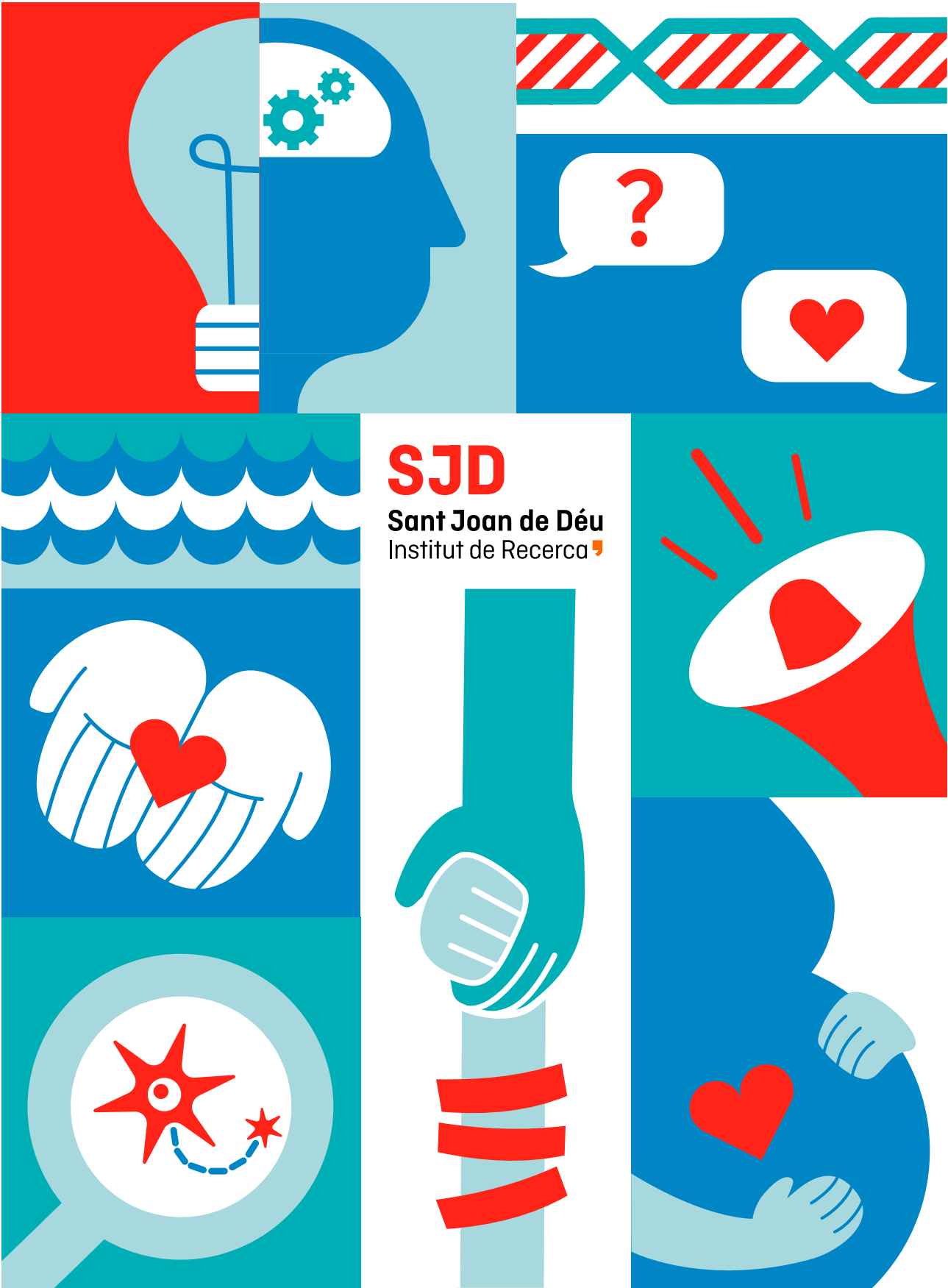


ANNUAL SCIENTIFIC REPORT

2021





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Institut de Recerca

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Sant Joan de Déu, Parc Sanitari Sant
Joan de Déu & Institut de Neurociències ·
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FOREWORD



Governance and structures
Global numbers 2021
Global numbers 2015-2021
Institutional awards



Governance and structures

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Dra. Marta Camprubí Camprubí

Dra. Roser Pueyo Benito

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Ms. Júlia Ribot Ballabriga



Global numbers 2021



506 PROJECTS

- 87** Competitive autonomic projects
- 236** Competitive national projects
- 58** Competitive international projects
- 10** Non competitive autonomic projects
- 100** Non competitive national projects
- 15** Non competitive international projects



720 RESEARCH STAFF

- 59** Group leader
- 280** Staff researcher
- 78** Postdoc - associate researcher
- 120** PhD student
- 183** Assistant researcher & Technician



321 CLINICAL TRIALS & OBSERVATIONAL STUDIES

- 26** Phase I
- 70** Phase II
- 115** Phase III
- 8** Phase IV
- 11** Other clinical trial
- 91** Observational studies



13 BUDGET*

- 68%** Private
- 32%** Public

* Thousand of euros



645 PUBLICATIONS

Articles & reviews published in scholarly journals indexed in the ISI Web of Science

- 3581** Impact Factor
- 64%** Publications in first quartile
- 5.55** Mean impact factor
- 52%** Articles with main contributing authors

Global numbers 2015-2021



PROJECTS

2015	251
2016	303
2017	360
2018	378
2019	470
2020	486
2021	506



RESEARCH STAFF

2015	321
2016	483
2017	552
2018	514
2019	594
2020	688
2021	720



CLINICAL TRIALS & OBSERVATIONAL STUDIES

2015	132
2016	124
2017	140
2018	147
2019	222
2020	231
2021	321



BUDGET*

2015	5.2
2016	6.5
2017	7.9
2018	9.2
2019	10.9
2020	12
2021	13

* Thousand of euros



PUBLICATIONS

2015	302 (1138 IF)
2016	376 (1442 IF)
2017	401 (1909 IF)
2018	443 (2287 IF)
2019	469 (2260 IF)
2020	590 (2652 IF)
2021	645 (3581 IF)

Institutional awards

HRS4R award

HRS4R Human Resources Strategy for Researchers

<https://www.irsjd.org/en/research/hrs4r-hr-excellence-research/>

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

CERCA

The Institut de Recerca Sant Joan de Déu (IRSJD) has been recognized by the board of trustees of the Institution for Research Centers in Catalonia (CERCA) as a CERCA centre in 2020

This award involves the development of a frontier research aimed at the scientific and economic impact and the improvement of the social and individual wellbeing.

The CERCA institute focuses its actions on the contribution to the international presence of research centres from Catalonia and to promote the cooperation and scientific exchange with the top centres and universities worldwide. Moreover, it promotes the adoption of joint policies in research management, scientific development and knowledge transfer. Also, it works in order to increase and ease knowledge transfer in the business sector and general public, promoting the involvement of these agents in the field of research.





2

STAFF



Research staff

Research staff

**720**

240 men (33%) 479 women (67%)

59 GROUP LEADER

31 men (53%) 28 women (47%)

280 STAFF RESEARCHER

115 men (41%) 165 women (59%)

78 POSTDOC ASSOCIATE RESEARCHER

24 men (31%) 54 women (69%)

120 PhD STUDENT

31 men (26%) 89 women (74%)

183 TECHNICIAN AND ASISTANT RESEARCHERS

40 men (22%) 143 women (78%)





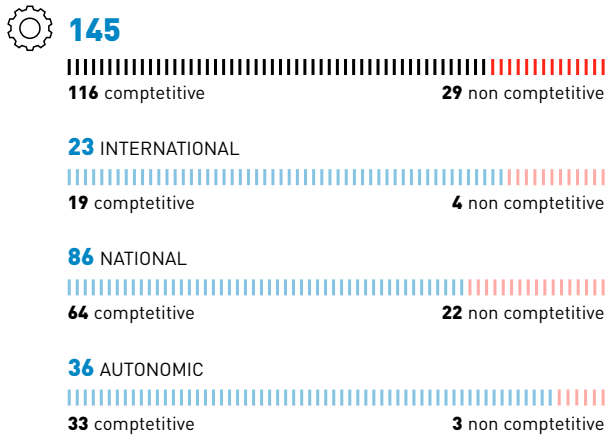
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PROJECTS AND CLINICAL TRIALS

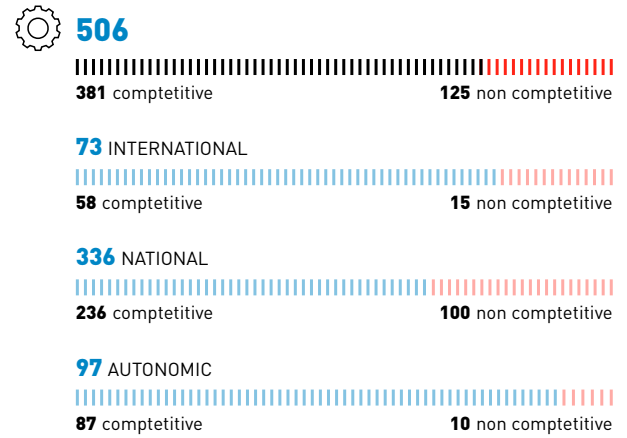


- New projects
- Active projects
- New clinical trials and observational studies
- Active clinical trials and observational studies

New projects

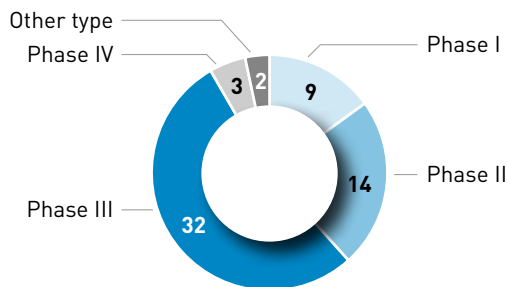


Active projects



New clinical trials and observational studies

Clinical Trials by Type



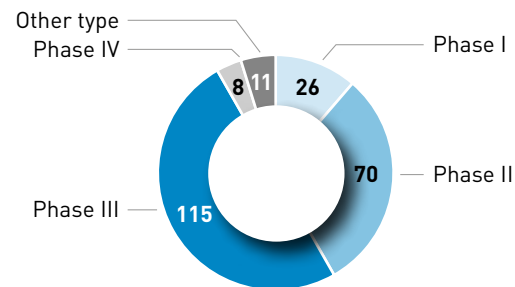
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CLINICAL TRIALS AND OBSERVATIONAL STUDIES BY RESEARCH AREA

- 12 NEUROLOGY**
(11 clinical trials & 1 observational study)
- 12 ONCOLOGY**
(12 clinical trials)
- 5 DERMATOLOGY**
(5 clinical trials)
- 4 CLINICAL HAEMATOLOGY**
(3 clinical trials & 1 observational study)
- 4 ALLERGY AND CLINICAL IMMUNOLOGY**
(2 clinical trials & 2 observational studies)
- 7 GASTROENTEROLOGY, HEPATOLOGY AND NUTRITION**
(5 clinical trials & 2 observational studies)
- 3 EMERGENCY ROOM AND INTERNAL MEDICINE**
(3 clinical trials)
- 1 NEPHROLOGY**
(1 clinical trial)
- 2 ORTHOPAEDICS AND TRAUMATOLOGY**
(1 clinical trial & 1 observational study)
- 1 PSYCHIATRY AND PSYCHOLOGY**
(1 clinical trial)
- 2 PNEUMOLOGY**
(2 clinical trials)
- 5 RHEUMATOLOGY**
(5 clinical trials)
- 2 INTENSIVE CARE UNIT**
(2 clinical trials)
- 7 INFECTIOUS DISEASES**
(7 clinical trials)
- 1 NURSERY**
(1 observational study)

Active clinical trials and observational studies

Clinical Trials by Type



230

CLINICAL TRIALS AND OBSERVATIONAL STUDIES BY RESEARCH AREA

- 3 PAEDIATRICS**
(2 clinical trials & 1 observational study)
- 63 NEUROLOGY**
(48 clinical trials & 15 observational studies)
- 16 CLINICAL HAEMATOLOGY**
(8 clinical trials & 8 observational studies)
- 12 EMERGENCY ROOM AND INTERNAL MEDICINE**
(9 clinical trials & 3 observational studies)
- 20 DERMATOLOGY**
(17 clinical trials & 3 observational studies)
- 13 ALLERGY AND CLINICAL IMMUNOLOGY**
(6 clinical trials & 7 observational studies)
- 11 GASTROENTEROLOGY, HEPATOLOGY AND NUTRITION**
(6 clinical trials & 5 observational studies)
- 8 GYNAECOLOGY AND OBSTETRICS**
(3 clinical trials & 5 observational studies)
- 5 NEONATOLOGY**
(5 clinical trials)
- 3 ANAESTHESIA**
(3 observational studies)
- 8 NEPHROLOGY**
(4 clinical trials & 4 observational studies)
- 6 PSYCHIATRY AND PSYCHOLOGY**
(5 clinical trials & 1 observational study)
- 5 ORTHOPAEDICS AND TRAUMATOLOGY**
(21 clinical trial & 3 observational studies)
- 6 PNEUMOLOGY**
(4 clinical trials & 2 observational studies)
- 3 OPHTHALMOLOGY**
(2 clinical trials & 1 observational study)
- 26 RHEUMATOLOGY**
(20 clinical trials & 6 observational studies)
- 4 CARDIOLOGY**
(3 clinical trials & 1 observational study)
- 2 REHABILITATION AND PHYSICAL MEDICINE**
(2 clinical trials)
- 14 ENDOCRINOLOGY**
(4 clinical trials & 10 observational studies)
- 3 DIGESTIVE MEDICINE**
(1 clinical trial & 2 observational studies)
- 24 INFECTIOUS DISEASES**
(21 clinical trials & 3 observational studies)
- 5 INTENSIVE CARE UNIT**
(5 clinical trials)



4

SCIENTIFIC PRODUCTION AND RESULTS



Scientific publications
Highest impact factor journals
Patents, utility models and spin-offs
Doctoral Theses

Scientific publications

	Output	Total IF	Mean IF	%Q1	%D1
Article	662	3440	5.5	64%	22%
Editorial material	7	25	3.6	0%	0%
Letter	22	173	7.9	68%	21%
Review	23	141	6.1	83%	0%
Others	40	365	9.1	72%	25%
Total	714	4143	5.8	64%	14%



Highest impact factor journals

ORIGINALS ARTICLES IN FIST DECIL

Number of original articles in first decil journals: **135**

Journal	Total articles	IF
NATURE MEDICINE	1	53,440
LANCET ONCOLOGY	1	41,316
EUROPEAN HEART JOURNAL	1	29,983
CIRCULATION	1	29,690
LANCET PSYCHIATRY	1	26,481
LANCET INFECTIOUS DISEASES	1	25,071
LANCET DIGITAL HEALTH	1	24,519
JOURNAL OF THE AMERICAN COLLEGE OF CARDIOLOGY	1	24,093
LANCET PUBLIC HEALTH	1	21,648
JAMA PSYCHIATRY	1	21,596
LANCET HAEMATOL	1	18,959
ADVANCED FUNCTIONAL MATERIALS	2	18,808
SCIENCE TRANSLATIONAL MEDICINE	2	17,992
CIRCULATION RESEARCH	1	17,367
ACTA NEUROPATHOLOGICA	2	17,088
NATURE COMMUNICATIONS	8	14,919
JOURNAL OF CLINICAL INVESTIGATION	1	14,808
BIOACTIVE MATERIALS	1	14,593
JOURNAL OF EXPERIMENTAL MEDICINE	1	14,307
JOURNAL FOR IMMUNOTHERAPY OF CANCER	1	13,751
NATURE METABOLISM	1	13,511
NATURE PROTOCOLS	1	13,491
ALLERGY	1	13,146
CLIN PSYCHOL REV	1	12,792
NEURO-ONCOLOGY	1	12,300
OPHTHALMOLOGY	2	12,079
TRENDS IN MOLECULAR MEDICINE	1	11,951
THERANOSTICS	1	11,556
MOLECULAR THERAPY	2	11,454
CLINICAL GASTROENTEROLOGY AND HEPATOLOGY	1	11,382

Highest impact factor journals

Journal	Total articles	IF
LANCET CHILD & ADOLESCENT HEALTH	2	11,288
ADDITIVE MANUFACTURING	2	10,998
ARTHRITIS & RHEUMATOLOGY	2	10,995
AGE AGEING	2	10,668
J NANOBIOTECHNOL	2	10,435
ANNALS OF NEUROLOGY	1	10,422
MOVEMENT DISORDERS	1	10,338
AMERICAN JOURNAL OF HEMATOLOGY	1	10,047
NEUROLOGY	2	9,910
ONCOGENE	1	9,867
BRITISH JOURNAL OF PSYCHIATRY	1	9,319
CLINICAL INFECTIOUS DISEASES	5	9,079
NEUROSCIENCE AND BIOBEHAVIORAL REVIEWS	1	8,989
GENETICS IN MEDICINE	2	8,822
BMC MEDICINE	1	8,775
ADV NUTR	1	8,701
METABOLISM-CLINICAL AND EXPERIMENTAL	1	8,697
AMERICAN JOURNAL OF OBSTETRICS AND GYNECOLOGY	1	8,661
CLINICAL CHEMISTRY	1	8,327
JOURNAL OF NEUROINFLAMMATION	1	8,322
CAN MED ASSOC J	1	8,262
ENVIRON POLLUT	1	8,071
JOURNAL OF MATERIALS SCIENCE & TECHNOLOGY	1	8,067
PLOS BIOLOGY	1	8,029
JOURNAL OF PATHOLOGY	1	7,996
MODERN PATHOLOGY	1	7,842
PSYCHOLOGICAL MEDICINE	1	7,723
OBSTET GYNECOL	1	7,661
JOURNAL OF MEDICINAL CHEMISTRY	1	7,446
ULTRASOUND IN OBSTETRICS & GYNECOLOGY	3	7,299
INTERNATIONAL JOURNAL OF EPIDEMIOLOGY	1	7,196

Highest impact factor journals

Journal	Total articles	IF
JOURNAL OF SPORT AND HEALTH SCIENCE	1	7,179
PEDIATRICS	2	7,125
ANALYTICAL CHEMISTRY	2	6,986
INT J BIOL MACROMOL	1	6,953
BIOINFORMATICS	1	6,937
EPIDEMIOLOGY AND PSYCHIATRIC SCIENCES	4	6,892
PLOS PATHOG	1	6,823
OSTEOARTHR CARTILAGE	1	6,576
NEUROIMAGE	1	6,556
BJOG-INT J OBSTET GY	1	6,531
BIOMEDICINE & PHARMACOTHERAPY	1	6,530
DEPRESSION AND ANXIETY	3	6,505
DEV COGN NEUROS-NETH	1	6,464
INTERNATIONAL JOURNAL OF BEHAVIORAL NUTRITION AND PHYSICAL ACTIVITY	2	6,457
PEDIATRIC ALLERGY AND IMMUNOLOGY	2	6,377
ANTIOXIDANTS	2	6,312
EUROSURVEILLANCE	1	6,307
TRAVEL MEDICINE AND INFECTIOUS DISEASE	1	6,211
J INTERPERS VIOLENCE	1	6,144
JOURNALS OF GERONTOLOGY SERIES A-BIOLOGICAL SCIENCES AND MEDICAL SCIENCES	1	6,053
BMJ GLOBAL HEALTH	2	5,558
JOURNAL OF MEDICAL INTERNET RESEARCH	2	5,428
MED SCI SPORT EXER	1	5,411
BRITISH JOURNAL OF GENERAL PRACTICE	1	5,386
AUTISM RESEARCH	1	5,216
DEVELOPMENTAL SCIENCE	1	5,131
EUROPEAN CHILD & ADOLESCENT PSYCHIATRY	2	4,785
JOURNAL OF PEDIATRICS	6	4,406
NEONATOLOGY	1	4,035
ARCH PHYS MED REHAB	1	3,966
JOURNAL OF ADVANCED NURSING	1	3,187

Highest impact factor journals

Originals articles D1 with IRSJD main contribution: 82

Original article	Journal	IF	Program
Rives S. CAR T cells in CNS-relapsed leukaemia: one step forward. LANCET HAEMATOL. 2021;8(10):675-676. IF:18,959. (Q1).	LANCET HAEMATOL	18,959	Paediatric Cancer
Ribó S, Sánchez D, Martínez L, García I, Ramon M, Tondo M et al. Increasing breast milk betaine modulates Akkermansia abundance in mammalian neonates and improves long-term metabolic health. SCI TRANSL MED. 2021;13(587):322IF:17,992. (Q1).	SCIENCE TRANSLATIONAL MEDICINE	17,992	Adaptive Response, Metabolism and Immunobiology
Demontis D, Walters RK, Rajagopal VM, Waldman ID, Grove J, Als TD et al. Risk variants and polygenic architecture of disruptive behavior disorders in the context of attention-deficit/hyperactivity disorder. Nat Commun. 2021;12(1):576-576. IF:14,919. (Q1).	NATURE COMMUNICATIONS	14,919	Precision Medicine in Rare Diseases
Esteve A, Anton J, Pino RM, Sánchez J, Fumadó V, Fortuny C et al. Similarities and differences between the immunopathogenesis of COVID-19-related pediatric multisystem inflammatory syndrome and Kawasaki disease. J CLIN INVEST. 2021;131(6):1-28. IF:14,808. (Q1).	JOURNAL OF CLINICAL INVESTIGATION	14,808	Neuroscience
Esteve A, Anton J, Pino RM, Sánchez J, Fumadó V, Fortuny C et al. Similarities and differences between the immunopathogenesis of COVID-19-related pediatric multisystem inflammatory syndrome and Kawasaki disease. J CLIN INVEST. 2021;131(6):1-28. IF:14,808. (Q1).	JOURNAL OF CLINICAL INVESTIGATION	14,808	Adaptive Response, Metabolism and Immunobiology
García C, Córdoba LC, Buxadera J, Marquina A, Jiménez E, Ginebra MP et al. Zn-Mg and Zn-Cu alloys for stenting applications: From nanoscale mechanical characterization to in vitro degradation and biocompatibility. Bioact Mater. 2021;6(12):4430-4446. IF:14,593. (Q1).	BIOACTIVE MATERIALS	14,593	Health Technologies and Innovation
Ruperez CH, Ferrer G, Cervera A, Florit L, Guitart M, Garrabou G et al. Meteorin-like/Meteorin-β protects heart against cardiac dysfunction. J Exp Med. 2021;218(5):20201206IF:14,307. (Q1).	JOURNAL OF EXPERIMENTAL MEDICINE	14,307	Adaptive Response, Metabolism and Immunobiology
Tornin J, Labay C, Tampieri F, Ginebra MP, Canal C. Evaluation of the effects of cold atmospheric plasma and plasma-treated liquids in cancer cell cultures. Nat Protoc. 2021;16(6):2826-2850. IF:13,491. (Q1).	NATURE PROTOCOLS	13,491	Health Technologies and Innovation
Pfaar O, Agache I, Bonini M, Brough HA, Chivato T, Del Giacco SR et al. COVID-19 pandemic and allergen immunotherapy-an EAACI survey. Allergy. 2021;76(11):3504-3516. IF:13,146. (Q1).	ALLERGY	13,146	Adaptive Response, Metabolism and Immunobiology
Palomer FX, Aguilar D, García R, Nistal JF, Vazquez M. Sirtuins: To Be or Not To Be in Diabetic Cardiomyopathy. Trends Mol Med. 2021;27(6):554-571. IF:11,951. (Q1).	TRENDS IN MOLECULAR MEDICINE	11,951	Adaptive Response, Metabolism and Immunobiology
Victor C, Schieber R, Raymond Y, Ségry B, Sabaté R, Kolandaivelu K et al. Solvent-cast direct-writing as a fabrication strategy for radiopaque stents. Additive Manufacturing. 2021;48(B):102392IF:10,998. (Q1).	ADDITIVE MANUFACTURING	10,998	Health Technologies and Innovation

Highest impact factor journals

Original article	Journal	IF	Program
Raymond Y, Thorel E, Liversain M, Riveiro A, Pou J, Ginebra M. 3D printing non-cylindrical strands: Morphological and structural implications. <i>Additive Manufacturing</i> . 2021;46:10,998. (Q1).	ADDITIVE MANUFACTURING	10,998	Health Technologies and Innovation
Smith L, Shin JI, Ghayda RA, Hijaz A, Sheyn D, Pope R et al. Physical multimorbidity and incident urinary incontinence among community-dwelling adults aged ≥50 years: findings from a prospective analysis of the Irish Longitudinal Study on Ageing. <i>Age Ageing</i> . 2021;50(6):2038-2046. IF:10,668. (Q1).	AGE AGEING	10,668	Mental Health and Society
Felez M, Haro JM, Stubbs B, Smith L, Koyanagi A. Moving more, ageing happy: findings from six low- and middle-income countries. <i>Age Ageing</i> . 2021;50(2):488-497. IF:10,668. (Q1).	AGE AGEING	10,668	Mental Health and Society
Tristan A, Borràs E, Molero M, Wassenberg T, Peters T, Verbeek MM et al. Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. <i>Mov Disord</i> . 2021;36(3):690-703. IF:10,338. (Q1).	MOVEMENT DISORDERS	10,338	Precision Medicine in Rare Diseases
Tristan A, Borràs E, Molero M, Wassenberg T, Peters T, Verbeek MM et al. Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. <i>Mov Disord</i> . 2021;36(3):690-703. IF:10,338. (Q1).	MOVEMENT DISORDERS	10,338	Neuroscience
Oliver L, Martin H, Reyes L, Nouredine F, Ada Cavalcanti E, Ginebra MP et al. An Engineered Biomimetic Peptide Regulates Cell Behavior by Synergistic Integrin and Growth Factor Signaling. <i>ADV HEALTHC MATER</i> . 2021;10(7):9,933. (Q1).	ADVANCED HEALTHCARE MATERIALS	9,933	Health Technologies and Innovation
Natera D, Foley AR, Domínguez C, Ortez CI, Jain M, Mebrahtu A et al. Association of Initial Maximal Motor Ability With Long-term Functional Outcome in Patients With COL6-Related Dystrophies. <i>Neurology</i> . 2021;96(10):1413-1424. IF:9,910. (Q1).	NEUROLOGY	9,910	Neuroscience
Crovetto F, Fátima B, Llorba E, Pascal R, Larroya M, Trilla C et al. Impact of Severe Acute Respiratory Syndrome Coronavirus 2 Infection on Pregnancy Outcomes: A Population-based Study. <i>Clin Infect Dis</i> . 2021;73(10):1768-1775. IF:9,079. (Q1).	CLINICAL INFECTIOUS DISEASES	9,079	Neuroscience
Brotons P, Launes C, Buetas E, Fumadó V, Henares D, Fernández de Sevilla M et al. Susceptibility to Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children and Adults: A Seroprevalence Study of Family Households in the Barcelona Metropolitan Region, Spain. <i>Clin Infect Dis</i> . 2021;72(12):1-8. IF:9,079. (Q1).	CLINICAL INFECTIOUS DISEASES	9,079	Neuroscience
García L, LÓpez M, GoncÉ A, Fortuny C, Salazar L, Valenzuela BI et al. Cardiac Remodeling and Hypertension in HIV-Uninfected Infants Exposed in utero to Antiretroviral Therapy. <i>Clin Infect Dis</i> . 2021;73(4):586-593. IF:9,079. (Q1).	CLINICAL INFECTIOUS DISEASES	9,079	Adaptive Response, Metabolism and Immunobiology
Crovetto F, Fátima B, Llorba E, Pascal R, Larroya M, Trilla C et al. Impact of Severe Acute Respiratory Syndrome Coronavirus 2 Infection on Pregnancy Outcomes: A Population-based Study. <i>Clin Infect Dis</i> . 2021;73(10):1768-1775. IF:9,079. (Q1).	CLINICAL INFECTIOUS DISEASES	9,079	Adaptive Response, Metabolism and Immunobiology

Highest impact factor journals

Original article	Journal	IF	Program
Brotons P, Launes C, Buetas E, Fumadó V, Henares D, Fernández de Sevilla M et al. Susceptibility to Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children and Adults: A Seroprevalence Study of Family Households in the Barcelona Metropolitan Region, Spain. <i>Clin Infect Dis.</i> 2021;72(12):1-8. IF:9,079. (Q1).	CLINICAL INFECTIOUS DISEASES	9,079	Adaptive Response, Metabolism and Immunobiology
Serra M, Radua J, Soriano C, Gómez A, Porta D, Marta R et al. Structural brain correlates in major depression, anxiety disorders and post-traumatic stress disorder: A voxel-based morphometry meta-analysis. <i>Neurosci Biobehav Rev.</i> 2021;129:269-281. IF:8,989. (Q1).	NEUROSCIENCE AND BIOBEHAVIORAL REVIEWS	8,989	Neuroscience
Dobaño C, Alonso S, Fernández de Sevilla M, Vidal M, Jiménez A, Pons G et al. Antibody conversion rates to SARS-CoV-2 in saliva from children attending summer schools in Barcelona, Spain. <i>BMC Med.</i> 2021;19(1):309-309. IF:8,775. (Q1).	BMC MEDICINE	8,775	Neuroscience
Dobaño C, Alonso S, Fernández de Sevilla M, Vidal M, Jiménez A, Pons G et al. Antibody conversion rates to SARS-CoV-2 in saliva from children attending summer schools in Barcelona, Spain. <i>BMC Med.</i> 2021;19(1):309-309. IF:8,775. (Q1).	BMC MEDICINE	8,775	Adaptive Response, Metabolism and Immunobiology
Dobaño C, Alonso S, Fernández de Sevilla M, Vidal M, Jiménez A, Pons G et al. Antibody conversion rates to SARS-CoV-2 in saliva from children attending summer schools in Barcelona, Spain. <i>BMC Med.</i> 2021;19(1):309-309. IF:8,775. (Q1).	BMC MEDICINE	8,775	Health Technologies and Innovation
Zarei M, Aguilar D, Palomer FX, Vazquez M. Revealing the role of peroxisome proliferator-activated receptor β/d in nonalcoholic fatty liver disease. <i>METABOLISM.</i> 2021;114:154342-154342. IF:8,697. (Q1).	METABOLISM-CLINICAL AND EXPERIMENTAL	8,697	Adaptive Response, Metabolism and Immunobiology
Peguero A, Herraiz I, Perales A, Melchor JC, Melchor I, Marcos B et al. Placental growth factor testing in the management of late preterm preeclampsia without severe features: a multicenter, randomized, controlled trial. <i>Am J Obstet Gynecol.</i> 2021;225(3):308IF:8,661. (Q1).	AMERICAN JOURNAL OF OBSTETRICS AND GYNECOLOGY	8,661	Adaptive Response, Metabolism and Immunobiology
Trifunov S, Paredes AJ, Badosa MC, Codina A, Montoya C, Ruiz E et al. Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. <i>Clin Chem.</i> 2021;67(8):1113-1121. IF:8,327. (Q1).	CLINICAL CHEMISTRY	8,327	Paediatric Cancer
Trifunov S, Paredes AJ, Badosa MC, Codina A, Montoya C, Ruiz E et al. Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. <i>Clin Chem.</i> 2021;67(8):1113-1121. IF:8,327. (Q1).	CLINICAL CHEMISTRY	8,327	Precision Medicine in Rare Diseases
Trifunov S, Paredes AJ, Badosa MC, Codina A, Montoya C, Ruiz E et al. Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. <i>Clin Chem.</i> 2021;67(8):1113-1121. IF:8,327. (Q1).	CLINICAL CHEMISTRY	8,327	Neuroscience

Highest impact factor journals

Original article	Journal	IF	Program
Afia AB, Vila È, MacDowell KS, Ormazabal A, Leza JC, Haro JM et al. Kynurenine pathway in post-mortem prefrontal cortex and cerebellum in schizophrenia: relationship with monoamines and symptomatology. <i>J Neuroinflammation</i> . 2021;18(1):198-198. IF:8,322. (Q1).	JOURNAL OF NEUROINFLAMMATION	8,322	Mental Health and Society
Afia AB, Vila È, MacDowell KS, Ormazabal A, Leza JC, Haro JM et al. Kynurenine pathway in post-mortem prefrontal cortex and cerebellum in schizophrenia: relationship with monoamines and symptomatology. <i>J Neuroinflammation</i> . 2021;18(1):198-198. IF:8,322. (Q1).	JOURNAL OF NEUROINFLAMMATION	8,322	Scientific base and approach to mental disorders
Afia AB, Vila È, MacDowell KS, Ormazabal A, Leza JC, Haro JM et al. Kynurenine pathway in post-mortem prefrontal cortex and cerebellum in schizophrenia: relationship with monoamines and symptomatology. <i>J Neuroinflammation</i> . 2021;18(1):198-198. IF:8,322. (Q1).	JOURNAL OF NEUROINFLAMMATION	8,322	Precision Medicine in Rare Diseases
Fernández D, Giné I, Liu I, Yucel R, Nai M, Morena M et al. Are environmental pollution and biodiversity levels associated to the spread and mortality of COVID-19? A four-month global analysis. <i>Environ Pollut</i> . 2021;271:116326-116326. IF:8,071. (Q1).	ENVIRON POLLUT	8,071	Mental Health and Society
Rodriguez A, Punset M, Calero JA, Javier F, Rupérez E, Manero JM. Powder metallurgy with space holder for porous titanium implants: A review. <i>J Mater Sci Technol</i> . 2021;76:129-149. IF:8,067. (Q1).	JOURNAL OF MATERIALS SCIENCE & TECHNOLOGY	8,067	Health Technologies and Innovation
Ferrer G, Guitart M, Ruperez CH, MATIAS R, Fátima B, Villarroya F et al. The protective effect of fibroblast growth factor-21 in alcoholic cardiomyopathy: a role in protecting cardiac mitochondrial function. <i>J Pathol</i> . 2021;253(2):198-208. IF:7,996. (Q1).	JOURNAL OF PATHOLOGY	7,996	Adaptive Response, Metabolism and Immunobiology
Jacob L, Smith L, McDermott D, Haro JM, Stickley A, Koyanagi A. Relationship between sexual orientation and psychotic experiences in the general population in England. <i>Psychol Med</i> . 2021;51(1):138-146. IF:7,723. (Q1).	PSYCHOLOGICAL MEDICINE	7,723	Mental Health and Society
Gonzalez E, Grau L, Ferrero SI, Hernandez A, Rebollo M, Gómez M et al. Pessary for Management of Cervical Varices Complicating Pregnancy. <i>Obstet Gynecol</i> . 2021;138(3):482-486. IF:7,661. (Q1).	OBSTET GYNECOL	7,661	Precision Medicine in Rare Diseases
Gonzalez E, Grau L, Ferrero SI, Hernandez A, Rebollo M, Gómez M et al. Pessary for Management of Cervical Varices Complicating Pregnancy. <i>Obstet Gynecol</i> . 2021;138(3):482-486. IF:7,661. (Q1).	OBSTET GYNECOL	7,661	Adaptive Response, Metabolism and Immunobiology
Basurto D, Fuenzalida J, Martinez RJ, Russo FM, Pertierra A, Martinez JM et al. Intrapulmonary artery Doppler to predict mortality and morbidity in fetuses with mild or moderate left-sided congenital diaphragmatic hernia. <i>Ultrasound Obstet Gynecol</i> . 2021;58(4):590-596. IF:7,299. (Q1).	ULTRASOUND IN OBSTETRICS & GYNECOLOGY	7,299	Adaptive Response, Metabolism and Immunobiology

Highest impact factor journals

Original article	Journal	IF	Program
Meler E, Mazarico E, Eixarch E, Gonzalez A, Peguero A, Martinez J et al. Ten-year experience of protocol-based management of small-for-gestational-age fetuses: perinatal outcome in late-pregnancy cases diagnosed after 32 weeks. <i>Ultrasound Obstet Gynecol.</i> 2021;57(1):62-69. IF:7,299. (Q1).	ULTRASOUND IN OBSTETRICS & GYNECOLOGY	7,299	Adaptive Response, Metabolism and Immunobiology
Sánchez A, Forero CG, Wu YT, Giné I, Prina M, De La Fuente J et al. Development of a common scale for measuring healthy ageing across the world: results from the ATHLOS consortium. <i>INT J EPIDEMIOL.</i> 2021;50(3):880-892. IF:7,196. (Q1).	INTERNATIONAL JOURNAL OF EPIDEMIOLOGY	7,196	Mental Health and Society
Barranco M, Solà P, Picart S, Kanaan S, Fonollosa J, Perera A. mWISE: An Algorithm for Context-Based Annotation of Liquid Chromatography-Mass Spectrometry Features through Diffusion in Graphs. <i>Anal Chem.</i> 2021;93(31):10772-10778. IF:6,986. (Q1).	ANALYTICAL CHEMISTRY	6,986	Health Technologies and Innovation
Tampieri F, Ginebra MP, Canal C. Quantification of Plasma-Produced Hydroxyl Radicals in Solution and their Dependence on the pH. <i>Anal Chem.</i> 2021;93(8):3666-3670. IF:6,986. (Q1).	ANALYTICAL CHEMISTRY	6,986	Health Technologies and Innovation
Babeli I, Puiggalí A, Roa JJ, Ginebra MP, José M, Alemán C. Hybrid conducting alginate-based hydrogel for hydrogen peroxide detection from enzymatic oxidation of lactate. <i>Int J Biol Macromol.</i> 2021;193(Pt B):1237-1248. IF:6,953. (Q1).	INT J BIOL MACROMOL	6,953	Health Technologies and Innovation
Picart S, Thompson WK, Buil A, Perera A. The effect of statistical normalization on network propagation scores. <i>Bioinformatics.</i> 2021;37(6):845-852. IF:6,937. (Q1).	BIOINFORMATICS	6,937	Health Technologies and Innovation
Ayuso JL, Morillo D, Haro JM, Olaya B, Lara E, Maria M. Changes in depression and suicidal ideation under severe lockdown restrictions during the first wave of the COVID-19 pandemic in Spain: a longitudinal study in the general population. <i>Epidemiol Psychiatr Sci.</i> 2021;30:1-27. IF:6,892. (Q1).	EPIDEMIOLOGY AND PSYCHIATRIC SCIENCES	6,892	Mental Health and Society
Cabello M, Rico LA, Martínez JC, Sánchez A, Caballero FF, Borges G et al. The role of ageing in the wish to be dead: disentangling age, period and cohort effects in suicide ideation in European population. <i>Epidemiol Psychiatr Sci.</i> 2021;30:1-27. IF:6,892. (Q1).	EPIDEMIOLOGY AND PSYCHIATRIC SCIENCES	6,892	Mental Health and Society
Jacob L, Kostev K. Osteoarthritis and the incidence of fracture in the United Kingdom: a retrospective cohort study of 258,696 patients. <i>Osteoarthritis Cartilage.</i> 2021;29(2):215-221. IF:6,576. (Q1).	OSTEOARTHR CARTILAGE	6,576	Mental Health and Society
Gorina N, Kurkela J, Hämäläinen J, Astikainen P, Escera C. Neural generators of the frequency-following response elicited to stimuli of low and high frequency: A magnetoencephalographic (MEG) study. <i>Neuroimage.</i> 2021;231:117866-117866. IF:6,556. (Q1).	NEUROIMAGE	6,556	Neuroscience
Peguero A, Fernandez L, Mazarico E, Benitez L, Gonzalez A, Youssef L et al. Added prognostic value of longitudinal changes of angiogenic factors in early-onset severe pre-eclampsia: a prospective cohort study. <i>BJOG.</i> 2021;128(2):158-165. IF:6,531. (Q1).	BJOG-INT J OBSTET GY	6,531	Adaptive Response, Metabolism and Immunobiology

Highest impact factor journals

Original article	Journal	IF	Program
Paredes AJ, Cesar S, Montero R, Latre C, Genovés J, Martorell L et al. Plasma idebenone monitoring in Friedreich's ataxia patients during a long-term follow-up. <i>Biomed Pharmacother.</i> 2021;143:112143-112143. IF:6,530. (Q1).	BIOMEDICINE & PHARMACOTHERAPY	6,530	Precision Medicine in Rare Diseases
Paredes AJ, Cesar S, Montero R, Latre C, Genovés J, Martorell L et al. Plasma idebenone monitoring in Friedreich's ataxia patients during a long-term follow-up. <i>Biomed Pharmacother.</i> 2021;143:112143-112143. IF:6,530. (Q1).	BIOMEDICINE & PHARMACOTHERAPY	6,530	Neuroscience
Paredes AJ, Cesar S, Montero R, Latre C, Genovés J, Martorell L et al. Plasma idebenone monitoring in Friedreich's ataxia patients during a long-term follow-up. <i>Biomed Pharmacother.</i> 2021;143:112143-112143. IF:6,530. (Q1).	BIOMEDICINE & PHARMACOTHERAPY	6,530	Adaptive Response, Metabolism and Immunobiology
Collado C, Navarro M, Adrián A, López Y, García J, Montero J. Effectiveness of mindfulness-based stress reduction and attachment-based compassion therapy for the treatment of depressive, anxious, and adjustment disorders in mental health settings: A randomized controlled trial. <i>Depress Anxiety.</i> 2021;38(11):1138-1151. IF:6,505. (Q1).	DEPRESSION AND ANXIETY	6,505	Mental Health and Society
Smith L, Il J, McDermott D, Jacob L, Barnett Y, López GF et al. Association between food insecurity and depression among older adults from low- and middle-income countries. <i>Depress Anxiety.</i> 2021;38(4):439-446. IF:6,505. (Q1).	DEPRESSION AND ANXIETY	6,505	Mental Health and Society
Felez M, Bort J, Ma R, Romano E, Faires M, Stubbs B et al. Light-intensity physical activity and mental ill health: a systematic review of observational studies in the general population. <i>Int J Behav Nutr Phys Act.</i> 2021;18(1):123-123. IF:6,457. (Q1).	INTERNATIONAL JOURNAL OF BEHAVIORAL NUTRITION AND PHYSICAL ACTIVITY	6,457	Mental Health and Society
Aznar I, Zabaleta E, Casajuana M, Sánchez A, Parody E, Bolívar B et al. Cost-effectiveness analysis of a multiple health behaviour change intervention in people aged between 45 and 75 years: a cluster randomized controlled trial in primary care (EIRA study). <i>Int J Behav Nutr Phys Act.</i> 2021;18(1):88-88. IF:6,457. (Q1).	INTERNATIONAL JOURNAL OF BEHAVIORAL NUTRITION AND PHYSICAL ACTIVITY	6,457	Mental Health and Society
García A, Buendia M, Deyà A, Lozano J, Serrano M, Van Den A RYM et al. Novel PGM3 compound heterozygous variants with IgE-related dermatitis, lymphopenia, without syndromic features. <i>Pediatr Allergy Immunol.</i> 2021;32(3):566-575. IF:6,377. (Q1).	PEDIATRIC ALLERGY AND IMMUNOLOGY	6,377	Precision Medicine in Rare Diseases
García A, Buendia M, Deyà A, Lozano J, Serrano M, Van Den A RYM et al. Novel PGM3 compound heterozygous variants with IgE-related dermatitis, lymphopenia, without syndromic features. <i>Pediatr Allergy Immunol.</i> 2021;32(3):566-575. IF:6,377. (Q1).	PEDIATRIC ALLERGY AND IMMUNOLOGY	6,377	Adaptive Response, Metabolism and Immunobiology
García R, Gavalda A, Villarroya F, Marfany G, Mirra S. Overexpression of CERKL Protects Retinal Pigment Epithelium Mitochondria from Oxidative Stress Effects. <i>Antioxidants (Basel).</i> 2021;10(12):2018IF:6,312. (Q1).	ANTIOXIDANTS	6,312	Precision Medicine in Rare Diseases

Highest impact factor journals

Original article	Journal	IF	Program
García R, Gavalda A, Villarroya F, Marfany G, Mirra S. Overexpression of CERKL Protects Retinal Pigment Epithelium Mitochondria from Oxidative Stress Effects. <i>Antioxidants (Basel)</i> . 2021;10(12):2018IF:6,312. (Q1).	ANTIOXIDANTS	6,312	Adaptive Response, Metabolism and Immunobiology
Durán Fernández C, Rodríguez J, López M, Hadley S, Cavia M, Muñoz P et al. Effects of Hypothermia and Allopurinol on Oxidative Status in a Rat Model of Hypoxic Ischemic Encephalopathy. <i>Antioxidants (Basel)</i> . 2021;10(10):1523IF:6,312. (Q1).	ANTIOXIDANTS	6,312	Adaptive Response, Metabolism and Immunobiology
Lamonja N, Dacosta R, López J, Prades L, Roig F, Castells A et al. Sex-Specific Protective Effects of APOE e2 on Cognitive Performance. <i>J Gerontol A Biol Sci Med Sci</i> . 2021;76(1):41-49. IF:6,053. (Q1).	JOURNALS OF GERONTOLOGY SERIES A-BIOLOGICAL SCIENCES AND MEDICAL SCIENCES	6,053	Neuroscience
Martinez A, Bekele D, Sabariego C, Rodríguez Á, Vorstenbosch E, Rico LA et al. The Structural and Intercultural Competence for Epidemiological Studies (SICES) guidelines: a 22-item checklist. <i>BMJ Glob Health</i> . 2021;6(4)IF:5,558. (Q1).	BMJ GLOBAL HEALTH	5,558	Mental Health and Society
Santini ZI, Koyanagi A, Stewart S, Perry BD, Marmot M, Koushede V. Cumulative risk of compromised physical, mental and social health in adulthood due to family conflict and financial strain during childhood: a retrospective analysis based on survey data representative of 19 European countries. <i>BMJ Glob Health</i> . 2021;6(3)IF:5,558. (Q1).	BMJ GLOBAL HEALTH	5,558	Mental Health and Society
Tyrovolas S, Giné I, Fernández D, Morena M, Koyanagi A, Janko M et al. Estimating the COVID-19 Spread Through Real-time Population Mobility Patterns: Surveillance in Low- and Middle-Income Countries. <i>J Med Internet Res</i> . 2021;23(6)IF:5,428. (Q1).	JOURNAL OF MEDICAL INTERNET RESEARCH	5,428	Mental Health and Society
Castells A, Roig F, Lamonja N, Torán P, Pera G, Montero P et al. Sex Matters in the Association between Physical Activity and Fitness with Cognition. <i>Med Sci Sports Exerc</i> . 2021;53(6):1252-1259. IF:5,411. (Q1).	MED SCI SPORT EXER	5,411	Neuroscience
Pijuan J, Ortigoza JD, Juan Jairo Ortiz G, Alcalá A, Calvo MJ, Cubells M et al. PLXNA2 and LRRC40 as candidate genes in autism spectrum disorder. <i>AUTISM RES</i> . 2021;14(6):1088-1100. IF:5,216. (Q1).	AUTISM RESEARCH	5,216	Precision Medicine in Rare Diseases
François C, Rodriguez A, Teixidó M, Agut T, Bosch L. Attenuated brain responses to speech sounds in moderate preterm infants at term age. <i>Dev Sci</i> . 2021;24(1):12990IF:5,131. (Q1).	DEVELOPMENTAL SCIENCE	5,131	Neuroscience
Calderon M, Vila R, Dolz M, Butjosa A, Barajas A, del Cacho N et al. Depressive symptoms and their relationship with negative and other psychotic symptoms in early onset psychosis. <i>Eur Child Adolesc Psychiatry</i> . 2021;30(9):1383-1390. IF:4,785. (Q1).	EUROPEAN CHILD & ADOLESCENT PSYCHIATRY	4,785	Mental Health and Society
Calderon M, Vila R, Dolz M, Butjosa A, Barajas A, del Cacho N et al. Depressive symptoms and their relationship with negative and other psychotic symptoms in early onset psychosis. <i>Eur Child Adolesc Psychiatry</i> . 2021;30(9):1383-1390. IF:4,785. (Q1).	EUROPEAN CHILD & ADOLESCENT PSYCHIATRY	4,785	Scientific base and approach to mental disorders

Highest impact factor journals

Original article	Journal	IF	Program
Zerón MF, Carpio TV, Estrella G, Díez A, Cambras T, Alda JA et al. ADHD subtypes are associated differently with circadian rhythms of motor activity, sleep disturbances, and body mass index in children and adolescents: a case-control study. <i>Eur Child Adolesc Psychiatry</i> . 2021;30(12):1917-1927. IF:4,785. (Q1).	EUROPEAN CHILD & ADOLESCENT PSYCHIATRY	4,785	Neuroscience
Calderon M, Vila R, Dolz M, Butjosa A, Barajas A, del Cacho N et al. Depressive symptoms and their relationship with negative and other psychotic symptoms in early onset psychosis. <i>Eur Child Adolesc Psychiatry</i> . 2021;30(9):1383-1390. IF:4,785. (Q1).	EUROPEAN CHILD & ADOLESCENT PSYCHIATRY	4,785	Neuroscience
García A, Arnaez J, Arca G, Agut T, Alarcon A, Martín A et al. Development, Reliability, and Testing of a New Rating Scale for Neonatal Encephalopathy. <i>J Pediatr</i> . 2021;235:83-91. IF:4,406. (Q1).	JOURNAL OF PEDIATRICS	4,406	Neuroscience
Iglesias I, Beardsall K. Strict glycemic control in very low birthweight infants using continuous glucose monitoring reduces dysglycemic. <i>J Pediatr</i> . 2021;238:338-342. IF:4,406. (Q1).	JOURNAL OF PEDIATRICS	4,406	Adaptive Response, Metabolism and Immunobiology
Martínez A, Baquero F, Santiago B, Fortuny C, Méndez A, Del Rosal T et al. Interferon-Gamma Release Assays Differentiate between <i>Mycobacterium avium</i> Complex and Tuberculous Lymphadenitis in Children. <i>J Pediatr</i> . 2021;236:211 IF:4,406. (Q1).	JOURNAL OF PEDIATRICS	4,406	Adaptive Response, Metabolism and Immunobiology
Aldecoa V, Velilla M, Teresa M, Balcells C, Herranz A, Sin M et al. Lung Ultrasound in Bronchopulmonary Dysplasia: Patterns and Predictors in Very Preterm Infants. <i>Neonatology</i> . 2021;118(5):537-545. IF:4,035. (Q1).	NEONATOLOGY	4,035	Neuroscience
Aldecoa V, Velilla M, Teresa M, Balcells C, Herranz A, Sin M et al. Lung Ultrasound in Bronchopulmonary Dysplasia: Patterns and Predictors in Very Preterm Infants. <i>Neonatology</i> . 2021;118(5):537-545. IF:4,035. (Q1).	NEONATOLOGY	4,035	Adaptive Response, Metabolism and Immunobiology
Jacob L, López GF, Kostev K, Schnitzler A, Haro JM, Koyanagi A et al. Sexual Orientation and Functional Limitations: Cross-sectional Analyses From the Adult Psychiatric Morbidity Survey. <i>Arch Phys Med Rehabil</i> . 2021;102(11):2117-2124. IF:3,966. (Q1).	ARCH PHYS MED REHAB	3,966	Mental Health and Society

Patents, utility models and spin-offs

New patents, license agreements and utility models

Elemento de fijación de dispositivos a nivel craneal para neonatos

Country: Spain
Number: EP3368044
Inventors: Nuria Carrera Blesa
Proprietors: Hospital Sant Joan de Déu Barcelona

Peptidic Conjugates of SN38 useful in the treatment of cancer

Date: 15/01/2021
Country: PCT
Number: PCT/EP2021/076546
Inventors: Macare Sánchez Navarro, Meritxell Teixidó Turà, Nagore Gené Olaciregui and Angel Montero Carcaboso
Proprietors: Hospital Sant Joan de Déu Barcelona, Fundació Privada Institut de Recerca Biomèdica de Barcelona and Universitat de Barcelona

Válvula para respiradores (PEEP)

Country: Germany and Europe
Number: 2021042107265100DE and EP213823131
Inventors: Enric Palau Forte, Arnau Valls Esteve, Martí Pons Odena, Carlos Alaez Vasconcellos, Lluís Font Vizcarra, Adrián Bermúdez Castel, Xoel Pena Pérez, Marc Murcia Laguna
Proprietors: Hospital Sant Joan de Déu Barcelona

Tratamiento de la enfermedad de Charcot-Marie-Tooth

Country: Spain
Number: P202130576
Inventors: Jose Manuel Cuerza Marcos, Cristina Nuevo Tapióles, Francesc Palau Martínez, Jorgina Satrutegui Gil-Delgado
Proprietors: Hospital Sant Joan de Déu Barcelona, Fundació General de la Universidad Autónoma de Madrid and Centro de Investigación Biomédica en Red (CIBER)

Válvula para dispositivos de ventilación (VENTURI)

Country: Europe
Number: EP21165556

Inventors: Francis de Zegher and Lourdes Ibáñez
Proprietors: Hospital Sant Joan de Déu Barcelona and University Leuven Kath

Treatment of hepatic and or visceral fat excess

Country: Spain
Number: P2021382313
Inventors: Lluís Font Vizcarra, Arnau Valls Esteve, Carlos Alaez Vasconcellos and Martí Pons
Proprietors: Hospital Sant Joan de Déu Barcelona

GD2 Administration regimen

Country: Europe
Number: EP21383131
Inventors: Jaume Mora Graupera, Nai-Kong Cheung, Modak Shakeel, Vignesh Rajah and Claus J Moller SanPedro
Proprietors: Y-mAbs Therapeutics, Inc

Betaine for prevention of obesity

Country: Europe
Number: EP3621605
Inventors: Josep Jiménez Chillaron, Carles Lerín and Marta Ramón
Proprietors: Hospital Sant Joan de Déu Barcelona

Treatment of hepatic steatosis related oligo-ovulation

Country: Europa, Australia and Rusia
Number: EP3368044, AU2016344737 and RU2745606
Inventors: Francis de Zegher and Lourdes Ibáñez
Proprietors: Hospital Sant Joan de Déu Barcelona and University Leuven Kath

A medical ventilator system and a method for predicting transient states of a ventilated patient upon changes in mechanical ventilator settings

Country: Spain
Number: EP21382381.8
Inventors: L. Serna, M.A. Mañanas, A. Hernández
Proprietors: Universitat Politècnica de Catalunya and Universidad de Antioquia

Portfolio of spin-offs

Ephion Health S.L.

2021 · <https://ephion.health/>
Co-funders: Eurecat, Hospital Sant Joan de Déu and The Collider
Ephion Health is the platform to develop and deploy clinical measures for patient mobility. They assist the complete parametrization of mobility for several diseases and conditions, to create a positive impact on the patient's diagnosis, treatment and quality of life. Ephion's technology allows to save time and resources in clinical care and clinical trials.

Gate2Brain

2020 · <https://gate2brain.com/>
Co-funders: Universidad de Barcelona (UB), Hospital Sant Joan de Déu (HSJD)
Gate2Brain is a biotech company focused on the development of therapeutics that efficiently cross biological barriers such as the blood-brain barrier. We pursue our goals using a radically innovative peptide-based patented technology platform.

OMADRE

2019 · <https://www.omadre.com/>
Dr. Jaume Pérez Payarols (IRSJD)
The company Omadre was created to provide hospitals with the tools required for the efficient management of mother's milk, increasing the safety of existing processes in this way.

ABLE Human Motion

2018 · <https://www.ablehumanmotion.com/>
Co-funders: Dr. Alfons Carnicero, Dr. Alex García & Dr. Josep Maria Font (IRSJD)
ABLE Human Motion is a medical device spin-off from Universitat Politècnica de Catalunya (UPC) that is developing and will commercialize robotic exoskeletons to enhance mobility and independence of people with lower-limb paralysis. We have

Patents, utility models and spin-offs

created ABLE, the first lightweight, easy-to-use and affordable exoskeleton that allows walking again after a spinal cord injury. It helps palliating health complications while boosting self-confidence and independence.

DBGEN Ocular Genomics

2018 · <https://dbgen.com/>
Co-funders: Dr. Roser González-Duarte and Dr. Gemma Marfany (IRSJD)

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

CEBIOTEX

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

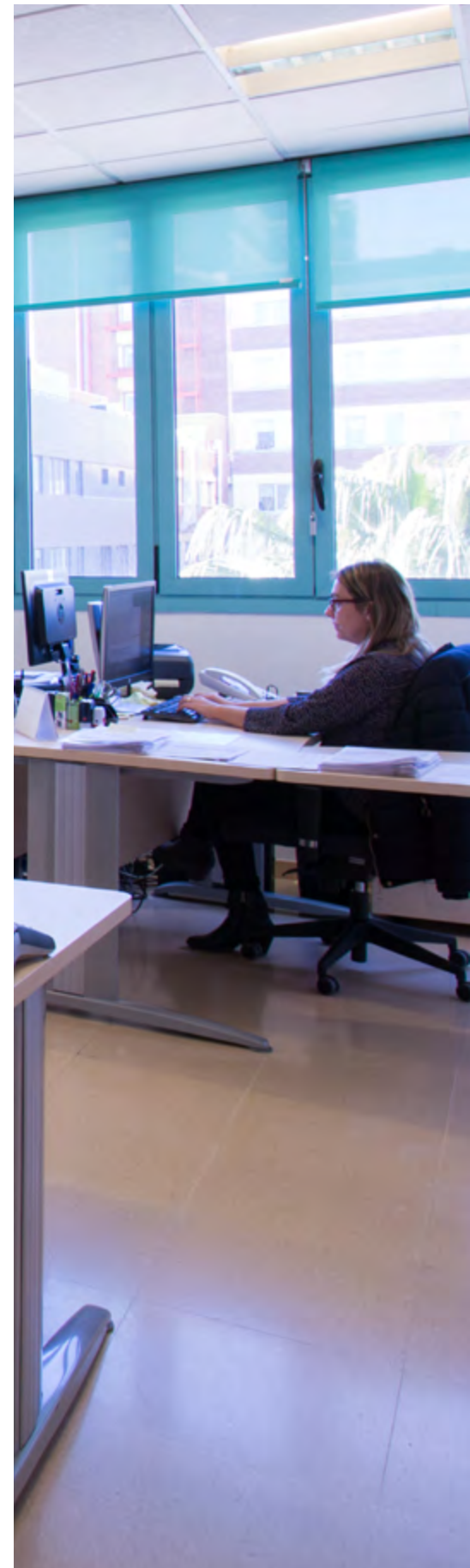
Cebiotex has developed and characterised at preclinical level, in vitro and in vivo, nonwoven nanofiber membranes for the local release of active ingredients in the local treatment of the surgical site after tumour excision surgery. The purpose of the local release of drugs is to achieve high concentrations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

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

BCN Innova

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


BCN Innova, a spin-off company of Sant Joan de Déu-Barcelona Children's Hospital, has developed a device (GazeLab) that substantially improves the diagnosis and treatment of strabismus, reducing the number of second operations that are now performed to achieve the expected results. GazeLab offers two main benefits: 1. It allows objective exploration on a revolutionary level of precision, with much better results than the subjective-measurement model that has prevailed until now. 2. It provides a 50% reduction in the number of second operations, obviating those which are surgically unnecessary.





Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	European mention
Eliana Carolina Gonçalves	Combined therapies for neuroblastoma based on the activation of the calcium sensing receptor	Universitat de Barcelona	Cinzia Lavarino and Silvia Mateo	January	
Maria Julieta Gonzalez	Avances en el estudio de la fenilcetonuria	Universitat de Barcelona	Jaume Campistol and Rafael Artuch	February	
Elena Borrego Domènech	La funció de CERKL a la retina: generació d'un model de ratolí i anàlisi de la seva implicació en la resposta a estrès oxidatiu	Universitat de Barcelona	Gemma Marfany	February	
Èlia Vidal	Development of metallic functionalized biomaterials with low elastic modulus for orthopedic applications	Universitat Politècnica de Catalunya	Daniel Rodríguez and Elisa Rupérez	February	
Núria Martínez-Gil	Role of Wnt pathway genes in complex and monogenic phenotypes of low and high bone mass	Universitat de Barcelona	Susanna Balcells and Daniel Grinberg	March	
Montserrat Bou	Papel de la microbiota intestinal en el tratamiento del síndrome de Prader-Willi	Universitat de Barcelona	Carles Lerin and Marta Ramon	April	
Marta Batllori	Investigació en biomarcadors de l'estat de la dopamina i la serotonina en pacients neuropediàtrics	Universitat de Barcelona	Marta Molero and Aida Ormazabal	April	
Anu Shivalikanjli	Shared and specific genetic basis of autism spectrum disorders and ADHD	Universitat de Barcelona	Bru Cormand and Noèlia Fernández	March	
América Vera Montecinos	Proteomic profil·le in postmortem brain in chronic schizophrenia	Universitat de Barcelona	Belen Ramos	March	
Jordina Tor	Riesgo de psicosis en població infanto-juvenil: característiques clíniques y cognitives	Universitat de Barcelona	Luis San Molina and Anna Sintès	March	
América Vera Montecinos	Proteomic profil·le in postmortem brain in chronic schizophrenia	Universitat de Barcelona	Belén Ramos	March	
Ivet Bayés Marín	Estudio de enfermedades crónicas de salud y funcionamiento en personas de mediana edad y mayores desde una perspectiva epidemiológica	Universitat de Barcelona	Beatriz Olaya and Josep Maria Haro	May	

Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	European mention
Carme Nolla Colomer	Anàlisi de patrons funcionals i estructurals en la regulació del calci en les cèl·lules cardíques	Universitat Politècnica de Catalunya	Raül Benítez and Leif Hove Madsen	May	
Laura Almeida	Modelo de síndrome alcohólico fetal en ratón según dos patrones de consumo humano y el papel de la epigallocatequina galato en su prevención. estudio de biodisponibilidad de la epigallocatequina galato en humanos	Universitat de Barcelona	V Andreu (buscar) and Maria Dolores Gómez Roig	June	
Silvia Simó	Implementació i resultats d'un programa d'optimització d'ús d'antimicrobians (PROA) pediàtric a un hospital infantil de tercer nivell	Universitat de Barcelona	Antoni Noguera	June	
Sara Bobillo	Utilidad de los biomarcadores en la respuesta inflamatoria sistémica y en la implementación de algoritmos de manejo de la antibioticoterapia en los pacientes críticos pediátricos	Universitat de Barcelona	Iolanda Jordan and Francisco Jose Cambra	June	
Joaquim Minguela	Surface characterization and cell instructive properties of superficially modified dental zirconia	Universitat Politècnica de Catalunya	Carles Mas-Moruno and Joan-Josep Roa	July	
Bernardo Sánchez Fernández	Diferencias de género en el adolescente con conducta suicida	Universitat Autònoma de Catalunya	Francisco Villar Cabeza, Mar Vila Grifoll, Mar and Anna Sintés Estévez	July	
Ester Antón Galido	Genetic and functional analyses of Autism Spectrum Disorder and ADHD	Universitat de Barcelona	Bru Cormand and Noèlia Fernández	September	
David Aguilar-Recarte	GDF15, a novel regulator of the AMPK-mediated antidiabetic actions of PPARbeta/delta and metformin	Universitat de Barcelona	Manuel Vázquez Carrera	September	
Santiago Raymond	Biomimetic Bone Grafts: from the lab to the clinic	Universitat Politècnica de Catalunya	Maria Pau Ginebra	September	
Joanna M. Konka	3D-Printed Biomimetic Bone	Universitat Politècnica de Catalunya	Maria Pau Ginebra and Montserrat Español	October	

Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	European mention
Alejandro Delgado Anglés	Experimental and human studies on aging of adipose tissues: role of Parkin	Universitat de Barcelona	Francesc Villarroya and Joan Villarroya	October	
Diego Torres	Desarrollo de un nuevo proceso basado en la inyección 3D de tintas con carga metálica para fabricar prótesis porosas de titanio bioactivas y con propiedades antibacterianas	Universitat Politècnica de Catalunya	José María Manero, Elisa Rupérez and José A. Calero (AMES).	October	
AnnaHuguet Miguel	Influencia de un programa de mindfulness sobre los síntomas nucleares, las funciones ejecutivas y el eje hipotálamo-hipófisis-adrenal (HPA), en niños recientemente diagnosticados de Trastorno por Déficit de Atención e Hiperactividad (TDAH). Un estudio aleatorizado	Universitat de Barcelona	José Ángel Alda	October	
Laura Castilla-Vallmanya	Combining exome sequencing and functional studies to identify causal genes of ultra-rare neurodevelopmental disorders	Unveritat de Barcelona	Susanna Balcells and Roser Urreiziti	November	
Marley Vega	Development and Applications of Neuromusculoskeletal Modeling Software for Personalized Treatment Design	Rice University (Houtson · USA)	Josep Maria Font and B.J. Fregly (Rice University)	November	
Natalia Diaz Garrido	Modulation of intestinal immune responses by microbiota-derived extracellular vesicles	Universitat de Barcelona	Laura Baldomà and Josefa Badia	November	
Estela Prada	Target Therapy in Rhabdomyosarcoma: Discovering new targets and opportunities	Universitat de Barcelona	Jaume Mora and Inmaculada Hernández (IRB Barcelona)	November	
Laia Villalta Macia	Emotional dysregulation, psychiatric disorders and trauma in adolescent	Universitat de Barcelona		November	
Teresa Ribas-Prats	Detection of abnormal neural encoding of speech sounds at birth using the frequency-following response	Universitat de Barcelona	Carles Escera	Desember	



5

CORE FACILITIES



Core facilities

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

Scientific core facilities

- Biological Sample Processing and Molecular Analysis
- Confocal Microscopy and Cellular Imaging Unit
- Clinical Trials Unit
- Library
- Methodology and statistics
- Neuroimaging
- Neurological tissues Biobank for research
- Neuropsychological evaluations
- Paediatric Biobank for research
- Patient Engagement in Research Area
- Sequencing Unit
- Animal housing facility
- Unit of Desing, Analysis and Interpretation of Omic data (DAIOmics)

Management support services

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

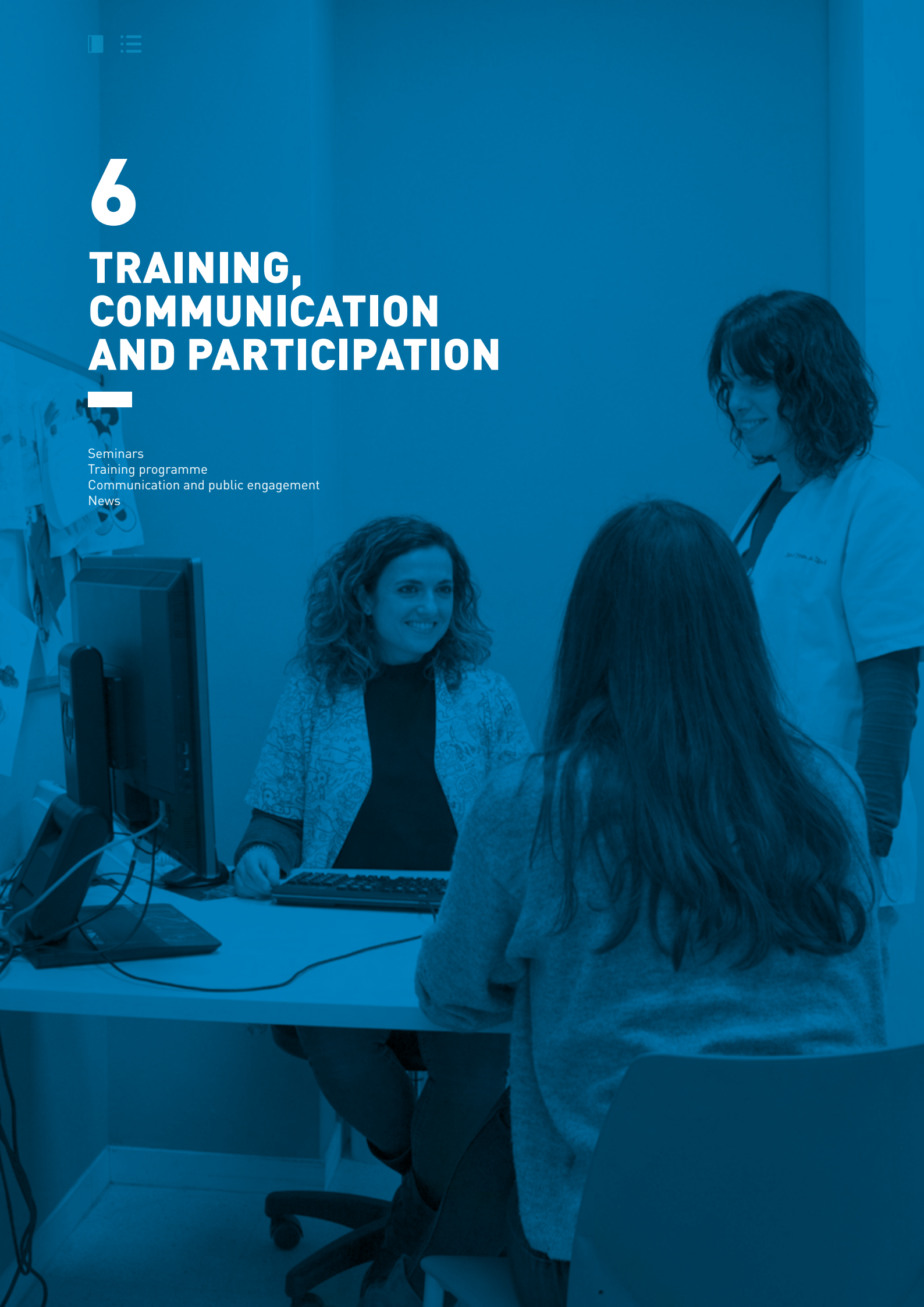




6

TRAINING, COMMUNICATION AND PARTICIPATION

Seminars
Training programme
Communication and public engagement
News



Seminars

22 SEMINARS

Relation of research seminars and IRSJD day of debates

21 JANUARY

Edició genètica amb CRISPR en Biomedicina: on estem actualment?

Dr. Lluís Montoliu, CSIC Centro
Nacional de Biotecnología (CNB)

9 FEBRUARY

Microbiota en l'entorn maternal- infantil: rellevància per a la salut

Dra. María Carmen Collado. CSIC
Institut d'Agroquímica i Tecnologia
d'Aliments (IATA)

16 FEBRUARY

Mitochondria-lysosome membrane contacts are defective in GDAP1- related Charcot-Marie-Tooth disease

Dra. Lara Cantarero Abad, IRSJD

23 FEBRUARY

Model de síndrome alcohòlic fetal en ratolí segons dos patrons de consum humà i el paper de la EGCG en la seva prevenció

Dra. Laura Almeida, IRSJD

27 FEBRUARY

Nano Rare Disease Day 2020

Organise: Nanomed Spain with IBEC
& IRSJD

16 MARCH

Similarities and differences between the immunopathogenesis of COVID- 19-related pediatric inflammatory multisystem syndrome and Kawasaki disease

Dra. Ana Esteve-Solé, IRSJD

30 MARCH

Impacte emocional i de la conducta de la pandèmia COVID19 en població infanto-juvenil

Dra. Esther Via & Dr. Xavier Estrada,
IRSJD

13 APRIL

Detecció de disfuncionalitats del sistema nerviós central en nounats mitjançant marcadors

neurofisiològics: la resposta de seguiment de freqüència (RSF)

Dra. Teresa Ribas-Prats, IRSJD

27 APRIL

CERKL, a retinal dystrophy gene, regulates mitochondrial function and dynamics in the mammalian retina

Dra. Serena Mirra, IRSJD.

11 MAY

Role of Wnt pathway genes in complex and monogenic phenotypes of low and high bone mass

Dra. Núria Martínez Gil, IRSJD

25 MAY

The spectrum of COVID-19 in pregnancy

Dra. Rosalia Pascal, IRSJD, & Dra.
Francesca Crovetto (IDIBAPS)

15 JUNE

Prevalença i factors explicatius de la no iniciació pediàtrica i l'impacte en costos de la no iniciació pediàtrica en antibiòtics i medicaments de salut mental

Cristina Carbonell, IRSJD, & Alba
Sánchez, IRSJD

29 JUNE

Addressing Clinical Psychology's WEIRD Problem: The cross- cultural validity of the Five-Facet Mindfulness Questionnaire across 16 countries

Dr. Juan Vicente Luciano, IRSJD.

13 JULY

Gas Plasma technology in Osteosarcoma: advances and challenges of a novel selective therapy based on oxidative-stress

Dra. Cristina Canal, IRSJD

15 SEPTEMBER

Epigenetic Genotyping (EpiGe): A novel approach for single cytosine methyl-genotyping and its application

in Pediatric Oncology

Dra. Soledad Gómez, IRSJD

28 SEPTEMBER

Simulation of Assisted Motion with Musculoskeletal Models

Dr. Albert Peiret Giménez, IRSJD

30 SEPTEMBER

Functionalizing the Cancer Genome Through Copy Number Engineering

Dr. Francisco Barriga, Memorial Sloan
Kettering Cancer Center. New York. USA

19 OCTOBER

Efecte de la nicotinamida, la forma amida de la vitamina B3, contra el desenvolupament d'obesitat

Dr. Josep Julve, IRHSCSP

26 OCTOBER

La no iniciació del tractament farmacològic des de la perspectiva dels pacients i els professionals

M^a Teresa Peñarrubia-Maria, IRSJD, &
Dra. Montserrat Gil Girbau, IRSJD

16 NOVEMBER

ANXA11: un gen, dues malalties neuromusculars i l'espectre fenotípic d'una proteinopatia

Dr. Daniel Natera, IRSJD, & Jonathan
Olival, IRSJD

30 NOVEMBER

Estrès psicosocial (trauma infantil) i factors protectors en l'aparició d'un primer episodis psicòtic

Regina Vila Badia, IRSJD, & Clara
Serra Arumí, IRSJD

14 DECEMBER

Relationship between the circadian rhythms disruption in children with neurodevelopmental disorder (ASD) and healthy controls with Body Mass Index (BMI) and an altered chronotype

Ares Sentenach, IRSJD

Training programme

18
COURSES IN 2021

250
ATTENDEES IN 2021

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

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

Courses

- How to publish a scientific article
- Treball en equips a distància
- Oportunitats de finançament competitiu
- Improving your paragraphs and sentences in english
- Organització i gestió del temps
- Estadística
- Writing Retreat (2ed)
- Per què és important la divulgació científica?
- Learn to write concise and impactful reports
- Lideratge (2 semestre)
- Excel nivell mig
- Excel nivell avançat
- R programació Basica
- Bones pràctiques en investigació
- Participació del pacient en recerca
- Sessió d'especialització: RGPD a Recerca
- Sensibilización en materia de igualdad
- Visibilidad y divulgación de la actividad investigadora

Communication and public engagement

Activities

18
ACTIVITIES ORGANIZED

170
RESEARCHERS

1.000
AUDIENCE REACHED

Participation in:

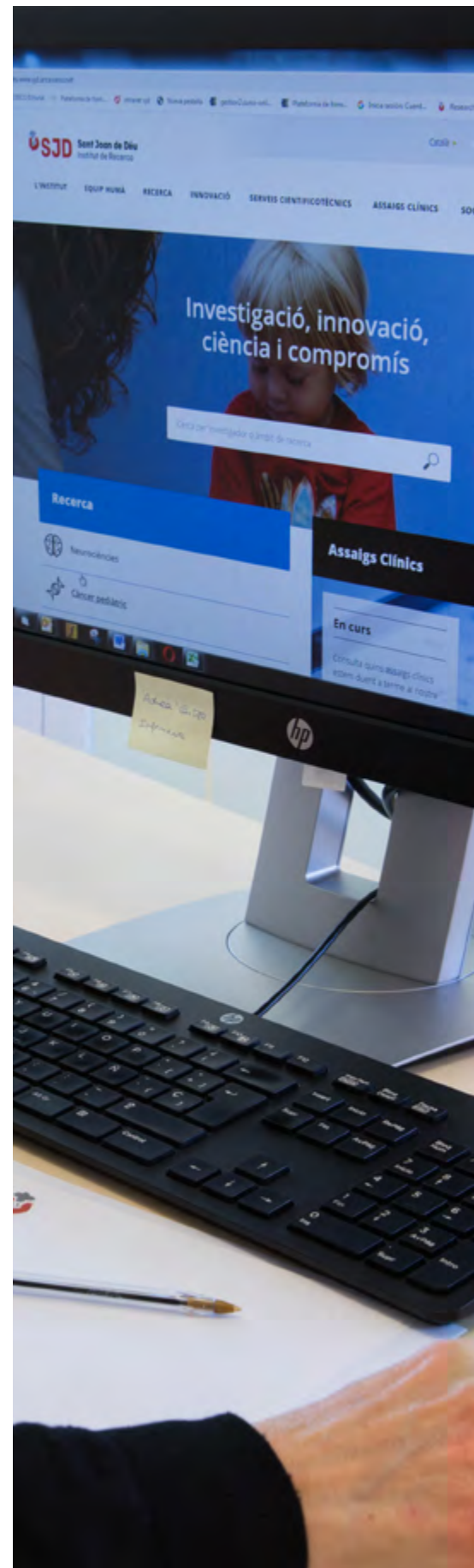
- International Day of women and girls in science
- Europeans Researcher's Night
- Setmana de la Ciència
- Festa de la Ciència

Internet and Social media

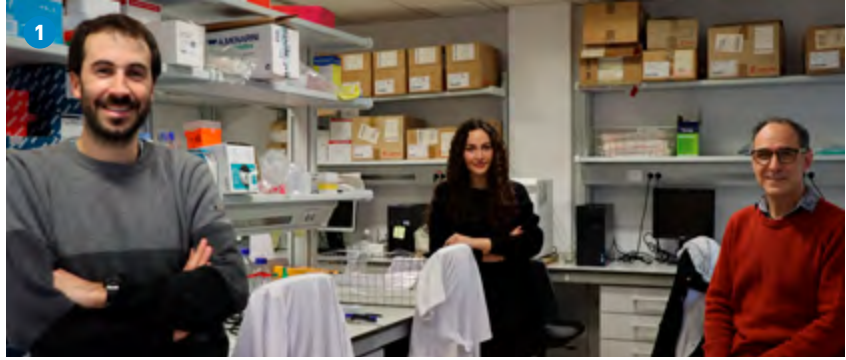
LINKEDIN
13.329 FOLLOWERS

TWITTER
937 FOLLOWERS

WEBSITE
47.275 USERS
209.231 NUMBER OF VISITS TO PAGES



News



January

We have seen that in mouse models, malnutrition during lactation is associated with alterations in the metabolism of the adult. (Photo 1)

February

Metacognitive training has beneficial effects on cognitive perception in patients with early psychotic episodes. The IRSJD is participating in a European project to assess the effects of the pandemic on mental health. A study analyses the risk genetic factors shared between ADHD and disruptive behaviour disorders. (Photo 2)



March

The conditions of 70% of people with chronic pain have worsened during the pandemic. (Photo 3)
Supplementing the maternal diet with betaine during lactation may reduce the risk of childhood obesity.



April

Can a newborn's brain discriminate speech sounds? (Photo 4)
A new facial recognition tool could make it easier for people with schizophrenia to socialize.

May

They draw a "map" of the enteroviruses that cause hand-foot-and-mouth disease in our country. Mutations in the GEMIN5 gene as the origin of rare genetic disorders in boys and girls.



June

The CERKL gene involved in mitochondrial regulation of retinal cells. (Photo 5)
Predicted prognosis of one type of childhood cancer by tumour engraftment in mice. (Photo 6)
More than 6% of Europeans suffer from depression. (Photo 7)



News

July

Dr. Lourdes Ibañez coordinates the European SPIOMET4HEALTH project: a new treatment for polycystic ovary syndrome in adolescents and young women. Survival rate of high-risk neuroblastoma patients treated with next-generation immunotherapy at SJD Barcelona Children's Hospital.



August

Study the factors associated with the appearance of depression during confinement.

IRSJD' researchers relate the concentration of certain chemical elements in the placenta with fetal growth. (Photo 8)



September

Researchers describe a new pathway of the energy metabolism in peripheral tissues regulated by cytokine GDF15. (Photo 9)

Detecting long QT syndrome early could help reduce the impact of sudden infant death.

October

A guide to improving pediatric patient participation in drug research. (Photo 10)
Institut de Recerca Sant Joan de Déu is holding its 3rd Scientific Conference. (Photo 11)



November

Brown adipose tissue activity is higher in girls during the first year of life.

The European project SARAS, towards cognitive robotics in surgery, is coming to an end.

Measuring antibodies in saliva is a useful and easy strategy to detect SARS-CoV-2 infections.

December

Study reveals that a high percentage of children and adolescents with early onset psychosis suffer from clinical depression.





7

SCIENTIFIC ACTIVITY

- 7.1. MEDICINE AND PEDIATRIC SCIENCE AREA
- 7.2. MENTAL HEALTH AREA
- 7.3. TRANSVERSAL PROGRAM





7.1

MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.1 Neuroscience

Applied research in neuromuscular diseases
Pediatric neurometabolism: neural communication mechanisms
and personalized therapies
Neonatal brain
Neuropsychology
Cognitive neuroscience
Attention, Perception and Language Acquisition (APAL)
Research in child and adolescent mental health



Applied research in neuromuscular diseases

Cecilia Jiménez Malebrera, PhD

MEMBERS

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TECHNICIAN

Ana Codina Bergadà

SELECTED PROJECTS

- Emerging biomarkers for spinal muscular atrophy. (ES-SPN-1188). Biogen International GmbH. 2021-2023. IP: Cecilia Jiménez Mallebrera.
- ArmTracker: A state-of-the-art wearable system to assess upper limb motor function in real-life conditions for patients with Duchenne muscular dystrophy and spinal muscular atrophy. (561/C/2020). Fundació La Marató de TV3. 2021-2024. IP: Daniel Natera de Benito.
- Medicina Personalizada para las Distrofias Musculares Congénitas: Desarrollo de terapias avanzadas, modelos fisiológicos y herramientas diagnósticas precisas. (PI19/00122). Instituto de Salud Carlos III (ISCIII). 2020-2022. IP: Cecilia Jiménez Mallebrera.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio abierto para evaluar la seguridad, la tolerabilidad y la eficacia de viltolarsén en niños con distrofia muscular de Duchenne (DMD), capaces e

incapaces de caminar. A Phase 2 Open-label Study to Assess the Safety, Tolerability, and Efficacy of Viltolarsén in Ambulant and Non-Ambulant Boys with Duchenne Muscular Dystrophy (DMD) Compared to Natural History Controls. (NS-065/NCNP-01-211). Medpace Spain, S.L.; Medpace, Inc; NS Pharma, Inc. 2021-. IP: Andrés Nascimento Osorio.

- Estudio de Fase 4 de Nusinersen (BIB058) en Pacientes con Atrofia Muscular Espinal que Recibieron Onasemnogene Apeparvovec. (232SM404). Biogen IDEC Research LTD; IQVIA RDS Spain SL. 2021-. IP: Andrés Nascimento Osorio.
- Estudio de extensión de fase 3, multicéntrico y abierto, para evaluar la eficacia y la seguridad de viltolarsén en niños con distrofia muscular de Duchenne (DMD) capaces de caminar. (NS-065/NCNP-01-302). Medpace Clinical Research, LLC; NS Pharma, Inc. 2021-. IP: Andrés Nascimento Osorio.

SELECTED PUBLICATIONS

- López A, Carrasco C, Fernández C, Santisteban P. Unraveling the Complex Interplay Between Transcription Factors and Signaling Molecules in Thyroid Differentiation and Function, From Embryos to Adults. *Front Endocrinol (Lausanne)*. 2021;12:654569-654569. IF:5,555. (Q1).
- Natera D, Aguilera S, Costa L, García M, Miranda MC, Rúbies J et al. COVID-19 in children with neuromuscular disorders. *J. Neurol*. 2021;268(9):3081-3085. IF:4,849. (Q1).
- Natera D, Foley AR, Domínguez C, Ortez CI, Jain M, Mebrahtu A et al. Association of Initial Maximal Motor Ability With Long-term Functional Outcome in Patients With COL6-Related Dystrophies. *Neurology*. 2021;96(10):1413-1424. IF:9,910. (Q1).
- Trifunov S, Paredes AJ, Badosa MC, Codina A, Montoya C, Ruiz E et al. Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. *Clin Chem*. 2021;67(8):1113-1121. IF:8,327. (Q1).

Pediatric neurometabolism: neural communication mechanisms and personalized therapies

M^a Angels García Cazoral, MD, PhD

Over the course of 2021, we have been able to advance in the characterisation of various significant aspects of neurotransmitter disorders. In the first place we described new markers relating to prognosis and disease mechanisms in dopaminergic neurotransmission defects. We have also described new elements of the clinical phenotype and neuroimaging of these disorders. This was possible thanks to the collaboration of many different centres through the international network I-NTD. Likewise, we carried out our own clinical trial with L-serine on patients with mutations due to loss of function in NMDA receptors. We also completed the preclinical concept test of the "Neuroprotect" formula designed in our laboratory, which has the purpose of improving various aspects of neurodevelopment in neuropaediatric disorders. Additionally, we obtained several grants and other funding to hire research personnel. With the help of these funds, we seek to develop and/or apply personalised treatments in a group of pathologies that include paediatric parkinsonism and neurometabolic disorders in general.

MEMBERS

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Roser Colomé Roura, Silvia Maria Meavilla Olivas, Alejandra Darling, Jordi Muchart López, M^adel Mar O'Callaghan Gordo, Federico Ramos, Anna López Sala, Maria del Carme Fons Estupiña

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Palacios

ASSISTANT RESEARCHER

Neus Guiu Gonzalez, Mariya
Sigatullina Bondarenko

TECHNICIAN

Anna Serradell Salís, Cristina Grau
Páez

SELECTED PROJECTS

- TREAT-AHC. Identification of compounds for the treatment of Alternating Hemiplegia of Childhood by drug repositioning. Molecular and pre-clinical evaluation and clinical outcome measure validation. [283/C/2020]. Fundació La Marató de TV3. 2021-2024. PI: Carme Fons Estupiña.
- Teràpies personalitzades pels trastorns cognitiu-conductuals

de les malalties hereditàries del metabolisme en l'edat pediàtrica. (2020 DI 103). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2021-2024. PI: M^aAngels García Cazorla.

- Developing patient-tailored therapeutic candidates for rare genetic forms of pediatric parkinsonism. (202012-30). Fundació La Marató de TV3. 2021-2024. PI: M^aAngels García Cazorla.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de la eficacia y seguridad de vatiquinona en el tratamiento de la epilepsia farmacorresistente en pacientes con enfermedad mitocondrial. Efficacy and Safety Study of Vatiquinone for the Treatment of Mitochondrial

Disease Patients with Refractory Epilepsy. (PTC743-MIT-001-EP). Parexel International S.L; PTC Therapeutics, Inc. 2021-. PI: M^adel Mar O'Callaghan Gordo.

- Estudio fase 3, multicéntrico, aleatorizado, doble ciego, de grupos paralelos y controlado con placebo, para evaluar la seguridad, la tolerabilidad y la eficacia de 2000 mg/kg de Trappsol® Cycloã.,ç (hidroxipropil-Î²-ciclodextrina) y tratamiento estándar comparado con placebo y tratamiento estándar en pacientes con la enfermedad de Niemann-Pick de tipo C1. A Phase III, Double-blind, Randomized, Placebo-controlled, Parallel-group, Multicenter Study to Evaluate the Safety, Tolerability, Pharmacokinetics, and Efficacy of 2000 mg/kg de Trappsol® Cycloã.,ç (Hydroxypropyl-Î²-cyclodextrin) and Standard of Care Compared to Placebo and Standard of Care in Patients with Niemann-Pick Disease Type C1. (CTD-TCNPC-301). Cyclo Therapeutics, Inc.; Worldwide Clinical Trials Ltd. 2021-. PI: M^adel Mar O'Callaghan Gordo.
- Estudio aleatorizado, de grupos paralelo, doble ciego, controlado con placebo, con extensión abierta, para evaluar la eficacia y la seguridad del vatiquinona para el tratamiento de la ataxia de Friedreich (MOVE-FA). A randomized, parallel-arm, double-blind, placebo-controlled study with open-label extension to assess the safety and efficacy of vatiquinone for the treatment of friedreich ataxia. (PTC743-NEU-003). CTI Clinical Trial & consulting services Spain S.L; PTC Therapeutics, Inc. 2021-. PI: Alejandra Darling.

SELECTED PUBLICATIONS

- De Oyarzabal AL, Musokhranova U, Barros LF, Garcia A. Energy metabolism in childhood neurodevelopmental disorders. EBioMedicine. 2021;69:103474. IF:8,143. (Q1).
- Kuseyri O, Horvath G, Cortés E, Yildiz Y, Mastrangelo M, Pons R et al. Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nat Commun. 2021;12(1):5529. IF:14,919. (Q1).
- Tristan A, Borràs E, Molero M,



Wassenberg T, Peters T, Verbeek MM et al. Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. *Mov Disord.* 2021;36(3):690-703. IF:10,338. (Q1).

Neonatal brain

**Thais Agut Quijano, MD, PhD
& Ana Alarcon Allen, MD, PhD**

The Neonatal Brain Group has maintained its commitment to generate knowledge on brain disorders of the foetal/neonatal period with an impact on children's health and capacities. The group was founded by Dr Alfredo García-Alix and directed by him until his recent retirement in April of 2021. Drs Ana Alarcón and Thais Agut succeeded him as principal investigators and the group has been consolidated with the incorporation of two pre-doctoral researchers, Drs Nuria Carreras and Mar Velilla, and the paediatric and perinatal pathologist Dr Sílvia Planas.

The group has continued with its line of work on pathogenic mechanisms and biomarkers in neonatal hypoxic-ischaemic encephalopathy and cerebral arterial infarction. Its other active primary lines are: (1) Brain maturation and assessment of the impact of acquired brain lesions on brain maturation (PI: Thais Agut); (2) Foetal/neonatal infections with an impact on the developing brain (PI: Ana Alarcón); and (3) Care practices for newborns with neurological risk and their families (PI: Nuria Herranz).

One of the most positive aspects has been the continued collaboration with other specialists and researchers in areas relating to foetal and neonatal neurology, and with other groups from the Sant Joan de Déu Barcelona Research Institute and from other institutions. The Group's projects have adopted several different approaches (clinical, including both medical and nursing approaches; basic science; biomedical

engineering; epidemiology), favouring the translational character of the research in all cases. Moreover, within the sphere of our research, we continued to participate in both national and European working groups with a global teaching and research outlook.

The group's achievements in 2021 included a total of 18 publications, with a total impact factor of 74.35. Likewise, Ms. Nuria Herranz coordinated three documents with evidence-based recommendations on aspects of neonatal care that influence the developing brain. Lastly, the group continued to promote pre-doctoral research, with one doctoral thesis that was presented and three in progress, in addition to three final master's degree projects.

MEMBERS

STAFF RESEARCHER

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POSTDOC ASSOCIATED RESEARCHERS

Nuria Carreras Blesa

ASSISTANT RESEARCHER

Jonathan Francisco Olival Tremaria

TECHNICIAN

Maria Jose Coll Lobarte, Nerea Liñan Rosa

SELECTED PROJECTS

- Estudio CRIB (Cribado de citomegalovirus congénito en Recién nacidos sanos e Ingresados – Barcelona). (PI19/00095). Instituto de Salud Carlos III (ISCIII). 2020-2024. PI: Ana Alarcón Allen.
- Elaboración de un atlas morfométrico normalizado de la maduración cerebral del recién nacido prematuro y valoración del impacto de la patología cerebral en el proceso madurativo. (PI18/00110). Instituto de Salud Carlos III (ISCIII). 2019-2023. PI: Thais Agut Quijano.
- Contratos Rio Hortega. (CM18/00258). Instituto de Salud Carlos III (ISCIII). 2019-2021. PI: Alfredo Garcia-Alix Pérez. Beneficiaria del contrato: Nuria Carreras Blesa.
- Rare Coding Genetic Variants Associated with neonatal arterial ischemic stroke. (201714.10). Fundació La Marató de TV3. 2018-2021. PI: Alfredo Garcia-Alix Pérez.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Randomized, Double-blind, Placebo-controlled Single-ascending Dose Trial to Evaluate the Safety, Tolerability, and Pharmacokinetics of GWP42003-P in Conjunction with Hypothermia in Neonates with Moderate or Severe Hypoxic Ischemic Encephalopathy. (GWEP1560). GW Research; PaidionResearch, Inc. 2019- PI: Ana Alarcon Allen.

SELECTED PUBLICATIONS

- Ades AE, Soriano A, Alarcon A, Bonafonte F, Thorne C, Peckham CS et al. Vertical transmission of Zika virus and its outcomes: a Bayesian synthesis of prospective studies. *Lancet Infect Dis.* 2021;21(4):537-545. IF:25,071. (Q1).
- François C, Rodriguez A, Teixidó M, Agut T, Bosch L. Attenuated brain responses to speech sounds in moderate preterm infants at term age. *Dev Sci.* 2021;24(1)IF:5,131. (Q1).
- García A, Arnaez J, Arca G, Agut T, Alarcon A, Martín A et al. Development, Reliability, and Testing of a New Rating Scale for Neonatal Encephalopathy. *J Pediatr.* 2021;235:83-91. IF:4,406. (Q1).
- Planas S, Cruz O, Miguel S, Albert A, Rovira C, Bombi JA. Extra-axial sacral soft tissue giant cell ependymoma affecting a child: Case report and review of the literature. *Neuropathology.* 2021;41(2):139-145. IF:1,906. (Q3).

Neuropsychology

Roser Pueyo Benito, PhD

In 2021, our group continued to progress with the study of the neuropsychological functioning of persons with risk factors (obesity), and with the study of the neuropsychological consequences of congenital, perinatal and/or acquired injuries (cerebral palsy and stroke). Likewise, we continued with projects financed in national

competitive calls, including the telethon La Marató de TV3, and we continued to hold the ICREA-Academia award, received by one of our researchers. We also continued to apply to European calls and we are awaiting the resolution of a COST Action in the field of cerebral palsy. Our projects and our selected publications in Q1 evidence our group's capacity to adopt a neuropsychology focus that allows us to achieve results ranging from the development of neuropsychological assessment guides to the pursuance of studies in greater depth on the underlying mechanisms of disorders (with analysis of neuroimages, genetics, microbiota, etc.). We have also studied the effectiveness of interventions addressed to improving cognitive and emotional functioning, such as physical exercise, mindfulness and cognitive training.

MEMBERS

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TECHNICIAN

Gemma Monté Rubio

SELECTED PROJECTS

- Guías basadas en la evidencia y consensuadas para la evaluación neuropsicológica de las personas con parálisis cerebral grave y parálisis cerebral discinética (PID2020-117163RB-I00). Ministerio de Economía y Competitividad. Convocatoria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2021-2024. PI: Roser Pueyo Benito.
- Ajut per incentivar i consolidar la recerca d'excel·lència ja existent a les universitats públiques de Catalunya. Programa ICREA

Academia 2018. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). Convocatòria del Programa Icrea Acadèmia. 2019-2023. PI: Maria Mataró Serrat.

- Efectes de teràpies combinades d'exercici, mindfulness i estimulació cognitiva en la cognició i la neuroplasticitat en pacients amb ictus isquèmic crònic. Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 2018-2022. PI: Maria Mataró Serrat.
- Inflamación, estrés y microbiota como mediadores entre la obesidad y el funcionamiento cerebral durante la adolescencia (PSI2017-86536-C2-1-R) Ministerio de Economía y Competitividad. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Projectes I+D+I. 2018-2022. PI: Maria Angeles Jurado Luque.

SELECTED PUBLICATIONS

- Castells A, Roig F, Dacosta R, Lamonja N, Sawicka AK, Torán P et al. Exercise and Fitness Neuroprotective Effects: Molecular, Brain Volume and Psychological Correlates and Their Mediating Role in Healthy Late-Middle-Aged Women and Men. *Front Aging Neurosci.* 2021;13:615247. IF:5,750. (Q1).
- Castells A, Roig F, Lamonja N, Torán P, Pera G, Montero P et al. Sex Matters in the Association between Physical Activity and Fitness with Cognition. *Med Sci Sports Exerc.* 2021;53(6):1252-9. IF:5,411. (Q1).
- Lamonja N, Dacosta R, López J, Prades L, Roig F, Castells A et al. Sex-Specific Protective Effects of APOE e2 on Cognitive Performance. *J Gerontol A Biol Sci Med Sci.* 2021;76(1):41-9. IF:6,053. (Q1).
- Ottino J, Baggio HC, Jurado MA, Segura B, Caldu X, Prats X et al. Alterations in Brain Network Organization in Adults With Obesity as Compared With Healthy-Weight Individuals and Seniors. *Psychosom Med.* 2021;83(7):700-6. IF:4,312. (Q1).

Cognitive neuroscience

Carles Escerca Micó, PhD

In 2021, our group continued to foster collaboration with the Foetal Environment and Obstetric Complications research group, led by Dr Gómez-Roig, for the purpose of promoting the identification of EEG-derived biomarkers obtained at birth, predicting neurodevelopment in the early stages of childhood. We have published two significant studies on this subject: one proposing a method for obtaining cerebral response to language sounds (frequency-following response – FFR), which allows identification of neural coding processes which are already mature at birth and of others which are not yet mature [Arenillas-Alcón *et al.*, 2021]; and a study establishing the protocols required to register the neonatal FFR [Lemos *et al.*, 2021]. These two publications open the way for the mass monitoring of the cerebral maturation allowing language processing in newborns. A third significant study conducted in this period identifies the brain structures responsible for FFR, pinpointing subcortical components of the auditory pathway [Gorina-Careta *et al.*, 2021]. Indeed, this study is very important since it reveals the basis for the use of FFR as a biomarker for language acquisition in persons at risk (for example, full-term low-weight newborns), thus providing a significant indicator of subcortical activity in the neonatal population, in which the cerebral cortex needs to undergo a far-reaching experience-dependent maturation. At present our studies are addressed to researching the consequences of high-risk pregnancy (low birthweight and high birthweight) by means of this indicator. Other studies have also been promoted in which we will be making use of brain signals associated with prediction errors, and glutamatergic neuromodulation —such as those used in Font-Alaminos *et al.* (2021)— in populations with neurodevelopment alterations.

MEMBERS

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PHD STUDENTS

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

SELECTED PROJECTS

- Ajut per incentivar i consolidar la recerca d'excel·lència ja existent a les universitats públiques de Catalunya. Programa ICREA Academia 2020. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). Convocatòria del Programa Icrea. 2021-2025. PI: Carles Escera.
- Trastornos del espectro alcohólico fetal (TEAF): Identificación precoz y biomarcadores de riesgo para alteraciones neurocognitivas mediante el potencial de seguimiento de frecuencia (PSF). Fundación Alicia Koplowitz. Convocatoria de Ayudas a Proyectos de Investigación en el área de Psiquiatría de la Infancia y Adolescencia y Neurociencias en el Niño. 2020-2022. PI: Carles Escera.
- Caracterización de la respuesta de seguimiento de frecuencia (RSF) en recién nacidos como posible biomarcador de desarrollo neurocognitivo (PGC2018-094765-B-I00). Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Modalitat Projectes d'R+D de Generació del Coneixement. 2019-2022. PI: Carles Escera.
- ARTSOUNDSCAPES - The sound of special places: exploring rock art soundscapes and the sacred (787842). European Research Council (ERC). Advanced Grant. 20218-2024. PI: Carles Escera.

SELECTED PUBLICATIONS

- Arenillas-Alcón S, Costa J, Ribas T, Gómez MD, Escera C. Neural encoding of voice pitch and formant structure at birth as revealed by frequency-following responses. *Sci*

Rep. 2021;11(1):6660. IF:4,379. (Q1).

- Font-Alaminos M, Ribas T, Gorina N, Escera C. Emergence of prediction error along the human auditory hierarchy. *Hear Res.* 2021;399:107954. IF:3,208. (Q1).
- Gorina-Careta N, Kurkela J, Hämäläinen J, Astikainen P, Escera C. Neural generators of the frequency-following response elicited to stimuli of low and high frequency: A magnetoencephalographic (MEG) study. *Neuroimage.* 2021;231:117866. IF:6,556. (Q1).
- Lemos FA, da Silva Nunes AD, de Souza CK, Escera C, Taveira KVM, Balen SA. Frequency-Following Response in Newborns and Infants: A Systematic Review of Acquisition Parameters. *J Speech Lang Hear Res.* 2021;64(6):2085-2102. IF:2,297. (Q1).

Attention, Perception and Language Acquisition (APAL)

Laura Bosch Galceran, PhD

Our research activity has been affected by the pandemic situation and by the change of location of the APAL group's laboratory to the facilities at Numància Street. We have advanced in three directions. Firstly, in the research line on the child population with bilateral profound deafness, we completed the sample of pre-implant patients and started up their 24-months post-implant reassessment with measurements of lexical learning and oral language level achieved (research in collaboration with the ORL unit [Dr Claveria and Dr Haag], Neuroradiology [M. Rebollo] and Psychology [D. Polo]). At the same time, we assessed two groups of children with normal hearing, at 10 months and at 18-24 months, who will form the control groups of this study, which is expected to end in 2022. Preliminary data suggest differences between groups at 10 months in social cognition tasks, and a great variability in post-implant performance. These

results will be analysed, taking into consideration the neuroimaging data obtained in the pre-implant period and other cognitive and communicative development variables that have been collected. We have also progressed in a second research line focused on the study of the audiovisual processing of speech from the age of 4 months in full-term and moderately premature children (the sample is pending completion, with the collaboration of Dr Agut). The purpose of this research line is to analyse the impact of a bilingual/multilingual family setting on the basic processes of recognition of the mother tongue, the association between the speaker and the speech ordinarily used, and the association between the voice of the speaker and speech. Lastly, we succeeded in completing the follow-up study of a sample of full-term low-weight (SGA) children with cognitive and speech data, and with data on school performance, at the age of 7-8 years (a study in collaboration with Dr Ibáñez's group). The preliminary results, which were presented at the 3rd Research Conference of the IRSJD, show differences between groups in the areas of attention and executive function, and minor differences in speech, which were more prominent in the phonological and articulatory domain, with a possible link to reading difficulties. We also worked on the connection between endocrine-metabolic variables and cognitive-linguistic performance, and on the search for catch-up patterns that may predict cognitive results to be achieved in the long term.

MEMBERS

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Joan Birulés Muntané, Clément François

ASSISTANT RESEARCHER

Jessica Sánchez-Galán Frauca

SELECTED PROJECTS

- Pròrroga del conveni de col·laboració per una recerca sobre cognició i percepció de llengües en nens. Fundació Privada Betània-Patmos. 2019-2023. PI: Ferran Pons Gimeno.
- Variabilidad en el procesamiento audiovisual del habla en la infancia:

rol de la experiència lingüística, maduració neurològica y capacitat sensorial (PGC2018-097487-B-I00). Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Modalitat Projectes d'R+D de Generació del Coneixement. 2019-2022. PI: Ferran Pons Gimeno i Laura Bosch Galceran.

- Resonancia magnética funcional e imagen de tensor de difusión en la valoración prequirúrgica del paciente pediátrico con hipoacusia prelingual neurosensorial profunda bilateral de detección en el cribado neonatal como potenciales biomarcadores de desarrollo de lenguaje oral tras la colocación de un implante coclear (PCP00215). Sociedad Española de Neuroradiología. SERN 2017. 2017-2022. PI: Mónica Rebollo.

SELECTED PUBLICATIONS

- François C, García-Alix A, Bosch L, Rodríguez-Fornells A. Signatures of brain plasticity supporting language recovery after perinatal arterial ischemic stroke. *Brain Lang.* 2021;212:104880. IF:2,381. (Q1).
- François C, Rodríguez-Fornells A, Teixidó M, Agut T, Bosch L. Attenuated brain responses to speech sounds in moderate preterm infants at term age. *Dev Sci.* 2021;24(1):12990. IF:5,131. (Q1).
- Martínez A, Sanz M, Pons F, de Diego R. Rethinking attention in time: Expectancy violations reconcile contradictory developmental evidence. *J Exp Child Psychol.* 2021;206:105070. IF:2,610. (Q2).

Research in child and adolescent mental health

Montserrat Dolz Abadia, MD, PhD

The primary purpose of our research group is the clinical

investigation of various psychiatric disorders, with special emphasis on neurodevelopment disorders and early-onset disorder. At present our efforts are focused on the study of childhood-onset psychosis (PI: Dr Dolz), of attention deficit hyperactivity disorder (ADHD) (PI: Dr Alda) and of autism spectrum disorder (ASD) (PI: Dr Mairena, Dr Aranbarri), as well as on promoting the prevention and detection of child witnesses to domestic violence (PI: Dr Álvarez, Dr Lacasa).

In recent years, our group has become consolidated through the implementation of various projects financed by public (FIS projects, MINECO) and private institutions, both national and European (European Commission), as well as through collaboration with various stable research groups of the CIBERSAM network for biomedical research on mental health, and with the G04 group of the Clinical and Provincial Hospital of Barcelona and the G01 group of the Gregorio Marañón Hospital in Madrid. Of special note is the recent award (2021) of the FIS grant "Multi-centre longitudinal study of markers for prediction of transition to psychosis of children and adolescents with risk of psychosis: The role of intrauterine stress" (PI: Dr Dolz; PI21/00090). Mention should also be made of the development of the TEA-CARE programme in collaboration with the Mas Casadevall Autism Foundation and financed by La Caixa Foundation, addressed to implementing a clinical programme of autism care based on research and to the education of children aged 0-4 years.

MEMBERS

STAFF RESEARCHER

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SELECTED PROJECTS

- Impacte de l'estressor ambiental COVID19 sobre les emocions i la conducta en població infanto-juvenil: Macro-Estudi longitudinal EmCoVID19 i EmCoVID19-unitats clíniques. (BML 2021_HSJD). O.H.S.J.D.-Curia Provincial. 2021-2022. IP: Esther Via Virgili.
- Beca Retorno Fundación Alicia Koplowitz_Alba Vilaplana. (Beca Retorno_Alba Vilaplana). Fundación Alicia Koplowitz. 2020-2021. IP: Montserrat Dolz Abadia.
- Beca Retorno Fundación Alicia Kpplowitz_Maria Andreu. (Beca Retorno_Maria Andreu). Fundación Alicia Koplowitz. 2020-2021. IP: Montserrat Dolz Abadia.
- Efectos del tratamiento farmacológico en pacientes con Trastorno por Déficit de Atención y Hiperactividad (TDAH). (Ayudas Investigación 2019). Banc de Sabadell, S.A. 2020-2021. IP: Miriam Sanabra Gonzalez.
- Disrupción de los ritmos circadianos en población infantil con Trastornos del Neurodesarrollo (TDAH y TEA) y su impacto sobre la salud y calidad de vida. (PI19/00113). Instituto de Salud Carlos III (ISCIII). 2020-2022. IP: José Ángel Alda Díez.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Ensayo controlado con placebo en sujetos con alto riesgo para la psicosis comparado con ácidos grasos omega-3 en Europa. PURPOSE / ABR54654. University Medical Center Utrecht. 2016-. PI: Montserrat Dolz Abadia.
- A Double-blind, Randomized, Psychoactive Placebo-controlled, Study to Evaluate the Efficacy and Safety of 3 Fixed Doses (28 mg, 56 mg and 84 mg) of Intranasal

Esketamine in Addition to Comprehensive Standard of Care for the Rapid Reduction of the Symptoms of Major Depressive Disorder, Including Suicidal Ideation, in Pediatric Subjects Assessed to be at Imminent Risk for Suicide. *ESKETINSUI2002*. Janssen - Cilag, S.A. 2018-. PI: Montserrat Dolz Abadia.

SELECTED PUBLICATIONS

- ENIGMA Clinical High Risk for Psychosis Working Group; Jalbrzikowski M, Hayes RA, Wood SJ, Nordholm D, Zhou JH, Fusar-Poli P et al. Association of Structural Magnetic Resonance Imaging Measures With Psychosis Onset in Individuals at Clinical High Risk for Developing Psychosis: An ENIGMA Working Group Mega-analysis. *JAMA Psychiatry*. 2021;78(7):753-766. IF:21,596. (Q1).
- Insa Pineda I, Via E, Estrada-Prat X, Gómez-González CL, Alda JA. Attention Deficit Hyperactivity Disorder (ADHD) & COVID-19: Do Caregivers and Children Report the Same?. *Int J Mental Health Psychiatry*. 2021;7(1).
- Serra-Blasco M, Radua J, Soriano-Mas C, Gómez-Benlloch A, Porta-Casteràs D, Carulla-Roig M et al. Structural brain correlates in major depression, anxiety disorders and post-traumatic stress disorder: A voxel-based morphometry meta-analysis. *Neurosci Biobehav Rev*. 2021;129:269-281. IF:8,989. (Q1).
- Zerón-Ruggerio MF, Carpio-Arias TV, Ferreira-García E, Díez-Noguera A, Cambras T, Alda JA et al. ADHD subtypes are associated differently with circadian rhythms of motor activity, sleep disturbances, and body mass index in children and adolescents: a case-control study. *Eur Child Adolesc Psychiatry*. 2021;30(12):1917-1927. IF:4,785. (Q1).



7.1

MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.2

Adaptive response, metabolism and immunobiology

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

Metabolic endocrinology

Lourdes Ibañez Toda, MD, PhD

During this year, we completed the preparations to begin the recruitment of patients for the SPIOMET4HEALTH (Horizon 2020, European Commission) multi-centre, multi-national, randomised double-blind European clinical trial. The final protocol was completed, the permits of the ethical committees and of the agencies of the participating countries were obtained, the electronic data collection logbook was developed, the communication and patient recruitment strategy was prepared, and the production and distribution of the drugs under study was completed. Likewise, we advanced in the study of adipose tissue biomarkers in newborns and babies, and in the study of their relation to body composition, which led to the publication of two articles in Q1 journals. We also continued to investigate the pathophysiological mechanisms determining the polycystic ovary syndrome, and the effects of different treatments on the intestinal microbiome, as well as the changes in the concentrations of growth and differentiation factor 15 (GDF15), which controls body weight by means of a central receptor. These studies led to the publication of two more articles in Q1. We completed the longitudinal follow-up of patients and began the analysis of the circulating exosomes and of their proteome in children with low weight and normal weight for gestational age, within the framework of the ISCIII project PI18/00109. We communicated all the results of these studies in guest conferences, notably including those of the 15th Congress of the European Association of Paediatric and Adolescent Gynaecology (EURAPAG, June 2021); the 22nd Congress of the Mexican Society for Paediatric Endocrinology (August 2021); and the 48th Annual Meeting of the British Society for Paediatric Endocrinology & Diabetes (November 2021).

MEMBERS

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Gemma Carreras Badosa, Esther Lizarraga Mollinedo, Judith Bassols Casadevall, Francis de Zegher, Abel López Bermejo

SELECTED PROJECTS

- SPIOMET4HEALTH_PCOS in adolescent girls and young women: Toward a treatment guided by pathophysiology. (899671). European Commission. 2021-2026. PI: Lourdes Ibañez Toda.
- Perfil longitudinal de exosomas circulantes y de su proteoma en niños nacidos con bajo peso y peso adecuado para la edad gestacional: Utilidad como biomarcador de síndrome metabólico. (PI18/00109). Instituto de Salud Carlos III (ISCIII). 2019-2023. PI: Lourdes Ibañez Toda.
- SGR 2017_Repercussió de les alteracions prenatales perinatals en el desenvolupament postnatal. Malalties d'origen fetal. (2017 SGR 01236). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2017-2021. PI: Lourdes Ibañez Toda.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio observacional, prospectivo y multicéntrico a largo plazo en una cohorte de niños con talla baja nacidos pequeños para su edad gestacional (PEG) que han sido tratados con Saizen®. (EMR 200098_008 / MER-GRH-2009-01). Merck KGaA. 2018-. PI: Lourdes Ibañez Toda.
- Treatment with recombinant growth hormone (rGH) in short patients born small-for-gestational-age (SGA) followed at Hospital Sant Joan de Déu: Analysis of early predictors of good response useful in clinical practice. (W1191603). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2014-. PI: Lourdes Ibañez Toda.
- Identification of circulating microRNAs in short children

born small for gestational age: Relationship with pre- and postnatal growth and metabolic parameters. (LOR-HOR-2013-01). López. 2014-. PI: Lourdes Ibañez Toda.

- GHLiquid-3676 (NNH-SOM-2012-01) Estudio internacional de desenlaces de NordiNet® (IOS de NordiNet®). (GHLiquid-3676 (NNH-SOM-2012-01)). Novo Nordisk Pharma, S.A. 2012-. PI: Lourdes Ibañez Toda.

SELECTED PUBLICATIONS

- de Zegher F, Díaz M, Villarroja J, Cairo M, López A, Villarroja F et al. The relative deficit of GDF15 in adolescent girls with PCOS can be changed into an abundance that reduces liver fat. *Sci Rep.* 2021;11(1):7018-7018. IF: 4,379. (Q1).
- Díaz M, Blasco A, Villarroja J, López A, de Zegher F, Villarroja F et al. Circulating diazepam-binding inhibitor in infancy: Relation to markers of adiposity and metabolic health. *Pediatr Obes.* 2021;16(11):12802. IF: 4,000. (Q1).
- García C, Malpique R, Carbonetto B, González P, Henares D, Brotons P et al. Gut microbiota in adolescent girls with polycystic ovary syndrome: Effects of randomized treatments. *Pediatr Obes.* 2021;16(4):12734-12734. IF: 4,000. (Q1).
- Inarejos E, Navarro OM, Navallas M, Enrique G, Colombo C, Suñol M et al. Omphalomesenteric Duct Anomalies in Children: A Multimodality Overview. *Radiographics.* 2021;41(7):2090-2110. IF: 5,333. (Q1).

Metabolic diseases of paediatric origin

Josep Jiménez Chillarón, PhD & Carles Lerin Martínez, PhD

Our research focuses on gaining a better understanding of the molecular mechanisms at play when nutrition and growth during very early stages of life affect the subsequent

development of childhood obesity or metabolic diseases in adulthood. This paradigm is known as the Developmental Origins Of Health and Disease (DOHaD). We have brought together a multidisciplinary group for this research effort including doctors, nutritionists, biologists, biochemists, and laboratory technicians. Together we carry out highly translational research involving:

1. The development and characterisation of experimental models (mouse) that effectively recapitulate human pathophysiology.
2. The design of clinical studies aimed at developing new prevention and treatment strategies.

Over the past few years, we have applied omic techniques to help further understand these processes and identify early biomarkers. We are currently evaluating the physiological role of these biomarkers through in vitro and in vivo animal cell models as well as clinical studies, including two ongoing clinical studies. The aim is to attain a better understanding of the possible implication in the pathophysiology associated with metabolic diseases and to design new nutritional-therapeutic interventions for the treatment of childhood obesity and associated metabolic disorders.

MEMBERS

STAFF RESEARCHER

Montserrat Amat Bou, Serafin Murillo Garcia, Marina Llobet Garces, Ruben Díaz Naderi, Marta Ramon Krauel

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Ivonne Palacios Marin

SELECTED PROJECTS

- Non-genomic Inheritance of environmentally-induced traits: Deciphering epigenetically-driven genetic variation in mammals. ID 62167. JOHN TEMPLETON FOUNDATION. 2021-2024. IP: José Carlos Jiménez Chillarón.
- Modulación de la dieta durante las primeras etapas de la vida como estrategia de prevención de la

obesidad infantil. (PDC2021-121816-100). Ministerio de Economía Y Competitividad (MINECO). 2021-2023. IP: Carles Lerin Martinez.

- Impact of maternal SARS-CoV-2 infection and vaccination on breast milk antibodies, milk bioactive compounds and maternal-child health (MilkCORONA). (202106-32). Fundació La Marató de TV3. 2021-2023. IP: Carles Lerin Martinez.
- El microbioma intestinal como diana terapéutica en el síndrome de Prader-Willi: efectos metabólicos y en el comportamiento de los pacientes. (PI20/00301). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Marta Ramon Krauel.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- El microbioma intestinal como diana terapéutica en el síndrome de Prader-Willi: efectos metabólicos y en el comportamiento de los pacientes. (PI20/00301). Instituto de Salud Carlos III (ISCIII). IP: Marta Ramon Krauel.
- Suplementación con betaína de la dieta materna durante la lactancia como estrategia de prevención de obesidad infantil. (SAF2017-88005-R). Ministerio de Economía Y Competitividad (MINECO). IP: Carles Lerin Martinez.

SELECTED PUBLICATIONS

- Ribas F, Parra M, Ramon M, Díaz R, Lerin C, Cambras T et al. Time-Restricted Feeding during Puberty Ameliorates Adiposity and Prevents Hepatic Steatosis in a Mouse Model of Childhood Obesity. *Nutrients*. 2021;13(10):3579. IF:5,717. (Q1).
- Ribas F, Ribó S, Parra M, Fernández A, Cebrià J, Guardiola M et al. Neonatal overfeeding during lactation rapidly and permanently misaligns the hepatic circadian rhythm and programmes adult NAFLD. *Mol Metab*. 2021;45:101162. IF:7,422. (Q1).
- Ribó S, Sánchez D, Martínez L, García I, Ramon M, Tondo M et al. Increasing breast milk betaine modulates Akkermansia abundance in mammalian neonates and improves long-term metabolic health. *SCI TRANSL MED*. 2021;13(587):322. IF:17,956. (Q1).

Molecular biology and gene regulation of adipose tissue and its pathologies

Francesc Villarroya Gombau, PhD

In 2021, our team continued its basic and translational research on the control mechanisms of adiposity and cardiometabolism, and their changes under pathological conditions such as obesity, diabetes, and other metabolic and cardiovascular diseases. We identified new adipokines and cardiokines which are predominantly produced by brown adipose tissue and heart and new intracellular control factors of adipose and cardiac plasticity in response to hormonal stimuli. In the translational field, we analysed the activity of the brown adipose tissue and the variations in the levels of new metabolic biomarkers in connection with pathophysiological determinants in the paediatric population, as well as we analyzed new cardiokines as molecular biomarkers of cardiac diseases.

MEMBERS

STAFF RESEARCHER

Aleix Gavalda Navarro, Joan Villarroya Terrade, Marta Giralt Oms, Roser Iglesias Coll, Teresa Mampel Astals, Anna Planavila Porta

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TECHNICIAN

Mercedes Morales Rueda, Gemma Moreno Besora, Albert Però Garcia

SELECTED PROJECTS

- ACBP, ¿una nueva cardiomiocina para la detección y tratamiento de la enfermedad cardíaca?. Sociedad Española de Cardiología. Becas de ayuda para proyectos de Investigación Básica o Traslacional

en Salud Cardiovascular de la Sociedad Española de Cardiología y Fundación Española del Corazón. 2021-2022. PI: Ana Planavila Porta.

- Descifrando el papel endocrino del tejido adiposo marrón: nuevos actores, nuevas funciones (PID2020-114112RB-I00). Ministerio de Ciencia e Innovación (MICINN). Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2021-2024. PI: Francesc Villarroya Gombau.
- Estudios preclínicos para determinar las alteraciones en el balance energético y/o la función adipocitaria subyacentes al incremento de peso corporal en personas que viven con VIH tratadas con INsTI y/o TAF (PI20/00106). Ministerio de Ciencia, Innovación y Universidades. Ajuts de l'Acció Estratègica en Salut: Subprograma de projectes d'investigació en salut (PI). 2020-2023. PI: Marta Giralt Oms.

evolution of their growth, metabolism and neurodevelopment.

With respect to the critical patient, we continue to research brain injury, especially in the patients affected by congenital heart defects. The projects we have conducted with non-invasive neuromonitoring studies have allowed us to collaborate on a European project for the development of a new integrated neuromonitoring device (TinyBrains). The use of a new optical technology will help us to determine non-invasively oxygen consumption at all times, haemodynamics-dependent changes in cerebral blood flow, and changes in brain electrical activity during the most vulnerable periods or at moments of greatest risk, such as during surgery or in the immediate postoperative period. Along this same line, our studies with biomarkers of brain injury and oxidative stress markers are opening up for us new clinical assessment strategies for the period of foetal life or at the patient's bedside, providing us with instruments that will help us to detect patients at risk in order to be able to implement neuroprotective strategies. All this is being done with a translational approach, expanding our knowledge through experimental animal models.

2023. IP: Martin Iriondo Sanz.

- Consecuencias en la infancia del fallo perinatal de crecimiento en Recién Nacidos Muy Prematuros: estudio clínico y metabólico. (PI20/00294). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Isabel Iglesias Platas.
- TinyBrains_Bio-photonics imaging of the infant brain, the missing link between the cellular brain damage and the neurovascular unit during acute illness. (101017113). European Commission. 2021-2024. IP: Marta Camprubi Camprubi/Juan Sánchez de Toledo.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Efecto del Alopurinol en pacientes con encefalopatía hipóxico-isquémica tratados con hipotermia. ALBINO ensayo clínico multicéntrico, aleatorizado, ciego, controlado con placebo. (H2020-PHC-18-2015-667224). Instituto de investigación Sanitaria Hospital La Fe. 2017-. IP: Marta Camprubi Camprubi.
- Evaluation of Real Time Continuous Glucose Monitoring in the very preterm infant. (REACT RCT). University of Cambridge. 2017-. IP: Isabel Iglesias Platas.

Neonatology research group

Martin Iriondo Sanz, MD, PhD

Our group has two main research lines: nutrition and prematurity, on the one hand, and the critical patient, on the other. In our nutrition and prematurity line, we have helped to demonstrate, within the framework of a European multi-centre study, the utility of the continuous monitoring of the blood glucose level in real time in premature patients under intensive care. We continued investigating the factors present in human milk that may have an impact on the development of very premature newborns (born sooner than the 32nd week of pregnancy), with special attention to microbiota and to the nutritional and hormonal content. We also studied the long-term consequences of our interventions and we are following our cohort of very premature newborns in the prepubertal period, focusing on the

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Ines Fernanda Medina Rivera

COLLABORATOR RESEARCHER

María Dolores Salvia Roiges, Mattia Bosio, Marta Thió Lluch, Cristina Borràs Novell, Ana Herranz Barbero

SELECTED PROJECTS

- Validación prospectiva mediante un estudio multicéntrico estatal (REDSAMID) de modelos predictivos de mortalidad neonatal en prematuros. (PI20/00295). Instituto de Salud Carlos III (ISCIII). 2021-

SELECTED PUBLICATIONS

- Aldecoa V, Velilla M, Teresa M, Balcells C, Herranz A, Sin M, Iriondo M, Salvia D. Lung Ultrasound in Bronchopulmonary Dysplasia: Patterns and Predictors in Very Preterm Infants. *Neonatology*. 2021;118(5):537-545. IF:4,035. (Q1).
- Basurto D, Fuenzalida J, Martinez RJ, Russo FM, Pertierra A, Martinez JM et al. Intrapulmonary artery Doppler to predict mortality and morbidity in fetuses with mild or moderate left-sided congenital diaphragmatic hernia. *Ultrasound Obstet Gynecol*. 2021;58(4):590-596. IF:7,299. (Q1).
- Beardsall K, Thomson L, Guy C, Iglesias I, van MM, Bond S et al. Real-time continuous glucose monitoring in preterm infants (REACT): an international, open-label, randomised controlled trial. *Lancet Child Adolesc Health*. 2021;5(4):265-273. IF:11,288. (Q1).
- Durán Fernández C, Rodríguez J,

López M, Hadley S, Cavia M, Muñoz P et al. Effects of Hypothermia and Allopurinol on Oxidative Status in a Rat Model of Hypoxic Ischemic Encephalopathy. *Antioxidants* (Basel). 2021;10(10):1523. IF:6,312. (Q1).

- Iglesias I, Beardsall K. Strict glycemic control in very low birthweight infants using continuous glucose monitoring reduces dysglycemic. *J Pediatr*. 2021;238:338-342. IF:4,406. (Q1).

Pharmacological targets in inflammation and metabolic diseases

Manuel Vázquez Carrera, PhD

The purpose of our line of research is to study new pharmacological targets for the treatment of insulin resistance and type 2 diabetes, as well as such associated disorders as non-alcoholic fatty liver disease and diabetic cardiomyopathy. More specifically, one of our group's research lines is the study of the molecular mechanisms by which the activators of the peroxisome proliferator-activated receptor (PPAR) β/δ improve insulin resistance. In 2021, our research group discovered that a large part of the antidiabetic and anti-inflammatory effects of the PPAR β/δ activators are mediated by the increase of the stress-induced cytokine GDF15 (growth differentiation factor 15), which is involved in many biological processes including the regulation of energy balance. In effect, these PPAR β/δ activators increase GDF15 by the AMPK-p53 pathway. What's more, this increase of GDF15 produced by PPAR β/δ activators is of fundamental importance for its metabolic effects, since these effects are significantly reduced in GDF15-deficient mice. Additionally, PPAR β/δ ligands require GDF15 for the activation of AMPK, although this activation does not appear to require the presence of GFRAL, which is the

central receptor responsible for the action of GDF15. This fact suggests the presence of other peripheral receptors of GDF15 which could be responsible for this action. Likewise, in collaboration with the Pharmaceutical Chemistry Unit of our department, we took part in a study in which nine compounds were synthesized which inhibit the soluble epoxide hydrolase that reduces the inflammation and stress of the endoplasmic reticulum in an animal model of acute pancreatitis, a disorder for which there is not yet any specific pharmacological treatment.

MEMBERS

STAFF RESEARCHER

F. Xavier Palomer Tarridas

POSTDOC ASSOCIATED RESEARCHERS

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

SELECTED PROJECTS

- Unveiling the role of the growth differentiation factor 15 (GDF15) in non-alcoholic fatty liver disease (NAFLD). (CIBERDEM 2021/PIM04). Projectes intramurals CIBERDEM. CIBERDEM. 2021-2022. PI: Emma Barroso, Rcardo Rodriguez and María Garcia Altares.
- Activación de PPAR β/δ y HRI: explorando nuevos mecanismos compensatorios para el tratamiento de la inflamación, la resistencia a la insulina y la esteatohepatitis no alcohólica (RTI2018-093999-B-I00). Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. Ministerio de Ciencia, Innovación y Universidades. 2019-2022. PI: Manuel Vazquez Carrera.
- Diabetes y enfermedades metabólicas (CB07/08/0003). Convocatòria d'ajuts destinats a finançar estructures estables d'investigació cooperativa, a l'àrea de biomedicina i ciències de la salut, en el marc del Programa Ingenio 2010, Programa Consolider, accions CIBER. Ministerio de Sanidad y Consumo. 2007-2022. PI: Manuel Vazquez Carrera.

SELECTED PUBLICATIONS

- Aguilar D, Barroso E, Gumá L, Pizarro J, Pena L, Ruart M et al. GDF15 mediates the metabolic effects of PPAR β/δ by activating AMPK. *Cell Reports*. 2021;36(6):109501-109501. IF:9,423. (Q1).
- Codony S, Calvó C, Valverde E, Osuna S, Morisseau C, Loza MI et al. From the Design to the In Vivo Evaluation of Benzohomoadamantane-Derived Soluble Epoxide Hydrolase Inhibitors for the Treatment of Acute Pancreatitis. *J Med Chem*. 2021;64(9):5429-5446. IF:7,446. (Q1).
- Palomer FX, Aguilar D, García R, Nistal JF, Vazquez M. Sirtuins: To Be or Not To Be in Diabetic Cardiomyopathy. *Trends Mol Med*. 2021;27(6):554-571. IF:11,951. (Q1).
- Zarei M, Aguilar D, Palomer FX, Vazquez M. Revealing the role of peroxisome proliferator-activated receptor β/δ in nonalcoholic fatty liver disease. *METABOLISM*. 2021;114:154342. IF:8,694. (Q1).

Study of diseases due to immune dysfunction in pediatrics (GEMDIP)

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

The group is actively participating in various European multicentre trials to identify new effective treatments for rheumatological, autoinflammatory and primary immunodeficiency diseases. We have secured various competitive projects to study the pathophysiological bases of these diseases and have published several high-impact articles. Our focus this year has been on inflammatory diseases (multisystem inflammatory syndrome and COVID-19 and systemic lupus erythematosus) and immunodeficiencies secondary to B-cell depletion therapies (CART-19 and Rituximab) in children.

MEMBERS

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Violeta Bittermann, Juan Manuel Mosquera Angarita, Ana M^a Plaza Martín, Andrea Zacarias Crovato, Rosa Bou i Torrent, Àngela Deyà Martínez, Estibaliz Iglesias Jimenez, Mónica Piquer Gibert, Judith Sánchez Manubens

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Jordi Yagüe Ribes, Manel Juan Otero, Juan Ignacio Arostegui Gorospe

SELECTED PROJECTS

- Characterization of the nasopharyngeal microbiota in Kawasaki disease. (PI20/00517). Instituto de Salud Carlos III (ISCIII). 2021-2023. PI: Jordi Antón López.
- PIVIT_Construcció d'una sala blanca per al seu ús en teràpies avançades en pacients pediàtrics, dirigit principalment a la producció de vectors virals per a teràpia gènica. (IU16-016824). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca; Departament de Salut - Generalitat de Catalunya. 2020-2022. PI: Laia Alsina Manrique de Lara.
- Contrato predoctoral de formación en investigación en salud_Beneficiario: Yiyi Luo. (FI19/00208). Instituto de Salud Carlos III (ISCIII). 2020-2023. PI: Laia Alsina Manrique de Lara.
- ImmunAID_Immunome project consortium for AutoInflammatory Disorders. (779295). European Commission. 2018-2023. PI: Jordi Antón López.
- Los nuevos criterios de PRINTO para la clasificación de la artritis idiopática juvenil. Un proyecto basado en la evidencia científica para la revisión de los criterios de la Liga Internacional contra el Reumatismo (ILAR). (ILAR). Accountancy Departament Instituto Giannina Gaslini. 2019 PI: Jordi Antón López.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio abierto, multicéntrico, de dos cohortes y un solo grupo para

evaluar la eficacia, la seguridad y la tolerabilidad, la farmacocinética y la farmacodinámica de emapalumab en niños y adultos con síndrome de activación de macrófagos (SAM) en la enfermedad de Still (incluida la artritis idiopática juvenil sistémica y la enfermedad de Still del adulto) o con SAM en el lupus eritematoso sistémico. A two-cohort, open-label, single arm, multicenter study to evaluate efficacy, safety and tolerability, pharmacokinetics (PK) and pharmacodynamics (PD), of emapalumab in children and adults with macrophage activation syndrome (MAS) in Still's disease (including systemic juvenile idiopathic arthritis and adult onset Still's disease) or with MAS in systemic lupus erythematosus. (NI-0501-14). Pharmaceutical Research Associates España SAU; Swedish Orphan Biovitrum AG. 2021-. PI: Jordi Antón López.

- Estudio abierto y de dosis múltiples ascendentes para evaluar la seguridad, tolerabilidad, farmacocinética de romosozumab en niños y adolescentes con osteogénesis imperfecta. A Randomized, Double-blind, Placebo-controlled, Ascending Multiple-dose Study to Evaluate Safety, Pharmacokinetics, and Pharmacodynamics of Romosozumab in Children and Adolescents With Osteogenesis Imperfecta. (20160227). AMGEN S.A. 2021-. PI: Rosa Bou i Torrent.

SELECTED PUBLICATIONS

- Brogan PA, Arch B, Hickey H, Anton J, Iglesias E, Baildam E et al. Mycophenolate Mofetil Versus Cyclophosphamide for Remission Induction in Childhood Polyarteritis Nodosa: An Open-Label, Randomized, Bayesian Noninferiority Trial. *Arthritis Rheumatol.* 2021;73(9):1673-1682. IF:10,995. (Q1).
- Deyà A, Alonso A, Garcia AP, Faura A, Torredadell M, Vlaga A et al. Kinetics of humoral deficiency in CART19-treated children and young adults with acute lymphoblastic leukaemia. *Bone Marrow Transplant.* 2021;56(2):376-386. IF:5,483. (Q1).
- Deyà A, Garcia AP, Vlaga A, Jordán I, Fumadó V, Fortuny C et al. Confirmed SARS-COV-2 Infection

In A Cohort Of Children And Young Adults With Moderate Or Severe Primary Immunodeficiencies. *J Clin Immunol.* 2021;41(SUPPL 1):107-109. IF:8,317. (Q1).

- Esteve A, Anton J, Pino RM, Sánchez J, Fumadó V, Fortuny C et al. Similarities and differences between the immunopathogenesis of COVID-19-related pediatric multisystem inflammatory syndrome and Kawasaki disease. *J Clin Invest.* 2021;131(6):1-28. IF:14,808. (Q1).

Infectious diseases and microbiome

Carmen Muñoz Almagro, MD, PhD

We were acknowledged as an emerging group in 2009, as a consolidated group in 2014, and as a consolidated group with AGAUR financing (SGR00742) in 2017. Likewise, we have belonged to the Consortium for Networked Biological Research on Epidemiology and Public Health (CIBERESP) since 2016. Our group is focused on the study of the clinical, molecular and epidemiological aspects of the principal paediatric infectious diseases, and on the diagnostic innovation of infectious diseases and the study of the human microbiome. The cumulative impact factor of our group has been 1176.34 over the last 5 years and it is following an upward trend. Ours is a multidisciplinary team formed by 16 doctors, 12 predoctoral researchers and 2 research technicians specialised in paediatrics, microbiology, biostatistics, epidemiology, and management. We have 7 consolidated research lines (infection in vulnerable patients, tuberculosis, vaccine-preventable diseases, respiratory infection, diagnostic innovation, molecular epidemiological vigilance, and microbiome). These seven lines are directed by doctors who combine their research activity with university teaching. Two of them are full professors (Dr Fortuny and Dr

Muñoz-Almagro), three are associate professors (Dr Jordán, Dr Noguera-Julián and Dr García-García), one is an adjunct lecturer (Dr Launes) and one is a contracted doctor (Dr Brotons). In 2021, we consolidated our human microbiome line, as is shown by our five publications on this subject. Also of note are our studies on the impact of the COVID-19 pandemic on the paediatric population, which were carried out thanks to the collaboration of numerous donors who supported our research. We published 16 articles in which we analysed epidemiological, clinical, immunological, microbiological, and diagnostic innovation factors influencing the SARS-CoV2 infection.

MEMBERS

STAFF RESEARCHER

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COLLABORATOR RESEARCHER

Quique Bassat, Emilia Sánchez Ruiz

SELECTED PROJECTS

- Tecnologías NGS para la Vigilancia Epidemiológica Molecular de *S. pneumoniae* y para determinar su invasividad. Quo vadis y Quo invades pneumococcus?. (PI19/00104). Instituto de Salud Carlos III (ISCIII). 2020-2022. IP: Carmen Muñoz Almagro.
- Estudio CRIB (Cribado de citomegalovirus congénito en recién nacidos sanos e ingresados - Barcelona). (PI19/00095). Instituto de Salud Carlos III (ISCIII). 2020-

2022. IP: Antoni Noguera Julián.

- Contract ECD.11486- Developing an infrastructure and performing vaccine effectiveness studies for COVID-19 vaccine in the EU/EEA. LOT3 (HCW). European Centre for Disease Prevention and Control (ECDC). 2021-2022. IP: Carmen Muñoz Almagro.
- Plataforma Kids Corona. Clientes Diversos. 2020-2022. IP: Juan José García García.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de fase II, sin enmascaramiento, no comparativo y multicéntrico para evaluar la seguridad, la tolerabilidad, la eficacia y la farmacocinética del sulfato de isavuconazonio en el tratamiento de la aspergilosis invasiva (AI) o la mucormicosis invasiva (MI) en pacientes pediátricos. (9766-CL-0107). Astellas Pharma Global Development, Inc.; Syneos Health UK Limited. 2021-. IP: Clàudia Fortuny Guasch.
- Estudio de fase 3, multicéntrico, aleatorizado, con enmascaramiento parcial y controlado con palivizumab para evaluar la seguridad, la eficacia y la farmacocinética de MK-1654 en lactantes y niños con un riesgo elevado de enfermedad grave por el VRS. Phase 3, Multicenter, Randomized, Partially Blinded, Palivizumab-Controlled Study to Evaluate the Safety, Efficacy, and Pharmacokinetics of MK-1654 in Infants and Children at Increased Risk for Severe RSV Disease. (MK-1654-007). Merck, Sharp & Dohme de España. S.A. 2021-. IP: Clàudia Fortuny Guasch.
- Estudio fase III, aleatorizado, doble ciego, controlado con placebo y multicéntrico para demostrar la eficacia de una dosis única de la vacuna materna no adyuvada frente al Virus Respiratorio Sincitial (VRS), administrada por vía IM a gestantes de 18 a 49 años de edad, para la prevención de la Enfermedad vías respiratorias Bajas (EVRB) asociada al VRS en sus hijos hasta los 6 meses de edad. (212171 (RSV MAT-009)). Glaxosmithkline, S.A. 2021-. IP: Victoria Fumadó.
- Estudio de inmunogenia y seguridad de una vacuna conjugada antimeningocócica tetravalente con

respecto a Nimenrix®, y cuando se administra sola o de manera concomitante con las vacunas 9vHPV-IPV en adolescentes sanos. (MEQ00071). Sanofi-Aventis S.A. 2021-. IP: Antoni Noguera Julián.

SELECTED PUBLICATIONS

- Brotons P, Launes C, Buetas E, Fumadó V, Henares D, Fernández de Sevilla M et al. Susceptibility to Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children and Adults: A Seroprevalence Study of Family Households in the Barcelona Metropolitan Region, Spain. *Clin Infect Dis.* 2021;72(12):1-8. IF:9,079. (Q1).
- Esteve A, Anton J, Pino RM, Sánchez J, Fumadó V, Fortuny C et al. Similarities and differences between the immunopathogenesis of COVID-19-related pediatric multisystem inflammatory syndrome and Kawasaki disease. *J Clin Invest.* 2021;131(6):1-28. IF:14,808. (Q1).
- Henares D, Brotons P, Fernández de Sevilla M, Fernández A, Hernández S, Pérez A et al. Differential nasopharyngeal microbiota composition in children according to respiratory health status. *Microb Genom.* 2021;7(10):661. IF:5,237. (Q1).
- Redin A, Ciruela P, Fernández de Sevilla M, Gomez F, Gonzalez S, Benitez MA et al. Serotypes and Clonal Composition of *Streptococcus pneumoniae* Isolates Causing IPD in Children and Adults in Catalonia before 2013 to 2015 and after 2017 to 2019 Systematic Introduction of PCV13. *Microbiol Spectr.* 2021;9(3) :e0115021. IF:7,171. (Q1).

Gut microbiota

Laura Baldomà Llavínés, PhD

Gut bacteria release extracellular vesicles as an intercellular communication mechanism that prime the host innate immune system. Previous studies of our group showed

that extracellular vesicles from gut resident *E. coli* activate dendritic cells (DCs) and subsequent T cell responses in a strain-specific manner. In addition to direct cell-to-cell contacts, DCs communicate with neighboring T cells through secreted mediators. This year we have provided new insights into the mechanisms used by microbiota to orchestrate host immune regulation. In particular, we have analysed the impact of microbiota vesicles on DC-secreted effectors including cytokines and exosomes. DC-derived exosomes were characterized in terms of costimulatory molecules and miRNAs cargo. Exosomes released by DCs activated by microbiota vesicles were enriched in surface proteins involved in antigen presentation and T cell activation but differ in the content of immune-related miRNA depending on the producer bacterial strain. These differences were consistent with the derived immune responses. To our knowledge, this is the first study showing the influence of microbiota extracellular vesicles on the immunomodulatory cargo of DC-derived exosomes.

In the context of our contribution to the study of biological activities of nanoparticles for drug delivery, we have collaborated with groups of the Nanobiotechnology Research Institute-UB. In particular, we have shown that thymol-loaded nanoparticles are an efficient alternative for the treatment of skin acne infection based on their antioxidant and antimicrobial activity against *Cutibacterium acnes*. We have also contributed to characterize the uptake and anti-inflammatory activity of lactoferrin-loaded liposomes as a drug delivery system for the treatment of dry eye disease.

MEMBERS

STAFF RESEARCHER

Rosa Giménez Claudia, Josefa Badia Palacin

SELECTED PROJECTS

- Evaluación de nuevas actividades de vesículas secretadas por probióticos y microbiota intestinal y su potencial aplicación como postbióticos en salud humana. (PID2019-107327RB-I00). Ministerio de Ciencia, Innovación y



- Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2020-2023. PI: Laura Baldoma Llavines.
- C-C Bond Formation Using Top Performing Enzymes (CC-TOP). (956631). Unió Europea. Horizon 2020 (2014-2020). P1. Marie Skłodowska-Curie Innovative Training Networks (H2020-MSCA-ITN-2020). 2021-2025. PI: Josefa Badia Palacin.

SELECTED PUBLICATIONS

- Díaz N, Badia J, Baldoma L. Microbiota-derived extracellular vesicles in interkingdom communication in the gut. *J Extracell Vesicles*. 2021;10(13):12161. IF:25,841. (Q1).
- Díaz N, Cordero C, Olivo Y, Badia J, Baldoma L. Cell-to-Cell Communication by Host-Released Extracellular Vesicles in the Gut: Implications in Health and Disease. *Int. J. Mol. Sci*. 2021;22(4):2213. IF:5,923. (Q1).
- Folle C, Marqués AM, Jorge G, Espina M, Sánchez E, Badia J et al. Thymol-loaded PLGA nanoparticles: an efficient approach for acne treatment. *J Nanobiotechnology*. 2021;19(1):359. IF:10,435. (Q1).

- López A, Jorge G, Cano A, Espina M, Badia J, Baldoma L et al. Development of Lactoferrin-Loaded Liposomes for the Management of Dry Eye Disease and Ocular Inflammation. *Pharmaceutics*. 2021;13(10):1698. IF:6,321. (Q1).

Environment and complications in pregnancy

Maria Dolores Gómez Roig, MD, PhD

We continued to study the influence of abusive substances and of the materno-foetal environment on foetal and newborn health. We completed the recruitment of the Barcelona Life Study Cohort (BISC), which comprises 1,100 mother-child pairs in the Greater Barcelona area, who are well phenotyped for environmental exposure. With this cohort, we will be able to continue conducting research along our line. Our areas of interest are the study of the influence of lifestyle and exposure to environmental contaminants on

foetal neurodevelopment, and on heart morphology and functionalism. We have also extended our studies to include new methodologies for the study of postnatal neurodevelopment such as frequency-following response (FFR). Another aspect of the work that we have carried out is the study of how the environment affects the placental function and foetal growth, and it may also be noted that we have increased our experimental knowledge about alcohol consumption during pregnancy.

MEMBERS

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TECHNICIAN

Àgueda García Meseguer

SELECTED PROJECTS

- Impacto de un programa de intervención prenatal estructurado, de disminución de consumo de alcohol, en los resultados perinatales y de neurodesarrollo. (PI20/00490). Instituto de Salud Carlos III (ISCIII). 2021-2023. PI: Maria Dolores Gómez Roig.
- Respuesta de seguimiento de frecuencia (RSD) como biomarcador de desarrollo neurocognitivo en recién nacidos con exposición prenatal al alcohol. Hospital Sant Joan de Déu - Esplugues HSJD. 2020-2023. PI: Maria Dolores Gómez Roig.
- ATHLETE_Advancing Tools for Human Early Lifecourse Exposome research and Translation. (874583). European Commission. 2020-2024. PI: Maria Dolores Gómez Roig.
- Traffic-related air pollution and birth weight: the roles of noise, placental function, green

space, diet, physical activity, and socioeconomic status (FRONTIER). (CR-83590201). Health Effects Institute (HEI). 2018-2022. PI: Maria Dolores Gómez Roig.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Hydroxychloroquine efficacy in preventing SARS-CoV-2 infection and CoVid-19 disease severity during pregnancy. (CoVid-Preg). Fundació Privada ISGLOBAL. 2020-. PI: Eduarne Mazarico Gallego.
- Tratamiento del crecimiento intrauterino restringido precoz con heparina de bajo peso molecular: Ensayo Clínico Randomizado (TRACIP). (FSJD-TRACIP-2017). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2017-. PI: Eduarne Mazarico Gallego.
- Tratamiento del cir precoz con heparina de bajo peso molecular (TRACIP): ensayo clínico randomizado. (PI16/00151). Instituto de Salud Carlos III (ISCIII). 2017-2021. PI: Eduarne Mazarico Gallego.

SELECTED PUBLICATIONS

- Almeida L, Andreu V, Aras R, García Ó, Martínez L, Gómez MD. Epigallocatechin Gallate Ameliorates the Effects of Prenatal Alcohol Exposure in a Fetal Alcohol Spectrum Disorder-Like Mouse Model. *Int. J. Mol. Sci.* 2021;22(2):715. IF:5,923. (Q1).
- Arenillas S, Costa J, Ribas T, Gómez MD, Escera C. Neural encoding of voice pitch and formant structure at birth as revealed by frequency-following responses. *Sci Rep.* 2021;11(1):6660-6660. IF:4,379. (Q1).
- Crovetto F, Fátima B, Llorba E, Pascal R, Larroya M, Trilla C et al. Impact of Severe Acute Respiratory Syndrome Coronavirus 2 Infection on Pregnancy Outcomes: A Population-based Study. *Clin Infect Dis.* 2021;73(10):1768-1775. IF:9,079. (Q1).
- Meler E, Mazarico E, Eixarch E, Gonzalez A, Peguero A, Martinez J et al. Ten-year experience of protocol-based management of small-for-gestational-age fetuses: perinatal outcome in late-pregnancy

cases diagnosed after 32 weeks. *Ultrasound Obstet Gynecol.* 2021;57(1):62-69. IF:7,299. (Q1).

Cardiovascular diseases in development

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

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Bosco Alejandro Moscoso Garrido

SELECTED PROJECTS

- CoALeS: Improving long term outcome in infants with coarctation through machine learning for data integration from fetus to childhood. (202016-31). Fundació La Marató de TV3. 2021-2024. IP: Joan Sánchez de Toledo.
- Antioxidantes y neuroprotección en un modelo animal pediátrico de circulación extracorporea y parada circulatoria: Nuevos horizontes terapéuticos. (PI20/00298). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Joan Sánchez de Toledo.
- ACCELERATE_AI Accelerator to "A Smart Hospital Care Pathway Engine. (101016902). European Commission. 2021-2024. IP: Joan Sánchez de Toledo.
- TinyBrains_Bio-photonic imaging of the infant brain, the missing link between the cellular brain damage and the neurovascular unit during acute illness. (101017113). European Commission. 2021-2024. IP : Joan Sánchez de Toledo.
- ECG neonatal sistemático para la

detección precoz de enfermedades cardíacas relacionadas con la muerte súbita del lactante. Sociedad Española de Cardiología. 2019-2021. IP: Georgia Sarquella Brugada.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio aleatorizado, multicéntrico, doble ciego, controlado con placebo, de grupos paralelos, basado en acontecimientos y con diseño secuencial por grupos con un periodo de ampliación abierto para evaluar la eficacia y seguridad de selexipag como tratamiento complementario al tratamiento de referencia en niños de 2 a <18 años con hipertensión arterial pulmonar. A Randomized, Multicenter, Double-Blind, Placebo-Controlled, Parallel-Group, Event-Driven, Group-Sequential Study with Open-Label Extension Period to Assess the Efficacy and Safety of Selexipag as Add-On Treatment to Standard of Care in Children Aged 2 to <18 years with Pulmonary Arterial Hypertension. [AC-065A310]. Janssen - Cilag, S.A. 2020-. IP: Joan Sánchez de Toledo.
- Estudio multicéntrico para evaluar la seguridad y tolerabilidad a largo plazo de un tratamiento en abierto con sacubitrilo/valsartán en pacientes pediátricos con insuficiencia cardíaca debida a disfunción sistólica de ventrículo izquierdo sistémico, que han completado el estudio CLCZ696B2319. [CLCZ696B2319E1 - Ext.]. Novartis Farmacéutica, S.A. 2020-. IP: Joan Sánchez de Toledo.
- Estudio multicéntrico, abierto, para evaluar la seguridad, tolerabilidad, farmacocinética y farmacodinámica de LCZ696, seguido de un estudio de 52 semanas, aleatorizado, doble ciego, paralelo controlado con fármaco activo para evaluar la eficacia y seguridad de LCZ696 comparado con enalapril en pacientes pediátricos con edades desde 1 mes hasta < 18 años, con insuficiencia cardíaca debida a disfunción sistólica del ventrículo izquierdo sistémico. [CLCZ696B2319]. Novartis Farmacéutica, S.A. 2019-. IP: Joan Sánchez de Toledo.

SELECTED PUBLICATIONS

- Esteve A, Anton J, Pino RM, Sánchez J, Fumadó V, Fortuny C et al. Similarities and differences between the immunopathogenesis of COVID-19-related pediatric multisystem inflammatory syndrome and Kawasaki disease. *J Clin Invest.* 2021;131(6):1-28. IF:14,808. (Q1).
- Sarquella G, Fernandez A, Cesar S, Arbelo E, Jordà P, García A et al. Pediatric Malignant Arrhythmias Caused by Rare Homozygous Genetic Variants in TRDN: A Comprehensive Interpretation. *Front Pediatr.* 2021;8:601708-601708. IF:3,418. (Q1).
- Sarquella G, Lorena A, Zambrano MD, Fernández A, Sailer S, Cesar S et al. Early Identification of Prolonged QT Interval for Prevention of Sudden Infant Death. *Front Pediatr.* 2021;9:704580-704580. IF:3,418. (Q1).
- Valverde I, Singh Y, Sanchez J, Theocharis P, Chikermane A, Di S et al. Acute Cardiovascular Manifestations in 286 Children With Multisystem Inflammatory Syndrome Associated With COVID-19 Infection in Europe. *Circulation.* 2021;143(1):21-32. IF:29,690. (Q1).

Immune and respiratory dysfunction in the critical child

Francisco José Cambra Lasaosa, MD, PhD

Notable aspects of 2021 included our research on the pathophysiology of severe illness caused by COVID in paediatric patients as well as in adults, considering the number of patients over age 18 years hospitalised in our intensive care unit during the COVID epidemic. We investigated various aspects in depth and produced several publications. Our research, which was led by Dr Iolanda Jordan, was conducted with a multidisciplinary

approach, involving various services of our hospital.

Our scientific output was maintained with several publications along our two priority research lines: the field of inflammatory response and that of respiratory insufficiency. Also during this period, Sara Bobillo Pérez presented her doctoral thesis "Utility of biomarkers in systemic inflammatory response and in the implementation of antibiotic-therapy handling algorithms in paediatric critical patients". The thesis was directed by Iolanda Jordan and Francisco José Cambra. University of Barcelona (UB). Grade: Excellent Cum Laude.

MEMBERS

STAFF RESEARCHER

Iolanda Jordán García, Martí Pns Ódena, Anna Solé Ribalta, Carmina Guitart Pardellans, Mónica Balaguer Gargallo, Elisabeth Esteban Torne, Mónica Girona Alarcón

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Jorgelina Lopez Zabala, Vanessa Andriño Estrecha, Nuria Millán García del Real, Elena Fresán Ruiz, Sergio Benito Fernández, Lluisa Hernandez Platero, Carme Alejandro Galobardes, Patricia Corniero Alonso, Aida Felipe Villalobos, Susana Segura Matute, David Vila Pérez, Sergi Huerta Calpe, Ricardo Suarez Ramirez

COLLABORATOR RESEARCHER

Montserrat Esquerda Aresté

SELECTED PROJECTS

- Implicación de la microbiota bacteriana de la nasofaringe en el desarrollo de neumonía asociada a ventilación mecánica en la unidad de cuidados intensivos pediátricos. Estudio MiNoReP. (PI19/00106). Instituto de Salud Carlos III (ISCIII). 2020-2022. IP: Iolanda Jordán García.
- Valoración del efecto neuroprotector de la dexmedetomidina en el modelo animal pediátrico de circulación extracorporea y parada circulatoria. Sociedad y Fundación Española de Cuidados Intensivos Pediátricos (SECIP). 2018-2022. IP: Sergio Benito Fernández.
- Estudio de seguridad de un dispositivo de presión positiva no mecánico en pacientes con insuficiencia respiratoria

hipoxémica secundaria a neumonía por SARS COV 2. [CPAP]. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2020IP: Martí Pons Òdena.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio aleatorizado con control activo para comparar la eficacia y la seguridad de isoflurano inhalado administrado por el AnaConDa-S (dispositivo de conservación de anestésico) con el midazolam intravenoso para la sedación en pacientes pediátricos con ventilación mecánica de entre 3 y 17 años (menos de 18 años). A Randomised Active-controlled Study to Compare Efficacy and Safety of Inhaled Isoflurane Delivered by the AnaConDa-S (Anaesthetic Conserving Device) to Intravenous Midazolam for Sedation in Mechanically Ventilated Paediatric Patients 3 to 17 (Less than 18) Years Old. (SED002). LINK Medical Research; Sedana Medical AB. 2021IP: Monica Girona Alarcon.
- Estudio abierto, de un solo brazo para evaluar la farmacocinética, seguridad y tolerabilidad, y para explorar los resultados clínicos del tratamiento con dosis única o múltiple de zanamivir IV en neonatos e infantes menores de 6 meses con infección por gripe confirmada complicada. An open label, single arm study to evaluate the pharmacokinetics, safety and tolerability of intravenous (IV) zanamivir in hospitalised neonates and infants under 6 months of age with confirmed or suspected influenza infection. (200925). Glaxosmithkline, S.A. 2020IP: Mónica Balaguer Gargallo.
- A Phase 1, open-label, non-comparative, multicenter clinical trial to evaluate the safety, tolerability, and pharmacokinetics of ceftolozane/tazobactam (MK-7625A) in pediatric participants with nosocomial pneumonia. (MK-7625A-036). Merck, Sharp & Dohme de España. S.A. 2019IP: Iolanda Jordán García.

SELECTED PUBLICATIONS

- Alejandre C, Guitart C, Balaguer M, Torrús I, Bobillo S, Cambra

FJ et al. Use of procalcitonin and C-reactive protein in the diagnosis of bacterial infection in infants with severe bronchiolitis. *Eur J Pediatr.* 2021;180(3):833-842. IF:3,183. (Q1).

- Bobillo S, Balaguer M, Jordán I, Batista A, Ramon M, Otero O et al. Delivery room ultrasound study to assess heart rate in newborns: DELIROUS study. *Eur J Pediatr.* 2021;180(3):783-790. IF:3,183. (Q1).
- Dobaño C, Alonso S, Fernández de Sevilla M, Vidal M, Jiménez A, Pons G et al. Antibody conversion rates to SARS-CoV-2 in saliva from children attending summer schools in Barcelona, Spain. *BMC Med.* 2021;19(1):309. IF:8,775. (Q1).
- Esquerda M, Palau F, Lorenzo-Izquierdo D, Cambra FJ, Bofarull M, Cusi V et al. Ethical questions concerning newborn genetic screening. *Clin Genet.* 2021;99(1):93-98. IF:4,438. (Q2).

Allergic diseases in childhood and adolescence

Montserrat Alvaro Lozano, MD, PhD

MEMBERS

STAFF RESEARCHER

Rosa Jiménez Feijoo, Jaime Lozano Blasco, Adrianna Machinena Spera

PHD STUDENTS

Olga Dominguez Sánchez

SELECTED PROJECTS

- Estudio de la alergenicidad de diferentes proteínas en la alergia alimentaria a pescado en población pediátrica. Sociedad Española de Inmunología Clínica, Alergia y Asma Pediátrica. 2021-2022. IP: Olga Dominguez Sánchez.
- Genetics of Food Allergy Barcelona Montserrat Alvaro. Max Delbrück Centrum für Molekulare medizin Berlin-Buch. 2021-2022. IP: Montserrat Alvaro Lozano.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de 52 semanas, aleatorizado, doble ciego, controlado con placebo, de grupos paralelos, multicéntrico sobre la eficacia y seguridad de la terapia adyuvante de GSK3511294 en participantes adultos y adolescentes con asma grave no controlada con un fenotipo eosinofílico. A 52-week, randomised, double-blind, placebo-controlled, parallel-group, multi-centre study of the efficacy and safety of GSK3511294 adjunctive therapy in adult and adolescent participants with severe uncontrolled asthma with an eosinophilic phenotype. (213744). Glaxosmithkline, S.A; IQVIA RDS Spain SL. 2021-. IP: Jaime Lozano Blasco.
- Estudio de 52 semanas de duración, aleatorizado, doble ciego, con doble simulación, de grupos paralelos, multicéntrico, de no inferioridad, para evaluar la tasa de exacerbación, las mediciones adicionales de control del asma y la seguridad en participantes adultos y adolescentes con asma grave con fenotipo eosinofílico tratados con GSK3511294 en comparación con mepolizumab o benralizumab. A 52-week, randomised, double-blind, double-dummy, parallel group, multi-centre, non-inferiority study assessing exacerbation rate, additional measures of asthma control and safety in adult and adolescent severe asthmatic participants with an eosinophilic phenotype treated with GSK3511294 compared with mepolizumab or benralizumab. (206785). Glaxosmithkline, S.A; IQVIA RDS Spain SL. 2021-. IP: Jaime Lozano Blasco.

SELECTED PUBLICATIONS

- Escarrer M, Julia JC, Quevedo S, Prieto Del A, Mónica Paola Sandoval R, Quesada F et al. Cambios en la epidemiología y en la práctica clínica de la alergia mediada por IgE en pediatría. *An. Pediatr.* 2021;95(1): 56.e1-56.e8. IF:1,500. (Q4).
- Pfaar O, Agache I, Bonini M, Brough HA, Chivato T, Del Giacco SR et al. COVID-19 pandemic and allergen immunotherapy-an EAACI survey. *Allergy.* 2021;76(11):3504-3516. IF:13,146. (Q1).



7.1

MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.3

Paediatric cancer

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



Translational genomics

Cecilia Emilia Lavarino, PhD

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Alba Parra Checa

TECHNICIAN

Sonia Paco Mercader, Isadora Lemos Das Neves

SELECTED PROJECTS

- Desarrollo de clasificadores epigenéticos para el Cáncer Pediátrico (ECPC). (PI20/00519). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Cinzia Emilia Lavarino.
- Platform of Epigenetic Classifiers for Childhood CAncer (PECA). (201921-30). Fundació La Marató de TV3. 2020-2023. IP: Cinzia Emilia Lavarino.
- Método para la clasificación en subgrupos moleculares de pacientes con Meduloblastoma. (PI17/00353). Instituto de Salud Carlos III (ISCIII). 2018-2022. IP: Cinzia Emilia Lavarino.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Ensayo de Fase I/II, Abierto y de un solo brazo para Evaluar la Farmacocinética, Seguridad/Tolerabilidad y Eficacia de la Formulación Granulada de Selumetinib en niños de 1 a < 7 Años con Neurofibromas Plexiformes (NP) Sintomáticos e Inoperables relacionados con Neurofibromatosis Tipo 1 (NF1) (SPRINKLE). (D1346C00004). Astrazeneca Farmaceutica Spain, S.A. 2021-. IP: Héctor Salvador Hernández.
- Ensayo fase I, de un solo brazo, secuencial, para evaluar el efecto de los alimentos sobre la seguridad y farmacocinética de selumetinib

después de dosis múltiples en niños adolescentes con neurofibromas plexiformes (NP) asociados a neurofibromatosis tipo 1 (NF1). A Phase I, Single-Arm, Sequential Study to Evaluate the Effect of Food on the Gastrointestinal Toxicity and Pharmacokinetics of Selumetinib after Multiple Doses in Adolescent Children with Neurofibromatosis Type 1 (NF1) Related Plexiform Neurofibromas (PN). (D1346C00015). Astrazeneca Farmaceutica Spain, S.A. 2021-. IP: Héctor Salvador Hernández.

SELECTED PUBLICATIONS

- Khan S, Solano P, Suwal T, Lu M, Al S, Ho B et al. Clinical phenotypes and prognostic features of embryonal tumours with multi-layered rosettes: a Rare Brain Tumor Registry study. *Lancet Child Adolesc Health*. 2021;5(11):800-813. IF:11,288. (Q1).
- Petit M, Guasp M, Armangue T, Lavarino C, Morales A, Saiz A et al. Absence of GluD2 Antibodies in Patients With Opsoclonus-Myoclonus Syndrome. *Neurology*. 2021;96(7): e1082-e1087. IF:9,910. (Q1).
- Oliván M, Garcia M, Suárez L, Guiu M, Gros L, Méndez O et al. Loss of microRNA-135b Enhances Bone Metastasis in Prostate Cancer and Predicts Aggressiveness in Human Prostate Samples. *Cancers (Basel)*. 2021;13(24):6202IF:6,639. (Q1).
- Sievers P, Henneken SC, Blume C, Sill M, Schrimpf D, Stichel D et al. Recurrent fusions in PLAGL1 define a distinct subset of pediatric-type supratentorial neuroepithelial tumors. *Acta Neuropathol*. 2021;142(5):827-839. IF:17,088. (Q1).

Sarcomas and histiocytosis

Jaume Mora Graupera, MD, PhD

In 2021, while we were still immersed in the pandemic, which has fortunately not adversely affected children and juveniles, our Cancer Group carried

on with its mission of expanding our knowledge of this disease, which is responsible for the greatest mortality in the child population of the advanced countries.

In the scientific sphere, mention should be made of the publications of the neuroblastoma immunotherapy team directed by Dr Jaume Mora, which has become consolidated as an international reference group in the treatment of high-risk neuroblastoma. The information collected in the "Pivotal phase 2 trial of antibody naxitamab (hu3F8) and granulocyte-macrophage colony stimulating factor (GM-CSF) in high-risk neuroblastoma patients with primary refractory disease or incomplete response to salvage treatment in bone and/or bone marrow (Y-MABS 201) Y-mAbs Therapeutics A/S. 2018", 50% of which was provided by patients of our centre, allowed the FDA to approve naxitamab for the treatment of relapsed/refractory neuroblastoma in bone marrow/bone on 20 November 2020.

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Soraya Morales Moreno, Óscar Muñoz Aznar, Noelia Salvador Marcos

COLLABORATOR RESEARCHER

Maria Inmaculada Hernandez Muñoz

SELECTED PROJECTS

- Evaluation of the effects of Oryzon Genomics proprietary compounds on pediatric patient tumors. Oryzon Genomics S.A. 2021-2023. IP: Jaume Mora Graupera.
- Ipatasertib in Rhabdomyosarcoma.

Roche Diagnostics International Ltd. 2019-2021. IP: Jaume Mora Graupera.

- Influencia del contexto epigenético en la edad como factor pronóstico en Ewing Sarcoma. (PI20/00179). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP:Jaume Mora Graupera.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Ensayo Clínico plataforma en fase II de inmuno-oncología de precisión con indicación tumor agnóstica para mutaciones somáticas específicas (TAPISTRY). Tumor-agnostic precision immuno-oncology and somatic targeting rational for you (Tapistry) phase II platform trial. (B041932). Roche Farma, S.A. 2021IP: Jaume Mora Graupera.
- An overarching study for children and adults with Frontline and Relapsed RhabdoMyoSarcoma. (FaR-RMS). Fundación Sociedad Española de Hematología y Oncología Pediátricas. 2021IP: Moira Garraus Oneca.
- Estudio de fase 1, multicéntrico, abierto, con intensificación de dosis y expansión de cohorte con Niraparib y Dostarlimab en pacientes pediátricos con tumores sólidos recurrentes o refractarios. A phase 1, multicentre, open-label, dose-escalation and cohort expansion study of niraparib and dostarlimab in paediatric patients with recurrent or refractory solid tumours. (213406). GlaxoSmithKline Research and Development Ltd. 2021IP: Jaume Mora Graupera.

SELECTED PUBLICATIONS

- Mora J, Castañeda A, Colombo MC, Gorostegui M, Fernando M, Mañe S et al. Clinical and Pathological Evidence of Anti-GD2 Immunotherapy Induced Differentiation in Relapsed/Refractory High-Risk Neuroblastoma. *Cancers (Basel)*. 2021;13(6): 164. IF:6,639. (Q1).
- Mora J, Castañeda A, Gorostegui M, Santa V, Garraus M, Muñoz JP et al. Naxitamab combined with granulocyte-macrophage colony-stimulating factor as consolidation for high-risk neuroblastoma patients in complete remission.

Pediatr Blood Cancer. 2021;68(10): e29121. IF:3,167. (Q1).

- Castillo H, Pascual G, Pérez S, Resa C, Vilà M, Monterrubio C et al. Prognostic value of patient-derived xenograft engraftment in pediatric sarcomas. *J Pathol Clin Res*. 2021;7(4):338-349. IF:5,638. (Q1).
- García DJ, Hajji N, Sánchez S, Figuerola E, de Pablos RM, Espinosa AM et al. Selective inhibition of HDAC6 regulates expression of the oncogenic driver EWSR1-FLI1 through the EWSR1 promoter in Ewing sarcoma. *Oncogene*. 2021;40(39):5843-5853. IF:9,867. (Q1).

Therapy development in pediatric oncology

Angel Montero Carcaboso, PhD

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TECHNICIAN

Ana Jiménez Cabaco, Mònica Vilà Ubach

COLLABORATOR RESEARCHERS

Guillermo Chantada, Ofelia Cruz Martínez, Andrés Morales La Madrid

SELECTED PROJECTS

- Desenvolupament preclínic del fàrmac G2B-001 i aplicació en el tractament de tumors sòlids pediàtrics. (2021 DI 008). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2021-2024. IP: Angel Montero Carcaboso.
- Inhibition of the PI3K/Akt pathway in pediatric high grade and diffuse intrinsic pontine gliomas (pHGG/DIPG). (201927-30). Fundació La

Marató de TV3. 2020-2023. IP: Angel Montero Carcaboso.

- Contrato predoctoral de formación en investigación en salud_ Beneficiario: Helena Castillo. (FI19/00138). Instituto de Salud Carlos III (ISCIII). 2020-2023. IP:Angel Montero Carcaboso.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- SIOP Ependimoma II - Programa clínico internacional para el diagnóstico y tratamiento de niños, adolescentes y adultos jóvenes con ependimoma. (ET-13-002). Sociedad Española de Hematología y Oncología Pediátricas. 2017-. IP: Ofelia Cruz Martínez.
- Phase Ib Clinical Trial on the Safety of Immunotherapy With Autologous Dendritic Cells Primed With Lysate Allogeneic Tumor Lines in Patients With Diffuse Intrinsic Pontine Glioma (DIPG). (FSJD-DIPG-DC). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2016-- IP: Andrés Morales La Madrid.
- Estudio prospectivo internacional en niños mayores de 3 a 5 años con meduloblastoma de riesgo estándar clínico de perfil biológico de bajo riesgo. (SIOP - PNET 5 MB - LR). Sociedad Española de Hematología y Oncología Pediátricas. 2016-. IP: Ofelia Cruz Martínez.

SELECTED PUBLICATIONS

- Aschero R, Francis JH, Ganiewich D, Gomez S, Sampor C, Zugbi S et al. Recurrent Somatic Chromosomal Abnormalities in Relapsed Extraocular Retinoblastoma. *Cancers (Basel)*. 2021;13(4):673IF:6,639. (Q1).
- Castillo H, Pascual G, Pérez S, Resa C, Vilà M, Monterrubio C et al. Prognostic value of patient-derived xenograft engraftment in pediatric sarcomas. *J Pathol Clin Res*. 2021;7(4):338-349. IF:5,638. (Q1).
- Mukkada S, Bhakta N, Chantada G, Chen Y, Vedaraju Y, Faughnan L et al. Global characteristics and outcomes of SARS-CoV-2 infection in children and adolescents with cancer (GRCC): a cohort study. *Lancet Oncol*. 2021;22(10):1416-1426. IF:41,316. (Q1).
- Sievers P, Henneken SC, Blume

C, Sill M, Schrimpf D, Stichel D et al. Recurrent fusions in PLAGL1 define a distinct subset of pediatric-type supratentorial neuroepithelial tumors. *Acta Neuropathol.* 2021;142(5):827-839. IF:17,088. (Q1).

Paediatric leukaemia and haematopoiesis

Mireia Camós Guijosa, MD, PhD

The Leukaemias and Other Paediatric Haemopathies group was very active in 2021, despite external difficulties.

We continued to work on the projects which had been previously begun and we obtained important grants for the start-up of new projects.

For example, some new projects were implemented in 2021 in relation to acute leukaemia. On the one hand, Dr Català is leading the GEMMA study for the complete characterisation of the myelodysplastic syndromes and hereditary acute myeloid leukaemias in childhood, a project funded by the Foundation La Marató de TV3.

On the other hand, the Unoentrecienmil (One in a Hundred Thousand) Foundation financed a project to facilitate the implementation of the ALLTogether treatment strategy for paediatric acute lymphoblastic leukaemia in Spain, which will become the next nationwide healthcare protocol.

Our activity in clinical trials was very intense in all fields in 2021, and our group leads and participates in trials with directed treatments and with various forms of immunotherapy in leukaemia and other types of haemopathies. Notably, Dr Rives continues to lead nationally and internationally the CART therapy for acute leukaemia, conducting various clinical trials and promoting new research projects.

MEMBERS

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Maximiliano Ezequiel Distefano Muñoz

TECHNICIAN

Cèlia Portillo Bog, Warda Suleman, Lidia Ruiz Bocanegra, Maria Camino Estella Aguado, Mercè Richarte Frnaqués

SELECTED PROJECTS

- New approaches for the identification and functional characterization of prognostic genetic biomarkers in inherited childhood myelodysplastic/acute leukemia syndromes Acronym: GEMMA. Fundació La Marató de TV3. 2021-2024. IP: Albert Català Temprano.
- Hacia la personalización de la profilaxis de la trombosis venosa profunda en pacientes pediátricos afectos de leucemia linfoblástica aguda: uso de test de generación de trombina. (PI17/00356). Instituto de Salud Carlos III (ISCIII). 2018-2022. IP: Rubén Berruero Moreno.
- Implementación de la estrategia de tratamiento Alltogether para leucemia linfoblástica aguda pediátrica en España. Fundación Uno entre Cien Mil. 2021-2022. IP: Susana Rives Solà.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de fase 1/2, abierto y de un solo brazo que evalúa la seguridad y eficacia del ponatinib para el tratamiento de leucemias o tumores sólidos recurrentes o refractarios en pacientes pediátricos. An Open-Label, Single-Arm, Phase 1/2 Study Evaluating the Safety and Efficacy of Ponatinib for the Treatment of Recurrent or Refractory Leukemias or Solid Tumors in Pediatric Participants. (INCB 84344-102). Incyte Biosciences International

Sàrl; Pharmaceutical Product Development Spain, S.L. 2021. IP: Albert Català Temprano.

- Estudio de fase 1/2 multicéntrico para evaluar la seguridad y eficacia de KTEX19 en sujetos pediátricos y adolescentes con leucemia linfoblástica aguda de precursores de linfocitos B recidivante/refractaria o linfoma no Hodgkin de linfocitos B recidivante/refractario (ZUMA-4). A Phase 1/2 Multi-Center Study Evaluating the Safety and Efficacy of KTE-X19 in Pediatric and Adolescents Subjects with Relapsed/Refractory B-precursor Acute Lymphoblastic Leukemia or Relapsed/Refractory B-Cell Non-Hodgkin Lymphoma (ZUMA-4). (KTE-C19-104 [Zuma - 4]). Kite Pharma Inc; Labcorp Drug Development Inc. 2021. IP: Susana Rives Solà.
- Estudio en fase III, aleatorizado, abierto, con control activo, multicéntrico, para evaluar la eficacia y la seguridad de Crovalimab frente a Eculizumab en pacientes con hemoglobinuria paroxística previamente con inhibidores del complemento. A Phase III, Randomized, Open-Label, Active-Controlled, Multicenter Study Evaluating The Efficacy And Safety Of Crovalimab Versus Eculizumab In Adult And Adolescent Patients With Paroxysmal Nocturnal Hemoglobinuria not previously treated with complement inhibitors. (BO42162). IQVIA RDS Spain SL; Roche Diagnostics International Ltd. 2021. IP: Albert Català Temprano.
- Un estudio de fase III, abierto, de un solo grupo, multicéntrico de Ravulizumab junto al mejor tratamiento sintomático en participantes pediátricos (de 1 mes a <18 años) con microangiopatía trombótica (MAT) después de un trasplante de células madre hematopoyéticas (TCMH). A Phase 3, Open-label, Single Arm, Multicenter Study of Ravulizumab in Addition to Best Supportive Care in Pediatric participants (from 1 month to < 18 years of age) with Thrombotic Microangiopathy (TMA) after Hematopoietic Stem Cell transplantation (HSCT). (ALXN1210-TMA-314). Alexion Pharmaceuticals Inc.; IQVIA RDS Spain SL. 2021. IP: Julia Marsal Ricomà.



SELECTED PUBLICATIONS

- Levine JE, Grupp SA, Pulsipher MA, Dietz AC, Rives S, Myers GD et al. Pooled safety analysis of tisagenlecleucel in children and young adults with B cell acute lymphoblastic leukemia. *J Immunother Cancer*. 2021;9(8):e002287. IF: 13,751. (Q1).
- Navarro S, Sevilla J, Rio P, Sanchez R, Zubizaray J, Galvez E et al. Hematopoietic Stem Cell Collection for the Gene Therapy of Fanconi Anemia Patients. *Mol Ther*. 2021;29(4):342-342. IF: 11,454. (Q1).
- Ortíz V, Rives S, Castellà M, Alonso A, Benítez D, Caballero M et al. CART19-BE-01: A Multicenter Trial of ARI-0001 Cell Therapy in Patients with CD19(+) Relapsed/Refractory Malignancies. *Mol Ther*. 2021;29(2):636-644. IF: 11,454. (Q1).
- Rives S. CAR T cells in CNS-relapsed leukaemia: one step forward. *Lancet Haematol*. 2021;8(10):e675-e676. IF: 18,959. (Q1).
- Sahoo SS, Pastor VB, Goodings C, Voss RK, Kozyra EJ, Szvetnik A et al. Clinical evolution, genetic landscape and trajectories of clonal hematopoiesis in SAMD9/SAMD9L syndromes. *Nat Med*. 2021;27(10):1806-1817. IF: 53,440. (Q1).
- Vega N, Pérez S, Esperanza E, Vicente C, Torredadell M, Jiménez A et al. Measurable Residual Disease Assessed by Flow-Cytometry Is a Stable Prognostic Factor for Pediatric T-Cell Acute Lymphoblastic Leukemia in Consecutive SEHOP Protocols Whereas the Impact of Oncogenetics Depends on Treatment. *Front Pediatr*. 2021;8:614521. IF: 3,418. (Q1).

Molecular pharmacology and experimental therapies

Marçal Pastor-Anglada, PhD

The bioavailability of certain anti-tumoural drugs whose mechanism of action is based on interference with nucleotide metabolism depends on transport proteins located in the cell membrane. The expression of these transporters is modified in both solid and liquid tumours and it is not uncommon to find that some specific subtypes responsible for the uptake of these drugs, such as the protein hCNT1, present negligible levels in tumours. This

circumstance, as well as being able to contribute to the tumoural metabolic reprogramming, can also be the cause of chemoresistance. Last year, our group's foremost contribution was the demonstration of the fact that the loss of expression of hCNT1 in a wide range of tumours has an epigenetic origin associated with the deregulation of the expression pattern of miRNAs in the tumours. The oncomiRs miR-106a and miR-17 negatively and directly regulate the nucleoside-derived drug transporter hCNT1. By silencing these miRNAs, we succeed in recovering the expression and part of the function of this drug transporter, and this contributes to the improvement of the cellular response to these anti-tumoural agents. We believe this is a new epigenetic mechanism that may condition the response to chemotherapy.

MEMBERS

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Sandra Perez-Torras

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Liska Caviedes Cárdenas, Jose Antonio Agüero Fernández, Laura García Vega, Aida Mata Ventosa, Nerea Urtasun Plans

PHD STUDENTS

Andrea Valentina Prieto Carruyo, Eloy Pena Rodríguez

SELECTED PROJECTS

- Desoxiribonucleòsids com a teràpia per als trastorns de la replicació de l'ADN mitocondrial: investigació dels mecanismes terapèutics i ampliació del tractament a les mutacions en POLG i altres gens (97/C/2020). Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 2021-2024. PI: Marçal Pastor Anglada.
- Descubrimiento y desarrollo de nuevos tratamientos Inmuno-Oncológicos (RTC2019-006865-1). Ministerio de Economía y Competitividad. Convocatòria Reptes-Col·laboració del Programa Estatal de Recerca, Desenvolupament i Innovació Orientada als Reptes de la Societat. 2020-2022. PI: Marçal Pastor Anglada.
- Doctorats industrials 2019. Empresa: Laboratorios Reig Jofré, S.A (2019DI005). Agència de Gestió

d'Ajuts Universitaris i de Recerca (AGAUR). Ajuts a universitats i a empreses per al desenvolupament d'un projecte de doctorat industrial, en qualsevol àmbit de coneixement i qualsevol sector empresarial. Doctorats industrials 2019. 2019-2022. PI: Sandra Pérez Torras

- Proteínas transportadoras de nucleósidos (NT), metabolismo de nucleótidos y cáncer (RTI2018-094655-B-I00). Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2019-2022. PI: Marçal Pastor Anglada

SELECTED PUBLICATIONS

- Boces C, Mata A, Martín M, Boix L, Gironella M, Pastor M et al. OncomiRs miR-106a and miR-17 negatively regulate the nucleoside-derived drug transporter hCNT1. *Cell Mol Life Sci.* 2021;78(23):7505-7518. IF:9,261. (Q1).
- Pena E, Lajarin M, Mata A, Perez S, Fernández F. Dexamethasone-Loaded Lipomers: Development, Characterization, and Skin Biodistribution Studies. *Pharmaceutics.* 2021;13(4):533. IF:6,321. (Q1).
- Pena E, Mata A, Laura V, Pérez S, Fernández F. The Physicochemical, Biopharmaceutical, and In Vitro Efficacy Properties of Freeze-Dried Dexamethasone-Loaded Lipomers. *Pharmaceutics.* 2021;13(8):1322. IF:6,321. (Q1).

Paediatric cancer epigenetics

Alexandra Avgustinva, PhD

Our research group was founded in late 2020 with the overall goal to identify specific epigenomic vulnerabilities of developmental tumours, and to exploit them in the pursuit of personalised therapies

to improve patient welfare and prognosis. We believe that one size does not fit all, and we need to treat each individual, not the disease. Currently we are focussing on malignant rhabdoid tumours (MRTs), highly malignant and heterogeneous paediatric cancers characterized by biallelic inactivation of the SWI/SNF chromatin remodelling complex subunit SMARCB1. During 2021 we established a plethora of tools necessary for our studies, including mouse models, cell lines and optimised genome editing protocols. We have been granted the "Programa Estatal de I+D+i Orientada a los Retos de la Sociedad" grant entitled "The rogue epigenome of malignant rhabdoid tumours – define to treat" (MINECO) to support and develop this project. Additionally, we have incorporated new members into the group and expanded our national and international collaborator network: together we will bring about change sooner.

MEMBERS

POSTDOC ASSOCIATED RESEARCHERS

Carlos Javier Rodriguez Hernandez

PHD STUDENTS

Àlex Cebrià Xart

SELECTED PROJECTS

- El epigenoma malvado de los tumores rhabdoides - definir para tratar. (PID2020-118241RA-I00). Ministerio de Economía Y Competitividad (MINECO). 2021-2024. IP: Alexandra Avgustinova.
- Ayudas para contratos Ramón y Cajal (RYC) 2019. Beneficiario: Alexandra Avgustinova. (RYC2019-027738-I). Ministerio de Economía Y Competitividad (MINECO). 2020-2025. IP: Alexandra Avgustinova.



7.1 MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.4 Precision medicine in rare diseases

- Metabolic and mitochondrial medicine
- Neurogenetics and molecular medicine
- Human molecular genetics I
- Human molecular genetics II
- Genomics for the diagnosis of rare diseases
- Diagnostic and therapeutic imaging



Metabolic and mitochondrial medicine

Rafael Artuch Iriberry, MD, PhD

Our research group participated in 32 scientific publications in 2021 and here we will mention those led by members of our team and those which are of special scientific significance. In the field of mitochondrial disorders, we should point out our participation in the first epidemiological study of this type of diseases at national level (Bellusci et al., Genes 2021), and the development of a new biomarker for the study of mitochondrial disorders (Paredes-Fuentes et al., Clin Chem 2021). We also renewed our research project on these pathologies, PI20/00340, for the Carlos III Health Institute (ISCIII). In the sphere of technological development, we culminated the generation of a new biosensor for the measurement of ammonium, which is currently under testing at our hospital, through a health technologies development project. Additionally, we consolidated our metabolomic platform for research on cellular and animal models, which has given rise to four collaborative high-impact publications with basic researchers. With respect to congenital disorders of glycosylation, we continued to expand our knowledge of the phenotypic complex of PMM2-CDG (PMID: 34140212; PMID: 34442375) and, in particular, we developed an international project on stroke-like episodes (PMID: 34708008), which led to a project funded by ISCIII 2022-2024 (AES 2021). The project called "Sigma. MicroRNA signature in cerebellar development: exploring early biomarkers of cerebellar disease" has allowed us to find a circulating microRNA signature which is characteristic of cerebellar atrophy associated with PMM2-CDG and which may be a diagnostic and prognostic biomarker in an accessible sample such as plasma. Lastly, we developed an international consensus on follow-up of the PGM1-CDG defect (PMID: 32681750). Likewise, our collaboration with groups from Pompeu Fabra University (UPF) (Dr

Fernández Fernández and Dr Gallego) allowed the development of a yeast and human cell model of RFT1-CDG deficiency.

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TECHNICIAN

Angela Yasmina Arias Dimas, Adriana Balen Valeiro, Yahel Calzadilla Borrás, Aroa Fernández de Miguel, Juan Moreno García, Tania Pardal Chofle

SELECTED PROJECTS

- Secuenciación de exoma para el diagnóstico de pacientes con enfermedades mitocondriales: investigación de aspectos fisiopatológicos y terapéuticos de las deficiencias de conezima Q10. (PI20/00340). Instituto de Salud Carlos III (ISCIII). 2021-2023. PI: Rafael Artuch Iriberry.
- PHENYLKETONURIA: from Childhood to Adults through Brain Functional Connectomics, Cardiovascular Changes, Metabolomic and Microbiota Characteristics. Fundació La Marató de TV3. PI: Aida Ormazabal Herrero.
- Programa MIMeS_Detección, prevención y tratamiento precoz de Salud Mental en pacientes con Malalties Minoritàries Cognitiu-Conductuals de base Genètica. (Programa MIMeS). OSSJD.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio abierto, multicéntrico para evaluar la seguridad, tolerabilidad, farmacocinética y farmacodinamia

de R07248824 en participantes con Síndrome de Angelman.

An open-label, multicenter study to investigate the safety, tolerability, pharmacokinetics and pharmacodynamics of R07248824 in participants with angelman syndrome. (BP41674). Roche Diagnostics International Ltd; Roche Farma, S.A. 2020- PI: Mercedes Serrano Gimaré.

- Estudio de fase IV, abierto y de una sola cohorte, de los resultados neurocognitivos (NC) a largo plazo en niños de 4 a 5 años con fenilcetonuria tratados con dihidrocloruro de sapropterina (Kuvan®) durante 7 años. (EMR 700773-002). Merck KGaA; Parexel International S.L. 2014- PI: Jaume Campistol Plana.
- RRHH03 - Ciberer. PI: Rafael Artuch Iriberry.
- Pruebas clínicas y básicas sobre la carencia de fosfomanomutasa (PMM2-CDG). (GLY-000). And Glycomine, Inc; Voisin Consulting. 2020- PI: Mercedes Serrano Gimaré.

SELECTED PUBLICATIONS

- Kour S, Rajan DS, Fortuna TR, Anderson EN, Ward C, Lee Y et al. Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nat Commun. 2021;12(1):2558-2558. IF:14,919. (Q1).
- Peña M, Roldán G, Perez J, Ortuño FM, Carmona R, Aquino V et al. CSVS, a crowdsourcing database of the Spanish population genetic variability. NUCLEIC ACIDS RES. 2021;49(D1): D1130-37. IF:16,971. (Q1).
- Picó S, Parras A, Santos M, Pose J, Castro M, Fraga E et al. CPEB alteration and aberrant transcriptome-polyadenylation lead to a treatable SLC19A3 deficiency in Huntington's disease. SCI TRANSL MED. 2021;13(613) IF:17,956. (Q1).
- Tristan A, Borràs E, Molero M, Wassenberg T, Peters T, Verbeek MM et al. Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Mov Disord. 2021;36(3):690-703. IF:10,338. (Q1).

Neurogenetics and molecular medicine

Janet Hoenicka, PhD
& Francesc Palau Martínez, MD, PhD

The Neurogenetics and Molecular Medicine (NeuroGene) Group does research on the genetic, pathophysiological and therapeutic aspects of disorders affecting the central and peripheral nervous systems, with special emphasis on rare diseases, genetic disorders and development disorders. The milestones reached in 2021 in the areas of basic and translational biomedicine and clinical research included the following:

- Characterisation of the interaction, in membrane contact sites, between mitochondria and lysosomes, and their role in the axonopathy of Charcot-Marie-Tooth disease.
- The relation between the dopamine gene ANKK1, related to addictions, and the susceptibility to Parkinson's disease.
- Discovery of new genes related to complex forms of autism spectrum (PLXNA and LRRC40) and neurodevelopmental syndrome disorders (ZBTB7A, MIM #619769).
- Inclusion of the Translational Diagnosis of Functional Validation of Gene Variants Programme in the Paediatric Institute of Rare Diseases (IPER) and the Genetic Medicine Service of Sant Joan de Déu Barcelona Children's Hospital (HSJD), and publication of the preliminary results.

The group's national and international collaboration is intense: (i) member of the Rare Diseases Networked Biomedical Research Centre (CIBERER); (ii) collaboration with the Genomic Sciences and Precision Medicine Center (GSPMC), Medical College of Wisconsin; (iii) member of the International Parkinson's Disease Genomics Consortium (IPDGC) and the only Spanish member of GP2 (Global Parkinson's Genetics Program); (iv) member of the Academy of Sciences of Latin America (ACAL); and (v) editor-in-chief, *Orphanet Journal of Rare Diseases*.

In the sphere of training activity, four final master's degree projects and



three final bachelor's degree projects were carried out by students of the University of Barcelona (UB), the Autonomous University of Barcelona (UAB) and Pompeu Fabra University (UPF).

MEMBERS

STAFF RESEARCHER

Joan Maynou Fernández, Mónica Roldan Molina, Guerau Fernandez Isern

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Mar Borregán Prats, Esther Cuatrecasas Capdevila, Cristina

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PHD STUDENTS

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TECHNICIAN

Daniel Castillo Mestres, Jordi Genovès Escarré

SELECTED PROJECTS

- Mitochondrial membrane dynamics and its interorganelle contact sites in the cellular and nerve pathophysiology of neurogenetic disease – NeuroContacts. (PID2020-114655RB-100). Agencia Estatal de Investigación – MICINN, 2021-2024. PI: Francesc Palau Martínez.
- Genes y fenotipos de síndromes parkinsonianos: estudio clínico y experimental. (PI19/00126). Instituto de Salud Carlos III (ISCIII). 2020-2022. PI: Janet Hoenicka.
- Infraestructura de Medicina de Precisión asociada a la Ciencia y Tecnología (IMPACT). Ciencia de datos. Instituto de Salud Carlos III (ISCIII). 2021-2023. PI: Joan Maynou Fernández.
- El cerebro clínico del síndrome Coffin-Siris: estudio integral de un trastorno del neurodesarrollo. Hospital Sant Joan de Déu - Esplugues HSJD. 2020-2023. PI: Francesc Palau Martínez.
- Proyecto de investigación traslacional sobre los aspectos fisiopatológicos y clínicos de la enfermedad de Menkes. (Amigos de Nono). Fundación amigos de nono contra la enfermedad de Menkes. 2017-2022. PI: Francesc Palau Martínez y Janet Hoenicka.
- Proyecto piloto sobre la implementación de un cribado neonatal genético en España. Fundación Ramón Areces. 2019-2022. PI: Francesc Palau Martínez.
- Application of Advanced multimodal microscopy and imaging to foster therapy development in neuromuscular diseases. (Turróns Vicens). Turróns Vicens SL. 2020-2023. PI: Mónica Roldan Molina.

SELECTED PUBLICATIONS

- Cantarero L, Juárez-Escoto E, Civera-Tregón A, Rodríguez-Sanz M, Roldan M, Benítez R et al. Mitochondria-lysosome membrane contacts are defective in GDAP1-related Charcot-Marie-Tooth disease. *Hum Mol Genet.* 2021; 29: 3589-3605. IF: 6,150. (Q1).
- Natera-De Benito D, Sola A, Sousa PR, Boronat S, Expósito J, Carrera-García L et al. Copper Toxicity Associated With an ATP7A-Related Complex Phenotype. *Pediatr Neurol.* 2021; 119: 40-44. IF: 3,372. (Q1).
- Pérez-Santamarina E, García-Ruiz

- P, Martínez-Rubio D, Ezquerro M, Pla-Navarro I, Puente J et al. Regulatory rare variants of the dopaminergic gene ANKK1 as potential risk factors for Parkinson's disease. *Sci Rep.* 2021; 11 (1): 9879-9879. IF: 4,379. (Q1).
- Pijuan J, Rodríguez-Sanz M, Natera-De Benito D, Ortez CI, Altimir A, Osuna M et al. Translational Diagnostics: an in-house pipeline to validate genetic variants in children with undiagnosed and rare diseases. *J Mol Diagn.* 2021; 23 (1):71-90. IF: 5,568. (Q1).

Human molecular genetics I

Daniel Raúl Grinberg Vaisman, PhD

In 2021 we described new genes and mutations responsible for intellectual disability in patients originally diagnosed as Opitez C or Bohring-Opitez (*PORCN*, *ZIC2*). We delved deeper into the immunohistochemical, transcriptomic and metabolomic analysis of fibroblasts of patients with Schaaf-Yang syndrome (caused by mutations that truncate the *MAGEL2* protein) or with TRAF7 syndrome, and we derived iPSCs from the former for the future derivation of brain neurons and organoids.

Likewise, we functionally analysed the effect of variants in the *SOST* gene in relation to osteoporosis, and mutations in the *CYP11A1* gene in patients with atypical femoral fracture. We also took part in multi-centre studies on monogenic disorders of high and low bone mass, and on functional validation in genetic diseases of the skeleton.

In a study of genetic factors involved in stroke recovery, we have identified a gene involved in the migration of neutrophils through the blood-brain barrier, in which the presence of missense variants would be protective. We have studied the genetic and epigenetic risk factors that lie behind various psychiatric and neurological disorders of complex nature. On the one hand, the genetic analysis of a murine model of cocaine dependence,

which allowed us to propose a new pharmacological target for relapse (the protein *PLCB1*), and on the other hand we have investigated the epigenetics of drug addiction, identifying genetic risk variants that alter the methylation of the genes *CTNBL1*, *SCP2* and *ECHDC2*. We also helped to define the genetic landscape of autism, ADHD, obsessive-compulsive disorder, disruptive behaviour disorders, migraine, Parkinson's disease, epilepsies, and aggressiveness.

In the field of lysosomal diseases, we have demonstrated that the inhibition of soluble epoxide hydrolase improves cognitive capacities in a murine model of Niemann-Pick disease, type C. Lastly, we made progress in the project of generating and studying neuronal models based on iPSCs cells derived from patients with Williams-Beuren syndrome or 7q11.23 microduplication syndrome.

MEMBERS

STAFF RESEARCHER

Susana Balcells Comas, Bru Cormand Rifà, Roser Corominas

POSTDOC ASSOCIATED RESEARCHERS

Raquel Rabionet Janssen, Noelia Fernández Castillo, Núria Martínez Gil

ASSISTANT RESEARCHER

Estefania Alcaide Consuegra

TECHNICIAN

Mónica Cozar Morillo

SELECTED PROJECTS

- GENIUS: GENetic Influences on functional outcome after Stroke. (Reg. 70/307 Proj. 201726). Fundació La Marató de TV3. 2018-2021. PI: Raquel Rabionet Janssen.
- Effects of Nutrition and Lifestyle on Impulsive, Compulsive, and Externalizing behaviors (Eat2beNICE) (728018). Unió Europea. HORIZON 2020 (2014-2020). P3. SC2: SUSTAINABLE FOOD SECURITY – RESILIENT AND RESOURCE-EFFICIENT VALUE CHAINS (H2020-SFS-2016-2017). 2017-2022. PI: Bru Cormand Rifa.
- Estudios genéticos y funcionales hacia aproximaciones terapéuticas para varias enfermedades óseas y síndromes de discapacidad intelectual (PID2019-107188RB-C21). Ministerio de Ciencia, Innovación y Universidades.

Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2020-2023. PI: Susanna Balcells Comas

- Modeling 7q11.23 aneusomy syndromes with iPSC-derived neurons (iNeuro7q) (RTI2018-101960-A-I00). Ministerio de Ciencia Innovación y Universidades. RETOS. 2019-2021. PI: Roser Corominas.

SELECTED PUBLICATIONS

- Cabana J, Martín E, Gallego A, Maldonado R, Fernandez N, Cormand B. Reduced cue-induced reinstatement of cocaine-seeking behavior in Plcb1 +/- mice. *Transl Psychiatry*. 2021;11(1):521. IF:6,222. (Q1).
- Castilla L, Gürsoy S, Giray Ö, Prat A, Bullich G, Matalonga L et al. De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family. *Int. J. Mol. Sci.* 2021;22(4):1549. IF:5,923. (Q1).
- Demontis D, Walters RK, Rajagopal VM, Waldman ID, Grove J, Als TD et al. Risk variants and polygenic architecture of disruptive behavior disorders in the context of attention-deficit/hyperactivity disorder. *Nat Commun*. 2021;12(1):576. IF:14,919. (Q1).
- Martinez N, Roca N, Cozar M, Garcia N, Ovejero D, Nogués X et al. Genetics and Genomics of SOST: Functional Analysis of Variants and Genomic Regulation in Osteoblasts. *Int. J. Mol. Sci.* 2021;22(2):489. IF:5,923. (Q1).

Human molecular genetics II

Gemma Marfany Nadal, PhD

Our group's two main lines of research comprise, on the one hand, the further development of the genetic testing of rare diseases of the retina, and on the other hand, the generation of animal models to study the function of the genes that cause such diseases, together with

the search for a possible precision therapy. We apply NGS techniques for genetic testing in retinal dystrophies within the spin-off generated as transfer of knowledge, achieving a success rate of up to 85%.

In order to study the molecular bases of hereditary blindness, we recently generated model mice by gene editing techniques (CRISPR-Cas9) on two genes causing retinitis pigmentosa, cone-rod dystrophy, and S-cone syndrome: CERKL and NR2E3. In the last few months, we have been conducting molecular studies in greater depth with a view to elucidating why the mutations in these genes cause hereditary blindness. For the gene NR2E3, we have identified isoforms generated by alternative splicing which had not been previously described; at present we are carrying out single-cell transcriptome analyses to characterise the photoreceptor populations of mutant strains with respect to the control group (collaboration with Dr Vaquerizas in Münster). As concerns CERKL, we have collaborated with other groups of the IBUB and the IRSJD (Dr Villarroya and Dr Artuch) to show that its depletion causes changes in the mitochondrial morphology, dynamics and metabolism in retinal neurons, photoreceptors, and ganglion cells (RGCs), as well as in retinal pigment epithelium cells (RPEs), opening new perspectives for a potential treatment by regulation of mitochondrial metabolism. A senior postdoc of the group has made stays in Naples (collaboration with Drs Ursini and Trapani) to explore potential gene therapy treatments by subretinal injection of adeno-associated viruses (AAVs). Lastly, we are investigating various genes which may contribute to the formation and stability of the sensory cilium, which means that they could be candidate causes of ciliopathies, of which retinal neurodegeneration is one of the symptoms.

In the future we plan to pursue in greater depth the new paths of research that have been opened during this year. 2021 has been a year of intense work, with the establishment of new national and international collaborations which, it is hoped, will soon be coming to fruition.

MEMBERS

POSTDOC ASSOCIATED RESEARCHERS

Serena Mirra

PHD STUDENTS

Izarbe Aisa Marin, Elena Borrego Domènech

SELECTED PROJECTS

- Explorando las vías de señalización neurosensoriales en enfermedades hereditarias de retina como posible aproximación a la terapia (PID2019-108578RB-I00). Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2020-2023. PI: Gemma Marfany Nadal, co-PI: Serena Mirra.
- Contractes de recerca/serveis/transferència (309710). DBGEN Ocular Genomics, S.L. 2018-2023. PI: Gemma Marfany Nadal.
- Enfermedades Raras (CB06/07/0041). Ministerio de Sanidad y Consumo. Convocatòria d'ajuts destinats a finançar estructures estables d'investigació cooperativa, a l'àrea de biomedicina i ciències de la salut, en el marc del Programa Ingenio 2010, Programa Consolider, accions CIBER. 2006 – 2022. PI: Gemma Marfany Nadal.

SELECTED PUBLICATIONS

- Aísa I, García R, Mirra S, Marfany G. The Alter Retina: Alternative Splicing of Retinal Genes in Health and Disease. *Int J Mol Sci*. 2021;22(4):1855. IF:5,923. (Q1).
- García-Arroyo R, Gavalda A, Villarroya F, Marfany G, Mirra S. Overexpression of CERKL Protects Retinal Pigment Epithelium Mitochondria from Oxidative Stress Effects. *Antioxidants (Basel)*. 2021;10(12):2018. IF:6,312. (Q1).
- Mirra S, Arroyo RG, Domènech EB, Gavalda A, Herrera C, Oliva C et al. CERKL, a retinal dystrophy gene, regulates mitochondrial function and dynamics in the mammalian retina. *Neurobiol Dis*. 2021;156:105405. IF:5,996. (Q1).
- Sánchez L, Toulis V, Marfany G. On the Wrong Track: Alterations of Ciliary Transport in Inherited Retinal Dystrophies. *Front. Cell. Dev. Biol*. 2021;9:623734. IF:6,684. (Q1).

Genomics for the diagnosis of rare diseases

Judith Armstrong Morón, PhD

The Genomics for Diagnosis of Rare Diseases group is a recently organised unit at the Sant Joan de Déu Barcelona Research Institute (IRSJD). It focuses on genomic tools and their potential for diagnosis, specifically within the framework of neurological disorders but also with respect to other rare diseases of genetic origin. NGS technology for the detection of mutations causing neurological pathologies began to be implemented at our hospital in 2013, and this experience has been extended to include the rest of the specialities treated at our institution. This technological knowledge and the knowledge of neurological disorders and other diseases led to creation of our research group. Our main goal is to transform clinical diagnosis into genetic-molecular diagnosis and to achieve our aim we use mass sequencing technology, employing not only genomic data but also including data of the transcriptome and proteome, taking a multi-omics approach. Mass sequencing generates a large quantity of data which are highly useful in the hospital sphere and this is why we also focus on and lead a research project conceived to allow us to strengthen inter-hospital networks that can enhance the interpretation of genomic data to improve the diagnostic process.

MEMBERS

STAFF RESEARCHER

Núria Brandí Tarrrau, Délia Yubero Siles

PHD STUDENTS

Ainhoa Pascual Alonso, Clara Xiol Viñas

TECHNICIAN

Carlota Ros Bonaventura, Paola Pacheco Fernández

SELECTED PROJECTS

- Catalan Interhospital Network of Genetic Variants to Improve Genetic Diagnosis in Rare Diseases.

(590/C/2020). Fundació La Marató de TV3. 2021-2024. PI: Délia Yubero Siles.

- Estudio de pacientes dentro del Espectro Rett mediante un enfoque multiómico integrativo: delucidar la huella molecular de la vía funcional. (PI20/00389). Instituto de Salud Carlos III (ISCIII). 2021-2023. PI: Judith Silvia Armstrong Morón.
- Convocatòria d'ajuts per a la contractació de personal investigador novell (FI-2020). Beneficiari: Ainhoa Pascual (co-financiado por el FSE). [2020 FI_B_00888]. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2020-2023. PI: Judith Silvia Armstrong Morón.
- FPU_Clara Xiol. (FPU18/02152). Ministerio Educación, Cultura y Deporte (MECD). 2019-2023. PI: Judith Silvia Armstrong Morón.

SELECTED PUBLICATIONS

- Barbosa S, Vázquez ME, Gonzalez E, Hermida Á, Valverde LL, Armstrong J et al. Characterization of a Novel Splicing Variant in Acylglycerol Kinase (AGK) Associated with Fatal Sengers Syndrome. *Int. J. Mol. Sci.* 2021;22(24):13484. IF:5,923. (Q1).
- Pascual A, Martínez T, Xiol C, Armstrong J. MECP2-Related Disorders in Males. *Int. J. Mol. Sci.* 2021;22(17):9610. IF:5,923. (Q1).
- Xiol C, Marina T, Pascual A, De Oyarzabal AL, Armstrong J. Technological Improvements in the Genetic Diagnosis of Rett Syndrome Spectrum Disorders. *Int. J. Mol. Sci.* 2021;22(19):10375. IF:5,923. (Q1).

Diagnostic and therapeutic imaging

Josep Munuera del Cerro, MD, PhD

MEMBERS

STAFF RESEARCHER

Marta Gómez Chiari, Emilio Inarejos Clemente, Mónica Rebollo Polo

SELECTED PROJECTS

- Sistema de ayuda a la decisión clínica para la evaluación y predicción de enfermedad vascular cerebral pediátrica: Radiómica a partir de RM vascular e Inteligencia Artificial (Vasculómica). (PI20/00296). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Jose Luis Munuera del Cerro.
- Plataforma ISCIII de apoyo a la I+D+I en Biomedicina y Ciencias de la Salud. Biomodelos-Impresión 3D. (PT20/00090). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Jose Luis Munuera del Cerro.
- Node d'imatge computacional pediàtrica (PEDIATRIC_ICON). (PR15-019756). Departament de Salut - Generalitat de Catalunya. 2020IP: Jose Luis Munuera del Cerro.

SELECTED PUBLICATIONS

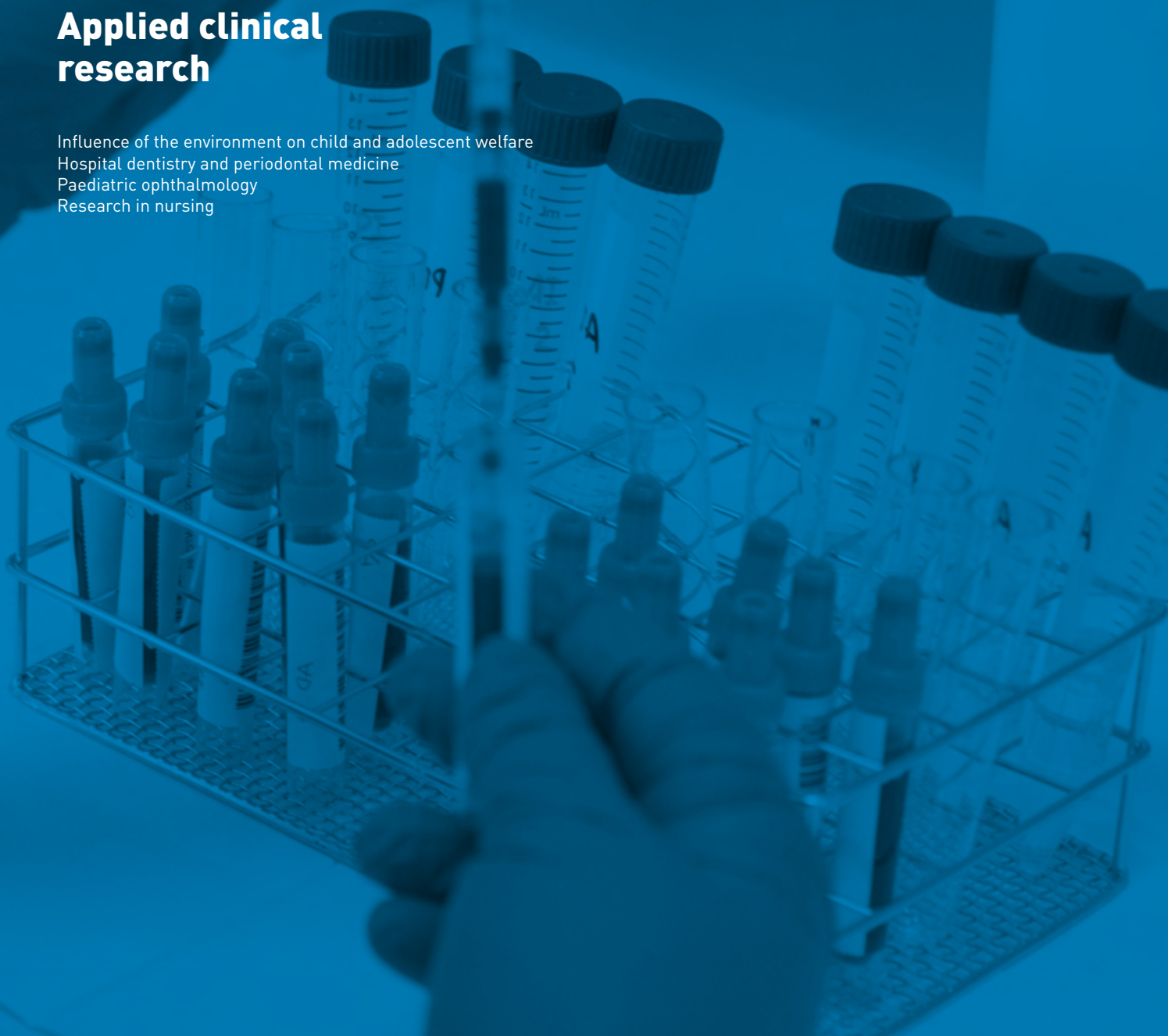
- Gonzalez E, Grau L, Ferrero SI, Hernandez A, Rebollo M, Marta C et al. Pessary for Management of Cervical Varices Complicating Pregnancy. *Obstet Gynecol.* 2021;138(3):482-486. IF:7,661. (Q1).
- Inarejos E, Navarro OM, Navallas M, Enrique G, Colombo C, Suñol M et al. Omphalomesenteric Duct Anomalies in Children: A Multimodality Overview. *Radiographics.* 2021;41(7):2090-2110. IF:5,333. (Q1).
- Panwar J, Tolend M, Redd B, Srinivasalu H, Colbert RA, Akikusa J et al. Consensus-driven conceptual development of a standardized whole body-MRI scoring system for assessment of disease activity in juvenile idiopathic arthritis: MRI in JIA OMERACT working group. *Semin Arthritis Rheum.* 2021;51(6):1350-1359. IF:5,532. (Q1).
- Serrano E, Zarco F, Gill AE, Hawkins CM, Macías N, Inarejos E et al. Percutaneous cryoablation of chondroblastoma and osteoblastoma in pediatric patients. *Insights Imaging.* 2021;12(1):106-106. IF:5,231. (Q1).



7.1 MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.5 Applied clinical research

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



Influence of the environment on child and adolescent welfare

Carles Luaces Cubells, MD, PhD

In 2021, our group also took part in two non-competitive research projects funded by private entities:

- Quick determination of heart fatty-acid binding protein (H-FABP) to rule out brain injuries in mild cranioencephalic trauma: a preliminary multi-centre study using a point-of-care instrument in traumatology and paediatrics emergency services. BIOTRABIS (BIOmarkers of TRAumatic Brain Injury- Spain). Promoter: Dr Joan Montaner Villalonga –Seville Biomedical Research Institute. Funding: Mutua Madrileña Foundation. 2020-22. PI: Carles Luaces / V Trenchs.
- Meningitis screening based on a novel, non-invasive, transfontanelar ultrasound device using machine learning techniques: a proof-of-concept study. Funding: New Born Solutions has provided funding. The participation of the Rabat HER in the study is co-financed by the Spanish International Development Cooperation Agency (2019/ACDE/001196). PI: Carles Luaces / D Muñoz-Santanach

It should also be pointed out that a significant part of the group's research has been focused on the impact of the SARS-CoV 2 pandemic on the paediatric population, placing special emphasis on its effects on psychological health. Moreover, along the same line, we collaborated actively on the promotion of the good handling of children and on the prevention of mistreatment during the lockdown period, participating in the publication of two decalogues endorsed by the Spanish Paediatric Emergencies Society ("Consejos para familias. Confinamiento saludable" ["Advice for Families. A Healthy Lockdown"], available at: https://seup.org/pdf_public/gt/Cons_fam.pdf, and "Decálogo de maltrato infantil para profesionales" ["A Decalogue on Child Mistreatment for Healthcare Professionals"]]).

MEMBERS

STAFF RESEARCHER

Gemma Claret Teruel, Ana Isabel Curcoy Barcenilla, Lidia Martínez Sánchez, Victoria Trenchs Sainz de la Maza

POSTDOC ASSOCIATED RESEARCHERS

Andrea Aldemira Liz, Vanessa Arias Constantí, Eva Gargallo Burriel, Cristina Parra Cotanda

PHD STUDENTS

David Muñoz Santanach, Sergi Navarro Vilarrubi, Marta Simó Nebot

SELECTED PROJECTS

- STEPS_Step by step standing up a Barnahus in Catalonia: A simulation-based program to avoid secondary victimization in the assessment and treatment of child sexual abuse. European Commission. 2020-2022. IP: Marta Simó Nebot.
- Estudio prospectivo observacional sobre la hidratación y nutrición en pacientes pediátricos con enfermedad avanzada. Sociedad Española de cuidados paliativos pediátricos (PEDPAL). 2019-2021. IP: Sergi Navarro Vilarrubi.
- La experiencia de los profesionales como segundas víctimas: estudio descriptivo. Sociedad Española de Urgencias de Pediatría. 2021. IP: Dra. Vanessa Arias Constantí.

SELECTED PUBLICATIONS

- Goldman RD, McGregor S, Marneni SR, Katsuta T, Griffiths MA, Hall JE et al. Willingness to Vaccinate Children against Influenza after the Coronavirus Disease 2019 Pandemic. *J Pediatr.* 2021;228:87-93. IF:4,406. (Q1).
- Goldman RD, Staubli G, Parra C, Brown JC, Hoeffe J, Seiler M et al. Factors associated with parents' willingness to enroll their children in trials for COVID-19 vaccination. *Hum Vaccin Immunother.* 2021;17(6):1607-1611. IF:3,452. (Q2).
- Ibrahim D, Miró Ó, Galicia M, Supervía A, Puiguiriguer J, Ortega J et al. Spanish Research Network on Drugs in Hospital Emergency Departments - the REDUrHE registry: general analysis and comparisons between weekend and weekday poisonings. *Emergencias.* 2021;33(5):335-344. IF:3,881. (Q1).
- Supervía A, Salgado E, Córdoba F,

García L, Martínez L, Moreno A et al. Poisoning cases in Catalonia: characteristics according to age groups in the Intox.28 study. *Emergencias.* 2021;33(2):115-120. IF:3,881. (Q1).

Hospital dentistry and periodontal medicine

Alejandro Rivera Baró, MD, PhD

We have kept up our efforts along our group's lines of work, starting up three new studies, one in each of our research lines: odontology (autotransplantation of teeth), periodontal medicine (gingival enlargement) and malformation medicine (presurgical orthopaedics). These studies were implemented after obtaining the Scientific Committee's endorsement and they are preparatory procedures for the doctoral theses of the first authors.

MEMBERS

STAFF RESEARCHER

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

SELECTED PROJECTS

- SGR 2017_Hospital Dentistry & Periodontal Medicine Research Group. (2017 SGR 00453). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2017-2021. IP: Lluís Brunet Llobet.

SELECTED PUBLICATIONS

- Moncunill J, Brunet LL, Cuadras D, Lorente N, Pascal R, Rovira C et al. Do the clinical criteria used to diagnose periodontitis affect the association with prematurity? *Odontology.* 2021;109(2):455-463. IF:2,634. (Q2).

Paediatric ophthalmology

Joan Prat Bartomeu, MD, PhD

MEMBERS

STAFF RESEARCHER

Elisa Carreras Bertran, Ester Casas Gimeno, Jaume Català Mora, Jesús Díaz Cascajosa, Ana Isabel Díaz Cortés, Carlos Fresno Cañada, Carmen García de Vicuña Muñoz, Enrique Jiménez Gañan, Ana Llorca Cardeñosa, Marta Morales Ballús, Núria Planas Domènech, M^a Josep Planas Tapias, Enric Puigventós, Gemma Romeu Cerrillo, Alicia Serra Castanera, Dowon, Simone Suh, Begonya Yeste Mayoral

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Ensayo clínico fase I, unicéntrico, abierto y de escalado de dosis, para evaluar la seguridad y la actividad del adenovirus oncolítico VCN-01 en pacientes con retinoblastoma refractario. (FSJD-RTB-2015). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; VCN Biosciences S.L. 2017-. IP: Jaume Català Mora.
- Estudio postautorización, multicéntrico, multinacional, longitudinal y observacional para el registro de la seguridad en pacientes tratados con voretigén neparovec. (CLTW888A12401/NOV-LUX-2019-01). Novartis Farmacéutica, S.A. 2020-. IP: Jaume Català Mora.

SELECTED PUBLICATIONS

- Fabian ID, Stacey AW, Foster A, Kivelä TT, Munier FL, Keren N et al. Travel burden and clinical presentation of retinoblastoma: analysis of 1024 patients from 43 African countries and 518 patients from 40 European countries. *Br J Ophthalmol.* 2021;105(10):1435-1443. IF:4,638. (Q1).
- Liu J, Ottaviani D, Sefta M, Desbrosses C, Chapeaublanc E, Aschero R et al. A high-risk retinoblastoma subtype with stemness features, dedifferentiated cone states and neuronal/ganglion

cell gene expression. *Nat Commun.* 2021;12(1):5578-5578. IF:14,919. (Q1).

- Stacey AW, Bowman R, Foster A, Kivelä TT, Munier FL, Cassoux N et al. Incidence of Retinoblastoma Has Increased: Results from 40 European Countries. *Ophthalmology.* 2021;128(9):1369-1371. IF:12,079. (Q1).
- Tomar AS, Finger PT, Gallie B, Kivelä TT, Mallipatna A, Zhang C et al. Global Retinoblastoma Treatment Outcomes: Association with National Income Level. *Ophthalmology.* 2021;128(5):740-753. IF:12,079. (Q1).

Research in nursing

Laura Lahuerta Valls, RN, PhD

Methodological support for nurse-led research projects in 2021.

2021 was a year of transition towards normalisation of the situation of coexistence with the pandemic. For us nurses who engage in clinical research and who form part of the associated clinical groups programme, this transition situation has allowed us to conduct translational research primarily focused on the areas of general paediatrics and paediatric oncology.

Cross-cutting projects:

- 13 communications in international congresses and conferences.
- 22 oral communications in national congresses and conferences.
- 20 posters in national congresses and conferences.
- 7 posters in international congresses and conferences.
- 5 publications in scientific journals.
- 2 awards.
- 2 competitive grants.

Initiatives promoted from the research area:

- #ViernesCientíficosSJD (ScientificFridaysSJD): In 2021, eight sessions were held with project-leader nurses to promote evidence-based knowledge and practice at institutional level. Coordination: Jordi Mitjà.

- Continuity of the process begun with the graduating class of resident intern nurses of 2019-2021, in the new approach to the final residency project led by the Research Nurse:
 - The nurse residents of the graduating class of 2020-2022 are introduced to research skills in the research lines established by the Nursing Management.
 - They submit to the Research Nurse the various parts of their research projects in order to receive assessment and orientation, for the purpose of achieving the required research skills.
 - They are introduced to the procedure for applying for the assessment and approval of the Research Committee of the Sant Joan de Déu Barcelona Research Institute (IRSJD), and for obtaining the assessment of the Ethics Committee for Research with Medicines (CEIm).
- Methodological support for projects: Effectiveness of a nursing staff training intervention for the handling of paediatric onco-haematology patients with ASD. PI: Claudia Nogales Gonzalez (in progress).

MEMBERS

STAFF RESEARCHER

Maribel Ferrer Orona, Concepción Agustín Mata, Silvia Ciprés Roig, Encarna Gómez Gamboa, Jordi Mitjà, Gemma Pérez Acevedo

POSTDOC ASSOCIATED RESEARCHERS

M^a Dolors Navarro Rubio

PHD STUDENTS

Marta Fajardo Samper

ASSISTANT RESEARCHER

Pau Miquel Diego, Ikram Allaoui, Montserrat Gutierrez Juarez, Ares Sentenach Carbó

COLLABORATOR RESEARCHER

Carmen Jerez Molina

SELECTED PROJECTS

- Seroprevalencia de la infección por SARS-COV-2 en los empleados del hospital materno-infantil Sant Joan de Déu. (PR-440/2020). Fundació Infermeria i Societat. 2021-2021. IP: Laura Lahuerta Valls.
- Anàlisi de l'espiritualitat, religiositat o filosofia de vida dels pares i mares d'infants amb

necessitats pal·liatives a Catalunya. (2018RELIG00008). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2019-2021. IP: M^a Dolors Navarro Rubio.

- Eficacia y seguridad de los ácidos grasos hiperoxigenados en la microcirculación de la púrpura fulminante en pacientes pediátricos con sepsis. (PR-242/17). Fundació Infermeria i Societat. 2018-2021. IP: Gemma Pérez Acevedo.
- Prevención de complicaciones de herida quirúrgica en paciente pediátrico: efectividad de la terapia de presión negativa incisional en cirugía de escoliosis no idiopática (Estudio TePreInPed). Beca Peris 2021-2025 (SLT017/20/000194). IP: Gemma Pérez Acevedo.
- Les narratives clíniques de la mort: una oportunitat per al desenvolupament ètic dels professionals? AJUT RELIG21 (Beca Grifols). IP: Pau Miquel i Diego.
- Anàlisi de l'impacte de les

cures infermeres en la qualitat assistencial i seguretat dels pacients, segons el model de Mitchell. Beca Enfermeria i Societat. IP: Immaculada Rodríguez Salamanca.

SELECTED PUBLICATIONS

- Moreno C, Roy C, Monforte C, González L, Navarro MD, Gallart A. Effectiveness of a nurse-led, face-to-face health coaching intervention in enhancing activation and secondary outcomes of primary care users with chronic conditions. *Res Nurs Health*. 2021;44(3):458-472. IF:2,228. (Q2).
- Perez A, Martinez A, Negre A, Lahuerta L. Pediatric Patient Transfer from a Tph Ward to Icu: How can we Improve our Communication?. *Bone Marrow Trasplantation*. 2021;55(s1): 752-753. IF: 5,483 (Q1).

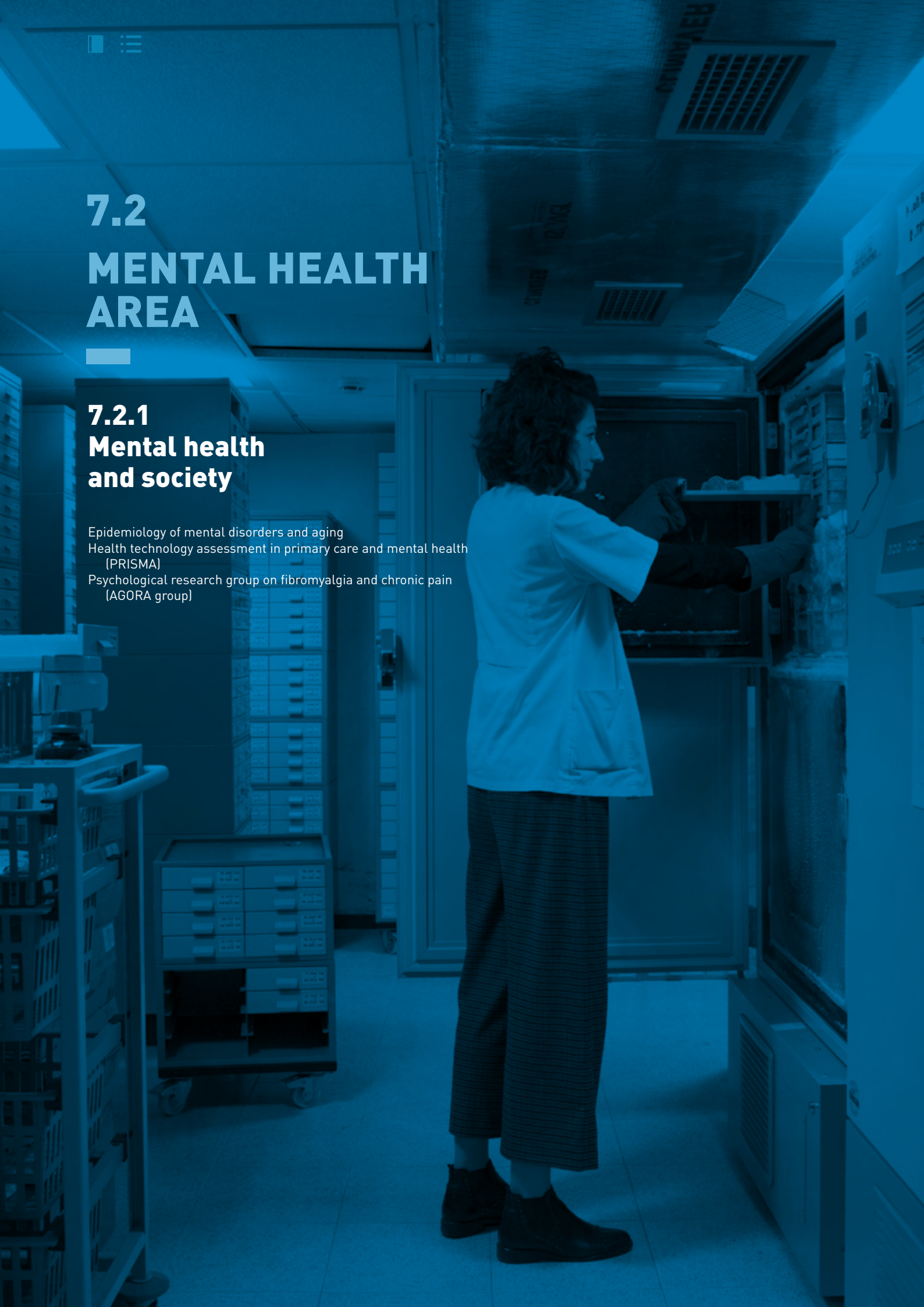


7.2

MENTAL HEALTH AREA

7.2.1 Mental health and society

Epidemiology of mental disorders and aging
Health technology assessment in primary care and mental health
(PRISMA)
Psychological research group on fibromyalgia and chronic pain
(AGORA group)



Epidemiology of mental disorders and aging

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

In 2021, the Epidemiology of Mental Health Disorders and Ageing group worked on various national and international projects. It coordinates two projects funded by the European Commission's Horizon 2021 programme: the EMPOWER project, which has the purpose of developing and testing a digital platform for the promotion and improvement of mental health at the workplace; and the SYNCHROS project, whose purpose is to establish recommendations for the standardisation of the cohort data harmonisation processes, and which will be completed around mid-2022. Dr Haro leads one of the 36 scientific research projects of excellence that have received funds raised by the 2020 telethon programme *La Marató* de TV3. The selected project is called REPICAL and its purpose is to achieve an understanding of the needs and working conditions of the staff in nursing homes with the aim of favouring the well-being of the elderly or disabled people who live there. Notable among the principal results obtained in 2021 are the main findings of the "Age with Health" study, which show that social support has an effect on feelings of loneliness, bringing about a significant reduction of the probabilities of relapse or prolongation of major depression episodes. In the MINDCOVID study, research has been conducted on feelings of disaffection towards social relations as one of the main risk factors of depression, anxiety and suicidal ideation during the pandemic. Consequently, the fact that young adults require more frequent and varied social relations than elderly persons has contributed to the circumstance that the former show a greater prevalence of mental health problems.

MEMBERS

STAFF RESEARCHER

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Carlota de Miquel Bleier, Anna Monistrol Mula, Aina Gabarrell Pascuet, Ivet Bayes Marín

ASSISTANT RESEARCHER

Iris Mauricio Arribas

SELECTED PROJECTS

- EDHEN: European Health Data and Evidence Network. European Commission. 2021-2022. PI: Josep Maria Haro Abad.
- Xarxa d'Innovació de Noves Tecnologies en Salut Mental - TECSAM. (2018 XARDI 00010). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2020-2022. PI: Josep Maria Haro Abad.
- EMPOWER_European platform to Promote Wellbeing and Health in the workplace. (848180). European Commission; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2020-2023. PI: Beatriz Olaya Guzmán.
- SYNCHROS: SYnergies for Cohorts in Health: integrating the Role of all Stakeholders. (82884). European Commission. 2019-2022. PI: Josep Maria Haro Abad.

SELECTED PUBLICATIONS

- Felez M, Haro JM, Stubbs B, Smith L, Koyanagi A. Moving more, ageing happy: findings from six low- and middle-income countries. *Age Ageing*. 2021;50(2):488-497. IF:10,668. (Q1).
- Fernández D, Giné I, Liu I, Yucel R, Nai M, Morena M et al. Are environmental pollution and biodiversity levels associated to the spread and mortality of COVID-19? A four-month global analysis. *Environ Pollut*. 2021;271:116326-116326. IF:8,071. (Q1).
- Jacob L, Smith L, McDermott D, Haro JM, Stickley A, Koyanagi

A. Relationship between sexual orientation and psychotic experiences in the general population in England. *Psychol Med*. 2021;51(1):138-146. IF:7,723. (Q1).

- Smith L, Shin JI, Ghayda RA, Hijaz A, Sheyn D, Pope R et al. Physical multimorbidity and incident urinary incontinence among community-dwelling adults aged ≥50 years: findings from a prospective analysis of the Irish Longitudinal Study on Ageing. *Age Ageing*. 2021;50(6):2038-2046. IF:10,668. (Q1).

Health technology assessment in primary care and mental health (PRISMA)

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

In 2021, the PRISMA group carried out over 30 projects. Additionally, it began to prepare a database of unit costs at national level to inform health economy studies, and it started up two European projects.

One of these European projects is funded by a starting grant from the European Research Council (ERC). Its purpose is to carry out a cluster clinical trial of an intervention to reduce initial medication non-adherence. All the preparations required to carry out the intervention have been performed during the year. The other project with European funding is focused on the performance of a study of the implementation cost and of the cost-effectiveness of an intervention to reduce the cancer diagnosis time in three Latin American countries.

As concerns our principal results in 2021, we demonstrated the efficiency of an intervention to improve healthy lifestyles, which was carried out in 8 autonomous regions in Spain. This study involved a major process of harmonisation of data from different healthcare systems.

Likewise, we demonstrated in a pilot study that the intervention about shared decision-making on IMA (Initial Medication Adherence) is accepted by patients and primary care professionals. Before conducting the study, we explored the reasons for which professionals thought that patients decided not to begin medication. We also established that an estimated 9% of the treatments prescribed to the paediatric population in Catalonia are not begun. We observed that the prevalence of initial medication non-adherence differed according to the age of the boys or girls concerned. Specifically, younger children and adolescents showed higher figures of initial medication non-adherence, while children between 5 and 11 years of age showed the lowest initial medication non-adherence figures.

A study carried out using qualitative methodology assessed the Mental Health Primary Care Support Programme in Catalonia based on the experience of primary care physicians and mental health professionals. We also carried out the project called "Design of an action plan for improving the quality of services and mental health in the Baix Llobregat Region", which was commissioned by the Barcelona Provincial Council, using a participative action research methodology.

Lastly, in 2021 we worked with Banc Farmacèutic (the Pharmaceutics Bank, an NGO whose purpose is to cover the price of medication for citizens who cannot afford it), to identify the profile of the beneficiaries of this organisation, and to determine the pattern of use of the covered medications and healthcare products.

MEMBERS

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Paula Arroyo Uriarte, Ana María Besoáin Cornejo, Cristina Carbonell

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María del Carmen Gallardo González, Montserrat Iracheta Todó, Ana Fernández Sánchez, Ana María Jové Massó, María Antonia March Pujol, Juan Manuel Mendive Arbeloa, María del Carmen Olmo, María Teresa Peñarrubia María, Ramon Sabés Figuera, Luís Salvador Carulla, Carles Vilaplana Carnerero

SELECTED PROJECTS

- EquityCancer-LA_Improving equity in access to early diagnosis of cancer: implementation research in different healthcare systems of Latin America. (965226). European Commission. 2021-2025. PI: Maria Rubio Valera.
- Evaluación de la adherencia a antipsicóticos a partir de datos de la práctica clínica real. (PI19/00111). Instituto de Salud Carlos III (ISCIII). 2020-2022. PI: Antoni Serrano Blanco.
- El problema de la no iniciación pediátrica: Evaluación con métodos mixtos. (PI19/00108). Instituto de Salud Carlos III (ISCIII). 2020-2022. PI: Maria Rubio Valera.
- IMA-cRCT_Effectiveness and cost-effectiveness of the Initial Medication Adherence intervention: cluster randomized controlled trial and economic model. (948973). European Commission. 2021-2025. PI: Maria Rubio Valera.

SELECTED PUBLICATIONS

- Arias J, Vilagut G, Ronaldson A, Serrano A, Martín V, Peters M et al. Prevalence and variability of current depressive disorder in 27 European countries: a population-based study. *Lancet Public Health*. 2021;6(10) IF:21,648. (Q1).
- Aznar I, Zabaleta E, Casajuana M, Sánchez A, Parody E, Bolívar B et al. Cost-effectiveness analysis of a multiple health behaviour change intervention in people aged between 45 and 75 years: a cluster randomized controlled trial in primary care (EIRA study). *Int J Behav Nutr Phys Act*. 2021;18(1):88-88. IF:6,457. (Q1).

- Iglesias M, Gil M, Peñarrubia MT, Blanco E, Fernández R, Serrano A et al. Barriers and opportunities for the treatment of mild-to-moderate depression with a watchful waiting approach. *Patient Educ Couns*. 2021;104(3):611-619. IF:2,940. (Q1).
- Rubio M, Marqués S, Peñarrubia MT, Urbanos RM, Borrell C, Bosch J et al. Who Suffers From Pharmaceutical Poverty and What Are Their Needs? Evidence From a Spanish Region. *FRONTPHARMACOL*. 2021;12:617687-617687. IF:5,810. (Q1).

Psychological research group on fibromyalgia and chronic pain (AGORA group)

Juan Vicente Luciano Devis, PhD

In 2021, the most important development for our team was the establishment of a double affiliation agreement of our PI to the Autonomous University of Barcelona (UAB) and the Sant Joan de Déu Barcelona Research Foundation (FSJD) thanks to the understanding reached between these two institutions. Since September 2021, Dr Luciano has been a teacher in the Clinical Psychology and Health Department of the UAB.

Over the course of the year, our group has mainly focused its efforts on two clinical trials: one on psychological therapies for patients with chronic low-back pain and depression (IMPACT Project), and the other on a new pharmacological therapy for fibromyalgia (INNOVA Project).

MEMBERS

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PHD STUDENTS

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ASSISTANT RESEARCHER

Jaime Navarrete Hidalgo

COLLABORATOR RESEARCHER

Albert Feliu Soler, Xavier Borràs Hernández

SELECTED PROJECTS

- Convocatòria d'ajuts per a la contractació de personal investigador novell (FI-2021). Beneficiari: Ariadna Colomer Carbonell. [2021_FI_B_00216]. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2021-2024. PI: Juan Vicente Luciano Devis.
- Contratos predoctorales de formación en investigación en salud (PFIS). Beneficiari: Juan Pablo Sanabria. (FI20/00034). Instituto de Salud Carlos III (ISCIII). 2021-2024. PI: Juan Vicente Luciano Devis.
- Effectiveness, cost-utility and neurobiological effects of low-dose naltrexone in patients with fibromyalgia (INNOVA project). (IC120/00080). Instituto de Salud Carlos III (ISCIII). 2021-2024. PI: Juan Vicente Luciano Devis.
- Coste-utilidad y efectos fisiológicos de la Terapia de Aceptación y Compromiso y la Activación Conductual en pacientes con dolor crónico y depresión mayor

comórbida (Proyecto IMPACT). (PI19/00112). Instituto de Salud Carlos III (ISCIII). 2020-2022. PI: Juan Vicente Luciano Devis.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Pacientes con espondiloartritis axial: Registro multinacional de características clínicas, incluida la progresión radiológica, y carga de la enfermedad durante 5 años en contextos de la vida real. Estudio PROOF. (10297). Abbvie Spain, S.L.U. 2014PI:Antoni Rozadilla Sacanell.

SELECTED PUBLICATIONS

- Pérez Aranda A, García J, Gude F, Luciano JV, Feliu A, González A et al. Impact of mindfulness and self-compassion on anxiety and depression: The mediating role of resilience. *Int J Clin Health Psychol.* 2021;21(2):100229-100229. IF:5,350. (Q1).
- Collado C, Navarro M, Adrián A, López Y, Garcia J, Montero J. Effectiveness of mindfulness-based stress reduction and attachment-based compassion therapy for the treatment of depressive, anxious, and adjustment disorders in mental health settings: A randomized controlled trial. *Depress Anxiety.* 2021;38(11):1138-1151. IF:6,505. (Q1).

- Martínez D, Martínez C, Monreal A, Barceló A, Campos D, Pérez Aranda A et al. Protective role of mindfulness, self-compassion and psychological flexibility on the burnout subtypes among psychology and nursing undergraduate students. *J ADV NURS.* 2021;77(8):3398-3411. IF:3,187. (Q1).
- Modrego M, López Y, García J, Pérez Aranda A, Navarro M, Beltrán M et al. Efficacy of a mindfulness-based programme with and without virtual reality support to reduce stress in university students: A randomized controlled trial. *Behav Res Ther.* 2021;142:103866-103866. IF:4,473. (Q1).
- Serrat M, Juan Pablo Sanabria M, Almirall M, Musté M, Feliu A, Méndez JL et al. Effectiveness of a Multicomponent Treatment Based on Pain Neuroscience Education, Therapeutic Exercise, Cognitive Behavioral Therapy, and Mindfulness in Patients with Fibromyalgia (FIBROWALK Study): A Randomized Controlled Trial. *Phys Ther.* 2021;101(12)IF:3,021. (Q1).



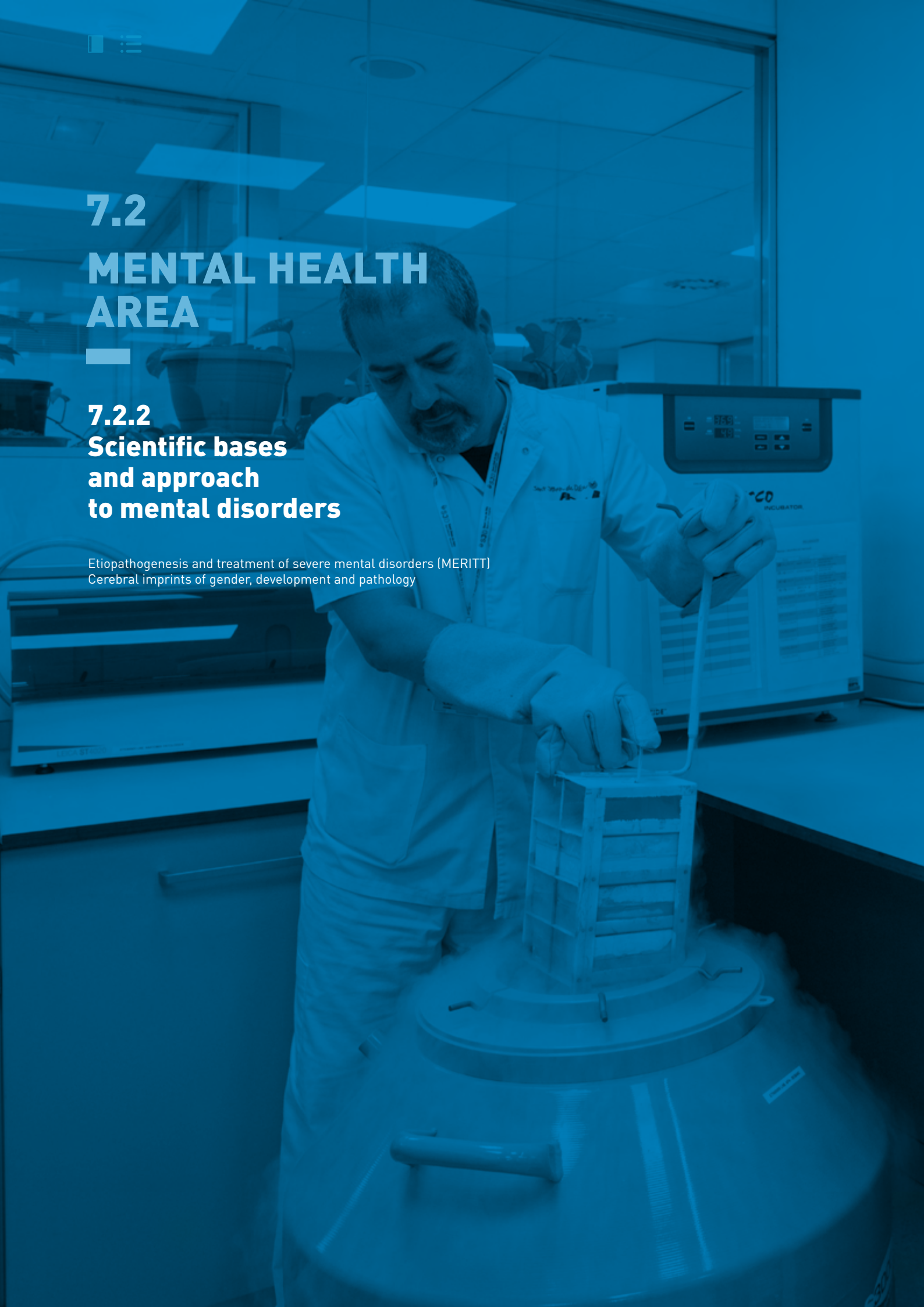
7.2

MENTAL HEALTH AREA

7.2.2

Scientific bases and approach to mental disorders

Etiopathogenesis and treatment of severe mental disorders (MERITT)
Cerebral imprints of gender, development and pathology



Etiopathogenesis and treatment of severe mental disorders (MERITT)

**Susana Ochoa Güerre, MD, PhD
& Judith Usall i Rodié, MD, PhD**

In 2021, our group participated actively in various financed research projects, published articles with a high impact factor, and collaborated with other centres. Moreover, we are leading different multi-centre projects in each of our research lines, the progress of which is reported in the resulting publications. The impact factor of our group grew with respect to previous years and the number of publications with the participation of other centres has increased as well.

With respect to new grants, it may be pointed out that we obtained three research projects in the call of the Carlos III Health Institute, which are addressed to the study of traumatic experiences, the effectiveness of a family metacognitive intervention, and the inflammatory effect of COVID on cognition.

A project of collaboration with an enterprise has been obtained to validate the potentiating effect of aquatic activities on psychological treatment.

The post-residence grant was awarded to a member of our group to allow continued training in research and the preparation of a doctoral thesis. Likewise, two other grants were awarded to two pre-doctoral members of our team so that they may make stays at other research centres abroad.

MEMBERS

STAFF RESEARCHER

Noemí del Prado Sánchez, José Manuel Fernández Rodríguez, Jaume Martínez Mas, Alvaro Muro Alvarez, Aitor-Eneko Olivé Albiztur, Jesús Pérez Pazos, María Ribas Siñol, Vicente Tort Herrando, María Luisa Barrigon Estevez, María José Escandell Morcillo, Belen Ramos JoseMaria, Antonio Manuel Alba Muñoz, Hilari Andres Mora, Belen Arranz Martí, Adela Berrozpe Blasco, Emma Casas Anguera,

Marta Coromina Sadurní, Jorge Cuevas Esteban, Ruth Cunill Clotet, Gemma Escartin Martin, Gemma Escuder Romeva, Alexandrina Foix Sanjuan, Mar García Franco, Marcia Irazábal Giménez, Francisco Martínez Zambrano, Eva Miquel Fernandez, María Trinidad Peláez Martínez, Elena Rubio Abadal, Luis San Molina, Sonia Vilamala Anton

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COLLABORATOR RESEARCHER

Joan Costa Escolà, Maria Elena Huerta Ramos, Raquel López Carrilero, Alfonso Monje Hernández

SELECTED PROJECTS

- Autoestigma i funcionament personal i social en persones amb primers episodis psicòtics. Beneficiari: Manu Abella. (AFR 2021). Parc Sanitari de Sant Joan de Déu -Serveis de Salut Mental. 2021-2022. IP: Judith Usall i Rodié.
- Influencia del estilo de apego en la cognición social y sesgos cognitivos en personas con primeros episodios psicóticos, esquizofrenia crónica y controles sanos. (PI18/00212). Instituto de Salud Carlos III (ISCIII). 2019-2023. IP: Susana Ochoa Güerre.
- Reguladores de la expresión génica como predictores de diagnóstico y biomarcadores de déficits cognitivos y cambios en el grosor cortical en psicosis de inicio reciente. (PI18/00213). Instituto de Salud Carlos III (ISCIII). 2019-2023. IP: Belen Ramos JoseMaria.

SELECTED PUBLICATIONS

- Afia AB, Vila È, MacDowell KS, Ormazabal A, Leza JC, Haro JM et al. Kynurenine pathway in post-mortem prefrontal cortex and cerebellum in schizophrenia:

relationship with monoamines and symptomatology. *J Neuroinflammation*. 2021;18(1):198. IF:8,322. (Q1).

- Traver-Edo D, Escuder-Romeva G, Talavera-Valverde MA, Moruno-Miralles P. Women in Forensic Mental Health Services: Lived Experiences and Meanings Attributed to Activities in Rehabilitation Programs. *Study Protocol. INT J QUAL METH*. 2021. IF:2,767. (Q2).
- Ferrer-Quintero M, Fernández D, López-Carrilero R, Birulés I, Barajas A, Lorente-Rovira E et al. Persons with first episode psychosis have distinct profiles of social cognition and metacognition. *NPJ Schizophr*. 2021;7(1):61. IF:5,200. (Q1).
- Vila-Badia R, Butjosa A, del Cacho N, Serra-Arumí C, Esteban-Sanjusto M, Ochoa S et al. Types, prevalence and gender differences of childhood trauma in first-episode psychosis. What is the evidence that childhood trauma is related to symptoms and functional outcomes in first episode psychosis? A systematic review. *Schizophr Res*. 2021;228:159-179. IF:4,939. (Q1).

Cerebral imprints of gender, development and pathology

Christian Stephan-Otto Attolini, PhD

Our group continues its interaction with clinical personnel in different fields. For example, the joint research line with Dr Alfredo García-Alix and Dr Thais Agut has allowed us to produce two scientific papers, one of which has already been accepted for publication in *Archives of Disease in Childhood*, while the other is currently under peer review. For its part, the research line on psychosis has produced a notable article, which reports various patterns of cognitive impairment in women, who are more susceptible to depression, and in men, who are more susceptible to anxiety. Other projects of interest are the joint analysis of

databases on neuroimaging in the psychiatric population, with the psychiatry research group of Hospital Sant Pau, and the collaboration with Dr Giovanni Frisoni (Fatebenefratelli, Italy) on the use of a database of brain data on Alzheimer's patients.

MEMBERS

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Antonio Callen Soto, Maria Rosa Cambra Martí, Christian Núñez Leánez

ASSISTANT RESEARCHER

Federica Lombardini

COLLABORATOR RESEARCHER

Gildas Yanin Guillaume Brebion

SELECTED PROJECTS

- Identificació dels principals trets cerebrals associats a les etapes més primerenques de diversos trastorns psiquiàtrics i neurològics. (05_2020). Parc Sanitari de Sant Joan de Déu -Serveis de Salut

Mental. 2021-2022. PI: Maria Rosa Cambra Martí.

- COGNIVITRA: COGNitive Vitality TRAIning at home. (AC18/00015). Instituto de Salud Carlos III (ISCIII). 2019-2022. PI: Antonio Callen Soto.
- SGR 2017: Cerebral imprints of gender, development and pathology. (2017 SGR 1297). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2017-2021. PI: Christian Stephan-Otto Attolini.

SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- ACSF2004 - RGS@Home. (RGS@Home). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2020 PI: Antonio Callen Soto.
- Estudio Prospectivo No Intervencionista De Pacientes Con Enfermedad De Alzheimer Leve A Moderada Y Sus Cuidadores En Cuatro Países Europeos. (MER-ALZ-2016-01). Merck,

Sharp & Dohme de España. S.A. 2016 PI: Antonio Callen Soto.

SELECTED PUBLICATIONS

- Brebion G, Nuñez C, Lombardini F, Carl S, Sánchez AM, Sara S et al. Subclinical depression and anxiety impact verbal memory functioning differently in men and women -an fMRI study. *J Psychiatr Res.* 2021;140:308-315. IF:4,791. (Q1).
- Martín Bejarano M, Ruiz B, Zamora B, Martínez de Aragón A, García C, Jiménez S et al. Actividad cerebral en jóvenes infectados por el virus de la inmunodeficiencia humana por transmisión vertical: estudio piloto de resonancia magnética funcional. *Rev Neurol.* 2021;72(10):343-351. IF:0,870. (Q4).
- Quintas J, Pais J, Martins AI, Santos H, Neves L, Sousa S et al. CogniViTra, a Digital Solution to Support Dual-Task Rehabilitation Training. *Electronics.* 2021;10(11) IF:2,397. (Q3).



7.3

TRANSVERSAL PROGRAM

7.3.1

Health technologies and innovation

Medical technologies: bioinformatics and biomedical signals
Medical technologies: computer graphics; serious games and gamification
Medical technologies: biomaterials and tissue engineering
Medical technologies: analysis for rehabilitation
Medical technologies: instrumentation and eHealth
Medical technologies: robotics and vision
Medical technologies: biomechanics
Patients engagement in research
Innovation in health technologies

Medical technologies: bioinformatics and biomedical signals

Alexandre Perera Lluna, PhD

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ASSISTANT RESEARCHER

Pol Gil Figuerola, Joshua Llano Viles, Pol Sola de los Santos, Laura Fincias Marin

TECHNICIAN

Joana Palés Huix, Enrico Manzini, Maria Maqueda

SELECTED PROJECTS

- CAIXAIMPULSE VALIDATE CI21-00186 Vincer. AI. Fundació La Caixa. 2021-2024. PI: Alexandre Perera Lluna.
- Development of new algorithms for analgesia management during general anesthesia (2020 DI 080). Agència De Gestió D'ajuts Universitaris I De Recerca (agaur). 2021-2024. PI: Montserrat Vallverdú.
- Development of an advisory system for the administration of anaesthetics and analgesics (2020 DI 102). Agència De Gestió D'ajuts Universitaris I De Recerca (agaur). 2021-2024. PI: Montserrat Vallverdú.
- Ús de la intel·ligència artificial i la biologia de sistemes per a la diagnosi i l'avaluació de risc personalitzat per a les malalties renals hereditàries, centrat en la síndrome d'Alport (IA&biologia per síndrome d'Alport). La Fundació La Marató de TV3. 2021-2024. PI: Montserrat Vallverdú.

SELECTED PUBLICATIONS

- Barranco M, Solà P, Picart S, Kanaan S, Fonollosa J, Perera A. mWISE: An Algorithm for Context-Based Annotation of Liquid Chromatography-Mass Spectrometry Features through Diffusion in Graphs. *Anal Chem.* 2021;93(31):10772-10778. IF:6,986. (Q1).
- Lopez A, Picart S, Perera A. Balancing Data on Deep Learning-Based Proteochemometric Activity Classification. *J Chem Inf Model.* 2021;61(4):1657-1669. IF:4,956. (Q1).
- Picart S, Thompson WK, Buil A, Perera A. The effect of statistical normalization on network propagation scores. *Bioinformatics.* 2021;37(6):845-852. IF:6,937. (Q1).
- Wei J, Yao J, Belke D, Guo W, Zhong X, Sun B et al. Ca(2+)-CaM Dependent Inactivation of RyR2 Underlies Ca(2+) Alternans in Intact Heart. *Circ Res.* 2021;128(4):63-83. IF:17,367. (Q1).

Medical technologies: computer graphics: serious games and gamification

Raül Benítez Iglesias, PhD

Our group has been granted a national project for the study of heart fibrillation phenomena at the cellular and molecular levels, a project in which we will be applying pattern recognition and deep learning tools to analyse fluorescence microscopy images. We have signed a collaboration agreement with Dr Thaïs Agut's Neonatal Brain research group at Sant Joan de Déu Barcelona Children's Hospital, and as a result of our collaboration a final master's degree project has been carried out. We participated actively in the 3rd edition of the course "Transversal Programme in Medical Image Postprocess. Training Professionals: 3D Printing to Research", financed by EIT Health. Additionally, we

published two articles in journals of acknowledged prestige in collaboration with the Neurogenetics and Molecular Medicine research group of the Sant Joan de Déu Barcelona Research Institute (IRSJD), coordinated by Dr Francesc Palau and Dr Janet Hoenicka. Lastly, a collaboration began with researchers of Amrita University (India) to develop new tools for interpretation of deep learning models.

MEMBERS

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Dolors Ayala Vallespí, Maria Amor Duch Guillen, Jose Luis Eguía Gómez, Mercè Ginjaume Egido, Robert Joan Arinyó, Núria Pla García, Josep Sempau Roma, Lluís Solano Albages, Daniela Tost Pardell, Marc Vigo Anglada

PHD STUDENTS

Núria Bonet Codina, Ariel von Barnekow

SELECTED PROJECTS

- Análisis automático de datos experimentales en fisiología cardiaca. SAF2017-88019-C3-3-R. MINECO. 2018-2021. PI: Raül Benítez
- Análisis computacional del impacto de riesgos genéticos y clínicos en la señalización molecular y disfunciones electrofisiológicas en fibrilación auricular. (PID2020-116927RB-C22). Ministerio de Ciencia e Innovación (MICINN). 2021-2024. PI: Blas Echebarría, Raül Benítez.
- Implications of Medical Low Dose Radiation Exposure. (MEDIRAD). (H2020-755523). H2020. 2017-2022. PI: Mercè Ginjaume.
- Estimaciones de dosis ocupacionales de cristalino en instalaciones sanitarias y de investigación (EDOCI). Propuestas de vigilancia radiológica individual. Consejo de Seguridad Nacional. 2019-2022. PI: Mercè Ginjaume.

SELECTED PUBLICATIONS

- Bonet N, von Barnekow A, Mata MT, Gomar C, Tost D. Three-Dimensional Game-Based Cardiopulmonary Bypass Training. *Clin Simul Nurs.* 2021;50:81. IF:2,391. (Q1).

- Cantarero L, Juárez E, Civera A, Rodríguez M, Roldan M, Benítez R et al. Mitochondria-lysosome membrane contacts are defective in GDAP1-related Charcot-Marie-Tooth disease. *Hum. Mol. Genet.* 2021;29(22):3589-3605. IF:6,150. (Q1).
- Civera A, Dominguez L, Martínez P, Serrano C, Vallmitjana A, Benítez R et al. Mitochondria and calcium defects correlate with axonal dysfunction in GDAP1-related Charcot-Marie-Tooth mouse model. *Neurobiol Dis.* 2021;152:105300. IF:5,996. (Q1).
- Pijuan J, Rodríguez M, Natera D, Ortez CI, Altimir A, Osuna M et al. Translational Diagnostics: An In-House Pipeline to Validate Genetic Variants in Children with Undiagnosed and Rare Diseases. *J Mol Diagn.* 2021;23(1):71-90. IF:5,568. (Q1).

Medical technologies: biomaterials and tissue engineering

Cristina Canal Barnils, PhD

We were granted 3 European projects in 2021, as coordinators in one case, as well as one AEI (Spanish Research Agency) project. Likewise, our group has an ERC Starting Grant, two H2020 projects and four projects from the AEI. We published a total of 38 articles in JCR indexed journals and 2 book chapters, in addition to presenting 11 guest lectures and 20 papers in international conferences. Our doctoral students presented a total of five theses, one in the form of industrial doctorate. All five theses were graded Excellent Cum Laude. Likewise, Dr Cristina Canal obtained an ICREA Academia Prize for her research career, and Dr Carles Mas-Moruno obtained the Marcial Moreno-Mañas 2020 Prize, awarded by the Catalan Section of the Spanish Royal Society of Chemistry (RSEQ). Several of our doctoral students also obtained prizes: J. M. Sadowska

received the Julia Polak European Doctorate Award 2020 from the European Society of Biomaterials (ESB) (Portugal, 06/09/2022); Lluís Oliver won the Audience Award in the competition "Present Your Thesis in 4 Minutes", of the Catalan Foundation for Research and Innovation (FCRI) (Barcelona, 22/06/2021); and Miguel Mateu obtained the prize for the "Best Thesis Presentation" at the 3rd IRSJD Scientific Conference (Barcelona, 28/10/2021). Lastly, our group has two University-Business Chairs with Soadco/Klockner and AMES Group, as well as a large number of agreements with companies of the biohealth and related sectors.

MEMBERS

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Andrea Malandrino, Albert Espona Noguera, José Manuel García Torres, Montserrat Español Pons, Francesc Xavier Gil Mur, Jordi Guillem Martí, Jose María Manero Planella, Carles Mas Moruno, Marta Pegueroles Neyra, Daniel Rodríguez Rius

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TECHNICIAN

Meritxell Sánchez Hervás, Noelia Aparicio Bádenas, Meritxell Molmeneu Trias, Miquel Punset Fusté

SELECTED PROJECTS

- ICREA Academia. ICREA. 2021-2025. PI: Cristina Canal.
- PlasTHER: Therapeutical Applications of Cold Plasmas (COST Action CA20114). European Commission. 2021-2025. PI: Cristina Canal.
- SMARTGEL: Bioactive and multifunctional peptide-based hydrogels obtained by bottom-up self-assembly for advanced biomedical applications (PID2020-114019RB-I00). Agencia Estatal de

Investigación, Ministerio de Ciencia, Innovación y Universidades. 2021-2024. PI: Carles Mas-Moruno.

SELECTED PUBLICATIONS

- García C, Córdoba LC, Buxadera J, Marquina A, Jiménez E, Ginebra MP et al. Zn-Mg and Zn-Cu alloys for stenting applications: From nanoscale mechanical characterization to in vitro degradation and biocompatibility. *Bioact Mater.* 2021;6(12):4430-4446. IF:14,593. (Q1).
- Oliver L, Martin H, Reyes L, Noureddine F, Ada Cavalcanti E, Ginebra MP et al. An Engineered Biomimetic Peptide Regulates Cell Behavior by Synergistic Integrin and Growth Factor Signaling. *Adv Healthc Mater.* 2021;10(7):e2001757. IF:9,933. (Q1).
- Raymond Y, Thorel E, Liversain M, Riveiro A, Pou J, Ginebra M. 3D printing non-cylindrical strands: Morphological and structural implications. *Additive Manufacturing.* 2021;46: 102129. IF:10,998. (Q1).
- Tornin J, Labay C, Tampieri F, Ginebra MP, Canal C. Evaluation of the effects of cold atmospheric plasma and plasma-treated liquids in cancer cell cultures. *Nat Protoc.* 2021;16(6):2826-2850. IF:13,491. (Q1).

Medical technologies: analysis for rehabilitation

Miquel Àngel Mañanas Villanueva, PhD

The BIOsignal Analysis for Rehabilitation and Therapy Research Group (BIOART) has been officially recognised by the Government of Catalonia since 2008. It is formed by four professors, three post-doctoral researchers, four PhD students, and medical specialists in motor and cognitive rehabilitation, respiratory

system, and internal medicine. The group belongs to the Biomedical Engineering Research Centre (CREB, from the Catalan) of the Polytechnic University of Catalonia (UPC). Between 2020 and 2022, our group has received financing from various competitive projects that has allowed us to strengthen our three research lines on motor, cognitive and respiratory system rehabilitation, and to launch a new line of research on artificial intelligence for the support of decision-making in the diagnosis, treatment and rehabilitation of high-impact disorders. Of special note are a concept test of the national R&D&I programme addressed to the challenges of society (Spanish Ministry of Science and Innovation), a TecnioSpring Industry project financed by the Marie Skłodowska-Curie Actions programme (Horizon Europe), and a Beatriu de Pinós post-doctoral grant, all three of which were obtained in 2021. Our group also receives funding from other projects such as Llabor i Producte (Seed and Product) (AGAUR- Government of Catalonia), CaixaImpulse (La Caixa Foundation), INNOTEC (ACCIÓ; Government of Catalonia), and the National Plan for Scientific and Technical Research and for Innovation (MINECO/MICINN). During the past year, nine articles were published in JCR indexed journals (six in the first quartile) and one patent was registered. Our group also received the Prize for the Best Innovation Patent for Its Application to the Market, awarded by the UPC's Social Council.

MEMBERS

STAFF RESEARCHER

Joan Francesc Alonso Lopez, Sergio Romero Lafuent

SELECTED PROJECTS

- Caixaimpulse validate CI21-00242 MV-Optimizer (CAIXAIMPULSE VALIDATE CI21-00242). Fundació La Caixa. 2021-2023. PI: Miquel Àngel Mañanas.
- MYOARM: tecnología inteligente para la rehabilitación motora (PDC2021-120818-I00). Agencia Estatal De Investigación. 2021-2023. PI: Miquel Àngel Mañanas.
- Tecnologías innovadoras para monitorizar y personalizar la

rehabilitación interdisciplinar de pacientes de cuidado intensivo (PID2020-117751RB-I00). Agencia Estatal De Investigación. 2021-2024. PI: Miquel Àngel Mañanas.

SELECTED PUBLICATIONS

- Marateb HR, Ziaie F, Mohebian MR, Sami R, Haghjooy S, Dehghan F et al. Automatic Classification Between COVID-19 and Non-COVID-19 Pneumonia Using Symptoms, Comorbidities, and Laboratory Findings: The Khorshid COVID Cohort Study. *Front Med (Lausanne)*. 2021;8:768467-768467. IF:5,091. (Q1).
- Migliorelli C, Romero S, Bachiller A, Aparicio J, Alonso JF, Mañanas MA et al. Improving the ripple classification in focal pediatric epilepsy: identifying pathological high-frequency oscillations by Gaussian mixture model clustering. *J Neural Eng*. 2021;18(4): 0460f2. IF:5,379. (Q1).
- Sarmiento CA, Hernandez AM, Serna LY, Mañanas MA. An integrated mathematical model of the cardiovascular and respiratory response to exercise: model-building and comparison with reported models. *Am J Physiol Heart Circ Physiol*. 2021;320(4): H1235-H1260. IF:4,733. (Q1).
- Tost A, Migliorelli C, Bachiller A, Medina IF, Romero S, García A et al. Choosing Strategies to Deal with Artifactual EEG Data in Children with Cognitive Impairment. *Entropy (Basel)*. 2021;23(8):1030. IF:2,524. (Q2).

García Gonzalez, Miguel J García Hernandez, Lexa Nescolarde Selva, Juan Ramos Castro, Pere Joan Riú Cost

SELECTED PROJECTS

- Integrating CARTo and multi-frequency impedance system. BiosenseWebster. 2019-2022. PI: Xavier Rosell.
- Electronic biopsy of in-vivo lung tissue based on electrical impedance spectroscopy (RTI2018-098116-B-C21). Agencia Eestatal de Investigación. 2019-2021. PI: Ramon Bragós Bardia.

SELECTED PUBLICATIONS

- Hernández JA, Nescolarde L, Roca E, Revuelta E, Ara J, Bayes A. Marathon Running Increases Synthesis and Decreases Catabolism of Joint Cartilage Type II Collagen Accompanied by High-Energy Demands and an Inflammatory Reaction. *Front Physiol*. 2021;12:722718-722718. IF:4,566. (Q1).
- Mateu M, Guede F, Rodriguez N, García MA, Ramos J, Fernández C. A non-contact camera-based method for respiratory rhythm extraction. *Biomed. Signal Process. Control*. 2021;66: 102443. IF:3,880. (Q2).
- Román E, Poca M, Amorós G, Rosell X, Gely C, Nieto JC et al. Phase angle by electrical bioimpedance is a predictive factor of hospitalisation, falls and mortality in patients with cirrhosis. *Sci Rep*. 2021;11(1):20415-20415. IF:4,379. (Q1).

Medical technologies: instrumentation and eHealth

Xavier Rosell Ferrer, PhD

MEMBERS

STAFF RESEARCHER

Ramon Bragós Badia, Mireya Fernández Chimeno, Miguel Angel

Medical technologies: robotics and vision

Alicia Casals Gelpi, PhD

In 2021m we successfully completed the SARAS project, which has entailed an important step forward

in the process of providing a certain autonomy to two robotic assistance arms for laparoscopic surgery that is carried out either manually (laparoscopy) or by telesurgery (robotics). This work has provided an advance in cognitive aspects which, with the knowledge of the steps of a specific surgical procedure and the capacity to perceive and to interpret the evolution in real time, allows the surgical assisting robots to carry out cooperatively auxiliary tasks that would otherwise need to be performed by one or more assistant surgeons.

In the field of intraluminal surgery, experimentation has already been conducted with teams of physicians on the interface resulting from the studies. We have studied the effect on the improvement achieved as a result of both the type of device used (endoscope) and the form of presentation of the information to the intervening professionals, as well as the effect of each type of assistance, either of perception or of provision of autonomy to the exploratory robotic device. Work was carried out on a colon simulator, which has been developed to allow progress in this line, and results on a significant sample of specialists are now available.

2021 began with a new project in the field of disability assistance, specifically involving the design of a robotised chair with the twofold function of providing mobility and assistance for the user's action as well as operativity in his or her surroundings. The project comprises two different parts. On the one hand, the design of the chair-exoskeleton itself, providing mobility, posture, and assistance for the action of the user's arms. This entails the challenge of making the human anatomy compatible with an external structure. And on the other hand, the project involves the design of an intelligent interface that will allow users with motor limitations to control their movement from one place to another, their posture, and their arm actions in an intuitive way that is adapted to their physical constraints.

Likewise, negotiations have been resumed to reach an understanding on our cooperation with the Norwegian company Topro, after the postponement of the new agreement

on this second phase of the process due to the pandemic. Now that the feasibility has been demonstrated of our systems for user monitoring support and walker control, which have already been produced and assessed, this agreement will allow our group to carry out the robotisation of Topro's walkers. We have also begun to work on the creation of the company that is to implement this project, on the development of its business plan, and on the process of its founding.

MEMBERS

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TECHNICIAN

Oriol Escoté Lluís

COLLABORATOR RESEARCHER

Antonio Benedico Blanes, Carlos Morata, Laureano Tinoco

SELECTED PROJECTS

- Plataforma híbrida cadira rodes-exoesquelet (PID2020-116091RB-I00). Ministerio de Economía, Industria y Competitividad. 2021-2024. PI: Alicia Casals.
- Smart . Autonomous RoboticAssistant Surgeon (SARAS) (H2020-ICT-2017 Ref 77981). H2020. 2018-2021. PI: Alicia Casals.
- Autonomous Intraluminal Surgery (ATLAS) (H2020-ITN 2018 REF: 813782). H2020. 2019-2023. PI: Alicia Casals.
- Caminadors elèctrics. TOPRO Industry. 2018-2022. PI: Antonio B Martínez.

SELECTED PUBLICATIONS

- Finocchiario M, Cortegoso P, Hernansanz A, Marino N, Amram D, Casals A et al. Training Simulators for Gastrointestinal Endoscopy: Current and Future Perspectives. *Cancers* [Basel]. 2021;13(6):1427. IF:6,639. (Q1).

Medical technologies: biomechanics

Josep Maria Font Llagunes, PhD

In 2021, the BIOMECH group received funding for a project of the telethon programme *La Marató* de TV3, in collaboration with Sant Joan de Déu Barcelona Children's Hospital, and we were also granted a TECNIO Spring Industry project, financed by the Marie Skłodowska-Curie Actions (Horizon Europe) programme. Likewise, during this period, our group led projects in progress financed by Spanish Research Agency (AEI) (Research Challenges), AGAUR (Product), and the European Institute of Innovation and Technology (EIT Health). Six articles were published in JCR indexed journals in this period, 10 papers were presented in scientific conferences, and a doctoral thesis co-directed by a researcher of our group was presented at Rice University (United States).

Additionally, our group coordinated the National Network of Excellence in Biomechanics Research, financed by Spanish Research Agency (AEI) and formed by 10 Spanish groups in the biomechanics field. We are also an active member of the innovation networks Xartec Salut and i4KIDS, led by CREB (Research Centre for Biomedical Engineering) and Sant Joan de Déu Barcelona Children's Hospital, respectively. Lastly, Dr Josep Maria Font has chaired the Technical Committee for Multibody Dynamics of the International Federation for the Promotion of Mechanism and Machine Science (IFTToMM).

In 2021, our research group received the Polytechnic University of Catalonia (UPC) Prize for Social Commitment in the Field of Inclusion, and a mention in the UPC Prize for Valorisation of Research, both of which are granted by the UPC's Social Council.

MEMBERS

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Giménez, Joan Lobo Prat

PHD STUDENTS

Jesús de Miguel Fernández, Michael Ray Epke, Antonio Rodríguez Fernández, Míriam Febrer Nafria

SELECTED PROJECTS

- ArmTracker: Un sistema portable per avaluar la funció motora de les extremitats superiors durant la vida diària per a pacients amb distròfia muscular de Duchenne i atròfia muscular espinal (202023-32). La Fundació La Marató de TV3. 2021-2024. PI: Josep Maria Font.
- ABLE Exoskeleton (EIT_HEALTH-20121-ABLE). EIT Health Innovation Programme (H2020, EU Commission). 2020-2022. PI: Josep Maria Font.
- ABLE: Exoesquelet robòtic que permet tornar a caminar de forma natural i intuïtiva després de patir una lesió medul·lar (2018-PROD-00059). AGAUR. 2019-2021. PI: Josep Maria Font.
- Design of personalized robotic and neuroprosthetic wearable systems for walking assistance using a predictive simulation framework (TAILOR) (RTI2018-097290-B-C33). Spanish Ministry of Science, Innovation and Universities (MICINN). 2020-2022. PI: Josep Maria Font.

SELECTED PUBLICATIONS

- Febrer-Nafria M, Pallarès-López R, Fregly BJ, Font-Llagunes JM. Prediction of three-dimensional crutch walking patterns using a torque-driven model. *Multibody Syst Dyn.* 2021;51(1):1-19. IF:3,109. [Q2].
- Gómez-Pérez C, Martori JC, Puig A, Medina J, Vidal J, Font-Llagunes JM. Gait event detection using kinematic data in children with bilateral spastic cerebral palsy. *Clin Biomech (Bristol, Avon).* 2021;90:105492. IF:2,063. [Q3].
- Koch MA, Font-Llagunes JM. Lower-Limb Exosuits for Rehabilitation or Assistance of Human Movement: A Systematic Review. *Appl Sci-Basel.* 2021;11(18): 105.492. IF:2,679. [Q2].
- Rodríguez-Fernández A, Lobo-Prat J, Font-Llagunes JM. Systematic review on wearable lower-limb exoskeletons for gait training in neuromuscular impairments. *J Neuroeng Rehabil.* 2021;18(1):22. IF:4,262. [Q1].

Patients engagement in research

Joana Claverol Torres, MSc & Begonya Nafria Escalera, MSc

In 2021, the Patient Participation in Research Area carried out its research activity in the framework of competitive projects with public financing as well as in projects conducted with our own funds, which have a direct impact on our institution. Notable among these activities was the development of the Patient Participation Plan, which will be implementing its first strategic plan in the period 2022-2024. This plan will allow the preparation and orientation of our institution's research activity towards a model centred on patients' needs. To this end, it will be essential to have patients' close involvement from the earliest phases of research to the final moment when the results of the projects are fed back to patients and disseminated.

Another initiative of note is the start of the Science Agora project, which has the primary goal of making our institution's research initiatives known, taking special care to highlight the importance of involving society in these initiatives and to present the results of the projects in clearly understandable language.

The scientific output of the Patient Participation Area was focused in 2021 on disseminating the results of projects financed by the European Commission in the field of collaborative research with patients and their families in relation to rare diseases (Share4Rare Project), the study of paediatric patient's preferences with respect to the oral formulation of medicines (EPTRI Project), the analysis of the impact of the quarantine on Spanish families (Kids Corona Project), the conceptualisation of a pan-European network of paediatric clinical trials (Conect4children Project), and the analysis of the needs of the various agents participating in research on new medicines (Paradigm Project).

MEMBERS

STAFF RESEARCHER

Nuria Noel Palacio, Estibaliz Urarte Rodríguez

TECHNICIAN

Elisa Mora de Checa

SELECTED PROJECTS

- Ágora de la Ciencia. (FCT-20-16045). Fundación Española para la Ciencia y la Tecnología. 2021-2022. IP: Begoña Nafria Escalera.
- YEAH: Youngsters EngAgement in Health science (20624). EIT Health e.V. CAMPUS Citizens. 2020-2021. IP: Begoña Nafria Escalera.
- EJP RD: European Joint Programme on Rare Diseases. (825575). European Commission. 2019-2023. IP: Joana Claverol Torres.
- c4c_conect4children (Collaborative Network for European Clinical Trials For Children). (777389). European Commission. 2018-2024. IP: Joana Claverol Torres.
- SHARE4RARE: Social media platform dedicated to rare diseases, using collective intelligence for the generation of awareness and advanced knowledge on this large group of diseases. European Commission. 2018-2021. IP: Begoña Nafria Escalera.

SELECTED PUBLICATIONS

- Ajanovic S, Jon A, Baro B, Balanza N, Varo R, Millat P et al. How Did the COVID-19 Lockdown Affect Children and Adolescent's Well-Being: Spanish Parents, Children, and Adolescents Respond. *Front Public Health.* 2021;9:746052. IF: 3,709. [Q1].
- Alessandrini E, Brako F, Scarpa M, Lupo M, Bonifazi D, Pignataro V et al. Children's Preferences for Oral Dosage Forms and Their Involvement in Formulation Research via EPTRI (European Paediatric Translational Research Infrastructure). *Pharmaceutics.* 2021;13(5):730. IF: 6,321. [Q1].
- Faulkner SD, Sayuri S, Pakarinen C, Somers F, Jose Vicente Edo M, Prieto L et al. Understanding multi-stakeholder needs, preferences and expectations to define effective practices and processes of patient engagement in medicine development: A mixed-methods study. *Health Expect.* 2021;24(2):601-616. IF:3,377. [Q1].

Innovation in health technologies

Jaume Pérez Payarols, MD, PhD

Our group's activity in 2021 notably included the inauguration of the Pediatric Innovation Hub (i4KIDS), which is formed by 26 institutions of excellence. They are all focused on strengthening paediatric research and innovation, and include hospitals, universities, biomedical research centres and technology centres. Likewise, we should point out the success achieved in terms of participation in the first edition of the Pediatric Innovation Day (PID). In order to comply with the new legislation, HSJD's 3D Unit (3D4H) has obtained the required operating licence to manufacture made-to-measure orthopaedic products from the competent authority of the Autonomous Region, which in our case is the Government of Catalonia (General Directorate for Health Planning and Regulation). This licence is valid for 5 years.

This year Sant Joan de Déu Barcelona Children's Hospital received the prize for the Best Innovating Hospital at the BHHSummit 2021 held by the Barcelona Health Hub on 28th October. Lastly, mention should also be made of our new spin-off Ephion Health S.L., a pioneer in the use of artificial intelligence to monitor the evolution of patients with disorders affecting their mobility, in cooperation with Eurecat Technology Centre and the Collider, the Mobile World Capital Barcelona (MWCcapital) innovation programme.

MEMBERS

STAFF RESEARCHER

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ASSISTANT RESEARCHER

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TECHNICIAN

Mercè Francisco Bordas, Xavier Puñet Ortiz, Ariadna Ballesteros Herruzo, Paula Cañal Vega

SELECTED PROJECTS

- Hub d'Innovació Pediàtrica. [2018 XARDI 00011]. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2020-2022. IP: Jaume Pérez Payarols.
- Ecosistema d' R+D+i per la implementació i adopció de la Fabricació Additiva / Impressió 3D a la indústria de Salut (QuirofAM). (COMRDI16-1-0011-06). Departament de Salut - Generalitat de Catalunya. 2018-2021. IP: Arnau Valls Esteve.
- ACADOM_Asegurant la continuïtat assistencial domiciliària. (COMRDI15-1-0026). Departament de Salut - Generalitat de Catalunya. 2016-2021. IP: Arnau Valls Esteve.
- Perfil técnico en las Unidades 3D sanitarias. El binomio Imagen para el Diagnóstico y Fabricación Mecánica, hibridación de futuro en FP. (AFP21/00052). Ministerio de educación. 2021-2023. IP: Arnau Valls Esteve.
- Families4Health (F4H), un proyecto para la formación en hábitos saludables para familias desfavorecidas. Fundación Ramon Prado i Pintó. 2020-2022. IP:Arian Tarbal Roquer.

SELECTED PUBLICATIONS

- COVIDSurg C, Dajti I, Valenzuela JI, Boccalatte LA, Gemelli NA, Smith DE et al. Machine learning risk prediction of mortality for patients undergoing surgery with perioperative SARS-CoV-2: the COVIDSurg mortality score. *Br J Surg.* 2021;108(11):1274-1292. IF:6,939. (Q1).
- Quiroz MJ, González H, Palazón A, Rubio J, Prat J. Amblyopia due a dentigerous cyst. Case report and literature review. *J FR OPHTALMOL.* 2021;44(8):1249-1255. IF:0,818. (Q4).
- Vanesa V, Irene MP, Ayats M, Parri Ferrandis F, Miguel S, Mireia RM et al. Accuracy of virtually planned mandibular distraction in a pediatric case series. *J Craniomaxillofac Surg.* 2021;49(2):154-165. IF:2,078. (Q3).
- Verduytsse HJR, Rubio J, Van de Castele E, Nadjmi N, De Praeter M, Dunaway D. Virtual planning in Le Fort III distraction osteogenesis: A case series. *J Craniomaxillofac Surg.* 2021;49(5):341-346. IF:2,078. (Q3).

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