. Service: Endocrinology. Start: 12/02/2019. Commercial

#### **BMN111-302**

Estudio de extensión de fase III, abierto, para evaluar la seguridad y la eficacia a largo plazo de BMN 111 en pacientes pediátricos con acondroplasia. Phase III. Promoter: BioMarin Pharmaceutical Inc. PI: Rosendo Ullot Font. Service: Orthopaedics and traumatology. Start: 11/02/2019. Commercial

#### **CP-NVK002-0001**

Atropina para la progresión de la miopía en niños (CHAMP): un estudio de fase 3 con tres grupos, aleatorizado, con doble enmascaramiento, controlado con placebo, sobre una solución oftálmica de atropina sulfato al 0,01% y 0,02%. Phase III. Promoter: NEVAKAR INC. PI: Isabel Ayet Roger. Service: Ophthalmology. Start: 08/02/2019. Commercial

#### **CL1-68587-001**

Estudio abierto, no comparativo, de fase 1 para evaluar la seguridad y la capacidad de UCART19 de inducir una remisión molecular en pacientes pediátricos con leucemia linfoblástica aguda de células B recurrente/refractaria. Phase I. Promoter: Laboratorios Servier, S. L. PI: Susana Rives Solà. Service: Oncology. Start: 07/02/2019. Commercial

#### CP40617

A Phase III, Multicenter, Randomized, Double-Blind, Active-Controlled Study to Assess the Efficacy and Safety of Baloxavir Marboxil in Combination with a Neuraminidase Inhibitor Versus a Neuraminidase Inhibitor Alone in Hospitalized Patients with Severe Influenza. Phase III. Promoter: F. Hoffmann- La Roche Ltd. PI: Iolanda Jordán García. Service: UCI. Start: 31/01/2019. Commercial

#### EFC13738

Open label, two cohort (with and without imiglucerase), multicenter study to evaluate pharmacokinetics, safety, and efficacy of eliglustat in pediatric patients with Gaucher disease type 1 and type 3. Phase III. Promoter: Sanofi-Aventis S. A. PI: M<sup>a</sup>del Mar O'Callaghan Gordo. Service: Neurology. Start: 14/01/2019. Commercial

#### GET-PEN-2016-01

Estudio prospectivo español de pacientes hematológicos pre- y posttrasplante hematopoyético que ingresan en unidades de cuidados intensivos: análisis de la práctica clínica, resultados en salud, factores pronósticos y utilización de recursos sanitarios. EPA-SP. Promoter: Grupo Español de Trasplantes Hematopoyéticos y Terapia Celular. PI: Julia Marsal Ricoma. Service: Oncology. Start: 02/01/2019. Non Commercial

#### Y-mAbs 101

A Multicenter Phase 2/3 Trial of the Efficacy and Safety of Intracerebroventricular Radioimmunotherapy using 131I-burtomab for Neuroblastoma Central Nervous System/Leptomeningeal Metastases. Phase II. Promoter: Y-mAbs Therapeutics A/S. PI: Jaume Mora

Graupera. Service: Oncology. Start: 20/12/2018. Commercial

#### CCTL01962201J

A phase II trial of tisagenlecleucel in first-line high-risk (HR) pediatric and young adult patients with B-cell acute lymphoblastic leukemia (B-ALL) who are minimal residual disease (MRD) positive at the end of consolidation (EOC) therapy. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Susana Rives Solà. Service: Oncology. Start: 18/12/2018. Commercial

#### I-CML-Ped Study

International study of chronic myeloid leukemia (CML) treatment and outcomes in children and adolescents. EPA-SP. Promoter: Poitiers University Hospital. PI: Anna Alonso Saladríguez. Service: Oncology. Start: 28/11/2018. Non Commercial

#### ESKETINSUI2002

A Double-blind, Randomized, Psychoactive Placebo-controlled, Study to Evaluate the Efficacy and Safety of 3 Fixed Doses (28 mg, 56 mg and 84 mg) of Intranasal Esketamine in Addition to Comprehensive Standard of Care for the Rapid Reduction of the Symptoms of Major Depressive Disorder, Including Suicidal Ideation, in Pediatric Subjects Assessed to be at Imminent Risk for Suicide. Phase II. Promoter: Janssen - Cilag, S. A. PI: Montserrat Dolz Abadia. Service: Psychiatry and psychology. Start: 27/11/2018. Commercial

#### CNT0148UC03003

A Phase 3 Randomized, Open-Label Study to Assess the Efficacy, Safety, and Pharmacokinetics of Golimumab Treatment, a Human anti-TNF<sub>α</sub> Monoclonal Antibody, Administered Subcutaneously in Pediatric Participants

with Moderately to Severely Active Ulcerative Colitis – PURSUIT 2. Phase III. Promoter: Janssen - Cilag, S. A. PI: Javier Martín De Carpi. Service: Gastroenterology, hepatology and nutrition. Start: 21/11/2018. Commercial

#### PFI-ADA-2018-01/B1801403

Uso de recursos sanitarios e impacto en la productividad laboral relacionada con la Artritis Idiopática Juvenil: estudio Retrospectivo. EPA-OD. Promoter: Pfizer, S. L. U. PI: Jordi Antón López. Service: Paediatrics. Start: 13/11/2018. Commercial

#### LJ501-CRH04

Estudio aleatorizado, controlado con placebo, doble ciego y multicéntrico de la eficacia y la seguridad de LJPC-501 en pacientes pediátricos de > 2 a 17 años de edad con hipotensión resistente a la catecolamina asociada a choque distributivo. Phase III. Promoter: La Jolla Pharmaceutical Company. PI: Iolanda Jordán García. Service: UCI. Start: 06/11/2018. Commercial

#### MT-1621-101

Estudio RETROspectivo de la combinación de nucleósidos pirimidínicos en pacientes con déficit de timidina quinasa 2 (TK2). EPA-OD. Promoter: Modis Therapeutics, Inc. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 02/11/2018. Commercial

#### GCP#05.01.020

A Multicenter, Randomized, Phase III Registration Trial of Transplantation of NiCord<sup>®</sup>, Ex Vivo Expanded, Umbilical Cord Blood-derived, Stem and Progenitor Cells, versus Unmanipulated Umbilical Cord Blood for Patients with Hematological Malignancies. Phase III. Promoter: Gamida-Cell LTD. Cellt Therapy Technologies. PI: Isabel Badell Serra.

Service: Oncology. Start: 04/10/2018. Commercial

#### AG348-C-008

Registro longitudinal internacional de deficiencia de piruvato cinasa / Pyruvate Kinase Deficiency Global Longitudinal Registry. EPA-SP. Promoter: Agios Pharmaceuticals, Inc. PI: Anna Ruiz Llobet. Service: Clinical haematology. Start: 03/10/2018. Commercial

#### EMR 200098\_008 / MER-GRH-2009-01

Estudio observacional, prospectivo y multicéntrico a largo plazo en una cohorte de niños con talla baja nacidos pequeños para su edad gestacional (PEG) que han sido tratados con Saizen<sup>®</sup>. EPA-LA. Promoter: Merck KGaA. PI: Lourdes Ibañez Toda. Service: Endocrinology. Start: 25/09/2018. Commercial

#### QGE031C2202

Estudio fase 2b de búsqueda de dosis, multicéntrico, aleatorizado, doble ciego, controlado con placebo para investigar la eficacia y la seguridad de ligelizumab (QGE031) en pacientes adolescentes con urticaria crónica espontánea (UCE). ". Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Montserrat Álvaro Lozano. Service: Allergy and clinical immunology. Start: 06/09/2018. Commercial

#### JCAR017-BCM-004

Estudio de fase Ib/II, multicéntrico, abierto, de un solo brazo y con varias cohortes para evaluar la seguridad y eficacia de JCAR017 en pacientes pediátricos con leucemia linfoblástica aguda de células B (LLA-B) y linfoma no Hodgkin de células B (LNH-B) recidivante/resistente (r/r). Phase II. Promoter: Celgene International II S. à. r. l. PI: Susana Rives Solà. Service: Oncology. Start: 28/08/2018. Commercial



#### **BP39056**

Estudio de continuidad en dos partes, abierto, multicéntrico, para investigar la seguridad, la tolerabilidad, la farmacocinética, la farmacodinámica y la eficacia de R07034067 en lactantes con atrofia muscular espinal de tipo 1. Phase II. Promoter: F. Hoffmann- La Roche Ltd. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 06/08/2018. Commercial

#### **DLA-ALT-2018-01**

Estudio observacional, transversal y multicéntrico para evaluar la prevalencia de sensibilización a Alternaria alternata y Alt a 1 en los pacientes con rinitis o rinoconjuntivitis y/o asma alérgicas. Estudio PREVALENCIA. EPA-OD. Promoter: Diater Laboratorio de Diagnóstico y Aplicaciones Terapéuticas S. A. PI: Ana Mª Plaza Martín. Service: Allergy and clinical immunology. Start: 01/08/2018. Commercial

#### **HC-O-H-1406**

Infusión de soluciones de gelatina en pacientes pediátricos de hasta 12 años. GPS Study. EPA-SP. Promoter: B. Braun Melsungen AG. PI: Silvia Serrano Casabon. Service: Anesthesia. Start: 26/07/2018. Commercial

#### **RG\_15-114 (PHITT)**

Paediatric Hepatic International Tumour Trial. Phase III. Promoter: University Of Birmingham. PI: Moira Garraus Oneca. Service: Oncology. Start: 25/07/2018. Non Commercial

#### **BEB-13**

Double-Blind, Randomised, Vehicle-Controlled, Phase III Efficacy and Safety Study with 24-Month Open-Label Follow-up of Oleogel-S10 in Patients with Inherited Epidermolysis Bullosa. Phase III. Promoter: AMRYT Research

Limited. PI: Mº Asunción Vicente Villa. Service: Dermatology. Start: 16/07/2018. Commercial

#### **ABÅ-CYSCARE II 2018**

Estudio exploratorio para evaluar la eficacia del producto AB-CYSCARE en la variación de patógenos en infecciones urinarias recurrentes. Prod. Alimenticio. Promoter: AB-BIOTICS S. A. PI: Lluís Amat Tardiu. Service: Gynaecology and obstetrics. Start: 09/07/2018. Commercial

#### **MNK14112096**

Estudio multicéntrico, aleatorizado, de grupos paralelos, doble ciego, de dosis múltiples y controlado con placebo para evaluar la eficacia y la seguridad de MNK-1411 en varones de 4 a 8 años de edad con distrofia muscular de Duchenne. Phase II. Promoter: Mallinckrodt ARD Inc. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 29/06/2018. Commercial

#### **54767414ALL2005**

Ensayo abierto, multicéntrico, de fase 2 que evalúa la eficacia y seguridad de Daratumumab en pacientes pediátricos y jóvenes adultos entre ≥1 y <30 años con leucemia linfoblástica aguda o linfoma linfoblástico de precursores de células B y T en recaída o refractarios al tratamiento. Phase II. Promoter: Janssen-Cilag International NV. PI: Susana Rives Solà. Service: Oncology. Start: 20/06/2018. Commercial

#### **CAIN457A2311**

Ensayo aleatorizado, abierto, multicéntrico para evaluar la eficacia de secukinumab subcutáneo después de doce semanas de tratamiento y evaluar la seguridad, tolerabilidad y eficacia a largo plazo en pacientes de 6 a menos de 18 años de edad con psoriasis crónica en placas de moderada a grave. Phase III. Promoter: Novartis Farmacéutica,

S. A. PI: Mº Asunción Vicente Villa. Service: Dermatology. Start: 08/06/2018. Commercial

#### **FAST REGISTRY**

Estudio prospectivo observacional sobre cohorte de fetos con Flutter auricular o taquicardia supraventricular. EPA-SP. Promoter: SickKids. PI: Eduard Gratacós Solsona. Service: Gynaecology and obstetrics. Start: 06/06/2018. Non Commercial

#### **NYADAPETRICS**

Ensayo clínico fase I, piloto, doble-cego, aleatorizado, controlado con placebo y enmascarado, para evaluar la tolerabilidad y inmunogenicidad del probiótico Nyaditum Resae® administrado en población pediátrica en contacto con la tuberculosis con o sin infección tuberculosa latente. Prod. Alimenticio. Promoter: Manresana de Micobacteriología S. L. PI: Antoni Noguera Julián. Service: Paediatrics. Start: 28/05/2018. Commercial

#### **AI444-423**

Ensayo abierto y con un solo grupo para evaluar la farmacocinética, la seguridad y la eficacia de daclatasvir (DCV) en combinación con sofosbuvir (SOF) en niños de entre 3 y menos de 18 años con infección por los genotipos del 1 al 6 del virus de la hepatitis C crónica (HCC). Phase II. Promoter: Bristol-Myers Squibb International Corporation (BMSIC). PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 15/05/2018. Commercial

#### **IntReALL HR 2010**

A randomized Phase III Study Conducted by the Resistant Disease Committee of the International BFM Study Group. Phase III. Promoter: Sociedad Española de Hematología y Oncología Pediátricas.

PI: Susana Rives Solà. Service: Oncology. Start: 23/03/2018. Non Commercial

#### **CA40192**

Estudio de Fase I, abierto y aleatorizado para evaluar la farmacocinética, la farmacodinamia y la seguridad de etrolizumab seguido de una extensión abierta y una vigilancia de la seguridad en pacientes pediátricos de entre 4 y menos de 18 años de edad con colitis ulcerosa o enfermedad de crohn moderada o grave. Phase I. Promoter: Roche Farma, S. A. PI: Javier Martín De Carpi. Service: Gastroenterology, hepatology and nutrition. Start: 21/03/2018. Commercial

#### **BP39055**

A two part seamless multi-center randomized placebo controlled study to investigate the safety, tolerability, pharmacokinetics, pharmacodynamics and efficacy of R07034067 in type 2 and 3 spinal muscular atrophy patients. Phase II. Promoter: F. Hoffmann- La Roche Ltd. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 14/03/2018. Commercial

#### **CMX001-999**

Un estudio abierto de dos brazos, paralelo, aleatorio, randomizado, multicéntrico para evaluar la seguridad, tolerabilidad y la actividad antiviral de BCV versus el cuidado habitual para el tratamiento de las infecciones de adenovirus en los receptores de trasplantes de células madre hematopoiética pediátrica. Phase II. Promoter: Chimerix, Inc. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 13/03/2018. Commercial

#### **BAY-FAC-2017-01**

Estudio de fase IV multinacional para evaluar el patrón de tratamiento en la

“vida real” en pacientes con hemofilia A tratados previamente que reciben KOVALTRY (octocog alfa) para la profilaxis habitual. TAURUS. EPA-SP. Promoter: Bayer Hispania, S. L. U. PI: Ruben Berrueto Moreno. Service: Clinical haematology. Start: 07/03/2018. Commercial

#### ALXN1210-aHUS-312

A Phase 3, Open-Label, Multicenter Study of ALXN1210 in Children and Adolescents with Atypical Hemolytic Uremic Syndrome (aHUS). Phase III. Promoter: Alexion Pharmaceuticals Inc. PI: Alvaro Madrid Aris. Service: Nephrology. Start: 06/03/2018. Commercial

#### Y-MABS 201

Ensayo de Fase pivotal 2 del anticuerpo Naxitamab (hu3F8) y factor estimulante de colonias de granulocitos y macrófagos (GM-CSF) en pacientes con Neuroblastoma de alto riesgo con enfermedad refractaria primaria o con respuesta incompleta al tratamiento de rescate en el hueso y/o médula ósea. Phase III. Promoter: Y-mAbs Therapeutics A/S. PI: Jaume Mora Graupera. Service: Oncology. Start: 05/03/2018. Commercial

#### CPZP034X2203 / VEG116731

Estudio fase II de pazopanib (GW786034, N° NSC 737754) en niños, adolescentes y adultos jóvenes con tumores sólidos refractarios. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Jaume Mora Graupera. Service: Oncology. Start: 21/02/2018. Commercial

#### ITCC-059

A phase I/II study of Inotuzumab Ozogamicin as a single agent and in combination with chemotherapy for pediatric CD22-positive relapsed/refractory Acute Lymphoblastic Leukemia Study ITCC-059. Phase I. Promoter:

Erasmus MC. PI: Susana Rives Solà. Service: Oncology. Start: 15/02/2018. Non Commercial

#### HSJD-HR-NB-DHA

Interventional, open label, single arm, institutional study to assess Docosahexaenoic acid triglyceride (DHA-TG) supplementation combined with induction chemotherapy in patients with high-risk neuroblastoma. Prod. Alimenticio. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Jaume Mora Graupera. Service: Oncology. Start: 12/02/2018. Non Commercial

#### TAMDM

Tamoxifen in Duchenne muscular dystrophy: A multicenter, randomised, double-blind, placebo-controlled, phase 3 safety and efficacy 48-week trial. Phase III. Promoter: Universitätskinderhospital beider Basel (UKBB). PI: Andrés Nascimento Osorio. Service: Neurology. Start: 02/02/2018. Non Commercial

#### 602041 NEOVANC

Estudio multicéntrico aleatorizado, abierto, fase IIb para comparar la eficacia, seguridad y farmacocinética (PK) de la dosis óptima y la dosis estándar de vancomicina en neonatos y niños de < a 90 días de vida con sospecha de sepsis o sepsis confirmada por gérmenes gram-positivos. NeoVanc. Phase II. Promoter: Fondazione Penta Onlus. PI: Ana Alarcon Allen. Service: Neonatology. Start: 30/01/2018. Non Commercial

#### XLH-QoL-2017-02

Carga de la enfermedad de la hipofosfatemia ligada al cromosoma X. No EPA. Promoter: KYOW KIRIN FARMACÉUTICA, S. L. U. PI: Silvia Marin Del Barrio. Service: Endocrinology. Start: 17/01/2018. Commercial

#### SED-INS-2017-01

Registro Nacional de Pacientes con Diabetes tipo 1. EPA-OD. Promoter: Sociedad Española de Diabetes. PI: Roque Cardona Hernandez. Service: Endocrinology. Start: 15/01/2018. Non Commercial

#### ZX008-1503

Ensayo de ampliación sin enmascaramiento para evaluar la seguridad a largo plazo de la solución oral ZX008 (clorhidrato de fenfluramina) como tratamiento complementario en niños y adultos jóvenes con síndrome de Dravet. Phase III. Promoter: Zogenix, Inc. PI: Alia Ramirez Camacho. Service: Neurology. Start: 12/01/2018. Commercial

#### EuroNet-PHL-C2

Second International Inter-Group Study for Classical Hodgkin's Lymphoma in Children and Adolescents. Phase III. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI: Veronica Celis Passini. Service: Oncology. Start: 12/01/2018. Non Commercial

#### GS-US-205-1850

Ensayo aleatorizado y doble ciego de fase IIIb para evaluar la seguridad y la eficacia de 2 regímenes de tratamiento con Aztreonam 75 mg polvo y disolvente para solución para inhalación por nebulizador / Aztreonam solución para inhalación (AZLI) en pacientes pediátricos con fibrosis quística (FQ) e infección/colonización de las vías respiratorias por Pseudomonas aeruginosa (PA) de nueva aparición. Phase III. Promoter: Gilead Sciences, Inc. PI: Jordi Costa i Colomer. Service: Paediatrics. Start: 21/12/2017. Commercial

#### ESTEVE-SANF-201

Phase I/II safety, tolerability and initial efficacy study of adeno-associated

viral vector serotype 9 containing human sulfamidase gene after intracerebroventricular administration to patients with MPSIIIA. (ESTEVE-SANF-201). Phase II. Promoter: Esteve Pharmaceuticals, S. A. PI: Mercè Pineda Marfà. Service: Neurology. Start: 04/12/2017. Commercial

#### NN7088-3908

Seguridad y eficacia de turoctocog alfa pegol (N8-GP) en pacientes con hemofilia A no tratados previamente. Phase III. Promoter: Novo Nordisk a/s. PI: Ruben Berrueto Moreno. Service: Clinical haematology. Start: 23/11/2017. Commercial

#### CP-4-006

A phase 3, open-label, randomized, multicenter, 12 months, efficacy and safety study of weekly MOD-4023 compared to daily Genotropin - therapy in pre-pubertal children with growth hormone deficiency. Phase III. Promoter: OPKO Biologics Ltd. PI: Lourdes Ibañez Toda. Service: Endocrinology. Start: 10/11/2017. Commercial

#### BMN111-301

A Phase 3 Randomized, Double-Blind, Placebo-Controlled, Multicenter Study to Evaluate the Efficacy and Safety of BMN 111 in Children with Achondroplasia. Phase III. Promoter: BioMarin Pharmaceutical Inc. PI: Rosendo Ullot Font. Service: Orthopaedics and traumatology. Start: 10/11/2017. Commercial

#### ET-13-002

SIOP Ependimoma II - Programa clínico internacional para el diagnóstico y tratamiento de niños, adolescentes y adultos jóvenes con ependimoma. Phase III. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI:



Ofelia Cruz Martínez. Service: Oncology.  
Start: 06/11/2017. Non Commercial

#### NI-0501-06

Estudio piloto abierto, multicéntrico y de un solo grupo para evaluar la seguridad, tolerabilidad, farmacocinética y eficacia de administraciones intravenosas de NI-0501, un anticuerpo monoclonal anti-interferón gamma (anti-IFN $\gamma$ ), en pacientes con artritis idiopática juvenil de inicio sistémico (AIJIS) que desarrollan síndrome de activación del macrófago/HLH secundaria (SAM/HLHs). Phase II. Promoter: Swedish Orphan Biovitrum AB (publ). PI: Jordi Antón López. Service: Paediatrics. Start: 06/11/2017. Commercial

#### M16-123

An Open-Label, Multicenter Study to Evaluate the Pharmacokinetics, Safety, and Efficacy of Glecaprevir/Pibrentasvir in Pediatric Subjects with Genotypes 1 & 6 Chronic Hepatitis C Virus (HCV) Infection. Phase III. Promoter: "AbbVie Deutschland GmbH & Co. KG "AbbiVie"". PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 03/11/2017. Commercial

#### FSJD-TRACIP-2017

Tratamiento del crecimiento intrauterino restringido precoz con heparina de bajo peso molecular: Ensayo Clínico Randomizado (TRACIP). Phase III. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Eudene Mazarico Gallego. Service: Gynaecology and obstetrics. Start: 02/11/2017. Non Commercial

#### EFC14153

Estudio aleatorizado, doble ciego, controlado con placebo, de grupos paralelos para evaluar la eficacia y seguridad de Dupilumab en niños de 6 a <12 años con asma persistente

no controlada. Phase III. Promoter: Sanofi-Aventis S. A. PI: María Cols Roig. Service: Paediatrics. Start: 18/10/2017. Commercial

#### MK-6072-0001

Ensayo clínico aleatorizado, doble ciego y controlado con placebo para evaluar la seguridad, la tolerabilidad, la farmacocinética y la eficacia de una única infusión de bezlotoxumab (MK-6072, anticuerpo monoclonal humano frente a la toxina B de C. difficile) en niños y adolescentes de 1 a menos de 18 años que reciben tratamiento antibacteriano para la infección por C. difficile (MODIFY III). Phase III. Promoter: Merck, Sharp & Dohme de España. S. A. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 09/10/2017. Commercial

#### FAE-PRO-2015-01

Registro de Hemangiomas de la AEDV. EPA-OD. Promoter: Fundación Piel Sana Academia Española de Dermatología y Venereología. PI: M<sup>o</sup> Asunción Vicente Villa. Service: Dermatology. Start: 03/10/2017. Non Commercial

#### WN40227 (CN001016)

Estudio aleatorizado, doble ciego y controlado con placebo para evaluar la eficacia, la seguridad y la tolerabilidad de BMS-986089 en niños ambulatorios con distrofia muscular de Duchenne. Phase III. Promoter: F. Hoffmann - La Roche Ltd. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 28/09/2017. Commercial

#### FSJD-RTB-2015

Ensayo clínico fase I, unicéntrico, abierto y de escalado de dosis, para evaluar la seguridad y la actividad del adenovirus oncolítico VCN-01 en pacientes con retinoblastoma refractario. Phase I. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu

- FSJD FSJD. PI: Jaume Català Mora. Service: Ophthalmology. Start: 06/09/2017. Non Commercial

#### MK7625A-035

Ensayo clínico fase 2, multicéntrico, aleatorizado, doble ciego y controlado con comparador activo para evaluar la seguridad y la eficacia de ceftolozano/tazobactam (MK-7625A) más Metronidazol versus Meropenem en sujetos pediátricos con infección intraabdominal complicada. Phase II. Promoter: Merck, Sharp & Dohme de España. S. A. PI: Antoni Noguera Julián. Service: Paediatrics. Start: 31/08/2017. Commercial

#### NOPHO-DBH AML 2012

Research study for treatment of children and adolescents with acute myeloid leukaemia 0-18 years. Phase III. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI: Susana Rives Solà. Service: Oncology. Start: 24/08/2017. Non Commercial

#### CV185-362

Estudio prospectivo, aleatorizado, abierto y multicéntrico de la seguridad y farmacocinética de Apixaban comparado con un antagonista de la vitamina K o HBPM en sujetos pediátricos con cardiopatías congénitas o adquiridas que requieren anticoagulación crónica para la prevención del tromboembolismo. Phase II. Promoter: Bristol-Myers Squibb International Corporation (BMSIC). PI: Ruben Berruero Moreno. Service: Clinical haematology. Start: 01/08/2017. Commercial

#### 4045-301

Estudio doble ciego, controlado con placebo y multicéntrico con una extensión abierta para evaluar la eficacia y la seguridad de SRP-4045 y SRP-4053 en

pacientes con distrofia muscular de Duchenne. Phase III. Promoter: Sarepta Therapeutics, Inc. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 19/07/2017. Commercial

#### CART19-BE-01

Estudio piloto de la infusión de linfocitos T autólogos modificados genéticamente para expresar anti-CD19 en pacientes con leucemia o linfoma CD19+ resistente o refractario a tratamiento. Phase I. Promoter: Hospital Clínic Barcelona. PI: Susana Rives Solà. Service: Oncology. Start: 04/07/2017. Non Commercial

#### NF1-EXCEL

Efecto de la lamotrigina en déficits cognitivos asociados con la neurofibromatosis tipo 1: un estudio de fase II aleatorio, controlado con placebo y llevado a cabo en diversos centros (NF1-EXCEL). Phase II. Promoter: Erasmus MC. PI: Federico Ramos. Service: Neurology. Start: 03/07/2017. Non Commercial

#### I1F-MC-RHCD

Estudio multicéntrico, doble ciego, aleatorizado, comparado con placebo, en el que se evalúan la seguridad, la tolerabilidad y la eficacia de ixekizumab en pacientes con psoriasis en placas de moderada a grave y entre 6 y < 18 años. Phase III. Promoter: Lilly S. A. PI: M<sup>o</sup> Asunción Vicente Villa. Service: Dermatology. Start: 30/06/2017. Commercial

#### PHA-HEM-2017-01

Estudio de mercado sobre los diferentes tratamientos para la hemofilia A en la práctica clínica habitual. EPA-OD. Promoter: Phamax AG. PI: Ruben Berruero Moreno. Service: Clinical haematology. Start: 16/06/2017. Commercial

#### CA209744

Ensayo de fase II abierto, basado en riesgo, adaptado a la respuesta, de nivolumab + brentuximab vedotina (N+Bv) en niños, adolescentes y adultos jóvenes con linfoma de Hodgkin clásico (LHc) CD30+ en recidiva/refractario (R/R) después del fracaso del tratamiento de primera línea seguido de brentuximab+bendamustina (Bv+B) en participantes con una respuesta subóptima bendamustine (Bv + B) for participants with a suboptimal response. CheckMate 744: CHECKpoint pathway and nivolumab clinical Trial Evaluation. Phase II. Promoter: Bristol-Myers Squibb International Corporation (BMSIC). PI: Jaume Mora Graupera. Service: Oncology. Start: 14/06/2017. Commercial

#### UX007G-CL202

Estudio de extensión abierto para evaluar la seguridad y eficacia a largo plazo de UX007 en sujetos con síndrome de deficiencia del transportador de glucosa de tipo 1 (SD Glut1). Phase II. Promoter: Ultragenyx Pharmaceutical, Inc. PI: M<sup>a</sup> Angels García Cazorta. Service: Neurology. Start: 05/06/2017. Commercial

#### FSJD-PMM2-2016

Ensayo clínico fase II, para evaluar la efectividad y la seguridad de la acetazolamida en el tratamiento del síndrome cerebeloso en pacientes con deficiencia de PMM2-CDG. Phase II. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Mercedes Serrano Gimarré. Service: Neurology. Start: 02/06/2017. Non Commercial

#### DSC/14/2357/48

Estudio multicéntrico, aleatorizado, doble ciego, controlado con placebo para evaluar la eficacia y la seguridad de givinostat

en pacientes ambulatorios con distrofia muscular de duchenne (DMD). Phase III. Promoter: Italfarmaco, S. A. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 31/05/2017. Commercial

#### CAIN457F2304

Estudio aleatorizado, doble ciego, controlado con placebo, compuesto de tres partes para investigar la eficacia y seguridad del tratamiento con secukinumab en subtipos de artritis idiopática juvenil de la artritis psoriásica y artritis relacionada con entesitis. Phase I. Promoter: Novartis Farmacéutica, S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 28/05/2017. Commercial

#### CA209-908

Ensayo clínico fase Ib/II de nivolumab en monoterapia y nivolumab en combinación con ipilimumab en sujetos pediátricos con neoplasias malignas primarias del SNC de alto grado. Phase II. Promoter: Bristol-Myers Squibb International Corporation (BMSIC). PI: Jaume Mora Graupera. Service: Oncology. Start: 22/05/2017. Commercial

#### CL010\_168

A Randomized, Double-Blind, Placebo-Controlled, Phase 3 Study to Evaluate the Safety and Efficacy of CCX168 (Avacopan) in Patients with Anti-Neutrophil Cytoplasmic Antibody (ANCA)-Associated Vasculitis Treated Concomitantly with Rituximab or Cyclophosphamide/Azathioprine. Phase III. Promoter: Chemocentryx. PI: Jordi Antón López. Service: Paediatrics. Start: 19/05/2017. Commercial

#### GEIS-39

Phase II trial of nab-paclitaxel for the treatment of desmoid tumors and multiply relapsed/refractory desmoplastic small round cell tumors and Ewing sarcoma.

Phase II. Promoter: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. Service: Oncology. Start: 18/05/2017. Non Commercial

#### SNT-III-012

A Phase III Double-blind, Randomized, Placebo-Controlled Study assessing the Efficacy, Safety and Tolerability of Idebenone in Patients with Duchenne Muscular Dystrophy Receiving Glucocorticosteroids - SIDEROS. Phase III. Promoter: Santhera Pharmaceuticals (Switzerland) Ltd., PI: Andrés Nascimento Osorio. Service: Neurology. Start: 12/05/2017. Commercial

#### HOSPIGRIP-2016

Estudio de la infección por virus influenza en niños hospitalizados en España en dos estaciones gripales consecutivas. Análisis de costos médicos directos. No EPA. Promoter: Asociación Instituto Bionostia. PI: Juan José García García. Service: Paediatrics. Start: 10/05/2017. Non Commercial

#### H2020-PHC-18-2015-667224

Efecto del Alopurinol en pacientes con encefalopatía hipóxica-isquémica tratados con hipotermia. ALBINO ensayo clínico multicéntrico, aleatorizado, ciego, controlado con placebo. Phase III. Promoter: Instituto de investigación Sanitaria Hospital La Fe. PI: Marta Camprubi Camprubi. Service: Neonatology. Start: 05/05/2017. Non Commercial

#### REACT RCT

Evaluation of Real Time Continuous Glucose Monitoring in the very preterm infant. Prod. Sanitario. Promoter: University of Cambridge. PI: Isabel Iglesias Platas. Service: Neonatology. Start: 04/05/2017. Non Commercial

#### CCTL019B2001X

Protocolo de tratamiento expandido de CTL019 para pacientes pediátricos/adultos jóvenes con leucemia linfoblástica aguda en recaída/refractaria. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Susana Rives Solà. Service: Oncology. Start: 21/04/2017. Commercial

#### LCH-IV-042011

LCH-IV International Collaborative Treatment Protocol for Children and Adolescents with Langerhans Cell Histiocytosis. Phase III. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI: Vicente Santa-María López. Service: Oncology. Start: 30/03/2017. Non Commercial

#### E2007-G000-311

Estudio abierto y multicéntrico con una fase de extensión para evaluar la seguridad, la tolerabilidad y la relación exposición-eficacia de perampanel en suspensión oral cuando se administra como tratamiento complementario en sujetos pediátricos (de 4 a menos de 12 años) con crisis de inicio parcial mal controladas o crisis tónico-clónicas generalizadas primarias. Phase III. Promoter: Eisai Limited. PI: María Victoria San Antonio Arce. Service: Neurology. Start: 22/03/2017. Commercial

#### GWEP1521

Estudio aleatorizado doble ciego controlado con placebo para investigar la eficacia y la seguridad del cannabidiol (GWP42003-P, CBD) como terapia complementaria en pacientes con complejo de esclerosis tuberosa que sufren crisis epilépticas inadecuadamente controladas. Phase III. Promoter: GW Research. PI: Federico Ramos. Service: Neurology. Start: 20/02/2017. Commercial

#### **NN7999-3895**

Seguridad y eficacia de nonacog beta pegol (N9-GP) en pacientes con hemofilia B no tratados previamente. Phase III. Promoter: Novo Nordisk a/s. PI: Ruben Berrueto Moreno. Service: Clinical haematology. Start: 08/02/2017. Commercial

#### **109MS306**

Estudio abierto, aleatorizado, multicéntrico, de múltiples dosis, comparativo con fármaco activo y con grupos paralelos, sobre la eficacia y la seguridad de BG00012 en niños de 10 a menos de 18 años de edad con esclerosis múltiple recurrente-remitente, con extensión abierta opcional. Phase III. Promoter: Biogen IDEC Research LTD. PI: Thais Armangue Salvador. Service: Neurology. Start: 30/12/2016. Commercial

#### **15-007**

A Phase 3, Randomized, Adaptive Study Comparing the Efficacy and Safety of Defibrotide vs Best Supportive Care in the Prevention of Hepatic Veno-Occlusive Disease in Adult and Pediatric Patients Undergoing Hematopoietic Stem Cell Transplant. Phase III. Promoter: Jazz Pharmaceuticals, INC. PI: Isabel Badell Serra. Service: Oncology. Start: 30/12/2016. Commercial

#### **PURPOSE / ABR54654**

Ensayo controlado con placebo en sujetos con alto riesgo para la psicosis comparado con ácidos grasos omega-3 en Europa. Phase III. Promoter: University Medical Center Utrecht. PI: Montserrat Dolz Abadia. Service: Psychiatry and psychology. Start: 05/12/2016. Non Commercial

#### **EFC14028**

Estudio de fase III aleatorizado, multicéntrico, multinacional y doble ciego

que compara la eficacia y la seguridad de infusiones bisemanales de neoGAA (GZ402666) y alglucosidasa alfa en pacientes con enfermedad de Pompe de inicio tardío sin tratamiento previo. Phase III. Promoter: Sanofi-Aventis S. A. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 30/11/2016. Commercial

#### **11/0499 (MYPAN)**

Ensayo clínico abierto controlado y aleatorizado de micofenolato mofetil (MMF) frente a ciclofosfamida para inducir la remisión en poliarteritis nodosa juvenil (PAN). Phase III. Promoter: University College London. PI: Jordi Antón López. Service: Paediatrics. Start: 25/11/2016. Non Commercial

#### **111-901**

Estudio clínico multicéntrico y multinacional de evaluación sobre pacientes pediátricos con acondroplasia. No EPA. Promoter: BioMarin Pharmaceutical Inc. PI: Rosendo Ullot Font. Service: Orthopaedics and traumatology. Start: 23/11/2016. Commercial

#### **DRI13926**

Estudio abierto, secuencial, de búsqueda de dosis ascendentes y repetidas de sarilumab administrado con inyección subcutánea (s.c.) a niños y adolescentes con edades comprendidas entre 1 y 17 años con artritis idiopática juvenil sistémica (AIJS), seguido de una fase de extensión. Phase II. Promoter: Sanofi-Aventis S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 25/10/2016. Commercial

#### **FSJD-ISON-2016**

Ensayo clínico abierto fase IIA de estudio de la absorción de una suspensión de isoniazida a 10 mg/mL para el tratamiento

de la infección tuberculosa en pacientes menores de 6 años. Phase II. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Antoni Noguera Julián. Service: Paediatrics. Start: 10/10/2016. Non Commercial

#### **54179060LYM3003**

A Randomized, Open-label, Safety and Efficacy Study of Ibrutinib in Pediatric and Young Adult Patients With Relapsed or Refractory Mature B-cell non-Hodgkin Lymphoma. Phase III. Promoter: Janssen Research and development LCC. PI: Veronica Celis Passini. Service: Oncology. Start: 05/10/2016. Commercial

#### **WA28029**

Estudio fase IV para evaluar la disminución de la frecuencia de la dosis en pacientes con artritis idiopática juvenil sistémica activa (aijs) que experimenten anomalías de laboratorio durante el tratamiento con Tocilizumab. Phase IV. Promoter: F. Hoffmann-La Roche Ltd. PI: Jordi Antón López. Service: Paediatrics. Start: 24/08/2016. Commercial

#### **PENTA 20**

Ensayo aleatorizado de tratamiento antirretroviral con dolutegravir (DTG) comparado con el tratamiento estándar (TE) en niños con infección VIH que inician tratamiento de primera línea o de segunda línea. Phase III. Promoter: Fondazione Penta Onlus. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 23/08/2016. Non Commercial

#### **BRI-EBP-2015-01**

Prácticas Europeas para el manejo de la punción dural accidental en obstetricia (Estudio EPIMAP Obstetrics). EPA-SP. Promoter: European Society of Anaesthesiology. PI: Alejandro Romero

Fernández. Service: Anesthesia. Start: 22/07/2016. Non Commercial

#### **ALXN1101-MCD-202**

Estudio abierto, multicéntrico y multinacional, de fase II/III, para evaluar la eficacia y la seguridad de ALXN1101 en recién nacidos con déficit del cofactor molibdeno (DCMo) de tipo A. Phase III. Promoter: Alexion Pharma Spain, S. L. PI: Carme Fons Estupiña. Service: Neurology. Start: 28/06/2016. Commercial

#### **MER-ACA-2015-01**

Evaluación del coste-efectividad del tratamiento con inmunoterapia en pacientes diagnosticados de asma con o sin rino-conjuntivitis por sensibilización a ácaros en España. EPA-SP. Promoter: Merck S. L. U. PI: Ana Mª Plaza Martín. Service: Allergy and clinical immunology. Start: 16/06/2016. Commercial

#### **FSJD-DIPG-DC**

Phase Ib Clinical Trial on the Safety of Immunotherapy With Autologous Dendritic Cells Primed With Lysate Allogeneic Tumor Lines in Patients With Diffuse Intrinsic Pontine Glioma (DIPG). Phase I. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Andrés Morales La Madrid. Service: Oncology. Start: 06/06/2016. Non Commercial

#### **IM101240**

"Registro observacional de Abatacept en pacientes con Artritis Idiopática Juvenil". EPA-LA. Promoter: Bristol-Myers Squibb, S. A. U. PI: Jordi Antón López. Service: Paediatrics. Start: 03/06/2016. Commercial

#### **LIN-DOX-2015-01**

Estudio sobre las condiciones de uso de Cariban® y de otros antieméticos en el

tratamiento de las náuseas y vómitos en el embarazo: Estudio prospectivo observacional longitudinal. EPA-SP. Promoter: Laboratorios Inibsa, S. A. PI: Jose Maria Boguñá Ponsa. Service: Gynaecology and obstetrics. Start: 02/06/2016. Commercial

#### **PR314/15**

Evaluación mediante hormona antimulleriana (AMH) de la reserva ovárica tras el tratamiento de los endometriomas mediante esclerosis con alcohol o cirugía. Estudio Piloto. No EPA. Promoter: García Tejedor, Amparo. PI: Santiago González Nuñez. Service: Gynaecology and obstetrics. Start: 01/06/2016. Non Commercial

#### **ISIS396443-CS11**

Estudio de extensión abierto para pacientes con atrofia muscular espinal que han participado previamente en estudios experimentales de ISIS 396443. Phase III. Promoter: Ionis Pharmaceuticals, Inc. PI: Andrés Nascimento Osorio. Service: Neurology. Start: 30/05/2016. Commercial

#### **ALX-HPP-501**

Registro observacional, longitudinal, prospectivo, a largo plazo de Pacientes con Hipofosfatasa (HPP). EPA-SP. Promoter: Alexion Pharmaceuticals Inc. PI: Rosa Bou i Torrent. Service: Paediatrics. Start: 17/05/2016. Commercial

#### **20110261**

Estudio Fase 1, multicéntrico, abierto de de-escalado de dosis para evaluar la seguridad y eficacia de Talimogene Laherparepvec en pacientes pediátricos con tumores avanzados que no sean del sistema nervioso central y que sean tratables con inyección directa. Phase I. Promoter: Amgen Inc. PI: Jaume Mora

Graupera. Service: Oncology. Start: 11/05/2016. Commercial

#### **DRI13925**

Estudio abierto, secuencial, de búsqueda de dosis ascendentes y repetidas de sarilumab administrado con inyección subcutánea (s. c. ) a niños y adolescentes con edades comprendidas entre 2 y 17 años con artritis idiopática juvenil de curso poliarticular (AIJP), seguido de una fase de extensión. Phase II. Promoter: Sanofi-Aventis S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 09/05/2016. Commercial

#### **EFC13957**

EDITION JUNIOR ? ?Estudio de 6 Meses, Multicéntrico, Aleatorizado, Abierto, de 2 Brazos Paralelos, para Comparar Eficacia y Seguridad de una Nueva Formulación de Insulina Glargina y Lantus® Inyectado Una Vez al Día en Niños y Adolescentes entre 6-17 años con Diabetes Mellitus Tipo 1, con un Periodo de Extensión de Seguridad de 6 meses?. Phase III. Promoter: Sanofi-Aventis S. A. PI: Larisa Suarez Ortega. Service: Endocrinology. Start: 06/05/2016. Commercial

#### **GTI1503**

Ensayo multicéntrico, abierto y con un solo grupo para evaluar la eficacia, la farmacocinética, la seguridad y la tolerabilidad de IGSC al 20 % en sujetos con inmunodeficiencia primaria. Phase III. Promoter: Instituto Grifols, S. A. PI: Laia Alsina Manrique de Lara. Service: Allergy and clinical immunology. Start: 05/05/2016. Commercial

#### **TBTC S31-A5349**

Ensayo clínico fase III, randomizado, abierto y controlado para evaluar el acortamiento del tratamiento de la tuberculosis pulmonar en regímenes con rifapentina. Phase III. Promoter:

Tuberculosis Trial Consortium. PI: Antoni Noguera Julián. Service: Paediatrics. Start: 28/04/2016. Non Commercial

#### **SIOP - PNET 5 MB - LR**

Estudio prospectivo internacional en niños mayores de 3 a 5 años con meduloblastoma de riesgo estándar clínico de perfil biológico de bajo riesgo. Phase II. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI: Ofelia Cruz Martínez. Service: Oncology. Start: 25/04/2016. Non Commercial

#### **M14-748**

Estudio abierto, multicéntrico para evaluar la farmacocinética, la seguridad y la eficacia de ombitasvir (OBV), paritaprevir (PTV), ritonavir (RTV), con o sin dasabuvir (DSV) y con o sin ribavirina (RBV) en pacientes pediátricos con infección crónica por el virus de la hepatitis C (VHC) genotipo 1 o 4 [ZIRCON]. Phase III. Promoter: Abbvie Spain, S. L. U. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 17/02/2016. Commercial

#### **FSJD-PIOSPIMET-2015**

Ensayo clínico prospectivo, abierto, aleatorizado, con dos grupos de pacientes para evaluar la eficacia y la seguridad de la combinación de Etinil-estradiol y levonorgestrel versus la combinación de pioglitazona, espironolactona y metformina a dosis bajas en adolescentes con hiperandrogenismo ovárico e hiperinsulinismo: efectos sobre la función ovulatoria, parámetros de inflamación crónica, marcadores de evolución y efectividad del tratamiento, y de desarrollo de diabetes tipo 2. Phase III. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Lourdes Ibañez Toda. Service: Endocrinology. Start: 11/02/2016. Non Commercial

#### **M11-001**

Un estudio observacional, no intervencionista, multicéntrico y multinacional de pacientes con Síndrome Hemolítico Urémico Atípico (registro de SHUa). EPA-LA. Promoter: Alexion Pharmaceuticals Inc. PI: María Ramos Cebrián. Service: Nephrology. Start: 25/01/2016. Commercial

#### **UX0076-CL201**

Un ensayo clínico doble ciego, controlado con placebo, de grupos paralelos para evaluar la seguridad y la eficacia de UX007 en sujetos con síndrome por deficiencia del transportador de glucosa de tipo 1. Phase II. Promoter: Ultragenyx Pharmaceutical, Inc. PI: M<sup>a</sup>Angels García Cazorla. Service: Neurology. Start: 07/01/2016. Commercial

#### **A0081042**

Estudio multicéntrico, de grupos paralelos, en doble ciego y controlado con placebo, para evaluar la eficacia y la seguridad de la Pregabalina como tratamiento adyuvante en niños de 1 mes a menos de 4 años de edad con crisis epilépticas parciales. Phase III. Promoter: PFIZER INC. PI: María Victoria San Antonio Arce. Service: Neurology. Start: 04/01/2016. Commercial

#### **CCTL019A2205B**

Seguimiento a largo plazo (LTFU) de pacientes tratados con la terapia celular CART dirigida a CD19 basada en vectores lentivirales. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Susana Rives Solà. Service: Oncology. Start: 22/12/2015. Commercial

#### **CCTL019B2202**

Ensayo de fase II, de un solo grupo, multicéntrico, para determinar la eficacia y la seguridad de CTL019 en pacientes

pediátricos con leucemia linfoblástica aguda de células B en recaída y refractaria. Phase II. Promoter: Novartis Farmacèutica, S. A. PI: Susana Rives Solà. Service: Oncology. Start: 21/12/2015. Commercial

#### **PENTA 17**

Estrategia para el mantenimiento de la supresión del VIH en niños con elvitegravir + darunavir/ritonavir. Phase II. Promoter: Fondazione Penta Onlus. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 09/12/2015. Non Commercial

#### **P11-292 / ABB-ADA-2014-01**

Un registro no intervencionista a largo plazo para evaluar la seguridad y efectividad de HUMIRA® (adalimumab) en pacientes pediátricos con enfermedad de Crohn (EdC) activa entre moderada y grave (CAPE). EPA-LA. Promoter: "AbbiVie Deutschland GmbH & Co.KG "AbbiVie". PI: Javier Martín De Carpi. Service: Gastroenterology, hepatology and nutrition. Start: 28/10/2015. Commercial

#### **SJD-RGH-2015-01**

Tratamiento con hormona de crecimiento recombinante (rGH) en pacientes con déficit de GH controlados en el Hospital Sant Joan de Déu: Análisis de marcadores precoces de buena respuesta de utilidad en la práctica clínica. EPA-OD. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Marta Ramon Krauel. Service: Endocrinology. Start: 26/10/2015. Non Commercial

#### **CAIN457A2310**

Ensayo multicéntrico, aleatorizado, doble ciego, controlado con placebo y fármaco activo para demostrar la eficacia de secukinumab subcutáneo en comparación con placebo y etanercept (en un brazo

simple ciego) después de doce semanas de tratamiento y evaluar la seguridad, la tolerabilidad y la eficacia a largo plazo en pacientes de 6 a menos de 18 años de edad con psoriasis en placas crónica grave. Phase III. Promoter: Novartis Farmacèutica, S. A. PI: Antonia González Enseñat. Service: Dermatology. Start: 15/10/2015. Commercial

#### **1645-CI-057**

Recién nacidos con edad gestacional extremadamente baja asignados al azar, a ciegas, para ser resucitados resucitado con un 21% frente al 60% de oxígeno: influencia sobre la mortalidad y las enfermedades crónicas en el periodo neonatal. Phase III. Promoter: Instituto de investigación Sanitaria Hospital La Fe. PI: Martín Iriondo Sanz. Service: Neonatology. Start: 06/10/2015. Non Commercial

#### **AZA-JMML-001**

Estudio de Fase 2, multicéntrico, abierto para evaluar la farmacocinética, farmacodinámica, seguridad y actividad de azacitidina y para comparar azacitidina con los controles históricos en sujetos pediátricos con nuevo diagnóstico de síndrome mielodisplásico avanzado o leucemia mielomonocítica juvenil antes de trasplante de células madre hematopoyéticas. Phase II. Promoter: Celgene International II S. à. r. l. PI: Susana Rives Solà. Service: Oncology. Start: 21/09/2015. Commercial

#### **SCI-SPAI-2015-01**

Estudio PID-RSV: Estudio epidemiológico sobre la incidencia de infecciones respiratorias agudas (IRA) causadas por el virus respiratorio sincitial (VRS) que requieren hospitalización en poblaciones pediátricas con inmunodeficiencia primaria (IDP). No EPA. Promoter: Abbvie Spain, S. L. U. PI: Laia Alsina Manrique de Lara. Service: Allergy and clinical immunology. Start: 04/09/2015. Commercial

#### **PTC124-GD-021e-CF**

Estudio de extensión de fase 3 de ataluren (ptc124) en pacientes con fibrosis quística mediada por mutación terminadora. Phase III. Promoter: PTC Therapeutics, Inc. PI: Jordi Costa i Colomer. Service: Paediatrics. Start: 28/07/2015. Commercial

#### **BAY59-7939/17992**

Ensayo de dosis única para evaluar la formulación de rivaroxaban en polvo seco para suspensión oral, en niños de 6 meses a 12 años con trombosis previa. Phase I. Promoter: Bayer HealthCare AG. PI: Ruben Berruero Moreno. Service: Clinical haematology. Start: 16/07/2015. Commercial

#### **MK5592-097**

Estudio de la seguridad, tolerabilidad y farmacocinética de dos formulaciones de posaconazol (POS), por vía intravenosa (IV) y en polvo para suspensión oral, en pacientes pediátricos inmunodeprimidos con neutropenia. Phase I. Promoter: Merck, Sharp & Dohme de España. S. A. PI: Clàudia Fortuny Guasch. Service: Paediatrics. Start: 07/07/2015. Commercial

#### **FSJ-ADE-2015-01**

Estudio multicéntrico retrospectivo-prospectivo nacional del valor de los igras en el diagnóstico de la adenitis por micobacterias no tuberculosas y de sus distintas aproximaciones terapéuticas. No EPA. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Antoni Noguera Julián. Service: Paediatrics. Start: 01/07/2015. Non Commercial

#### **ORP-CYS-2014-01 (E-HOD-CSP)**

Protocolo de vigilancia de Cystadane de Orphan Europe, en colaboración

con la Red Europea y registro para las homocistinurias y defectos de metilación (E-HOD). EPA-LA. Promoter: Centro de Investigación Biomédica en Red (CIBER) Antiguo CIBERSAM. PI: Silvia Maria Meavilla Olivas. Service: Neurology. Start: 27/05/2015. Non Commercial

#### **IGR2012/1883**

Estudio fase I-II de Vinblastina en combinación con Nilotinib en niños, adolescentes, y adultos jóvenes con glioma de bajo grado refractario o recurrente. Phase II. Promoter: Gustave Roussy. PI: Ofelia Cruz Martínez. Service: Oncology. Start: 14/05/2015. Non Commercial

#### **RG 13-277**

International Randomised Controlled Trial of Chemotherapy for the Treatment of Recurrent and Primary Refractory Ewing Sarcoma. Phase II. Promoter: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. Service: Oncology. Start: 23/04/2015. Non Commercial

#### **SJD-ENT-2014-01**

Utilización de Entonox en procedimientos dolorosos fuera de quirófano. EPA-SP. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Marina Perelló Riera. Service: Anesthesia. Start: 07/04/2015. Non Commercial

#### **PFI-ETA-2013-01**

Influencia de la inmunogenicidad de los fármacos anti-TNF en la eficacia del tratamiento en Artritis Idiopática Juvenil usando la calprotectina sérica como marcador de actividad de la enfermedad. Estudio multicéntrico. "Estudio ITACA". EPA-SP. Promoter: Pfizer, S. L. U. PI: Jordi Antón López. Service: Paediatrics. Start: 19/02/2015. Commercial



#### **TBT-ANT-2014-01**

TBTC Estudio 36 - Plataforma de evaluación de los resultados del tratamiento de tuberculosis. Estudio observacional de los individuos tratados con tuberculosis pulmonar (TB). EPA-SP. Promoter: Agència de Salut Pública de Catalunya. PI: Antoni Noguera Julián. Service: Paediatrics. Start: 06/02/2015. Non Commercial

#### **BAY 81-8973/13400**

Estudio de Fase III, multicéntrico, abierto y no controlado para evaluar la seguridad de BAY 81-8973 en niños con hemofilia A grave en tratamiento profiláctico. Phase III. Promoter: Bayer Hispania, S.L.U. PI: Ruben Berruero Moreno. Service: Clinical haematology. Start: 09/01/2015. Commercial

#### **FSJD-FUNNY FRIENDS-2014**

Estudio de la eficacia de un programa de incentivación de fisioterapia respiratoria domiciliar mediante videojuego (Funny Friends) en pacientes pediátricos afectados de fibrosis quística. Phase II. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Enrique Del Campo Garcia-Ramos. Service: Rehabilitation and physical medicine. Start: 20/11/2014. Non Commercial

#### **W1191603**

Treatment with recombinant growth hormone (rGH) in short patients born small-for-gestational-age (SGA) followed at Hospital Sant Joan de Déu: Analysis of early predictors of good response useful in clinical practice. EPA-OD. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Lourdes Ibañez Toda. Service: Endocrinology. Start: 14/11/2014. Non Commercial

#### **LOR-HOR-2013-01**

"Identification of circulating microRNAs in short children born small for gestational age: Relationship with pre- and postnatal growth and metabolic parameters". EPA-SP. Promoter: López Bermejo, Abel. PI: Lourdes Ibañez Toda. Service: Endocrinology. Start: 24/10/2014. Non Commercial

#### **20101221**

Estudio abierto, de un solo brazo para evaluar la seguridad y eficacia a largo plazo de romiplostim en sujetos Pediátricos Trombocitopénicos con Púrpura Trombocitopénica Inmune (PTI). Phase III. Promoter: AMGEN S. A. PI: Ruben Berruero Moreno. Service: Clinical haematology. Start: 14/10/2014. Commercial

#### **20130173**

Estudio prospectivo, multicéntrico y de un solo brazo para evaluar la eficacia, seguridad y farmacocinética de denosumab en niños con osteogénesis imperfecta. Phase III. Promoter: AMGEN S. A. PI: Rosa Bou i Torrent. Service: Paediatrics. Start: 07/10/2014. Commercial

#### **BRF116013**

Estudio Fase I/IIa de dos partes, multicéntrico, abierto, con un solo grupo para determinar la seguridad, tolerabilidad y farmacocinética de dabrafenib oral en niños y adolescentes con tumores sólidos avanzados con mutación BRAF V600 positiva. Phase I. Promoter: Novartis Farmacéutica, S. A. PI: Jaume Mora Graupera. Service: Oncology. Start: 12/09/2014. Commercial

#### **BAY59-7939/14372**

Ensayo clínico multicéntrico, aleatorizado, abierto, controlado con comparador activo,

para evaluar la eficacia y la seguridad de rivaroxaban a un régimen de dosis ajustado según la edad y el peso, en niños con tromboembolismo venoso agudo. Phase III. Promoter: Bayer Pharma AG. PI: Ruben Berruero Moreno. Service: Clinical haematology. Start: 04/09/2014. Commercial

#### **SEA-OMA-2014-01**

Estudio retrospectivo de casos de pacientes alérgicos a alimentos (leche y/o huevo y/o vegetales) tratados con omalizumab como tratamiento único o asociado a inmunoterapia oral mediante un registro multicéntrico (OmaBase). EPA-OD. Promoter: Sociedad Española de Inmunología Clínica, Alergia y Asma Pediátrica SEICAP. PI: Ana Mª Plaza Martín. Service: Allergy and clinical immunology. Start: 06/08/2014. Non Commercial

#### **EST-SFA-2013**

Observational, Ambispective Natural History Study of Patients with Mucopolysaccharidosis Type IIIA (MPS IIIA) or Sanfilippo Syndrome Type A. No EPA. Promoter: Esteve Pharmaceuticals, S. A. PI: Mercè Pineda Marfà. Service: Neurology. Start: 16/07/2014. Commercial

#### **WA29231**

Estudio de extensión a largo plazo para evaluar la seguridad y la eficacia de tocilizumab administrado por vía subcutánea en pacientes con artritis idiopática juvenil de curso poliarticular y artritis idiopática juvenil sistémica. Phase I. Promoter: Roche Farma, S. A. PI: Rosa Bou i Torrent. Service: Paediatrics. Start: 30/06/2014. Commercial

#### **HSJD-HR-NB-Ch14.18**

Ensayo Clínico de Fase II unicéntrico, no controlado, de Dinutuximab (CH 14. 18) con las citoquinas factor estimulante de

granulocitos y macrófagos (GM-CSF) e IL-2, en pacientes con neuroblastoma de alto riesgo que no son candidatos para otros ensayos de inmunoterapia. Phase II. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Jaume Mora Graupera. Service: Oncology. Start: 26/05/2014. Non Commercial

#### **Y-52-52120-153**

Estudio de fase III, multicéntrico, doble ciego, prospectivo, aleatorizado, controlado, de tratamiento múltiple para evaluar la eficacia y la seguridad de Dysport en el tratamiento pediátrico de la espasticidad de las extremidades superiores. Phase III. Promoter: Ipsen Innovation. PI: Xenia Alonso. Service: Neurology. Start: 09/05/2014. Commercial

#### **A1481316**

Estudio multicéntrico, aleatorizado, controlado con placebo, en doble ciego, de dos grupos paralelos, para evaluar la eficacia y la seguridad del sildenafil IV en el tratamiento de neonatos con hipertensión pulmonar persistente del recién nacido (HPPRN) o insuficiencia respiratoria hipóxica y en riesgo de HPPRN, con un seguimiento a largo plazo para investigación de la evolución de su desarrollo 12 y 24 meses después de la finalización del tratamiento del estudio. Phase III. Promoter: PFIZER INC. PI: Julio Moreno Hernando. Service: Neonatology. Start: 06/05/2014. Commercial

#### **FUN-ISO-2014-01**

Validación de la dosis única diaria de isoniazida a 10 mg/kg de peso en el lactante menor de 3 meses de vida. EPA-OD. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Antoni Noguera Julián. Service: Paediatrics. Start: 16/04/2014. Non Commercial

#### **SER-UVE-2013-02**

Registro Español de Uveítis Asociada a Artritis Idiopática Juvenil. No EPA. Promoter: Sociedad Española de Reumatología. PI: Jordi Antón López. Service: Paediatrics. Start: 01/04/2014. Non Commercial

#### **EMR 700773-002**

Estudio de fase IV, abierto y de una sola cohorte, de los resultados neurocognitivos (NC) a largo plazo en niños de 4 a 5 años con fenilcetonuria tratados con dihidrocloruro de sapropterina (Kuvan®) durante 7 años. Phase IV. Promoter: Merck KGaA. PI: Jaume Campistol Plana. Service: Neurology. Start: 05/02/2014. Commercial

#### **Sobi. NTBC**

Estudio postautorización de seguridad (EPAS) no intervencional para evaluar la seguridad a largo plazo del tratamiento con Orfadin en la Tirosinemia hereditaria tipo 1 (TH-1) en la práctica clínica estándar. EPA-SP. Promoter: Swedish Orphan Biovitrum AB (publ). PI: Camila García Volpe. Service: Gastroenterology, hepatology and nutrition. Start: 03/02/2014. Commercial

#### **SPAIN-CCSS**

Creación de una cohorte española de supervivientes de cáncer infantil: cohorte SPAIN-CCSS. No EPA. Promoter: Fundació Privada ISGLOBAL. PI: Héctor Salvador Hernández. Service: Oncology. Start: 20/01/2014. Non Commercial

#### **SENEP-EPI-2013-01**

Estudio Epidemiológico para evaluar la gravedad de la infección por VRS en pacientes con enfermedad neurológica severa. No EPA. Promoter: Sociedad Española de Neurología Pediátrica (SENEP). PI: Andrés Nascimento Osorio.

Service: Neurology. Start: 17/01/2014. Non Commercial

#### **ISG-GEIS-05-2**

Estudio prospectivo observacional de la expresión de ABCB1/P-glicoproteína como factor para la estratificación biológica del osteosarcoma no metastático de las extremidades. EPA-SP. Promoter: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. Service: Oncology. Start: 07/01/2014. Non Commercial

#### **NI-0501-05**

Estudio multicéntrico de seguimiento a largo plazo de pacientes con LHH que han recibido tratamiento con NI-0501, un anticuerpo monoclonal anti-interferón gamma. Phase II. Promoter: Novimmune SA. PI: Jordi Antón López. Service: Paediatrics. Start: 18/11/2013. Commercial

#### **GEI-SAR-2012-01**

Registro de sarcomas raros: Herramienta de ayuda para evaluar el número de casos de cada subtipo y su orientación terapéutica por el Grupo Español de Investigación en Sarcomas (GEIS). EPA-SP. Promoter: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. Service: Oncology. Start: 14/11/2013. Non Commercial

#### **CRAD001A2314**

Estudio multicéntrico, abierto, aleatorizado, controlado, de 12 meses de seguimiento para evaluar la eficacia, tolerabilidad y seguridad de la introducción temprana de everolimus, con ICN reducido, y eliminación temprana de esteroides en comparación con un régimen estándar de ICN, micofenolato mofetilo y esteroides en receptores pediátricos de trasplante renal con un seguimiento de seguridad adicional de

24 meses. Phase II. Promoter: Novartis Farmacéutica, S. A. PI: Yolanda Calzada Baños. Service: Nephrology. Start: 30/09/2013. Commercial

#### **BEL114055**

Estudio multicéntrico, aleatorizado, de grupos paralelos, controlado con placebo, doble ciego para evaluar la seguridad, eficacia y farmacocinética de belimumab, un anticuerpo monoclonal humano anti-BLyS, más terapia estándar en pacientes pediátricos con lupus eritematoso sistémico (LES). Phase II. Promoter: Glaxosmithkline, S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 30/09/2013. Commercial

#### **IM101-301**

Ensayo clínico fase III, multicéntrico, abierto, para evaluar la farmacocinética, eficacia y seguridad de abatacept administrado por vía subcutánea (s. c. ) en niños y adolescentes con artritis idiopática juvenil poliarticular (AIJp) activa y respuesta inadecuada (RI) a fármacos antirreumáticos modificadores de enfermedad (FAME) biológicos o no biológicos. Phase III. Promoter: Bristol-Myers Squibb International Corporation (BMSIC). PI: Jordi Antón López. Service: Paediatrics. Start: 17/09/2013. Commercial

#### **CRAD001MIC03 (NOV-TOR-2012-01)**

Un registro internacional de enfermedad que recoge datos sobre manifestaciones, intervenciones y resultados en pacientes con complejo esclerosis tuberosa -TOSCA (NOV-TOR-2012-01). EPA-SP. Promoter: Novartis Farmacéutica, S. A. PI: Jaume Campistol Plana. Service: Neurology. Start: 19/11/2012. Commercial

#### **MCB-INM-2012-01. MCB-INM-2012-01**

Estudi MOMO Monitorización humoral tras modificación del tratamiento inmunosupresor en trasplante renal

y cardiaco. Estudio MOMO. EPA-SP. Promoter: Fundación Instituto Mar de Investigaciones Médicas (Fundación IMIM). PI: Anna Vila Santandreu. Service: Nephrology. Start: 24/10/2012. Non Commercial

#### **A0221047**

Periodo aleatorizado y abierto de 12 semanas con comparador activo seguido de un periodo de extensión de seguridad de 12 semanas para evaluar la seguridad y la eficacia de fesoterodina en sujetos de 6 a 16 años y >25kg con síntomas de hiperactividad del detrusor asociada con un trastorno neurológico (hiperactividad neurógena del detrusor). Phase III. Promoter: Pfizer, S. L. U. PI: Lluís García Aparicio. Service: Surgery. Start: 22/10/2012. Commercial

#### **N01266**

Estudio de seguimiento, multicéntrico, abierto, a largo plazo, para evaluar la seguridad y la eficacia de brivaracetam como tratamiento adyuvante en pacientes pediátricos con epilepsia. Phase II. Promoter: UCB Pharma S. A. PI: Alia Ramírez Camacho. Service: Neurology. Start: 09/10/2012. Commercial

#### **GHLiquid-3676 (NNH-SOM-2012-01)**

"GHLiquid-3676 (NNH-SOM-2012-01) Estudio internacional de desenlaces de NordiNet® (IOS de NordiNet®)". Promoter: Novo Nordisk Pharma, S. A. PI: Novo Nordisk España. PI: Lourdes Ibañez Toda. Service: Endocrinology. Start: 04/10/2012. Commercial

#### **BAX-OCT-2011-01**

Base de datos de resultados en la hemofilia A con ADVATE (AHEAD). Promoter: Baxalta Innovations GmbH. PI: Ruben Berruero Moreno. Service: Clinical haematology. Start: 18/07/2012. Commercial



**IGR2009/1593/Inter-B-NHL ritux 2010**

"Estudio intergrupo para niños o adolescentes con linfoma no-Hodking B o Leucemia Linfoblástica de Células B; Evaluación de eficacia y seguridad de rituximab en pacientes de alto riesgo". Phase III. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI: Veronica Celis Passini. Service: Oncology. Start: 02/07/2012. Non Commercial

**B1801023**

Estudio abierto de extensión para evaluar la seguridad a largo plazo de Etanercept en niños y adolescentes con artritis idiopática juvenil oligoarticular extendida, artritis asociada a entesitis o artritis psoriásica, que participaron anteriormente en el protocolo 0881A1-3338-WW (B1801014). Phase II. Promoter: Pfizer, S.L.U. PI: Jordi Antón López. Service: Paediatrics. Start: 24/01/2012. Commercial

**HUMORAL-KTx**

Registro nacional de rechazo humoral: Estudio epidemiológico multicéntrico observacional prospectivo para evaluar las características clínicas, serológicas e histológicas y la evolución a 5 años del rechazo humoral post-trasplante renal en España. Promoter: Astellas Pharma S. A. PI: Anna Vila Santandreu. Service: Nephrology. Start: 08/06/2011. Commercial

**P10-262/ABB-ADA-2008-02**

Registro observacional post-comercialización, multicéntrico y longitudinal de evaluación de la seguridad y la eficacia a largo plazo de Humira® (adalimumab) en niños con artritis idiopática juvenil (AIJ) poliarticular o de evolución poliarticular de actividad entre moderada y grave. [ESTUDIO STRIVE]. Promoter: Abbott Laboratories, S. A. PI: Jordi Antón López. Service: Paediatrics. Start: 12/05/2010. Commercial

**EMR 700773-001**

Registro europeo pediátrico materno de adultos de Kuvan® (KAMPER). Promoter: BioMarin International Limited. PI: Jaume Campistol Plana. Service: Neurology. Start: 16/03/2010. Commercial

**IPS-MEC-2008-01**

"Base de datos del foro europeo de Increlex [mecasemina (derivada del ADN $\alpha$ ) inyectable] en el retraso del crecimiento: Registro de pacientes europeos para vigilar la eficacia y la seguridad a largo plazo de Increlex. Eu-IGFD". Promoter: Ipsen Pharma, S. A. PI: Marta Ramon Krauel. Service: Endocrinology. Start: 02/02/2009. Commercial

**IPS-SOM-2007-03**

Estudio Cooperativo Internacional del Crecimiento. Promoter: Ipsen Pharma Accounting Department/Comptabilité Fournisseur. PI: Marta Ramon Krauel. Service: Endocrinology. Start: 22/10/2007. Commercial