ANNUAL SCIENTIFIC REPORT 2017
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- Influence of the environment on child and adolescent welfare
- Foetal environment and obstetrical complication
- Impact of congenital heart disease on development: from foetus to adult
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- Molecular biology and gene regulation of adipose tissue and its pathologies
- Human molecular genetics I
- Human molecular genetics II
- Molecular pharmacology and experimental therapies
- Pharmacological targets in inflammation and metabolic diseases
- Microbiota intestinal
- Stem Cell and Neural Plasticity

4.2. Neuropsychology, cognitive and development Neuroscience
- Neuropsychology
- Cognitive Neuroscience
- Developmental Neuroscience

4.3. Biomedical engineering
- Bioinformatics and data analysis platform
- Computer graphics
- Biomaterials and tissue engineering
- Processing and interpretation of biomedical signals
- Instrumentation and bioengineering
- Robotics and Vision
- Medical radiophysics
- Biomechanical Engineering

4.4. Epidemiology
- Epidemiology of aging: factors associated with healthy aging
- Impact, risk and prevention of mental disorders
- Healthcare technologies and results in primary care and mental health [PRISMA]
1.1. Introduction

Our thanks to all the researchers who have made it possible, once again this year, for our research institute to continue to raise its high standard of expertise and to enhance the quality of its knowledge, as well as the translation of this knowledge to clinical practice. The intense rigorous work carried out by the Institut de Recerca Sant Joan de Déu (IRSJD) is unquestionably the most important contribution that we can offer to our patients.

IRSJD is a young institution that was formally created in 2015 and as such it has many challenges to face. One of these is to achieve the Public Administration’s recognition of the Institute’s high level of research quality. In 2017, IRSJD was assessed by the prestigious CERCA institution of the Government of Catalonia and the outcome was highly encouraging, not only because of the positive result of the appraisal but also because it gave us the chance to receive a broad series of recommendations which will help us to grow and to improve.

The year 2017 has also been marked by the effective incorporation of some of the research groups of the Parc Sanitari Sant Joan de Déu within the structure of IRSJD. The Parc Sanitari Sant Joan de Déu is an institution which counts among its strategic goals the integration of mental health as a natural aspect of our society. The mental health research groups provide a high level of quality research and scientific output to our Institute. We welcome them and sincerely hope that our scientific collaboration will prove very beneficial.

Drawing up a balance of the results obtained in 2017, we find that the Institute continues to follow a trend of sustained growth, having achieved more clinical trials and more competitive projects as well as a substantial increase of scientific publications reflecting qualitative growth and an impact factor of over 450 points, which goes to show the high level of competence of our researchers. Our joint collaborative high-quality endeavours have allowed us to improve some basic infrastructures such as the new Daniel Bravo Centre for Rare Disease Diagnosis and Research, to be a reference institution in paediatric clinical trials under European financing, to have increased our participation in financed European research projects and networks, and to publish significant papers in leading scientific journals of great prestige and impact.

This work in collaboration of the IRSJD research groups, which belong to highly regarded universities and hospitals, focuses scientific and technical solutions on the clinical problems of people and especially of those who are most vulnerable, like children and adolescents, and mental patients, improving the results of the medicine which we seek to practise and of the science which we seek to develop.
1.2. Governing and advisory bodies

**Board of trustees**
- Joan Elias García: Rector, Universitat de Barcelona (UB)
- Domènec Espriu Climent: Vice-Rector for Research, Universitat de Barcelona
- Esther Real Saladrigas: Vice-Rector for Knowledge Transfer, Universitat Politècnica de Catalunya (UPC)
- Francesc Palau Martínez: Director, Institut de Recerca Sant Joan de Déu
- Emili Bargalló Angerri: Director, Research Foundation Sant Joan de Déu
- Enric Mangas: Director, Parc Sanitari Sant Joan de Déu
- Manuel del Castillo Rey: General Manager, Hospital Sant Joan de Déu
- Jaume Pérez Payarols: Director of Innovation, Hospital Sant Joan de Déu

**Executive Committee**
- Francesc Palau Martínez: Director, Institut de Recerca Sant Joan de Déu
- Rafael Artuch Iriberri: Scientific Deputy Director, Institut de Recerca Sant Joan de Déu
- Emili Bargalló Angerri: Director, Research Foundation Sant Joan de Déu
- Josep M. Haro: Director of Research, Innovation and Teaching, Parc Sanitari Sant Joan de Déu
- Jaume Pérez Payarols: Director of Innovation, Hospital Sant Joan de Déu
- Daniela Tost Pardell: Director, Centre de Recerca en Enginyeria Biomèdica (CREB) - Universitat Politècnica de Catalunya (UBP)
- Marçal Pastor-Anglada: Director, Institut de Biomedicina - Universitat de Barcelona (IBUB)
- Carles Escera i Micó: Institut de Neurociències - Universitat de Barcelona (UBNeuro)
- Júlia Ribot Ballabriga: Scientific manager, Institut de Recerca Sant Joan de Déu

**Scientific Advisory Council**
- Joaquín Arenas Barbero: Instituto de Investigación Sanitaria Hospital 12 de Octubre
- Jordi Cami i Morell: Parc Recerca Biomèdica de Barcelona, Fundació Pasqual Maragall - Universitat Pompeu Fabra
- Mara Dierssen Soto: Genomic Regulation Centre of Barcelona
- Michael B. King: London’s Global University, Division of Psychiatry
- Romà Pallarès Giner: Deputy Director, General Management of the Biomedical Research Institute of Bellvitge (IDIBELL)
- Carlos Rodriguez Galindo: St. Jude Children’s Research Hospital
- Montserrat Vendrell i Rius: General Director, BIOCAT
- Susan Webb: Institut de Recerca de l’Hospital de la Santa Creu i Sant Pau - Universitat Autònoma de Barcelona

**Research commission**
- Rafael Artuch Iriberri, president
- Asteria Albert Cazalla
- Pere Caminal Magrans
- Carmen de Torres Gómez-Palleto
- Carles Escera i Micó
- Claudia Fortunky Guasch
- Alfredo García-Alíx
- Àngels García Cazorla
- Juan José García-García
- Maria Dolores Gómez Roig
- Josep M. Haro Abad
- Lourdes Ibáñez Toda
- Josep Jiménez Chilavert
- Àngel Montero Carcaboso
- Carmen Muñoz Almagro
- Susana Ochoa Guerre
- Francesc Palau Martínez
- Marçal Pastor Anglada
- Antoni Serrano Blanco
- Judith Usall Rodie
- Júlia Ribot Ballabriga, secretary
1.3. Services and infrastructures

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**
- Clinical Trials Unit
- Sequencing Unit
- Confocal Microscopy Unit
- Animal housing facility
- Biobank
- Methodological and statistical support
- Methodological support in neuroimaging
- Scientific and technical evaluations service
- Library

**Other scientific and technical services**
- UBNNeuro Services
- IBUB Services
- CREB Services

**Management support services**
- Administration and human resources
- Clinical Research Ethics Committee
- Research management and Promotion
- Knowledge management and communication
2 GLOBAL NUMBERS

2.1. Global numbers 2017
2.2. Global numbers 2015-2017
2.3. Research and innovation outputs
   Scientific publications
   Patents and utility models
2.4. Training
   Doctoral theses
   Scientific seminars
2.1. Global numbers 2017

- **Research Staff**: 552
  - 51 Group leader
  - 209 Researcher
  - 64 Postdoc – associate researcher
  - 105 PhD Student
  - 66 Assistant researcher
  - 34 Technician
  - 23 Collaborator researcher

- **Projects**: 360
  - 243 Competitive national project
  - 32 Competitive international project
  - 85 Non competitive

- **Clinical Trials & Observational Studies**: 140
  - 15 Phase I
  - 35 Phase II
  - 48 Phase III
  - 4 Phase IV
  - 37 Observational Study

- **Publications**: 401
  - 1909 Impact Factor
  - 61 % Publications in first quartile
  - 4.8 Mean impact factor
  - 48 % Articles with main contributing authors

- **Budget**: 7.9 €
2.2. Global numbers 2015-2017

- RESEARCH STAFF
  - 2015: 321
  - 2016: 483
  - 2017: 552

- PROJECTS
  - 2015: 251
  - 2016: 303
  - 2017: 360

- CLINICAL TRIALS & OBSERVATIONAL STUDIES
  - 2015: 132
  - 2016: 124
  - 2017: 140

- PUBLICATIONS
  - 2015: 1138 IF
  - 2016: 1442 IF
  - 2017: 1909 IF

- BUDGET
  - 2015: 5.2
  - 2016: 6.5
  - 2017: 7.9
2.3. Research and innovation outputs

**Scientific publications**

- **401 ARTICLES & REVIEWS WITH IMPACT FACTOR**
- **1909 IMPACT FACTOR**
- **61% % ARTICLES IN THE FIRST QUARTILE**
- **4,8 MEAN IMPACT FACTOR**
- **48% % ARTICLES WITH MAIN CONTRIBUTING AUTHORS**

% publications in the first quartile: % of publications published in journals ranked in the first quartile (top 25%)
Mean impact factor: Mean impact factor of all the scientific publications

**Highest impact factor journals**

The list reflects the 98 original articles and reviews published in the most relevant scientific journals (1st decil)

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<thead>
<tr>
<th>Journal</th>
<th>Journal</th>
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<tbody>
<tr>
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<tr>
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<td>JAMA PSYCHIATRY</td>
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<tr>
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<td>NATURE GENETICS</td>
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<td>JOURNAL OF EXPERIMENTAL MEDICINE</td>
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<td>ANNALS OF ONCOLOGY</td>
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<td>LANCET DIABETES &amp; ENDOCRINOLOGY</td>
<td>TRENDS IN ENDOCRINOLOGY AND METABOLISM</td>
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<td>CIRCULATION</td>
<td>EUROPEAN RESPIRATORY JOURNAL</td>
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<td>LANCET RESPIRATORY MEDICINE</td>
<td>JAMA PEDIATRICS</td>
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## 2.3. Research and innovation outputs

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<th>Journal</th>
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<td>AMERICAN JOURNAL OF OBSTETRICS AND GYNECOLOGY</td>
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<td>DEPRESSION AND ANXIETY</td>
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<td>CORTEX</td>
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<td>J APPL RES INTELLECT</td>
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Patents and utility models

**Nonwoven Membrane as a drug delivery System**
- **Date of grant**: 07/02/2017
- **Country of protection**: United States
- **Grant number**: 9,561,190
- **Applicants**: Universitat Politècnica de Catalunya & Hospital Sant Joan de Déu
- **Inventors**: Montero Carcaboso A – Hospital Sant Joan de Déu. Others

**Treatment of hepativ steatosis related oligo-ovulation**
- **Date of grant**: 04/05/2017
- **Grant number**: WO 2017/072243
- **Applicants**: Katholieke Universiteit Leuven & Hospital Sant Joan de Déu
- **Inventors**: Ibáñez L – Hospital Sant Joan de Déu. Others

**Immunotherapy for CaSR-expressing cancer (e.g., neuroblastoma)**
- **Date of grant**: 26/01/2017
- **Grant number**: WO 2017/013276 A1
- **European patent**: Nº 15756941.9
- **Applicants**: Hospital Sant Joan de Déu, Barcelona & Fundació Cellex, Barcelona
- **Agent**: Herrero & Asociados.
- **Inventors**: de Torres C – Hospital Sant Joan de Déu

**System to assist walking / Sistema para asistir a caminar**
- **Date of grant**: 17/10/2017
- **Country of protection**: Spain
- **Grant number**: P201600886. PCT extension: PCT/EP2017/076482
- **Owner**: UPC, UDC, UEX
- **Inventors**: Lugris U, Cuadrado J, Font-Llagunes JM, Clos D, Alonso FJ, Romero F - CREB

**Sistema de contención y estimulación sensorial para bebés prematuros**
- **Date of grant**: 04/04/2017
- **Grant number**: ES1180334
- **Applicants**: Fundació Privada Elisava Escola Universitària i Hospital Sant Joan de Déu
- **Inventors**: Díaz Mecchia E, del Corral AM, Gómez Colominas M, García Fernández M
## 2.4. Training

### Doctoral theses

A list provided below the 32 doctoral theses presented during 2017 in original language.

<table>
<thead>
<tr>
<th>Thesis Author</th>
<th>Title</th>
<th>Supervisors</th>
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<tbody>
<tr>
<td>Angélica Ivone Avilés Rivero</td>
<td>Estimating and understanding motion: from diagnostic to robotic surgery</td>
<td>Alicia Casals</td>
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<tr>
<td>Ignacio Azanar Lou</td>
<td>El problema de la no iniciación del tratamiento farmacológico: evaluación con métodos cuantitativos</td>
<td>Maria Rubio Antoni Serrano</td>
</tr>
<tr>
<td>Carles Bautista-Rodríguez</td>
<td>Relación entre la enfermedad neumocócica invasiva y el déficit genético de MBL (mannose-binding lectin)</td>
<td>Carmen Muñoz-Almagro Carles Launes Iolanda Jordán</td>
</tr>
<tr>
<td>Anna Butíosa Molines</td>
<td>Stressful life events and first-episode psychosis</td>
<td>Susana Ochoa Guerre Juana Gómez</td>
</tr>
<tr>
<td>Judit Buxadera</td>
<td>Development of polymeric coatings with combined antifouling/antibacterial properties for titanium dental implants</td>
<td>Daniel Rodríguez Rius Xavier Gil</td>
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<tr>
<td>Montserrat Cairó Calzada</td>
<td>Autofagia en la activación termogénica del tejido adiposo marrón</td>
<td>Francesc Villarroya Joan Villarroya</td>
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<tr>
<td>María Alexandra Cañas Pacheco</td>
<td>Comunicación microbiota-huésped: internalización y señalización intracelular de vesículas de membrana externa de cepas probióticas y comensales de E. coli en células de epitelio intestinal</td>
<td>Laura Baldomà Josefa Badia</td>
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<tr>
<td>María Isabel Castellanos Arboleda</td>
<td>Functionalized CoCr surfaces with adhesive molecules to improve endothelialization</td>
<td>Marta Pegueroles José M. Manero</td>
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<tr>
<td>Marta de Castro Miró</td>
<td>Metodologies d’alt rendiments per a la identificació de noves mutacions i gens causants de distròfies de retina hereditàries</td>
<td>Roser González-Duarte Bru Cormand Rifa</td>
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<tr>
<td>Albert Català Temprano</td>
<td>Caracterització de les alteracions moleculars de FLT3 en pacients pediàtrics afectes de leucèmia aguda. Correlació amb l’activitat i expressió dels transportadors de fàrmacs derivats de nucleósids</td>
<td>Mireia Camós Guijosa Marçal Pastor Anglada</td>
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<tr>
<td>Anna Diez-Escudero</td>
<td>Tuning the biological performance of calcium phosphates through microstructural and chemical modifications</td>
<td>Maria Pau GinebraMontserrat Español</td>
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<tr>
<td>Behjat Gholami</td>
<td>Functional analyses of candidate genes for osteoporosis: RUNX2 and LRP5</td>
<td>Susana Balcells Daniel Grinberg</td>
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<tr>
<td>Soledad Gómez-González</td>
<td>Developmental Tumors, Epigenetics and microRNAs</td>
<td>Cinzia Lavarino</td>
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<td>Marcia Irazabal Jiménez</td>
<td>Carga e impacto familiar de las cuidadoras y los cuidadores de personas adultas con discapacidad intelectual con o sin trastorno mental asociado</td>
<td>Susana Ochoa Guerre Crescencia Pastor Vicente</td>
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<td>Kanupriya Khurana</td>
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<td>María Jesús Leal-Witt</td>
<td>Efecto de una intervención del estilo de vida en el perfil metabólico de sujetos prepuberales con obesidad</td>
<td>Carles Lin</td>
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<td>Hadiseh Mahdavi</td>
<td>A contribution to unobtrusive measurement methods for sleep monitoring based on magnetic induction technique</td>
<td>Javier Rosell Ferrer</td>
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<td>Ricardo Moure Ortega</td>
<td>Alteraciones en la señalización de FGF21 asociadas a la terapia antiretroviral y la adiposidad</td>
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<tr>
<td>Diana Carolina Muñoz Lasso</td>
<td>Fisiopatología de la ataxia de Friedreich: transporte y degeneración axonal</td>
<td>Francesc Palau Martínez Pilar González Cabo</td>
</tr>
<tr>
<td>María del Mar O’Callagan</td>
<td>Nuevos aspectos clínicos, bioquímicos, genéticos y terapéuticos en la encefalomiopatías mitocondriales en la edad pediátrica</td>
<td>Mercé Pineda Rafael Artuch</td>
</tr>
<tr>
<td>Juan Darío Ortigoza Escobar</td>
<td>Estudio de defectos en el transporte y el metabolismo de tiamina asociados a encefalopatías recurrentes en la infancia</td>
<td>Belén Pérez Dueñas</td>
</tr>
<tr>
<td>Monica Piquer-Gisbert</td>
<td>Fenotips d’immunodeficiencia comuna variable (IDCV) en pacientes pediátricos. Correlació genotip-fenotip en l’evolució clínica i el tractament dels pacients</td>
<td>Mª Anunciación Martín Mateos Manel Juan Otero</td>
</tr>
<tr>
<td>Sara Principi</td>
<td>Development of methodologies for estimating the dose to the eye lens in Interventional radiology. Operational implications of the eye lens new dose limit</td>
<td>Mercé Ginjaume</td>
</tr>
<tr>
<td>Alejandra Margarita Ríos Hernández</td>
<td>Diet and sleep in children and adolescents with Attention-Deficit Hyperactivity Disorder</td>
<td>María Luz Izquiero Pulido José Ángel Alda Díez</td>
</tr>
<tr>
<td>Alaleh Safari</td>
<td>Time dependent flow of biolubricant and suspended particles behavior within total hip replacement</td>
<td>Maria Pau Ginebra Nazanin Emami Michel J. Cervantes</td>
</tr>
<tr>
<td>Romain Schieber</td>
<td>Novel functionalized and patterned surfaces for cardiovascular applications</td>
<td>Marta Pegueroles Frank Mücklich</td>
</tr>
<tr>
<td>Carina Shianya Álvarez</td>
<td>Modulación de la barrera epitelial intestinal por vesículas de membrana y factores solubles secretados por cepas probióticas y comensales de Escherichia coli</td>
<td>Laura Baldomà Josefa Badia</td>
</tr>
<tr>
<td>Sara Siddi</td>
<td>Neurociencia de las alucinaciones: mecanismos neurocognitivos, neurofuncionales y neuroestructurales</td>
<td>Gildas Brebion Christian Stephan-Otto</td>
</tr>
<tr>
<td>Antonio Soriano Asrandes</td>
<td>Influencia de los cambios migratorios en la epidemiología de la transmisión vertical del virus de la inmunodeficiencia humana en Catalunya (periodo 2000-2014)</td>
<td>Clàudia Fortuny Antoniu Noguera</td>
</tr>
<tr>
<td>Maria Teixidó Ibañez</td>
<td>Constraints on early word segmentation and mapping</td>
<td>Laura Bosch Galceran</td>
</tr>
<tr>
<td>Albert Viel Oliva</td>
<td>Determinants estructurals i implicacions funcionals de l’interactoma protèic d’hCNT1</td>
<td>Sandra Pérez Torras Marçal Pastor Anglada</td>
</tr>
<tr>
<td>David Vila-Pérez</td>
<td>Evaluación de los factores pronósticos de sepsis en la edad pediátrica</td>
<td>Iolanda Jordán</td>
</tr>
</tbody>
</table>
**Scientific seminars**

- **January, 24.** Neuroprotection and regeneration/repair by combined therapy (hypothermia + allopurinol) in an animal model of hypoxic-ischaemic brain injury
- **February, 7.** Evolution of nosocomial infection in paediatric intensive care units: ENVIN-HELICS study
- **February, 21.** Sudden death in children and adolescents in Catalonia. How genetics helps us and implications for the family
- **March, 7.** Insulin receptor and endothelial adenosine axis in gestational diabetes mellitus
- **April, 4.** New generation genetic sequencing of patients with radiological anomalies of the basal ganglia
- **April, 24.** Assessment of effectiveness of a multiple intervention addressed to improving early empirical antibiotherapy in child sepsis: paediatric ABISS-EDUSEPSIS study
- **May, 9.** New generation genetic sequencing of patients with radiological anomalies of the basal ganglia
- **May, 16.** Best practices for project management
- **May, 23.** A journey from the gene to the phenotype
- **June, 6.** Studies of disorders using advanced tools in systems biomedicine: derivation of multi-level maps
- **June, 20.** Imprinting in humans: the role of imprinting in the placenta and in cancer
- **October, 3.** Our preclinical path to a phase I trial on advanced therapy in retinoblastoma
- **October, 17.** Influence of nurse supervision on adherence to treatment with antituberculosis drugs in the paediatric population
- **October, 31.** European Financing Opportunities in Research in the Health Field - 2018
- **November, 14.** ABLE: A low-cost robotic exoskeleton for gait rehabilitation
- **November, 11.** Neonatal ECG screening for early detection of long QT syndrome
- **November, 29.** RNA therapies for inherited retinal diseases: design & proof of concept studies
- **December, 1.** Identification and characterization of the molecular pathway controlling SAMHD1 function: implication in HIV infection and beyond
- **December, 12.** Design and assessment of innovative arm supports for children and adults with Duchenne muscular dystrophy
3 VERTICAL AREAS

3.1. Paediatric neuroscience
3.2. Molecular and cellular biology of developmental tumour
3.3. Adult-age disorders originating in the foetal period or in the early years of life
3.4. Infectious diseases and systemic inflammatory response in paediatrics
3.5. Mental Health
3.6. Foetal / Paediatric Diseases and environment, metabolic and genetic factors
3.1. Paediatric neuroscience

Coordinator: Jaume Campistol and Rafael Artuch

GLOBAL NUMBERS OF AREA

PERSONAL RESEARCHER

- 74
- Group leader: 7
- Staff researcher: 18
- Postdoc – associate researcher: 12
- PhD Student: 9
- Assistant researcher: 16
- Technician: 10
- Collaborator researcher: 2

PROJECTS

- 50
- Competitive national project: 33
- Non-competitive: 17

CLINICAL TRIALS & OBSERVATIONAL STUDIES

- 22
- Phase II: 4
- Phase III: 11
- Phase IV: 1
- Observational Study: 6

PUBLICATIONS

- 74
- Article & review indexats: 50
- Articles & reviews with Impact Factor: 46
- Impact Factor: 273,374
- % publications in the first quartile: 54
- Mean impact factor: 5,942
- % articles with main contributing authors from area: 41
PKU AND OTHER AMINO ACID GENETIC DISEASES
Jaume Campistol Plana, MD, PhD

The fact that our hospital institution is a reference centre for phenylketonuria in Catalonia has allowed us to study in depth the pathophysiological mechanisms involved in this disease in our patients and to test new treatments aimed to improve their metabolic control and neurologic prognosis. Our studies have led to the conclusion that some neuropsychological and neurological alterations are associated, on the one hand, with metabolic control, and on the other with antioxidant deficiencies. We have begun new treatments (with tetrahydrobiopterin) on our patients with phenylketonuria that have provided a better metabolic control and the possibility of freeing them from the special diet they were following. The monitoring of this treatment will be an important subject of study for the next three years. With respect to other disorders of the intermediate metabolism, mention should be made of our work on disorders of the metabolism of homocysteine and other amino acids, primarily addressed to characterising new disorders and new phenotypic presentations.

Members
Staff researcher
Roser Colomé Roura, Carme Fons Estupiña, Camila García Volpe, Rosa Gassió Subirachs, María Julieta González, Silvia Maria Meavilla Olivas
Assistant researcher
Pol Canal Noguer, Irene Galceran Nou, Leticia Diana Pias Peleteiro

Selected projects

Selected clinical trials
• EMR 700773-002. Phase IV. Open single-cohort phase IV study of long-term neurocognitive (NC) results in children aged 4-5 years with phenylketonuria treated with sapropterin dihydrochloride (Kuvan®) for 7 years. EUDRACT: 2009-015844-41. Promoter: Merck KGaA. PI: Jaume Campistol Plana. 05/02/2014 -

Selected publications

Networks
• CIBERER
• TREAT-NMD
MISIONHONDRIAL ENERGY METABOLISM AND DEVELOPMENT OF BIOMARKERS FOR THE INVESTIGATION OF INBORN ERRORS OF METABOLISM

Rafael Artuch Iriberri, MD, PhD

We have implemented a metabolomic platform for the phenotyping of patients with genetically-based rare metabolic disorders, using animal and cellular models. This activity has been possible thanks to the acquisition of a UHPLC system coupled in tandem to a mass spectrometer, financed by the Daniel Bravo Foundation.

Members
Staff researcher
Mercedes Casado Rio, Raquel Montero Sánchez, Cristina Sierra March
Postdoc – associate researcher
Aida Ormazabal Herrero, Délia Yubero Siles
Technician
Yahel Calzadilla Borras, Aroa Fernández de Miguel, Juan Moreno García, Tania Pardal Chofle

Selected projects
• Study of trace elements (Cu, Zn, Se and Mn) in cerebrospinal fluid in paediatric patients and analysis of related biomarkers. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00032. PI: Aida Ormazabal Herrero. 2015 - 2018.

Selected publications

Networks
• CIBERER
• ERN MetabERN. European reference networks
We coordinate the national register (CIBERER Database of Neuromuscular Disorders) of patients with congenital myasthenias and with collagen VI deficiency. On the latter group of patients we are conducting studies of natural history (in collaboration with the HSJD Rehabilitation Service, Dr. J. Medina) and of prognostic factors (in collaboration with several international centres: Prof. C. Bonnemann, NINDS-NIH, Washington, USA; Dr. Nicholas Deconinck, Ghent University, Belgium).

In 2017 Dr. Daniel Natera obtained his PhD at the University of Barcelona for the study “Congenital myasthenic syndromes in the Spanish paediatric population: Clinical characterisation, genetics and therapeutic response”, and in 2018 he obtained a Río Ortega contract for the study of clinical and genetic aspects of congenital myopathies and congenital myasthenias (coordinated by Dr. Palau, IRSJD). In June 2017 we began a project in collaboration with the group of Pros. Daniel Grinberg and Susana Balcells (IBUB) for the purpose of editing dominant mutations in collagen VI genes by means of CRISPR/Cas9 technology and other gene editing methodologies. The project is funded by the Noelia Foundation for Collagen VI Deficiency Myopathies. In this project (in which Dr. Jimenez-Mallebrera and Mari Carmen Badosa participate) we also collaborate with the group of Dr. Francesc Palau and Janet Hoenicka (IRSJD) and with Dr. Mônica Roldán of the Confocal Microscopy Service of the Daniel Bravo Centre (IRSJD).

Members

Staff researcher
Carlos Ignacio Orterez Gonzalez, Jaume Colomer

Postdoc – associate researcher
Daniel Natera de Benito

Assistant researcher
Maria del Carmen Badosa Gallego, Anna Lia Frongia, Irene Beatriz Zschaech Luzado, Anna Codina

PhD Student
Andres Nascimento Osorio, Cristina Jou Muñoz

Collaborator researcher
Pol Castellano Escuder, Susana Graciela Kalko Witruk

Selected projects


Selected clinical trials

• DSC/14/2357/48. Phase III: Randomised, double blind, placebo controlled, multicentre study to evaluate the efficacy and safety of givinostat in ambulant patients with Duchenne Muscular Dystrophy. EUDRACT: 2016-000401-36. Promoter: Italfarmaco,S.A. Pl: Andrés Nascimento Osorio. 31/05/2017 -
• 4045-301. Phase III: A Double-Blind, Placebo-Controlled, Multicenter Study With an Open-Label Extension to Evaluate the Efficacy and Safety of SRP-4045 and SRP-4053 in Patients With Duchenne Muscular Dystrophy. EUDRACT: 2015-002069-52. Promoter: Sarepta Therapeutics, Inc. Pl: Andrés Nascimento Osorio. 19/07/2017 -

Selected publications


Networks

• CIBERER
• TREAT-NMD
NEUROMETABOLIC DISORDERS AND SYNAPTIC STUDIES IN RARE NEUROGENETIC CONDITIONS
Mª Àngels García Cazorla, MD, PhD

In 2017 we consolidated the study models available to us at the laboratory (cellular models for the study of tyrosine hydroxylase deficiency and Rett syndrome) and we developed new techniques for the study of brain function (connectivity). We have had a very active participation in the European networks, which took shape in the coordination of a disorders subgroup (neurotransmitter defects) in the metabERN network. Likewise, we have made a significant contribution to the development of clinical guides and to the description of a new cause of early Parkinsonism and of new molecular aspects of known neurometabolic disorders.

Members
Postdoctoral researcher
Alfonso Luis De Oyarzabal Sanz
Staff researchers
Anna López Sala, Mª del Mar O’Callaghan Gordo, Federico Ramos
PhD Students
Elisenda Cortès-Saladelafont, Alba Tristan Noguero
Assistant researcher
Inés Fernanda Medina Rivera

Selected projects

Selected clinical trials
• UX007G-CL201. Phase II. A double-blind placebo-controlled clinical trial of parallel groups to assess the safety and effectiveness of UX007 in subjects with glucose transporter type 1 deficiency syndrome. Codi EUDRACT: 2013-003771-35. PI: Mª Angels García Cazorla. 07/01/2016 -
• UX007G-CL202. Phase II. Open extension study to assess the long-term safety and effectiveness of UX007 in subjects with glucose transporter type 1 deficiency syndrome [Glut1 DS]. EUDRACT: 2015-000389-69. Promoter: Ultragenyx Pharmaceutical, Inc. PI: Mª Angels García Cazorla. 05/06/2017 -

Selected publications

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
MOVEMENT DISORDERS OF GENETIC AND NEUROMETABOLIC ORIGIN
Belén Pérez Dueñas, MD, PhD

In 2017, our group published 11 articles with a total impact factor of 66.743; it was engaged in 13 active projects (including 2 FIS projects, 1 TV3 Marathon grant, and 1 Rio Hortega grant, among others) and the reading of one doctoral thesis [doctoral candidate Juan D. Ortigoza-Escobar]. Dr. Serrano led a clinical trial [Phase II] on acetazolamide in patients with PMM2-congenital disorder of glycosylation. The unit also belongs to several national research networks (CSUR) and European ones (ERN-NMD).

Members

Staff researchers
Jordi Muchart López, Mercedes Serrano Gimaré

Postdoc – associate researcher
Alejandra Darling, Juan Darío Ortigoza Escobar

PhD Students
Laura Martín Sanchez, María Isabel Vanegas Grisales

Selected projects


Selected clinical trials

- FSJD-PMM2-2016. Phase II clinical trial to evaluate the effectiveness and safety of acetazolamide in the treatment of cerebellar syndrome in patients with PMM2-CDG deficiency. EUDRACT: 2017-000810-44. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Mercedes Serrano Gimaré. 2017 -

Selected publications


Networks

- CIBERER
- TREAT-NMD
- Unitat integrada de referència del sistema nacional de salut en “enfermedades raras que cursan con trastornos del movimiento (csur_53)
- “Health care provider” European Reference Network on Rare Neurologic Diseases
NEUROGENETICS AND MOLECULAR MEDICINE
Francesc Palau Martínez, MD, PhD & Janet Hoenicka Blanco, MD, PhD

The scientific and translational endeavours of the Neurogenetics and Molecular Medicine research group (NeuroGene) unfold in the field of experimental biomedicine on neurogenetic and neuropsychiatric disorders, specifically Charcot-Marie-Tooth neuropathy (CMT), Friedreich’s ataxia (FRDA), Parkinson’s disease (PD) and neurodevelopmental disorders, as well as in the clinical field of the Paediatric Institute for Rare Diseases (IPER). The scientific landmarks achieved in 2017 notably include: (1) determination of the role of ANKK1 protein in the cell cycle during neurodevelopment [España-Serrano et al., Cereb Cortex 2017]; (2) determination of the differential effects on calcium homeostasis of recessive GDAP1 mutations vs. dominant mutations in CMT neuropathy [González-Sánchez et al., Sci Rep 2017]; and (3) application of NGS mass sequencing in the genetic diagnosis and study of a long series of patients with Rett or Rett-type syndrome [Vidal et al., Sci Rep 2017]. In December 2017, F. Palau was proposed by France’s INSERM as editor-in-chief of the Orphanet Journal of Rare Diseases.

Members
Staff researchers
Judith Silvia Armstrong Morón
Postdoc – associate researcher
Lara Cantarero Abad, Alba Aina Castells Santamaria, Laura Domínguez Berzosa, Loreto Martorell Sampol, Mª del Mar Pérez Irbarne, Isabel Plensa Nebot
Assistant researcher
Cesar Arjona Fernandez, Núria Brandi Tarrrau, Antonio Martínez Monseny, Mireia Osuna López, Ainhoa Pascual Alonso, Judith Reina Castilión,Montserrat Roura Llerda, Silvia Vidal Falcó
PhD Student
Azahara Civera Tregon, Elena Juárez Escoto, Abel Sola Bacaica
Technician
Daniel Castillo Mestres, Jordi Genoveses Escarré, Dolores López López, Montserrat Naudó Lahoz, Paola Pacheco Fernández, Johanna Troya Batseca

Selected projects

Selected publications
• España-Serrano L, Guerra Martín-Palanco N, Montero-Pedrauzela A, Pérez-Santamarina E, Vidal R, García-Consuegra I, et al. The Addiction-Related Protein ANKK1 is Differentially Expressed During the Cell Cycle in Neural Precursors.. Cereb Cortex 2017. 27: 2809-2819. FI: 6,559(Q1).

Networks
• CIBERER
• CIBERSAM
• TREAT-NMD
• ACTION - Data and Policies for Rare Diseases
• European Reference Network for Rare Neuromuscular Diseases
• Orphanet-Spain
3.2. Molecular and cellular biology of developmental tumour

Coordinator: Jaume Mora

Global numbers of area

PERSONAL RESEARCHER

- 53 Personal researcher

PROJECTS

- 26 Projects

CLINICAL TRIALS & OBSERVATIONAL STUDIES

- 41 Clinical trials & observational studies

PUBLICATIONS

- 43 Publications
- 40 Articles & reviews with Impact Factor
- 372,316 Impact Factor
- 69,767 % publications in the first quartile
- 9,3079 Mean impact factor
- 20 % articles with main contributing authors from area
A treatment of retinoblastoma with the VCN-01 oncolytic adenovirus reached the clinical phase (phase 1 trial) at Sant Joan de Déu Barcelona Children’s Hospital after carrying out all the preclinical phases at our laboratory. We thus achieved a goal pursued over the last 4 years. Part of the results of another two 4-year projects were published in 2017. In the first paper we described a new strategy for studying the intratumoural distribution of drugs targeting paediatric cancer and its application in neuroblastoma. The results were published in the Journal of Controlled Release. In the second study, which was published in the same journal, we showed that the barrier between the blood and the retina works by restricting the entry of drugs into the retinoblastoma tumour and can be pharmacologically modulated to counteract this effect.

Selected publications


Networks

- Innovative Therapies for Children with Cancer
3.2. Molecular and cellular biology of developmental tumour
Coordinator: Jaume Mora

LEUKEMIA AND NON-MALIGNANT BLOOD DISORDERS
Mireia Camós Guijosa, MD, PhD

In 2017 the group combined an intense clinical research activity with translational research. Dr. Susana Rives continued the clinical trials involving cell therapy with T CAR19 lymphocytes, one with the pharmaceutical industry and the other academic. Our centre was the first in Spain to administer this therapy and the one that recruited the largest number of patients in Europe, and in 2017 it passed the inspection of the U.S. Food and Drug Administration (FDA). The results of this trial were published in the New England Journal of Medicine. In the translational field, this year we obtained several competitive grants, including the FIS grant of Dr. Berrueco, the first in the line of haemostasis, which seeks to reduce the risk of thrombosis in patients with acute leukaemia. In 2017 Dr. Albert Català defended his thesis and is now training in Canada where he is carrying out a project on bone marrow failures.

Members
Staff researchers
Isabel Badell Serra, Ruben Berrueco Moreno, Albert Català Temprano, Susanna Gassiot Riu, Julia Marsal Ricoma, Susana Rives Solà, Anna Ruiz Llobet, Montserrat Torrebadell Burriel PhD Student.
Anna Alonso Saladrigues, Elena Esperanza Cebollada, Montserrat Mesegue Meda, Sandra Pont Martí, Maria Trabajo del Castillo, Nerea Vega García
Technician
Maria Camino Estella Aguado, Lidia Ruiz Bocanegra

Selected projects
- Pilot study on the infusion of autologous T lymphocytes genetically modified to express anti-CD19 in patients with chemotherapy resistant or refractory CD19+ leukaemia or lymphoma. Instituto de Salud Carlos III (ISCIII) - PCI. IC114/00224. PI: Susana Rives Solà. 2015 - 2018.

Selected clinical trials
- CCTL019B2001X Phase II. CTL019 expanded treatment protocol for paediatric/young adult patients with relapsing/refractory acute lymphoblastic leukaemia. EUDRACT: 2016-001991-31. Promoter: Novartis Farmacéutica, S.A. PI: Susana Rives Solà. 21/04/2017 -
- CART19-BE-01 Phase I. Pilot study of the infusion of autologous T lymphocytes genetically modified to express anti-CD19 in patients with chemotherapy resistant or refractory CD19+ leukaemia or lymphoma. EUDRACT: 2016-002972-29. Promoter: Hospital Clínic Barcelona. PI: Susana Rives Solà. 04/07/2017 -

Selected publications

Networks
- Innovative Therapies for Children with Cancer
NEUROBLASTOMA
Carmen de Torres Gómez-Pallete, MD, PhD

The group continued the study of the pathway of the calcium sensor receptor and PTHrP as potential therapeutic tools in neuroblastoma. Financing was achieved in 2017 for a European project for the study of nanosensors as detection systems for proteins excreted to the environment using the PTHrP model in neuroblastoma as a study model. The group continued to form part of the European network for the study of CasR, which includes the training of researchers shared by various laboratories.

Members
Postdoc – associate researcher
Marta García López, Silvia Mateo Lozano, Carlos Javier Rodriguez Hernandez
PhD Student
Eliana Carolina Gonçalves Alves
Technician
Noelia Salvador Marcos

Selected projects
• Assessment of the sensor receptor of calcium and parathyroid hormone-related protein as new therapeutic targets in neuroblastoma. Instituto de Salud Carlos III (ISCIII) - FIS. PI14/00048. PI: Carmen De Torres Gómez-Pallete. 01/01/2015 - 31/12/2017.
• NACANCELL_Development of a Nanodiagnostic platform for monitoring of Cancer cell secreted proteins. Instituto de Salud Carlos III (ISCIII) - Acciones complementarias. AC16/00026. PI: Carmen De Torres Gómez-Pallete. 01/01/2017 - 31/12/2019.

Selected publications

Networks
• CaSR_Marie Curie International Training Network - CaSR Biomedicine_Calcium-Sensing Receptor Therapeutics for Non-Communicable Diseases.
• 2016 - CaSR_Marie Curie International Training Network - CaSR Biomedicine_Calcium-Sensing Receptor Therapeutics for Non-Communicable Diseases. MSCA-ITN 2015.
SARCOMA AND HISTIOCYTOSIS
Jaume Mora Graupera, MD, PhD

2017 saw the publication of the original study of the GEIS 21 trial on the first multi-institutional clinical trial on both adult and paediatric patients of the GEIS group on Ewing's sarcoma directed at our centre. This study was presented at the ASCO Annual Meeting in 2016 and it formed the basis of the GEIS group for the study and development of new Ewing's sarcoma treatments. The new clinical trial of the GEIS group on Ewing’s sarcoma, GEIS 39, began in 2017, following up the findings of our laboratory on the activity of nab-paclitaxel in these tumours. The treatment of patients with refractory and relapsing neuroblastoma began at our centre in June of 2017 using the Hu3F8 antibody for the first time outside MSKCC, thanks to the relation established with the company Ymabs, which was founded by the father of a neuroblastoma survivor patient whom we followed up at our centre. Ymabs has acquired all the rights on the antibodies developed at MSKCC for the treatment of paediatric tumours with the aim to register them and make them commercially available. The clinical trials of these new treatments will be conducted jointly at MSKCC and at our centre. Because of this plan, many patients from around the world have begun to come to our centre to receive these unique treatments.

Members
Staff researcher
Verónica Celis Passini, Moira Garraus Oneca, María Inmaculada Hernández Muñoz, Vicente Santa-Maria, Alicia Cañada Heredia

Postdoc – associate researcher
Sara Sánchez Molina

Assistant researcher
Elisabet Figuerola Bou

PhD Student
Estela Prada Varela, Alejandro Manzanares Quintela, Sara Pérez Jaume

Technician
Maria Jesus Nagel Moyano

Selected projects

Selected clinical trials
- HSJD-HR-NB-Ch14.18. Phase II single arm institutional study to assess Dinutuximab combined with the cytokines granulocyte-macrophage colony stimulating factor (GM-CSF) and IL-2 in patients with high-risk neuroblastoma not eligible to other immunotherapy trials. EUDRACT: 2013-004864-69. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Jaume Mora Graupera. 26/05/2014 -

Selected publications

Networks
- Innovative Therapies for Children with Cancer
Cinzia Lavarrino’s research group developed a method that allows the quick, simple and precise classification of patients with medulloblastoma, a malignant tumour of the cerebellum. This method is based on the analysis of a small panel of epigenetic markers specific to each of the four main types of medulloblastoma (wingless, sonic hedgehog, group 3 and group 4), which are characterised respectively by different clinical behaviours and genetics. This biomarker panel has been developed and tested using databases of over 1,500 samples. The value of this classification method lies in its applicability to small tumoural biopsies (frozen or preserved), as well as its speed, reproducibility of results, and low financial cost.
3.3. Adult-age disorders originating in the foetal period or in the early years of life

Coordinator: Lourdes Ibañez
Foetal growth restriction may have irreversible consequences. The study of the various stages from intrauterine life to adolescence will allow us to advance in the prevention of this condition and in the treatment and prevention of the complications derived from it.

Members
Postdoc – associate researcher
Judith Bassols Casadevall, Paula Casano Sancho, Francis de Zegher, Marta Díaz Silva, Abel López Bermejo, Rita Maria Martins de Sousa Maia Malpique, Giorgia Sebastiani
Assistant researcher
Esther Lizarraga Molledo
PhD Student
Cristina García Beltran

Selected projects

Selected clinical trials
• CP-4-006. Fase III. 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. EUDRACT: 2016-003874-42. Promoter: OPKO Biologics Ltd. PI: Lourdes Ibañez Toda. 10/11/2017 -

Selected publications

Networks
• CIBERDEM
EARLY ORIGINS OF LONG-TERM RISK OF OBESITY AND METABOLIC DISEASE

Josep Jiménez Chillarón, PhD

The hypothesis of the foetal origin of adult disorders postulates that an adverse intrauterine environment may affect the growth of the foetus by producing permanent changes of gene expression in key cells and/or tissues. Such changes would persist after birth and could contribute to the development of chronic and complex disorders typical of adults, such as type 2 diabetes or obesity. Our laboratory’s goal is to understand the molecular mechanisms that associate nutrition during intrauterine development with the risk of acquiring chronic disorders as an adult, with special emphasis on diabetes and obesity. In order to understand the underlying molecular mechanisms, in our group we have developed an experimental model of foetal malnutrition that recapitulates the metabolic phenotype described in humans: malnourished mice in the uterus show low weight at birth and develop obesity, intolerance to glucose, and diabetes as from the 4th month of life.

Selected projects
• Miguel Servet II_Beneficiario: Dr. Josep Jimenez. Instituto de Salud Carlos III (ISCIII) - Miguel Servet II. MSII16/00046. PI: José Carlos Jiménez Chillarón. 2017 - 2020.

Selected observational studies
• “Increlex European Forum Database [mecasemin (derived from injectable rDNA)] on growth retardation: European patient registry for monitoring the long-term efficacy and safety of Increlex. EU-IGFDD”. Promoter: Ipsen Pharma, S.A. PI: Marta Ramon. 2009 -
• Recombinant growth hormone (rGH) treatment in patients with controlled GH deficiency at Hospital Sant Joan de Déu: Analysis of early markers of good response useful in clinical practice. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Marta Ramon. 2015 -

Selected publications

Networks
• CIBERDEM
3.3. Adult-age disorders originating in the foetal period or in the early years of life  
Coordinator: Lourdes Ibañez

VER Vertical Areas

NEONATAL PHYSIOPATHOLOGY
Alfredo Garcia-Alix Pérez, MD, PhD

Foetal growth restriction may have irreversible consequences. The study of the various stages from intrauterine life to adolescence will allow us to advance in the prevention of this condition and in the treatment and prevention of the complications derived from it.

Members
Staff researcher
Thais Agut Quijano, Miguel Mª Alsina Casanova, Marta Camprubi Camprubi, Ruth Del Rio Florentino, Isabel Iglesias Platas, Martin Iriondo Sanz, Ana Martín Ancel, Julio Moreno Hernando, Ana Morillo Palomo, Africa Perttierra Cortada, Ana Riverola De Veciana, Javier Rodríguez Fanjul
Technician
Miriam López Abad

Selected projects

Selected clinical trials
- H2020-PHC-18-2015-667224. Effect of Allopurinol in addition to hypothermia for hypoxic-ischemic Brain Injury on Neurocognitive Outcome – a blinded randomized placebo-controlled parallel group multicenter trial for superiority (Phase III). EUDRACT: 2016-000222-19. Promoter: University Hospital Tübingen PI: Marta Campriu Camprubi 05/05/2017 -
- REACT RCT. Producte Sanitari. Evaluation of Real Time Continuous Glucose Monitoring in the very preterm infant. PromotEr; University of Cambridge. PI: Isabel Iglesias Platas. 04/05/2017 -
- A1481316. Phase III: A multi-centre, randomized, placebo-controlled, double-blind, two-armed, parallel group study to evaluate efficacy and safety of iv sildenafil in the treatment of neonates with persistent pulmonary hypertension of the newborn (PPHN) or hypoxic respiratory failure and at risk for PPHN, with a long term follow up investigation of developmental progress 12 and 24 months after completion of study treatment. Eudract: 2012-002619-24. Promoter: Pfizer Inc. PI: Julio Moreno Hernando. 2014 -

Selected publications

Networks
- BCNAtal
3.4. Infectious diseases and systemic inflammatory response in paediatrics

Coordinator: Carmen Muñoz-Almagro

Global numbers of area

PERSONAL RESEARCHER

- 38 Personal researcher

PROJECTS

- 43 Projects

CLINICAL TRIALS & OBSERVATIONAL STUDIES

- 51 Clinical trials & observational studies

PUBLICATIONS

- 53 Publications

- 50 Articles & reviews with Impact Factor
- 222,245 Impact Factor
- 49,05 % publications in the first quartile
- 4,449 Mean impact factor
- 48 % articles with main contributing authors from area
3.4. Infectious diseases and systemic inflammatory response in paediatrics
Coordinator: Carmen Muñoz-Almagro

VACCINE-PREVENTABLE DISEASES
Carmen Muñoz Almagro, MD, PhD

Notable achievements during the year included the clinical and molecular characterisation of the outbreak of encephalitis caused by enterovirus A71 in Catalonia in 2016, and the implementation of the human microbiome studies in the healthy and diseased states in children.

Members
Staff researcher
Pedro Brotons de los Reyes, Juan José García García, Iolanda Jordán García, Cristian Launes Montañà
Postdoc – associate researcher
Cristina Esteva Afonso, Mariona Fernández de Sevilla Estrach, Natalia Timoneda Sole, Ana Valero Rello
Assistant researcher
Susana Hernández Bou, Ivette Rubio Martínez, Miguel García Fernandez
PhD Student
Didac Casas Alba, Desiree Henares Bonilla, Miriam Triviño Rodríguez
Technician
Carles Cisneros Camps, Amareesh Pérez Argüello, Jesica Saucedo Verdes

Selected projects

Selected clinical trials
- CE01-203. A Phase 2/3, Randomised, Partly blinded (blinded investigator), Multi-center, Comparative Study to Determine the Safety and activity of Solithromycin (intravenous and oral) in paediatric patients with suspected or confirmed community acquire bacterial pneumonia (CABP). EUDRACT: 2014-004039-37. Promoter: Cempra Pharmaceuticals, Inc. PI: Juan José García García. 12/09/2016 -

Selected publications

Networks
- 2016 - CIBERESP
- 2013 - SPIDNET
- 2015 - PERTINENT
3.4. Infectious diseases and systemic inflammatory response in paediatrics
Coordinator: Carmen Muñoz-Almagro

INFECTIONS OF VERTICAL TRANSMISSION AND INFECTIONS IN VULNERABLE PATIENTS
Clàudia Fortuny Guasch, MD, PhD

In 2017 we completed the analysis of the results of the FIS PI13/01738 project, which studied the mitochondrial toxicity secondary to exposure to human immunodeficiency virus (HIV) and to antiretroviral drugs, focused particularly on paediatric patients and on the potential impact of the depletion of mitochondrial DNA and its mutations. We are currently preparing the publications on this study. We have also been awarded another FIS project (PI17/00359), addressed to studying the potential toxicity of valganciclovir/ganciclovir in patients with congenital CMV infection who receive this drug.

Members
Staff researcher
Vicky Fumadó, Silvia Simó Nebot

Selected projects
• Longitudinal study of the mutations of mitochondrial DNA in children exposed to HIV (infected and non-infected) and to antiretrovirals. Instituto de Salud Carlos III (ISCIII). FIS. PI13/01738. PI: Clàudia Fortuny Guasch. 03/11/2017 -

Selected clinical trials

Selected publications

Networks
• CoRISPE-cat and NENEXP Cohorts for Paediatric HIV Infection in Children and Mother-to-Child Transmitted HIV Infection in Catalunya [Spain]
• PENTA-Id Network
• CIBERESP, grupo de investigación CB15/00067


ANNUAL SCIENTIFIC REPORT 2017
Sant Joan de Déu Institut de recerca
Research on the adherence and toxicity associated with first-line anti-tuberculosis drugs in the paediatric age, on the pharmacokinetic characteristics of these drugs on newborns and pre-school children, on the new tuberculosis diagnostic techniques in healthy children, on children from high tuberculosis endemic areas and on immunosuppressed children, and on the clinical-epidemiological characteristics of tuberculosis in childhood in our area.

Members
PhD Student
Maria Goretti López Ramos, Eneritz Velasco, Joan Vincent Genestar

Selected projects

Selected clinical trials
- 7625A-035. A Phase 2, Randomized, Active Comparator-Controlled, Multicenter, Double-Blind Clinical Trial to Study the Safety and Efficacy of Ceftolozane/Tazobactam (MK-7625A) Plus Metronidazole Versus Meropenem in Pediatric Subjects with Complicated Intra-Abdominal Infection. EUDRACT: 2016-004820-41. Promoter: Merck, Sharp & Dohme de España. S.A. PI: Antoni Noguera Julián. 31/08/2017 -
- FSJD-ISON-2016. Open clinical trial phase IIA to study the isoniazid suspension absorption of 10mg/ml for the tuberculosis infection treatment in patients under the age of 6. EUDRACT: 2016-002000-31. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Antoni Noguera Julián. 10/10/2016 -

Selected publications

Networks
- Tuberculosis Trials Consortium (TBTC), de los CDC de Atlanta [EEUU]; sub-investigador del site 31 [Barcelona]
- PENTA Steering Committee (Paediatric European Network for the Treatment of AIDS)
- CIBER en Salud Pública (CIBERESP), Grupo de Investigación CB15/0006
- pTBnet (Red Europea para Estudio de la Tuberculosis Pediátrica).
This year the group’s pre-consolidated status has been recognised by AGAUR. The team led by Dr. Antón has carried on with the projects on the study of autoimmune diseases [such as juvenile idiopathic arthritis, juvenile dermatomyositis and scleroderma], autoinflammatory diseases [genetic and epigenetic studies] and the aetiology of Kawasaki disease, likewise taking part in various clinical trials. As a result of this research, several papers have been communicated and published. The group led by Dr. Alsina centres its research on primary/congenital and secondary immunodeficiencies, focusing on 1) primary immunodeficiencies of the IL-12 /IFN-γ pathway generating susceptibility to intracellular infections [mycobacteria, leishmanias, among others]; 2) primary immunodeficiencies with a predominant immune dysregulation component [inflammation, lymphoproliferation, autoimmunity], with particular interest in the study of the cells possessing a regulatory role [regulatory T and B lymphocytes]; 3) immunodeficiencies secondary to the use of monoclonal antibodies, especially anti-TNF alphas during pregnancy. The aim is to learn about the bases of immune dysfunction in the three pathology groups and to identify biomarkers and new therapeutic targets.

Members
Staff researcher
Laia Alsina Manrique de Lara, Rosa Bou i Torrent, Àngela Deyá Martínez, Estibaliz Iglesias Jimenez, Mónica Piquer Gibert, Juan Manuel Mosquera Angarita, Judith Sánchez Manubens, Andrea Zacarias Crovato
Assistant researcher
Violeta Bittermann, Clara Isabel Giménez Roca
PhD Student
Joan Calzada Hernández, Anna Esteve Solé

Selected projects
- Study of the effect of intrauterine exposure to anti-TNF-alpha drugs in the immune system of children with hereditary Periodic Fevers (TRAPS, HIDS, or crFMF), with subsequent randomized withdrawal/dosing frequency reduction and open-label long term treatment epochs. EUDRACT: 2013-004291-35. Promotor: Novartis Farmacéutica, S.A. PI: Jordi Antón López. 2014 -
- WA28118. Fase I. A phase Ib, Open-Label, Multicenter study to investigate the pharmacokinetics, pharmacodynamics, and safety of tocilizumab following subcutaneous administration to patients with systemic juvenile idiopathic arthritis. EUDRACT: 2012-003490-26. Promotor: F. Hoffmann-La Roche Ltd. PI: Jordi Antón López. 2015 -

Selected clinical trials
- WA28118. Fase I. A phase Ib, Open-Label, Multicenter study to investigate the pharmacokinetics, pharmacodynamics, and safety of tocilizumab following subcutaneous administration to patients with systemic juvenile idiopathic arthritis. EUDRACT: 2012-003490-26. Promotor: F. Hoffmann-La Roche Ltd. PI: Jordi Antón López. 2015 -

Selected publications

Networks
- 2000 - Jordi Anton member of PRINTO (Pediatric Rheumatology International Trials Organization)
- 2017 - Laia Alsina member of Comité del Registro Español de Inmunodeficiencias Primarias REDIP
3.5. Mental Health

Coordinator: Judit Usall and Susana Ochoa

Global numbers of area

- Personal Researchers: 67
  - Group leader: 5
  - Staff researcher: 37
  - Postdoc – associate researcher: 2
  - PhD Student: 7
  - Assistant researcher: 11
  - Collaborator researcher: 5

- Projects: 40
  - Competitive national project: 32
  - Competitive international project: 2
  - Non competitive: 6

- Clinical Trials & Observational Studies: 4
  - Phase III: 1
  - Observational Study: 3

- Publications: 43
  - Article & review indexats: 38
  - Articles & reviews with Impact Factor: 128,726
  - Impact Factor: 44,19
  - % publications in the first quartile: 4,388
  - Mean impact factor: 55
    - % articles with main contributing authors from area
3.5. Mental Health
Coordinator: Judit Usall and Susana Ochoa

VERTICAL AREAS

CLINICAL, COGNITIVE AND PSYCHOSOCIAL BASES OF SEVERE MENTAL DISORDERS
Susana Ochoa Güerre, MD, PhD

The evaluation of the clinical, psychosocial and neuropsychological characteristics of people with a severe mental disorder will allow us to identify more precisely the treatment needs in each case. The main goal of this line is to evaluate the various clinical, social and neuropsychological aspects, among others, of the people who begin to manifest the first episodes of severe mental disorder. In order to carry out good evaluations in these areas, though, suitable measuring instruments must be available.

Members

Staff researcher
Antonio Manuel Alba Muñoz, Hilari Andres Mora, Emma Casas Anguera, Jorge Cuevas Esteban, Gemma Escuder Romeva, Alexandrina Foix Sanjuan, Marcia Irazabal Jiménez, Francisco Martínez Zambrano, María Trinidad Peláez Martínez

Assistant researcher
Anna Butjosa Molines, Núria del Cacho Ortega, Ingrid Tortadès Arumí, Regina Vila Badia

Selected projects


Selected publications


Networks

- CIBERSAM
- Consolidated research group in AGAUR
3.5. Mental Health
Coordinator: Judit Usall and Susana Ochoa

MENTAL HEALTH INTERVENTIONS
Judith Usall Rodie, MD, PhD

The current pharmacological and psychological treatments are not effective in all the people who suffer schizophrenia. This research group conducts clinical trials on the treatment of resistant schizophrenia or schizophrenia with predominant negative pharmacological or psychological symptoms. Four clinical trials have been carried out independently of the pharmaceutical industry, with financing from public bodies and from private foundations (Stanley Foundation). Gender- and sex-sensitive psychopharmacology is also taken into consideration in the research that is conducted.

Members
Staff researcher
Belen Arranz Martín, Adela Berrozpe Blasco, Marta Coromina Sadurní, Ruth Cunill Clotet, Gemma Escartin Martin, Mar García Franco, Eva Miquel Fernandez, Elena Rubio Abadal, Luis San Molina, Sonia Vilamala Anton
Assistant researcher
Marta Ferrer Quintero, Gabriel Ledesma Iparraguirre, Raquel López Carrilero, Silvia Marcó Garcia
Collaborator researcher
Maria Elena Huerta Ramos

Selected projects

Selected observational studies

Selected publications

Networks
- CIBERSAM
In 2017 we published several articles on our usual research subjects, some in first decile journals. We also developed a new line of research based on the quantification of brain asymmetry. Likewise, we took on a master’s degree student who is exploring another line of research on body asymmetry in relation to schizophrenia symptoms.
CHILD AND YOUTH MENTAL HEALTH RESEARCH
Montserrat Dolz Abadia, MD, PhD

The general goal of our research group is to conduct clinical research on various psychiatric disorders, with special emphasis on neurodevelopmental disorders. At present our efforts are focused on the study of initial psychoses in childhood (PI: Dr. Dolz), attention deficit hyperactivity disorders (ADHD; PI: Dr. Alda), and autism spectrum disorders (ASD; PI: Dr. Mairena).

In recent years the group has become consolidated, carrying out various projects financed by both public institutions (FIS projects) and private ones, and collaborations with different stable research groups of the CIBERSAM mental health biomedical research network, the G04 group from Hospital Clínic i Provincial de Barcelona and the G01 group from Hospital Gregorio Marañón in Madrid. The year was marked by the award of the European project Focus Locus centred on ADHD and gamification systems. Within the national sphere, a number of prizes were won for various research studies. Our group was awarded a research prize which allows it to continue to study ADHD and mindfulness by means of virtual reality. Likewise, we have received awards for papers and posters on intervention in social skills in children with autism, and for the identification of differential biomarkers of ADHD characterised by sluggish cognitive time.

Members
Staff researcher
José Ángel Alda Diez, Mar Alvarez Segura, Ester Camprodon Rosanas, Maria Diez Juan, Neus Elisàs Carbonell, Fernando Lacasa Saludes, Ma Àngeles Mairena Garcia de la Torre, Josep Lluís Matai Costa, Daniel Muñoz Samons, Jordi Navarra Ordoñó, Marta Pardo Gallego, Isabel Rueda Barcena, Bernardo Sánchez Fernández, Eduardo Serrano Troncoso, Anna Sintes Estevez

Assistant researcher
Marina Fabrega Ribera, Andrea González Contijoch, Carlota Saumell Andreu

PhD Student
Claudia Caprile Elola - Olaso, Anna Huguet Miguel, Ania Perez Racana, Laia Portoles, Jordina Tor Fabra

Collaborator researcher
Carme Tello

Selected projects

Selected clinical trials & observational studies

Selected publications

Networks
- CIBERSAM
The AGORA research Group, coordinated by Juan V. Luciano PhD, has been formally accredited as an Emerging Group (2017 SGR 667) by the AGAUR (Agency for Management of University and Research Grants; Government of Catalonian) and it forms part of the Spanish themed network RETICS-RedIAPP (Institute of Health Carlos III). This multidisciplinary group (5 psychologists, 1 general practitioner, 1 rheumatologist, 1 anesthesiologist, and 1 statistician) is interested in chronic pain, more specifically, in what psychological treatments work, for whom, and why. The group has experience in research and teaching of clinical psychology, psychometrics, health economics, and more specifically in the economic evaluation of different pharmaceutical and non-pharmaceutical treatments and services designed to help people with chronic pain. Along 2017, the group has published 10 manuscripts in indexed scientific journals and has obtained 3 competitive contracts (2 postdocs + 1 predoc).

Current research topics of the group:
- Cost-effectiveness of psychological treatments for fibromyalgia and chronic pain.
- Process and mechanism oriented research in chronic pain.
- Neurobiological underpinnings of chronic pain.
- Psychometric analysis of patient-reported outcome measures.

Members

Staff researcher
Antoni Rozadilla Sacanell, Lluis Torruella Barraquer
Postdoc - associate researcher
Albert Feliu Soler
PhD Student
Laura Andrés Rodríguez, Adrián Pérez Aranda
Collaborator researcher
Jaume Aguado Carné, Xavier Borràs Hernández, Eugenia Cardeños Valera

Selected projects


Selected observational studies


Selected publications


Networks

- Coordinator participates in the redIAPP (ISCIII; RD16/0007/0012)
3.6. Foetal / Paediatric Diseases and environment, metabolic and genetic factors

Coordinator: Maria Dolores Gómez-Roig
PAEDIATRIC INTENSIVE CARE UNIT RESEARCH PROJECT GROUP
Francisco José Cambra Lasaosa, MD, PhD

Completion of the PFIS0116 project: Towards a body area network for measurement of stress levels. Drafting of final report. The results are currently in the process of publication.

PFIS0116 project. Analysis of a diagnostic algorithm for severe pneumonia in critical paediatric patients by means of lung ultrasound scanning and procalcitonin as a plan for improving healthcare quality.

It is unfolding appropriately within the envisaged timeframe.

Members

Staff researcher
Monica Balaguer Gargallo, Elisabeth Esteban Torne, Iolanda Jordán García, Martí Pons Òdena
Assistant researcher
Carme Alejandre Galobardes, Georgina Armero Campos, Sergio Benito Fernández, Sara Bobillo Perez, Patricia Cormero Alonso, Aida Felipe Villalobos, Elena Fresán Ruiz, Lluisa Hernandez Platero, Nuria Millán García del Real, Luciana Rodríguez Guerineau, Susana Segura Matute, David Vila Pérez
Collaborator researcher
Montserrat Esquerda Areste

Selected projects

• Towards a body area network for measurement of stress levels. Instituto de Salud Carlos III (ISCIII) - FIS-2012. PI12/00533. PI: Francisco José Cambra Lasaosa.

• Analysis of a diagnostic algorithm for severe pneumonia in critical paediatric patients by means of lung ultrasound scanning and procalcitonin as a plan for the improvement of healthcare quality. Instituto de Salud Carlos III (ISCIII) - Proyectos de investigación en salud. PI16/01040. PI: Iolanda Jordán García.

Selected publications


Networks

• Multicenter study; prophylaxis against thrombosis practice (PROTRACT).

• Groupe français respiratoire GFRUP

• PALICC (Pediatric Acute Lung Injury Consensus Conference)

• Respiratory group of ESPNIC society
INFLUENCE OF THE ENVIRONMENT ON CHILD AND ADOLESCENT WELFARE

Carles Luaces Cubells, MD, PhD

Group focused on research in the following spheres:
- Study of the epidemiologic and clinical factors of childhood accidents.
- Differential diagnosis of child mistreatment.
- Early detection and secondary prevention of social-health issues in adolescence: drugs, sexuality and violence.
- Impact of external agents (radiation, pollution, etc.) on child health.
- Improvement of pediatric patient care in medical visits: presence of parents, analgesia, differentiated care by age (children/adolescents), etc.

Members

Staff researcher
Andrea Aldemira Liz, Gemma Claret Teruel, Ana Isabel Curcoy Barcenilla, Eva Gargallo Burriel, Lidia Martínez Sánchez, Sergi Navarro Vilarrubi, Marta Simó Nebot, Victoria Trenchs Sainz de la Maza

Assistant researcher
David Muñoz Santanach

Selected clinical trials

- D3820C00016. A Phase I, Open-Label, Multicentre Study to Assess the Pharmacokinetics and Safety of Naloxegol in Paediatric Patients Ages ≥ 6 Months to < 18 Years Receiving Treatment with Opioids. EUDRACT: 2013-003935-32. Promoter: Astrazeneca Farmaceutica Spain, S.A. PI: Sergi Navarro Vilarrubi. 07/07/2014 -

Selected publications


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)
FOETAL ENVIRONMENT AND OBSTETRICAL COMPLICATION

Maria Dolores Gómez Roig, MD, PhD

With the creation of BCNatal (2013), we have achieved the integration of healthcare in our speciality with research and teaching. We continue to conduct research within the Maternal-Children’s Health and Development Network (SAMID Network) financed by ISCIII. We are in charge of the line of research “Effects of environment on foetal growth” in Fetal I+D, Foetal Medicine Foundation Barcelona, and the research group “Foetal environment and obstetrical complications” of the Sant Joan de Déu Research Institute. Our research path and objectives are focused on the foetal environment, placental pathology and foetal growth restriction. We are member experts of the Work Group on Reproductive and Environmental Health of FIGO (International Federation of Gynaecology and Obstetrics, 2016-2019). Likewise, we cooperate with IsGlobal (Barcelona Institute for Global Health). It may be noted that we have now obtained the acceptance of the HEI Research Committee (Health Effects Institute, USA) for issue RFA 17-1 (November 2, 2017): “Traffic-related air pollution and birth weight: the roles of noise, placental function, green space, diet, physical activity, and socioeconomic status [FRONTIER]”.

Members

Staff researcher
Isidora Andujar Tore, Jose María Bogoña Ponsa, Sergio Cabré Gili, Carolina Esteve Matanza, Jaume Ignasi Miñano Masip, Elisabet Palacios, María José Tojo Grandio

Postdoc – associate researcher
Silvia – Irena Ferrero Martínez, Edurne Mazarico Gallego, Miriam Pérez Cruz, Joan Sabrià Bach

PhD Student
Laura Almeida Toledano, Edda Marimon Garcia, Isabel Miró Sotelo

Selected projects


Selected clinical trials & observational studies


Selected publications


• Sabra S, Malmqvist E, Saborit A, Gratacós E and Gómez-Roig MD. Heavy metals exposure levels and their correlation with different clinical forms of fetal growth restriction. PLoS One 2017. FI: 2,806 (Q1).

• Sabria-Bach J, Guirado L, Miró I, Gómez-Roig MD and Borrell A. Crown-rump length audit plots with the use of operator-specific PAPP-Á and beta-hCG median MoM. Prenat Diagn 2017. 37: 229-234. FI: 2,523 (Q2).

Networks

• 2009 - Red SAMID-RETIC
• 2013 - BCNatal
3.6. Foetal / Paediatric Diseases and environment, metabolic and genetic factors
Coordinator: Maria Dolores Gómez-Roig

IMPACT OF CONGENITAL HEART DISEASE ON DEVELOPMENT: FROM FOETUS TO ADULT
Joan Sanchez de Toledo, MD, PhD

Selected publications
- Pérez-Casares A, Cesar S, Brunet-Garcia L and Joan Sanchez de Toledo. Echocardiographic Evaluation of Pericardial Effusion and Cardiac Tamponade. Front Pediatr 2017. 5: 79-0. FI: 2,172(Q2).

PAEDIATRIC ARRHYTHMIAS, SUDDEN DEATH AND CARDIOVASCULAR GENETICS
Georgia Sarquella Brugrada, MD, PhD

Members
Staff researcher
Esther Carro Fernandez, Sergio César Diaz, Isaac Moll Adrian

Selected clinical trials
- Syn201401. Open phase IV multicentre study to assess the diagnostic effectiveness of Nuubo nECGband prior to Holter monitor implantation. EUDRACT: N/A. Promoter: Smart Solutions Tecnologies S.L. PI: Georgia Sarquella Brugada. 11/03/2015 -

Selected publications
4 CROSS-SECTIONAL AREAS

4.1. Genetics, molecular biology and gene therapy
4.2. Neuropsychology, cognitive and development neuroscience
4.3. Biomedical engineering
4.4. Epidemiology
4.1. Genetics, molecular biology and gene therapy

Coordinator: Marçal Pastor-Anglada
4 CROSS-SECTIONAL AREAS

4.1. Genetics, molecular biology and gene therapy
Coordinator: Marçal Pastor-Anglada

MOLECULAR BIOLOGY AND GENE REGULATION OF ADIPOSE TISSUE AND ITS PATHOLOGIES
Francesc Villarroya Gombau, PhD

In 2017 we kept the greater part of our research focused on the identification and characterisation of new molecules capable of acting as regulators in the pathophysiology of metabolism and adiposity. We have developed new approaches based on transcriptomics and bioinformatic prediction in both animal models and samples of cohorts of human patients and volunteers, including study cohorts at Hospital Sant Joan de Déu. We have expanded our international cooperation and new national and international cooperative projects have been obtained.

Members

Group leader
Marta Giralt Oms, Anna Planavila Porta

Staff researcher
Roser Iglesias Coll, Teresa Mampel Astals, Marion Peyrou, Anna Planavila Porta

Postdoc – associate researcher
Montserrat Cairó Calzada, Rubén Cairó Calzada, Alex Gavalda Navarro, Ricardo Moure Ortega, Joan Villarroya Terrade

PhD Student
Laura Campderrós Traver, Gemma Ferrer Curriu, Samantha Morón Ros, Tania Quesada López, Celia Helena Rupérez Gonzalo

Technician
Mercedes Morales Rueda, Gemma Moreno Besora, Albert Però Garcia

Selected projects

- Role of the novel cardiomyokines FGF21 and Metri on cardiac damage induced by alcoholism and arterial hypertension. Fundació La Marató de TV3 - Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 20153330. PI: Ana Planavila Porta. 2016 - 2018.

Selected publications


Networks

- CIBEROBN
- 2016 - 2017 - ADIPOPLAST (BFU2015-70454-REDT)
The research of this group includes the study of complex genetic diseases. Studies are currently being made of association with various pathologies using gene markers and candidate loci in order to identify genetic risk factors and to determine the molecular basis of their contribution to pathology.

We investigate: Lysosomal Diseases, Costello Syndrome, Retinitis pigmentosa, Homocystinurias, Multiple Osteochondromatosis, Osteoporosis, Migraines, Autism, Attention Deficit Hyperactivity Disorder, Opitz C Syndrome.

Selected projects


Selected publications


Networks

- CIBERER
- GEFOS/GENOMOS (international consortium for the study of the genetic bases of osteoporosis)
- European Network and Registry for Homocystinurias and Methylation Defects
- ImPACT (International Multicenter Persistent ADHD Collaboration)
- IHGC (International Headache Genetics Consortium)
- PGC (Pschiatric Genomics Consortium)
- ECNP (European College of Neuropsychopharmacology) Network: ADHD across the lifespan
HUMAN MOLECULAR GENETICS II
Roser Gonzàlez Duarte, PhD

This group concerns monogenic disorders associated to different degrees of genetic heterogeneity, from those of lysosomal origin to retinitis pigmentosa. Our first aim was to identify new candidate genes, disease-causing mutations and to analyse the structural and functional features of the mutant alleles. As a continuation of our work, we aim to perform functional studies at the cellular and tissular level, the construction of animal models and new strategies for diagnosis and therapy.

Members
Staff researcher
Gemma Marfany Nadal
PhD Student
Elena Borrego Domènech, Marta de Castro Miró, Vasileios Toulis

Selected projects
• Dissecting protein trafficking in retinal neurodegeneration by super-resolution imaging on animal models and human iPSCs. Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 20141730. PI: Roser Gonzalez Duarte. 2016 - 2018.

Selected publications
• Marfany, G. Genoma i exoma clínic (WGS i WES): retorn dels resultats genètics sol.licitats, quan, com i per què?. Treballs de la Societat Catalana de Biologia. 67, pp. 33 - 37. (España): Societat Catalana de Biologia, 2017. ISSN 0212-3037

Networks
• CIBERER- UNITAT 718
• 2013 - 2018 - COST ACTION BM1307 - PROTEOSTASIS (Member of the Core Management Group)
MOLECULAR PHARMACOLOGY AND EXPERIMENTAL THERAPIES
Marçal Pastor-Anglada, PhD

Our research group is studying diverse aspects of the cellular and molecular biology of membrane transporters, essentially in epithelial cells and in cells of the immune system. Two of the transporter gene families currently under study are CNT and ENT transporters responsible for the uptake of natural nucleosides and a wide range of drugs used in the treatment of cardiovascular diseases and antitumor therapies and antivirals.

Members
Staff researcher
Sandra Perez-Torras
Postdoc – associate researcher
Nerea Urtasun Plans
PhD Student
Hamsa Banjer, Clara Boces Pascual, Olga Casulleras-Gras, Liska Caviedes Cárdenas, Aida Mata Ventosa, Sandra Moro-Villa, Catalina Perelló Reus, Albert Viel Oliva

Selected projects

Selected publications

Networks
• CIBER-EHD
In 2017 our group has reported that very low-density lipoproteins (VLDL) and apolipoprotein CIII, which are elevated in diabetic states, induce endoplasmic reticulum stress and inflammation and attenuate insulin signalling via Toll-like receptor 2 in mouse skeletal muscle cells. In addition, we have demonstrated that that upregulation of small heterodimer partner (SHP) upon high-fat feeding leads to lipid accumulation, insulin resistance and inflammation in cardiomyocytes.
MICROBIOTA INTESTINAL
Laura Baldomà Llavinés, PhD

Our research group has as objective the study of several phenomena related to the communication or dialogue between the microbiota and the intestinal epithelium. The study is framed within a very current theme that tries to elucidate the complex interactions between the host and the bacterium, as well as its association with pathophysiological phenotypes. The relevant implication of the microbiota in health and disease suggests that the modulation of its composition by different means may constitute a new therapeutic approach.

Members

Staff researcher
Josefa Badia Palacín, Rosa Giménez Claudio

Postdoc – associate researcher
Laura Aguilera Gil

PhD Student
Carina Shianya Alvarez Villagómez, Mª Alexandra Cañas Pacheco, Mª José Fábrega Fernández

Selected projects


• Cooperation agreement for the performance of the research project “Study for the regulation of the NHD gene in Escherichia coli”. Universidad Nacional de Tucumán - Sense. PI: Laura Baldoma Llavines. 2009 - 2018.

Selected publications


STEM CELL AND NEURAL PLASTICITY
Antonella Consiglio, PhD

Our laboratory is interested in adult neuroplasticity and regeneration in the brain. We are interested in understanding the function and the potential of these newly born cells in physiology and disease. The main goal pursued in our laboratory is to identify part of the multiple signals that are responsible for endogenous precursor division, differentiation, circuit integration and survival in different regions of the brain, and to validate the possibility to manipulate neural precursors toward therapeutic neuronal repopulation for the cure of different degenerative CNS pathologies.

Members
Postdoc – associate researcher
Neus Bayo Puxan
Assistant researcher
Armida Faella, Irene Fernández Carasa
PhD Student
Carles Calatayud Aristoy, Giulia Carola, Angelique Di Domenico

Selected projects

Selected publications
4.2. Neuropsychology, cognitive and development neuroscience

Coordinator: Carles Escera

Global numbers of area

- **PERSONAL RESEARCHER**
  - 32
  - 3 Group leader
  - 11 Staff researcher
  - 4 Postdoc – associate researcher
  - 12 PhD Student
  - 1 Technician
  - 1 Collaborator researcher

- **PROJECTS**
  - 22
  - 18 Competitive national project
  - 4 Non competitive

- **CLINICAL TRIALS**
  - 1

- **PUBLICATIONS**
  - 16 Article & review indexats
  - 16 Articles & reviews with Impact Factor
  - 65,684 Impact Factor
  - 87,50 % publications in the first quartile
  - 4,105 Mean impact factor
  - 56 % articles with main contributing authors from area
Neuropsychology implies a clinical application of this knowledge, because it studies how brain injuries or disorders affect cognitive functions and behaviour. Research groups working in this area analyse changes in cognition associated with aging and degenerative illness, as well as the mechanisms of brain reorganization after a traumatic brain injury. We aim to investigate how cognitive functions can be improved in patients with mild cognitive impairment using non-invasive brain stimulated techniques. Also, we study the effects of cerebrovascular diseases and the neurobiological mechanisms of neuroprotective interventions, like exercise and cognitive training. Other research lines from this area focus on the differences in central nervous system function between normal-weight and obese people, as well as the coexistence of substance use disorders and severe mental disorders. In addition, we analyse several learning disorders, for instance dyslexia or nonverbal learning disability. Also, we work with patients suffering from cerebral palsy and we study the major impacts in cognitive functions. In these studies we use neuroimaging techniques, genetic studies and cognitive tests.
Major achievements of the cognitive neuroscience research group during 2017 were in understanding the neural underpinning of basic cognitive brain functions. In particular, in collaboration with University of Salamanca (Dr. M.S. Malmierca) we demonstrated that prediction errors in auditory perception emerge in a hierarchically organized pattern along the auditory system (Parras et al., 2017, Nature Comm.), and that conscious and attentive organization of the auditory scene relies on entrainment of neural oscillations to the tagging of auditory event patterns (Costa-Faidella et al., 2017, Neuroimage). We have started a new collaborative project with the Women’s Area of the Hospital (Dr. Gómez-Roig) to implement the so-called Frequency-Following Response (FFR) as a candidate biomarker for neurocognitive development in fetal growth restrictions. Our group has been recognized and financed as Grup de Recerca Consolidat by the Generalitat de Catalunya (SGR2017-974).

Members

**Staff researcher**
Inma Clemente Lapena, María José Corral López, Maribel Nuñez Peña, Iria San Miguel Insúa, Hans Supér, Marc Via García

**Postdoc – associate researcher**
Raffaele Cacciaglia, Miriam Cornella Griful

**PhD Student**
Natàlia Gorina Careta, Alejandro Lerer, Teresa Ribas Prats

**Selected projects**

**Selected publications**
- López-Caballero F and Escera C. Binaural Beat: A Failure to Enhance EEG Power and Emotional Arousal. FRONT HUM NEUROSCI 2017. 11: 557-0. FI: 3,209(Q1).
DEVELOPMENTAL NEUROSCIENCE
Laura Bosch Galceran, PhD

The group has carried on with the research activities focused on speech perception and language learning: a) the role of audiovisual information in language discrimination in the first year of life; b) the capacity to segment words and to associate them with a referent (6-9 months) in full-term populations and premature populations of different gestational age; c) the response to vowel/consonant changes at three moments in the first year of life (newborns, 6 months and 12 months) with EEG/ERP measurements, comparing full-term and premature infants followed up longitudinally (studies with Dr. T. Agut of neonatology), and d) assessment of language level achieved at 4 years and 7-8 years in different risk populations followed up from the first year of life (patients with neonatal cerebral infarction and children born with low weight, research studies in collaboration with Dr. A. Garcia-Alix and Dr. L. Ibáñez, respectively.

Members
Staff researcher
Ferran Pons Gimeno, Marta Ramon Casas
Postdoc – associate researcher
Clément François
PhD Student
Joan Birulés Muntané, Maria Teixidó Ibáñez, Anna Martínez Álvarez
Assistant researcher
Jessica Sánchez-Galán
Collaborator researcher
Jorgina Solé Montoliu

Selected projects

Selected publications
- François C, Teixidó M, Takerkart S, Agut-Quijano T, Bosch L and Rodríguez-Fornells A. Enhanced Neonatal Brain Responses to Sung Streams Predict Vocabulary; Outcomes by Age 18 Months. Sci Rep 2017. 7: 12451-0. FI: 4,259(Q1).

Networks
- sGR-1413, 2015-2017, IP: Dr. Antoni Rodríguez-Fornells
4.3. Biomedical engineering

Coordinator: Pere Caminal

Global numbers of area

PERSONAL RESEARCHER
- Group leader: 8
- Staff researcher: 52
- Postdoc – associate researcher: 8
- PhD Student: 19
- Assistant researcher: 1
- Technician: 7
- Collaborator researcher: 4

PROJECTS
- Competitive national project: 33
- Competitive international project: 6
- Non competitive: 19

PUBLICATIONS
- Article & review indexats: 49
- Articles & reviews with Impact Factor: 46
- Impact Factor: 168,255
- % publications in the first quartile: 59
- Mean impact factor: 3.66
- % articles with main contributing authors from area: 37
4.3. Biomedical engineering
Coordinator: Pere Caminal

BIOINFORMATICS AND DATA ANALYSIS PLATFORM
Pere Caminal Magrans, PhD

It is considered essential to propose a platform for the analysis of data in the broadest sense, including bioinformatics and biosignals. The goal is to establish a bioinformatics platform for the IR-SJD with a twofold aim: to provide the set of research groups with support on massive data analysis and to carry out an active programme of research, providing added value to the platform.

Members

Staff researcher
Alexandre Perera Lluna, Raul Benítez Iglesias, Jesús Escrivà Muñoz, Giovana Elizabeth Gavidia Bovadilla, Pedro Gomis Román, Alexander Vallmitjana Lees, Montserrat Vallverdú Ferrer
PhD Student
Xavier Marimon Serra, Sergi Picart Armada
Technician
Maria Maqueda

Selected projects


Selected publications

COMPUTER GRAPHICS
Daniela Tost Pardell, PhD

The activity of the group has focused on the development of three sets of serious games and a gamified app. On one hand, a new educational video-game of the series “Personatges en Joc” has been designed. A pilot study on the efficacy of education through play for children with ADT was carried on with Dr. Alda from HSJD based on the use of this game. On the second hand, a set of video-game for the education of transversal skills at the workplace for persons with intellectual disability was initiated, as the natural continuation of the former project IntegraGame. Finally, a project on the training the perfusion technique was initiated, funded by the Spanish Association of perfusionists. In the project ADALT, funded by Eurecat, the team developed a gamified app to query the emotional state and level of pain of patients with hemiparesis of the upper limb.

Members
Staff researcher
Dolors Ayala Vallespí, 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
- Smile@Work: serious games for cross-skills training in the occupational sphere for persons with intellectual disability, financed by the 1st Call for Grants to Research Projects Applied to Development of Accessible Technologies. (Indra y Fundación Universia) PI: Daniela Tost Pardell, 2017 - 2018.

Selected publications
BIOMATERIALS AND TISSUE ENGINEERING

Maria Pau Ginebra Molins, PhD

The Tissue Engineering and Biomaterials (TEB) group published 25 articles in high-impact index journals and two book chapters in 2017. The TEB members obtained an ERC project, an MSCA, an H2020 NMBP project, an ERA-NET, a MINECO project for young researchers and designation as an SGR group financed by the Government of Catalonia. In the course of the year, the group members presented 17 papers and 10 guest conferences at international congresses. Likewise, 6 doctoral theses directed by TEB members were read in 2017.

Members

Staff researcher
Cristina Canal Barnils, Jose María Manero Planella, Carles Mas Moruno, Judith Buxadera Palomero, Montserrat Español Pons, Anna Diez Escudero, Roberta Fraioli, Kanupriya Khurana, Marta Pegueroles Neyra, Daniel Rodriguez Rius, Elisa Rupérez de Gracia, Zhitong Zhao

Postdoc – associate researcher
Jordi Guillem Martí, Cédric Labay, Giuseppe Scionti, Anna Khlyustova, Juan Tornín, Aranzazu Villasante

Assistant researcher
Xavier Gil Mur

PhD Student
Albert Barba Serrahima, Mireia Hoyos Nogués, Joanna Maria Sadowska, Elia Vidal Girona, Romain Hugues Marie Schieber, Mar Bonany, Inês Hamouda, Joanna Konka, Joaquim Minguela, Santiago Raymond, Diego Torres, Miguel Mateu, Lluís Oliver

Technician
Marc Avilés Palau, Meritxell Molmeneu Trias, Mónica Ortiz Hernández, Miquel Punset Fusté, Sergi Torrent Camarero

Selected projects


Selected publications


Networks

PROCESSING AND INTERPRETATION OF BIOMEDICAL SIGNALS
Miquel A. Mañanas Villanueva, PhD

Analysis and interpretation of signals of cerebral origin obtained by means of non-invasive techniques such as electroencephalography (EEG) and magnetoencephalography (MEG). Analysis of signals of muscular origin as a tool for motor diagnosis and rehabilitation. Modelling and simulation of the respiratory system in order to predict the ventilatory response to different respiratory stimuli and in various conditions which may be the result of pulmonary disorders.

Members
Staff researcher
Joan Francesc Alonso Lopez, Sergio Romero Lafuente

Selected projects

Selected publications
INSTRUMENTATION AND BIOENGINEERING

Xavier Rosell Ferrer, PhD

Design and characterisation of medical and biotechnological instrumentation, of ultrasonic transducers, and of control and monitoring equipment; redesign of equipment and facilities. Guidance on the effect of non-ionising radiations and on compliance with EMC and electrical safety standards and directives.

Members

Staff researcher

Ramon Bragós Badia, Mireya Fernández Chimeno, Miguel Angel García Gonzalez, Miguel J García Hernández, Tomás García Sánchez, Lexa Nescolarde Selva, Juan Ramos Castro, Pere Joan Riu Costa

Selected projects


Selected publications

4.3. Biomedical engineering
Coordinator: Pere Caminal

ROBOTICS AND VISION
Alicia Casals Gelpí, PhD

Guidance on process automation, adaptation and robotisation of facilities, and on different types of facilities, development of vision systems for inspection and guiding of robots, development of image processing systems, design of mobile robots (intelligent walkers, prams, etc.), and study and design of technical aids for persons with physical and sensory disabilities, as well as guidance on and development of advanced control systems.

Members
Staff researcher
Joan Aranda López, Josep Fernández Ruzafa, Manel Frigola Bourlon, Enric Xavier Martín Rull, Antonio Martínez Velasco, Eduard Bergès Martín
Postdoc – Associate researcher
Albert Hernansanz Prats
PhD Student
Narcís Sayols Baixeras, Manuel Vinagre Ruiz
Collaborator researcher
Antonio Benedito Blanes, Carlos Morat, Laureano Tinoco, Alejandro Veiga López
Technician
Lluís Oriol Escoté

Selected projects

Selected publications

Networks
• 2016 - 2017 - Hypernet: Network of excellence on neuroprosthetics and neurorobbing for functional compensation and rehabilitation of movement disorders
• 2015 - Clúster de Salut Mental de Catalunya
4.3. Biomedical engineering
Coordinator: Pere Caminal

CROSS-SECTIONAL AREAS

MEDICAL RADIOPHYSICS
Mercè Ginjaume Egido, PhD

1. Development of radiotherapy treatment (RT) planning systems based on the use of Monte Carlo (MC) simulation, in particular the Penelope programme. The principal novelties notably include the introduction of proton applications, the study of small fields and the study of the skin dose in IMRT and SBRT.

2. Within the framework of the H2020 MEDIRAD project, a Monte Carlo programme based on the use of GPU (MC-GPU) cards is being optimised to obtain a real-time dose control system capable of minimising the probability of radiogenic skin lesions resulting from fluoroscopy-guided procedures.

3. Based on the foregoing programme, a new project has been started that allows estimation of the effective dose in organs of interest, particularly the lens of the eye and the brain, of healthcare staff participating in radiology and interventional cardiology.

Members

Staff researcher
Maria Amor Duch Guillen, Youri Alexandrovich Koubychine Merkulov, Xavier Ortega Aramburu, Josep Sempau Roma
Postdoc – associate researcher
Sara Principi

Selected projects


Selected publications


Networks

- 2000 - EURADOS, European Radiation Dosimetry Group, www.eurados.org - Voting Member - Investigadors WG12 medical imaging
- SGR 2014, SGR 2017 Dosimetria i Radiofísica mèdica
BIOMECHANICAL ENGINEERING
Josep Maria Font Llagunes, PhD

Kinematic and dynamic measurements of human movement in the biomechanics laboratory, dynamic analysis of human movement, mechanical design of care and rehabilitation devices.

Members

Staff researcher
Ana Barjau Condomines, Daniel Clos Costa, Rosa Pàmies Vilà, Miriam Febrer Nafría, Gil Serrancolí Masferrer, Alfons Carricero Carmona

Selected projects


Selected publications


Networks

- JM Font- Member of the Technical Committee for Multibody Dynamics, International Federation for the Promotion of Mechanism and Machine Science, IFToMM.
- JM Font- Vice-President of the Spanish Chapter of the European Society of Biomechanics, ESB.
- JM Font- PI of the Spanish Network of Biomechanics, Ministry of Economy and Competitiveness, Spain.
- JM Font- Member of the European Society of Biomechanics, ESB.

Biomedical engineering
Coordinator: Pere Caminal
4.4. Epidemiology

Coordinator: Josep Maria Haro

Global numbers of area

PERSONAL RESEARCHER
- 36 researchers
- Group leader: 3
- Staff researcher: 9
- Postdoc – associate researcher: 1
- Assistant researcher: 13
- Collaborator researcher: 10

PROJECTS
- 30 projects
- Competitive national project: 22
- Competitive international projects: 7
- Non competitive projects: 1

CLINICAL TRIALS & OBSERVATIONAL STUDIES
- 2 studies
- Observational Study: 2

PUBLICATIONS
- 104 articles
- Articles & reviews with Impact Factor: 97
- Impact Factor: 426,580
- % publications in the first quartile: 42,540
- Mean impact factor: 4,398
- % articles with main contributing authors from area: 43

ANNUAL SCIENTIFIC REPORT 2017
Sant Joan de Déu Institut de recerca
4.4. Epidemiology
Coordinator: Josep Maria Haro

Epidemiology of Aging: Factors Associated with Healthy Aging
 Josep Maria Haro Abad, MD, PhD

The interest of this research line lies in determining the impact (prevalence and disability) of mental disorders in the general population and the health care received by the persons who suffer such disorders. Epidemiologic studies are necessary for the planning and management of resources and to evaluate the existence and impact of factors associated with the possibility of suffering a mental disorder. The various projects of this line are focused on the adult population, the child and adolescent population, and elderly people.

Members

Staff researcher
Albert Sánchez Niubó, Stefanos Tyrovolas
Assistant researcher
Joan Domènech Abella, Laia Egea Cortes, Blanca Mellor Marsa, Ziggi Santini, Sara Siddi, Elisabeth Catharina Vorstenbosch

Selected projects


Selected publications


Networks

- CIBERSAM
- European Innovation Partnership on Active and Healthy Ageing (the Partnership)
4.4. Epidemiology
Coordinator: Josep Maria Haro

IMPACT, RISK AND PREVENTION OF MENTAL DISORDERS
Ai Koyanagi, MD, PhD

Mental disorders are the group of illnesses with the greatest impact on the population’s health in the most economically-developed countries, as has been stated by the World Health Organisation and in the Global Burden of Disease study. Between 20% and 30% of all people suffer a mental disorder at some point in their life. In many cases, this disorder is highly incapacitating. Our interdisciplinary group, which includes psychiatrists, psychologists, statisticians, sociologists and basic researchers, studies the factors that provide protection against or increase the risk of mental disorders, and treatments for the people who suffer them, for the purpose of improving preventive and therapeutic strategies. In recent years, aware of the importance of mental health in the achievement of a physically, mentally and socially healthy ageing process, we are devoting special attention to the study of health histories in order to determine the factors which favour active healthy ageing and at exactly which moment in time it is necessary to intervene in order to improve the course of each individual’s health. We seek to translate all this knowledge into healthcare innovations which can be easily incorporated into our healthcare and social support systems.

Selected projects


Selected publications


Networks

- CIBERSAM
Researchers of the PRISMA group estimated the costs of failure to begin medication and the impact of co-payment on such behaviour. A review was made of the evidence on the effectiveness of antidepressants as compared to active monitoring in primary care. Likewise, they typified agitation episodes, their interventions and their costs in a mental health hospital. The group also organised the 11th Course on Socio-Health Policy and Financial Assessment at the Loyola University Andalucia in collaboration with the King’s College of London and the London School of Economics and Political Science.

Members
Staff researcher
Maria Rubio Valera, Luisa Baladon Higuera, Vanessa Barneda, Laura González Sufner, Maria Iglesias Gonzalez, Cristina López Ortiz, Enric Vicens Pons
Postdoc – associate researcher
Ignacio Aznar Lou
Assistant researcher
Maria Montserrat Gil Girbau, Ana Gabriela Murrugarra Centurion, Carles Vilaplana Carnerero
Collaborator researcher
Ana Fernández Sánchez, Ana Maria Jove Masso, Martin Knapp, Maria Antonia March Pujol, Paul McCrone, Juan Manuel Mendive Arbeloa, Maria Teresa Peñarrubia, Ramon Sabes Figuera, Lluís Salvador Carulla

Selected projects

Selected observational studies

Selected publications

Networks
- 2017 - CIBERESP
- Red IAPP

ANNUAL SCIENTIFIC REPORT 2017
Sant Joan de Déu Institut de recerca
76
The team members of areas and groups presents all information of the group including external collaborators. The global numbers of the institution does not include external collaborators.

The number of projects shows the total number of projects managed and active during 2017.

Clinical trials and observational studies correspond to all those active during 2017.

Publications include original articles and reviews published during 2017 (with assigned pagination) and authored by members of research groups during their membership. The impact factor and quartile assigned corresponds to the Journal Citation Reports of the Web of Science of 2016.

The Budget is the total resources managed.