CURRICULUM VITAE
Aurora Pujol, M.D., PhD
Bellvitge Biomedical Research Institute (IDIBELL)

AURORA PUJOL M.D, PhD, FACMG January 10 2023

DOB: 15th May 1968, Barcelona, Spain
Spanish Citizenship / USA Greencard
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Summary

I am a physician-scientist, Board Certified Clinical and Laboratory Geneticist by USA and European Boards. I hold a Genetics and Neuroscience Ful Professorship for the Catalan Government Scientific Council (ICREA).

I am the Director of the Neurometabolic Diseases Laboratory at IDIBELL Research Institute in Barcelona, Spain, since 2005. I serve as Medical Geneticist at the Neuromuscular and Rare Disorders Unit at the Bellvitge University Hospital, since 2008. I am passionate about improving the disease management of rare brain disorders through Genomic Medicine, from diagnostics and gene discovery to rational treatments, designed after integrative multiomics approaches. On top of genomics, we are applying epigenomics, scRNAseq, metabolomics/lipidomics and proteomics, both for diagnosis and for deciphering of molecular basis of disease for rational therapeutic design. We are developing our own algorithms for improving diagnostic yields and for integrating -omics data. My lab is pioneering Genomic Medicine in Spain, and I am recognized internationally as a key opinion leader in the inherited metabolic brain disorders and medical genomics fields.

I am leading a matrixed team of excellent bioinformaticians, molecular and cellular biologists, biochemists and clinicians since 2005. We have translated our work into four licensed patents on repurposed drugs, four orphan drug designations, and four phase II/ III clinical trials for rare leukodystrophies. We are also proud of having diagnosed over 500 families affected with rare neurological diseases using WES and WGS, attaining a diagnostic rate of 70% when incorporating functional validation of variants, which allowed the discovery of 12 novel syndromes and causative disease-gene associations. Our shared vision of integrating functional and clinical genomics is our recipe for delivering timely solutions to our rare disease families. On a more fundamental note, we are interested in deciphering the role of complex lipids and membrane integrity in aging and neurodegeneration.

It is my honor to serve at the scientific boards of the European Society of Human Genetics (ESHG), the American College of Medical Genetics (ClinGen Peroxisomal Working Group and Metabolic Working Group), and the Undiagnosed Disease Network International (UDNI, NIH USA, Functional Study Group). I am also honored to serve on the Editorial Boards of leading journals such as Neurobiology of Disease and Journal of Neuroscience Research. Is my pleasure to participate in the Scientific Boards of the patients' associations European Leukodystrophy Association (ELA-Spain) and the French Association for Friedreich Ataxia and Hereditary Spastic Paraplegias.

I hold an honorary position of Associate Professor in the Department of Neurology of Weill Cornell Medicine NYC, and I am frequently invited to lecture and chair sessions at international conferences (European Society of Human Genetics (ESHG), Gordon Research Conference, European Glia Meeting, FASEB, International Society of Neurochemistry Meetings). I am a regular consultant for the pharmaceutical industry on rare and neurodegenerative diseases.

I have received several international scientific recognitions, including the Young Investigator Award from the ESHG (2004), the Grifols Albus Award for a novel application of plasmapheresis to adrenoleukodystrophy (2019), and the prestigious Prize of the Society for the Study of Inborn Errors of Metabolism (SSIEM, London) in 2019 and 2020, for gene discovery and treatment of two novel brain diseases, HLD-18 leukodystrophy and SHMT2 deficiency respectively. My most relevant national awards include the prizes of the Royal House of Spain (Hesperia Foundation) (2011), Prize of the Pharmacists College of Seville (2014), and the XIV National Prize of the Physicians College of Córdoba (2016). My group's work is regularly featured in national and international press.

Education

07/19/1993 **Medical Degree,** with honors. Full License as General Practitioner
Medical School of the Autonoma University of Barcelona, Spain

02/05/1998 **PhD in Cellular and Molecular Biology**, summa cum laude
Applied Tumor Virology, German Cancer Research Center (DKFZ)
Ruprecht-Karls University of Heidelberg, Germany

09/28/2001 **French Board Certification in Medical Genetics (Medical Specialty)**
Concours National de Praticien Hospitalier en Génétique Médicale,
Louis Pasteur Medical School, Strasbourg, France.

09/01/2017 **American Board Certification in Clinical Molecular Genetics and Genomics, ABMGG.**
Fellow of the American College of Medical Genetics (FACMG)
NHGRI/ NIH, Bethesda MD, USA.

04/15/2019 **European Board Certification as Clinical Laboratory Geneticist (ErCLG).**

Languages

Bilingual in Catalan and Spanish.
Full Professional Proficiency in English, French, and German.
French, Diplôme Supérieur de Langue Française, Institut Français de Barcelone-Toulouse, June 1986.
German, Oberstufe, Goethe Institut, May 1994
English, TOEFL, September 2014.

Positions

>07/ 20/1993-01/ 07/1994
Attending Clinician in Family Medicine (Médico Adjunto en Medicina de Familia)
Catalan Institutes of Health, Barcelona, Spain

>08/ 1994-02/ 1998
Predoctoral fellow, Applied Tumor Virology, DKFZ (**German Cancer Research Center, Heidelberg**, Germany). Lab Director: Jean Rommelaere, Professor of Virology. Human Capital & Mobility Fellowship, European Commission Fellowship

>03/1998- 12/2003
Postdoctoral scientist, Human Molecular Genetics Department, **IGBMC, Strasbourg,** France
Director: Jean Louis Mandel, Professor of Human Genetics.

>05/1998- 09/2001
Resident in Clinical Genetics, Strasbourg University Hospital, France
Director: Jean Louis Mandel, Professor of Human Genetics, Chair de Génétique du College de France

>10/2001-12/2004
Attending Clinician in Medical Genetics (Praticien Hospitalier en Génétique Médicale, Attaché), Department of Human Genetics**, Strasbourg University Hospital**, France
Director: Jean Louis Mandel, Professor of Human Genetics, Chair de Génétique du College de France

>01/01/2004- 12/31/2004
Junior Group Leader, Human Molecular Genetics Department, **IGBMC, Strasbourg,** France
Director: Jean Louis Mandel, Professor of Human Genetics.

>01/01/2005-Present
Professor at ICREA, Catalan National Research Council.
Director of the Neurometabolic Diseases Laboratory, IDIBELL, Barcelona, Spain.
www.neurometabolic-lab.org

>11/01/2008-Present
Attending Clinician in Medical Genetics (Adjunto en Genética Médica)
Hospital Duran i Reynals, Barcelona, Spain

Neuromuscular and Rare Disorders Unit, Neurology Division, Bellvitge University Hospital, Hospitalet de Llobregat, Barcelona, Spain.

>09/01/2011-08/31/2012
Visiting Professor at Neurology and Neurosciences Department), **Weill Cornell Medicine**, New York City, NY. Sabbatical stay
Director: Flint Beal, Professor of Neurology

>06/21/2013-Present
Adjunct Associate Professor of Neuroscience at Brain and Mind Research Institute of **Weill Cornell Medicine**, New York City, NY.

>12/01/2014-05/31/2015
Visiting Professor at the Center for Mitochondrial and Epigenomic Medicine (CMEM),
Children's Hospital of Philadelphia (CHOP), University of Pennsylvania. Sabbatical stay
Head of the Lab: Douglas Wallace / Marni Falk, Professors of Human Genetics

>09/01/2015-06/30/2017
Fellow of the Clinical Molecular Genetics Program at NHGRI/NIH, Bethesda, Maryland, USA
Program Directors: Suzanne Hart, Maximillian Muenke

>09/01/2018-Present
Associate Professor for Clinical Genetics and Genomics, Pompeu Fabra University (UPF), Barcelona, Spain

>07/01/2022
Sr Distinguished Scientist, Rare and Neurological Diseases Area,
SANOFI, Boston, USA

Honors and Prizes

09/1992 **Clinical Fellow. EC Fellowship "Erasmus student exchange program",** for 6-month Surgery training at the Cardiac Surgery Department, Ruprecht-Karls University of Heidelberg, Germany.

01/1994 **Predoctoral fellowship.** EC PhD Fellowship "Human Capital and Mobility" program of the European Commission.

03/1998 **Postdoctoral fellowship EMBO,** resigned in favor of the "Training and Mobility of Researchers" fellowship.

03/1998 **Postdoctoral fellowship,** "Training and Mobility of Researchers" program of the European Commission FP4 Program (contract N. FMBICT983163).

04/2000 **Postdoctoral fellowship,** Foundation pour la Recherche Médicale (FRM).

06/2004 **Prize "Young Investigator Award" of the European Society of Human Genetics,** at the 36th European Human Genetics Conference, Munich, Germany.

01/2005 **ICREA Full Professorship,** to date.

05/2006 **President of the Scientific Commitee,** European Leukodystrophy Association-(ELA-Spain), Madrid Spain.

09/2010 **Scientific Committee Board**, IDIBELL, 2010-2011

05/2011 **Salvador de Madariaga Award from the Spanish Ministry of Education** for one sabbatical year stage at Weill Cornell Medical College, NYC, USA.

05/2011 **Prize of the Hesperia Foundation,** Princes of Spain.

02013 **Adjunct Research Associate Professor in Neurology and Neurosciences,** Brain and Mind Research Institute, Weill Cornell Medical College, NYC, USA, to date.

06/2014 **National Prize of the Pharmaceutical College of Seville, Spain.**

09/2014 **BAE Award from the Spanish National Institutes of Health (ISCiii)** for 6 months sabbatical stage at Children's Hospital of Philadelphia, USA.

04/2016 **XIV National Research Prize of the College of Physicians of Cordoba, Spain.** 1st Position with unanimity of the Jury.

12/2017 **Member of the Undiagnosed Disease Program International (UDNI),** NIH Bethesda USA.

03/2018 **Member, Functional Study Group** at UDNI, Undiagnosed Disease Network International NIH, Bethesda, USA.

07/2019 **Scientific Council Board, AFAF** (French Association Friedreich Ataxia); **CSC** (Knowing the Cerebellar Syndromes); **AS-L HSP** (Strumpell-Lorrain Association, Hereditary Spastic Paraplegias).

09/2019 **Metabolism Working Group, ClinGen consortium**, Bethesda Maryland. (Director Meredith Weaver, American College of Medical Genetics and Genomics, ACMGG) 2019-

05/2019 **Scientific Committee Board Member, European Society of Human Genetics,** ESHG, Vienna, Austria (2019-2023).

09/2019 **Late Breaking Award, Society for the Study of Inherited Errors of Metabolism** (SSIEM, London), at 2019 International Congress, Rotterdam, The Netherlands.

10/2019 **Prize Albus Award, Grifols International,** Barcelona, Spain

11/2020 **Late Breaking Award, Society for the Study of Inherited Errors of Metabolism** (SSIEM, London), at the 2020 International Virtual Congress.

Advisory Boards (for profit)

2014 **Advisory Board,** MedDay Parmaceuticals, Paris. Consulting as KOL for brain metabolism in white matter diseases including multiple sclerosis and the leukodystrophies.

2019- **Advisory Board,** SwanBio, Philadelphia USA. Consulting as KOL for preclinical models of leukodystrophies and motorneuron disorders.

2019 **Advisory Board,** Bluebird Bio, Boston USA. Consulting as KOL for biomarkers and natural history of adrenoleukodystrophy.

2020 **Advisory Board,** Inflectis Bioscience. Consulting as KOL for biomarkers and preclinical models of leukodystrophies.

Membership of Research and Clinical Networks

IMPaCT Genomica: Spanish Research Network for Personalized Medicine, 2021-Spanish National Institutes of Health (Lead by Angel Carracedo, La Coruña Spain).

UDNI- **Undiagnosed Rare Disease Network International, 2018-**National Institutes of Health, NIH (Lead by Dr William Gahl, NIH/NHGRI, Bethesda, MD USA)
Functional Analysis Working Group

URD-Cat **Undiagnosed Rare Neurological Disease Program of Catalonia 2017-2019**. Coordinator of IDIBELL node.

SGR **Research Group of Excellence of the Catalan Government (AGAUR).**
Group leader of unit GRC 085: Neurometabolic Diseases**,** since 2009.

CIBERER **National Biomedical Network on Rare Diseases**, from National Health Institute Carlos III
Spanish Ministry of Science and Innovation.
Group leader of unit U759 since 2008.

ANEP **National Agency for Research Evaluation and Foresight**
Advisory Committee of the Spanish Government for Evaluation of Research Grants; Biomedicine Study Section in 2010, 2012 and 2014.

AGAUR **Catalan Agency for Evaluation of Research**
Advisory Committee of the Catalan Government for Evaluation of Research.

ACSUCyL **Agency for Quality of the Universitary System of Castilla and Leon**
Advisory Panel for Evaluation of Research Grants of the Castillean Government; Spain.

SERGAS **Health System of Galicia**
Advisory Panel for Evaluation of Research Grants of the Government of Galicia; Spain.

Membership of Scientific Societies

2001-	Spanish Association for Biochemistry and Molecular Biology (SEBBM)	Member
2004-	European Society of Human Genetics (ESHG)	Member
2005-	Spanish Association for Human Genetics (AEGH)	Member
2017-	American College of Medical Genetics (ACMG)	Fellow

Managerial Activities

Organisation of International Congress and Workshops

1. Recent advances on diagnosis and therapeutics of Leukodystrophies. Invited Speakers: Profs Jaap Valk, Odile Boespflug-Tanguy, Patrick Aubourg, Nathalie Cartier.1st ELA-España Symposium, in conjunction with the 23th Annual Meeting of the Spanish Society for Neuropediatry. Alicante, Spain, 24th May 2007.
2. Novel therapeutic strategies for X-Adrenoleukodystrophy. Invited Speakers: Prof Patrick Aubourg, Dr Nathalie Cartier. Hospital Vall d'Hebron, Barcelona, 5th July 2007.

3. International Course on Diagnostic and Therapeutic Approaches for Leukodystrophies. Co-organised with Odile Boespflug-Tanguy and Salvador Martínez. Spanish Center of Reference for Patients with Rare Diseases. Burgos, Spain, 9-11th November 2010.
4. Symposium on Systematic Approaches to Glia. In collaboration with Hauke Werner (Göttingen, Germany), at the XII European Meeting on Glial cells in Health and Disease. Bilbao, Spain, 15-18th of July 2015.
5. Scientific Program of the European Society of Human Genetics, Berlin, Germany, June 6-8th 2020.
6. Scientific Program of the European Society of Human Genetics, Glasgow, UK, June 12-15th 2021.
7. Scientific Program of the European Society of Human Genetics, Vienna, Austria, June 13-17th 2022.

Chairman at International Congress

1. Chairman at the Symposium "Targeted therapies for cancer and genetic disease". **40th European Society for Human Genetics (ESHG).** Barcelona 1st of June 2008
2. Chairman at the Session "Therapeutics for Leukodystrophies**". 2nd European Leukodystrophy Association (ELA) Research Foundation congress**. Luxembourg, 27th of June 2009
3. Chairman at the Session "Peroxisomal Disorders". **Open European Peroxisomal Meeting**, Dijon, France 4-5th of July 2012
4. Chairman and organizer at the Session: Biomarkers, Targets and Therapies for Leukodystrophies. **XI European Meeting on Glial Cells in Health and Disease**, Berlin 2nd July 2013-
5. Chairman and organizer at Sessions: C05 Elucidating the Function of Cardiac Genes; C11 Multiple Malformation Syndromes; C20 Neurogenetics, C28 Neurodevelopment.

   European Society of Human Genetics ESHG 2020. Virtual Meeting, June 6-9 2020.
6. Chairman and organizer at Sessions: Polygenic Risks and Me; Metabolic and Mitochondrial Symposium; Novel Therapeutics Symposium;

   European Society of Human Genetics ESHG 2021. Virtual Meeting, June 12-15th-.
7. Chairman and organizer at Sessions: Multiomics for Diagnosis; Novel Therapeutics Symposium;
8. **European Society of Human Genetics ESHG 2022. Vienna,** June 13-17h
9. Chairman at CNS and Sensory Session

   European Society of Gene and Cell Therapy 2022. Virtual Congress October 19th-22nd

Editorial Boards and Activities

2010- **Invited Co-Editor for Brain Pathology (Wiley Online Library).** Minisymposium on X-linked adrenoleukodystrophy. Review series.
2013- Editorial Board (Associate), Journal of Neuroscience Research (Wiley).
2017- Editorial Board, Genética Médica, Medigene Press, Valencia. Spain.
2018- Editorial Board, Neurobiology of Disease (Elsevier).

2020- Invited Editor for Neurobiology of Disease. Special Issue on Lipid Signaling in Neurodegeneration.

Ad-hoc Reviewer

Journals: Nature Neuroscience, Nature Communications, Lancet Neurology, Acta Neuropathologica, American Journal of Human Genetics, Human Molecular Genetics, Brain, Journal of Neuroscience, Experimental Neurology, FASEB Journal, FEBS letters, Neurobiology of Disease, European Journal of Human Genetics, Neuroscience Letters, Molecular Genetics and Metabolism, Biochemical Biophysical Acta, Nucleic Acids Research, Proteomics, Journal of Neuroscience Research, PLoS One, PLoS Medicine, PLoS Genetics, Journal of Lipid Research.

Agencies: Spanish National Institutes for Health ISCIII, Spain; National Agency for Research Evaluation and Foresight (ANEP), Spain; Catalan Agency for Evaluation of Research (AGAUR), Spain; European Leukodystrophy Association (ELA), France; Association Française contre les Myopathies (AFM), France; Fondation pour la Recherche Médicale (FRM), France; Research Council of the University of Leuven, Belgium; The Netherlands Organisation for Scientific Research, The Netherlands Princess Beatrix Funds; Weill Cornell Medicine, NYC, Johns Hopkins University, Baltimore USA.

Teaching and Mentoring Experience

Teaching is the bread and butter of an academic career. In Spain, like in France, the best scientists are freed of teaching obligations at the University, since it is considered that the teaching hours are in detriment of the very competitive research projects.

I wish to highlight the remarkable accomplishment of two of our PhD students, both women, who received the **Extraordinary Thesis prize of the University of Barcelona in 2013** (Dr López-Erauskin) **and 2015** (Dr Morató). We are particularly proud of the work of Dr Lopez-Erauskin, who is deciphering molecular pathogenesis of ALS in Don Cleveland's lab (USCS), and has recently published outstanding articles in Nature Neuroscience and Neuron (PMID:30344044; PMID:30643298). Dr Morató is studying mitochondrial metabolism involvement in anxiety and depression disorders and has currently a first manuscript as leading author under review. Both were extraordinarily productive students in our lab.

I also find remarkable that 9/10 PhD students chose to pursue a career in research and embarked on a postdoctoral fellowship abroad. Our lab is actively collaborating with a former PhD Student, Dr Devesh Pant, who is currently in Nazarko's lab in Georgia. The project deals with the zebrafish modelling of rare leukodystrophies of the ceramide synthesis pathway, which will prepare Dr Pant for an independent career.

I am also glad that of the growing interest that Genomic Medicine has sparked among the students and the clinicians. We have seen this at the classroom and in the multiplication of requests for PhD and postdocs positions, and the high interest on MD/PhD programs with high content on clinical genomics. I am currently mentoring the genomic medicine PhD projects of three bright neurologists and clinical biochemists who represent the next-generation of physicians-scientists.

Mentoring of Junior Faculty

2009- Successful application of Dr Esther Dalfó for a "Miguel Servet Researcher" Contract (tenure-track position equivalent to Assistant Professor, granted by the Spanish National Institutes for Health). Currently Associate Professor at Universitat de Vic, Barcelona

2011- Successful application of Dr Stéphane Fourcade for a "Miguel Servet Researcher" Contract (tenure-track position equivalent to Assistant Professor, granted by the Spanish National Institutes for Health), after six years of postdoc in our lab. Currently Miguel Servet type II (CPII16/00016) at IDIBELL, Barcelona

Mentoring of Postdocs

2007-2009-European Commission fellowship contract (associated to project) to Dr Montse Ruiz, who obtained in 2010 a permanent contract as Senior Research Fellow of the Spanish Research Network for Rare Diseases (CIBERER), up to date.

2008-2010-Sara Borrell fellowship to Dr Nathalie Launay, French postdoc who obtained in 2011 on a permanent contract as Senior Research Fellow of the Spanish Research Network for Rare Diseases (CIBERER), up to date.

2016-2022- Juan de la Cierva I and II fellowships obtained by Dr Edgard Verdura, postdoc who trained as PhD student in France.

Direction of Master Thesis (M Sc)

2008: Jorge Galino Barrés, Master in Neurosciences, University of Barcelona
2008: Jone Lopez-Erauskin, Master in Neurosciences, University of Barcelona
2009: Laia Morató Fornaguera, Master in Neurosciences, University of Barcelona
2012: Pablo Ranea-Robles, Master in Neurosciences, University of Barcelona
2013: Sandra Franco Iborra, Master in Biomedicine, University Pompeu Fabra, Barcelona
2017: Sergio Ramos Sorigué, Master in Bioinformatics, Open University of Catalonia, Barcelona
2017: Arty Mistry, Master in Genetics and Molecular Biology, Saint George University London
2018: Laura Planas, Master in Neuroscience, Autonomous University of Barcelona
2018: Peio Aristu Zabalza, Master in Genetics and Genomics, University of Barcelona
2018: Paula del Pozo, Master in Biochemistry, Molecular Biology and Biomedicine, Autonomous University of Barcelona
2018: Laura Argelich, Master in Biochemistry, Molecular Biology and Biomedicine, Autonomous University of Barcelona
2019: Raquel Ramos, Master in Genetics and Genomics, University of Barcelona
2020: Noelia Carrión, Master in Neurosciences, University of Barcelona

Direction of Doctoral Thesis (PhD)

2005: Carme Camps, PhD in Biochemistry and Molecular Biology. University Autonoma de Barcelona "Biochemical and molecular dissection of the X-ALD mouse models". Co-direction with Marisa Giròs, Institut de Bioquimica Clinica, Barcelona, Spain. 22 October 2005. Cum laude. Currently Associate Researcher at Jenny Taylor lab, Wellcome Centre Human Genetics, University of Oxford, UK.

2011: Jorge Galino, PhD in Biomedicine, University of Barcelona. "Impact of oxidative stress and mitochondrial dysfunction in the energetic homeostasis of adrenoleukodystrophy: therapeutic

implications". 31st May 2011. Excellent Cum laude. Currently Senior Researcher in Leitat Technological Center (Barcelona), after five years as post-doctoral researcher at David Bennett lab, University of Oxford, UK.

2013: Jone López-Erauskin, PhD in Biomedicine, University of Barcelona. "Mitochondrial dysfunction and oxidative stress in the physiopathogenesis of X-adrenoleukodystrophy: from molecular basis to therapy". Excellent Cum laude. 15th March 2013. **Extraordinary PhD Thesis Prize of the University of Barcelona 2013**. Currently at Don Cleveland lab, Ludwig Cancer Research, UCSD La Jolla CA, USA.

2015: Laia Morató Fornaguera, PhD in Biomedicine, University of Barcelona. "Molecular mechanisms underlying mitochondrial dysfunction in X-linked adrenoleukodystrophy: therapeutic potential of SIRT1/PGC-1a activation". 16th March 2015. European Thesis Quality Label. Excellent Cum laude. **Extraordinary PhD Thesis Prize of the University of Barcelona 2015**. Currently post-doctoral researcher at Carmen Sandi lab, EPFL, Lausanne, Switzerland.

2015: Andrea Coppa, PhD in Biomedicine, University of Barcelona. "Modelling X-linked adrenoleukodystrophy in C. Elegans and beyond: a tale of fatty acids and oxidative stress" 2nd October 2015: Excellent Cum Laude. Co-direction with Dr Esther Dalfó. Currently Quality Assurance Manager at BeForPharma, Bari, Italy.

2015: Sanjib Guha, PhD in Biomedicine, University Pompeu Fabra. "A C. elegans model for X-linked Adrenoleukodystrophy: roles of pmp-4 fatty acid transporter in the nervous system", 21st December 2015: *Excellent cum Laude.* Co-direction with Dr Esther Dalfó. Currently post-doctoral research scholar at Pankaj Kapahi Lab, Buck Institute for Research on Aging, Novato, CA, USA.

2016: Patrizia Bianchi, PhD in Biomedicine, University of Barcelona: "Impairment of Mitochondrial Dynamics in X-linked adrenoleukodystrophy", 10th June 2016: Excellent Cum Laude. Co-direction with Dr Stéphane Fourcade. Currently post-doctoral researcher at Albert Quintana lab, Autonomous University of Barcelona.

2017: Pablo Ranea, PhD in Biomedicine, University of Barcelona: "NRF2 and RIP140 as new therapeutic targets for X-linked adrenoleukodystrophy (X-ALD): Control of redox/metabolic homeostasis ", 7th July 2017: Excellent Cum Laude. Co-direction with Dr Stéphane Fourcade. Currently post-doctoral researcher at Sander Houten Lab, Icahn School of Medicine at Mount Sinai, New York, USA.

2018: Janani Parameswaran, PhD in Biomedicine, University Pompeu Fabra: "Cross-talk between inflammation & mitochondria in X-ALD", 9th March 2018: Excellent Cum Laude. Currently post-doctoral researcher at Jie Jiang lab, Emory University, Atlanta, USA.

2018: Devesh Pant, PhD in Biomedicine, University Pompeu Fabra: "Identification of the sphingolipid desaturase DEGS1 as a novel gene for a leukodystrophy with therapeutic hope", 5th October 2018. Excellent Cum Laude. Currently post-doctoral researcher at Taras Nazarko lab, Georgia State University, USA.

2021: Leire Goicoechea, PhD in Biomedicine, University of Barcelona: "Regulation of lipid and redox metabolism in X-linked Adrenoleukodystrophy (X-ALD): therapeutic implications", 16th June 2021. Excellent Cum Laude. Currently post-doctoral researcher at Fernández-Checa lab, CSIC; Barcelona

2022: Agustí Rodriguez-Palmero, MD. PhD in Pediatrics, Obstetrics and Gynecology, University Autonoma of Barcelona: "Diagnostic of brain white matter disorders and novel gene discovery

through next-generation sequencing". 13th May 2022. Excellent Cum Laude. Currently Neuropediatrician consultant at Germans Trias Hospital of Badalona.

2022: Valentina Vélez-Santamaría, MD. PhD in Medicine, University of Barcelona. "Diagnosis of spastic paraplegias and ataxias through next-generation sequencing". 21th June 2022. Excellent Cum Laude. Currently Neurologist consultant at Hospital Universitario de Bellvitge, Barcelona

Doctoral Thesis (PhD) on progress

>2018-PhD thesis start of Laura Planas-Serra, MSc on progress

>2020-PhD thesis start of Lorenzo Torreni MD, Clinical Biochemist, on progress

>2022-PhD thesis start of Ayelen Artuch, MSc, on progress

Graduate and Undergraduate Teaching

2007- to date Master of Biomedicine Program. Translational research in rare diseases: leukodystrophies. University of Barcelona. Faculty of Medicine. 4 h/semester.

2008-2009 Master of Neurosciences Program. Animal models of diseases that affect myelin. Pathophysiology of leukodystrophies and therapeutic advances. University of Barcelona. Faculty of Biology. 4 h/ semester.

2013-to date Master of Biomedicine Program. Model organisms in Biomedical Research. University of Pompeu Fabra, Barcelona. 2h/ semester.

2018, 2019 Undergraduate, Clinical Genetics Course for Medicine and Biology Faculties at University Pompeu Fabra, Associate Professor for Barcelona. 30h/ year.

Outreach Activities

>Crowdfunding campaigns for research on adrenoleukodystrophy (Arnau 97) in 2016, 2017, 2018 and 2019, Taradell, Barcelona.

>Interviews in national press: La Razón (21 Oct 2006); Diario Médico (26 May 2006, 10 Mar 2010, 28 Feb 2013); Correo de Burgos (12 Nov 2010); La Vanguardia (3 Dec 2014 and 19 Jan 2016); Farmaco Salud (16 Aug 2016); Diario de Córdoba (22 Nov 2016); Diario ABC Catalunya (10 Jan 2019).
Extensive coverage on Covid genomics on Science publications, in Catalan and national TV (TV3, Tele5, La Sexta, Antena 3, Canarias7, Euskal Telebista); national radio (Cadena Ser, Onda Regional de Murcia along with major Spanish newspapers (El Periódico, La Vanguardia, Heraldo, La Voz de Galicia, Diario 16, 20 minutos, El Norte de Castilla).
International coverage in ForbesCentroamerica, Televisa Mexico among others (24 Sep 2020).

>National TV interviews: TV1 (22 Jun 2012); TV3 (26 Feb 2013).
Presentation of posters and children games for AFM-Téléthon outreach campaigns on rare diseases from 1999 till 2004, Strasbourg France.

>National Radio interviews: Onda Cero, Antena 3 (17 Dec 2009); Catalunya Radio (9 Dec 2009).

>Interviews in international press: Wired Science “Gene Therapy Halts fatal brain disease” (5 Nov 2009).

>Since 2006, yearly organization and participation in the Families-Scientists 2 day meetings of European Leukodystrophy Association-Spain (ELA-Spain).

> Presentation of posters and children games for AFM-Téléthon outreach campaigns on rare diseases from 1999 till 2004, Strasbourg France.

Research Support

Active Research Support (as Principal Investigator)

Governmental Funding

1-CIBERER Group leader of Unit U759: 2.100.000 € (2008-to date) Program Project of the Spanish National Institutes of Health Carlos III on Rare Diseases Network.

2-AGAUR: Group Leader of Excellence Group of Catalan Research Council SGR 1206; 410.000 € since 2009

3-The Undiagnosed Rare Disease Program of Catalonia (URDCat). Genomics for Undiagnosed Neurological Diseases, Peris SLT002/16/00174. Consortium of 16 PIs, Coordinator: Luis Pérez-Jurado. PI 1.2 M € total, (2017-2020).

4-Precision Medicine for Adrenoleukodystrophy: Identification of Predictive Biomarkers and Neuroinflammation Pathomechanisms.
Spanish National Institute for Health Carlos III. FIS PI17/00916; 372.000 € (2018-2020)

5-Inflammatory Lipid signaling in adrenoleukodystrophy: Multiomics integration for identification of biomarkers and novel therapeutic targets.
CIBERER ACCI 2018: Spanish Network for Rare Diseases; 165.000 € (2019-2020)

6-Genomic Medicine for tackling hyperinflammation in the young and healthy Covid-19 patients-Easi-Genomics EU H2020 grant, 100.000 €. (2020-2021). Leading national Consortium of 8 members.

7-Genomic risk biomarkers in patients with infection of coronavirus SARS-COV**-** Spanish National Institutes for Health Carlos III. FIS PI20/02375, 605.999 € (2020-2021). Consortium of 35 members. Coordinators Pablo Lapunzina and Angel Carracedo.

8-The human gut microbiome as modifying factor of the variable disease severity of X-linked adrenoleukodystophy. Spanish National Institutes for Health Carlos III. FIS: PI19/01008 186.000 € (2020-2023).

9- Genomic medicine for optimizing diagnosis of hereditary spastic paraplegias and ataxias: long-read genomes and functional genomics. Spanish National Institutes for Health Carlos III. FIS: PI20/00758: 205.700 € (2021-2023)

10- Deciphering variable disease expression in adrenoleukodystrophy: toward precision medicine through multiomics and brain organoids. Spanish National Institutes for Health Carlos III. FIS: PI20/00759: 304.315 € (2021-2023)

11- Multiomic medicine for understanding adrenomyeloneuropathy: randomized placebo-controlled phase IIb/III study for clinical and molecular study of dimethyl fumarate action. Spanish National Institutes for Health Carlos III. FIS: ICI21/00085: 898.700 € (2022-2025)

12- The human genetic and immunological determinants of the clinical manifestations of SARS-CoV-2 infection: Towards personalized medicine. HORIZON-HLTH-2021-Disease-04- Total of 6.7 M € (77.345 € for our group) (2022-2026)

13- Immunologic and Predictive Features of MIS-C. NIH-USA. Total of 3,3M € (173.753 for our group) (2022-2027)

14- Methylation repository of Spanish population and improvement of diagnostic of undiagnosed patients (Epi-ENoD)/ CIBERER Total of 60.200 € (2022-2026)

Private Funding

1- Genomic Medicine for diagnosis and gene discovery in neurogenetic disorders; Hesperia Foundation (Royal House of Spain).150.000 € (2015-2020)

2- Crowd-funding campaign Arnau'97 for identification of novel treatments and biomarkers for adrenoleukodystrophy (Barcelona, Spain). 58.000 € (2017-open)

3- Mechanism of action of High Dose Biotin in mouse and cellular models of adrenoleukodystrophy.
MedDay Therapeutics (Paris, France). Research Contract. 380.198 € (2017-2020)

4- Effects of plasma exchange and albumin replacement in Adrenomyeloneuropathy: unicentric, single arm, proof of concept trial. Grifols Albus Award (Barcelona, Spain) 50.000 € (2020-2021)

5- Advanced Genomics and Lipidomics for the discovery of novel genetic causes of movement disorders. La Marató de TV3 (Barcelona, Spain), 399.000 € (2021-2024)

6- Gene therapy by AAV-9 delivery for adrenomyeloneuropathy: preclinical study.
SwanBio Therapeutics (Philadelphia, USA). 252.000 € (2020-2022).

7- Multisystemic inflammatory syndrome associated with COVID-19 in children (MIS-C): genetic, epigenetic and immunopathogenic bases. La Marató de TV3 (Barcelona, Spain), 399.984 € (2021-2024)

8- Inhibitors of Elongase-1 for the treatment of adrenoleukodystrophy, 70.253 € (2022-2023), SANOFI USA

Completed Research Support (as Principal Investigator)

1-Functional analysis and therapy for mouse models for adrenomyeloneuropathy (X-ALD/X-AMN). Association Française contre les Myopathies (AFM). 25.000 € (2003-2004).

2-Characterization and functional analysis of mouse models for X-linked adrenoleukodystrophy (X-ALD) Transcriptome analysis of X-ALD mouse models. European Leukodystrophy Association (ELA). 80.000 € (2003-2004).

3-Insight into the molecular basis in X-adrenoleukodystrophy: Comparative genomics of the peroxisomal proteome and related fatty acid metabolic pathways. Association Française contre les Myopathies (AFM). 42.000€ (2003-2005).

4-Characterization and functional analysis of mouse models for X-linked adrenoleukodystrophy (X-ALD). Transcriptome analysis of X-ALD mouse models. European Leukodystrophy Association (ELA). 35.000€ (2004-2005).

5-X-linked Adrenoleukodystrophy (X-ALD): pathogenesis, animal model and therapy. STREP project LSHM-CT2004-502987 6th PCRD framework, European Union. I.P.: Dr. Aurora Pujol (Coordinator of Work Package 3, 7, 8). 354.000 € (2004-2007).

6-Insight into the physiopathogenesis of X-Adrenomyeloneuropathy. ABCD2 and ABCD4 peroxisomal transporters as therapeutic targets. Association française contre les myopathies (AFM). 60.000€ (2005-2007).

7-Fisiopatogénesis de la Adrenoleucodistrofia ligada al X. La diana terapéutica ABCD2.**:** Instituto de Salud Carlos III PI051118. 181.594€ (2005-2008).

8-Physipathogenesis of X-linked adrenoleukodystrophy. European Leukodystrophy Association (ELA). 181.000€ (2006-2009).

9-Mitochondrial dysfunction and oxidative stress underlying X-linked adrenoleukodystrophy physiopathogenesis. European Leukodystrophy Association (ELA). 2008-040C4. 354.000 € (2008-2011)

10- MYELINET: Myelin orphan diseases in health, COST Action BM0604 from the European Science Foundation (2007-2011).

11-Role of oxidative stress and mitochondria in the physiopathogenesis of X-linked adrenoleukodystrophy. Design of rational pharmacological therapies.
Spanish Institute for Health Carlos III. FIS: PI080991; 244.178€ (2008-2011).

12-Novel Gene Therapy and Pharmacological approaches for adrenomyeloneuropathy. The Myelin Project/ Oliver's Army. 129.240€ (2010-2012).

13-Therapeutic challenge in Leukodystrophies: translational and ethical research towards clinical trials. European Commission Program Grant. LEUKOTREAT: FP7-241622; 390.000€ for Pujol's group (2010-2013).

14-Clinical Trial in adrenomyeloneuropathy: validation of biomarkers of oxidative stress and tolerance of the combination of antioxidants N-acetylcysteine, lipoic acid and vitamin E.
Spanish Ministry for Health and Social Policy, Independent Clinical Investigation Grant EC10-137; 100.000€ (2011-2014).

15-Clinical Trial in adrenomyeloneuropathy: validation of biomarkers of oxidative stress and tolerance of the combination of antioxidants N-acetylcysteine, lipoic acid and vitamin E.
Hesperia Foundation (Royal House of Spain, Queen and King of Spain). 125.000€ (2011-2017)

16-Impact of oxidant stress on mitochondrial biogenesis, energetic homeostasis and proteolysis and its regulation: therapeutic implications in adrenoleukodystrophy
Spanish Institute for Health Carlos III. FIS: PI11/01043; 278.213 € (2012-2014).

17-Advanced chemical technologies and predictive experimental models leading to new treatments against rare diseases**.** EuroTransBio 7th Call European Commission ETB-2012-18; 320.250 € (2012-2015). Project Coordinator: Minoryx Therapeutics

18-Functional proteomics approach towards deciphering molecular pathology of adrenomyeloneuropathy
Oliver's Army grant. London UK, 106.000 € (2013-2014),

19- Pharmacological strategies for myelin regeneration and axon protection in X-linked Adrenoleukodystrophy.
WalkOnProject. Bilbao, Spain 198.000 € (2013-2014),

20- Phase II international, multicentric clinical trial with MD1003 for adrenomyeloneuropathy patients.
MedDay Therapeutics Research Contract, Paris France. 97.000 € (2014-2017).

21- Systems Biomedicine for identification of novel genes and modeling cortical motorneuron diseases. Marató de TV3 Catalonia, Spain (345/C/2014) 398.000 € (2014-2018)

22- Bioenergetics regulation as therapeutic target for adrenoleukodystrophy: dissection of E2F1/RIP140 pathways and protection of the mitochondria in the murine model of X-ALD.
Spanish Institute for Health Carlos III. FIS: PI14/00410; 304.315 € (2015-2017).

23- Epigenetic and environmental factors bracing cognitive impairment and late-onset depression in elderly and early stages of Alzheimer disease.
Spanish Institute for Health Carlos III.; Total funding 605.000 € for a consortium of 10 research groups (2015-2018).

24-Efficacy of pioglitazone administered to patients with adrenomyeloneuropathy: a phase II, single-arm, multicentric trial.
Spanish Institute for Health Carlos III. ICI14/00076; 157.300 € (2015-2019).

Clinical Trials

I generated the preclinical data, designed, carried out and obtained the funding for the following phase II clinical trials for adrenoleukodystrophy:

1- Validation of oxidative stress biomarkers, efficacy and tolerance of the combination N-acetyl cysteine, alpha lipoic acid and vitamin E. Phase IIb
Principal Investigator and Promotor: Aurora Pujol. Co-investigator: Carlos Casasnovas, Neurology Department, Bellvitge Hospital. 2012-2015
Registered EUDRACT Number: 2010-024084-40. ClinicalTrials.gov: NCT01495260

2- Efficacy of pioglitazone administered to patients with adrenomyeloneuropathy: a phase II, single-arm, multicentric trial. Phase II/III
Trial Coordinator: Aurora Pujol. Co-investigators: Carlos Casasnovas, Neurology Department, Bellvitge Hospital and Adolfo Lopez de Munain, Donostia Hospital, 2015-2018.

Registered EUDRACT Number: 2011-006113-34

3- MD1003 for adrenomyeloneuropathy: an international, multicentric clinical trial. Phase II/III
Sponsor: MedDay Pharmaceuticals, France.
Trial Coordinator: Patrick Aubourg, INSERM Paris
Principal Investigator: Aurora Pujol. Co-investigator: Carlos Casasnovas, Neurology Department, Bellvitge Hospital 2014-2016.
Registered EUDRACT Number: 2014-000698-38. ClinicalTrials.gov: NCT02961803

4- Effects of plasma exchange and albumin replacement in Adrenomyeloneuropathy: unicentric, single arm, proof of concept trial
Principal Investigator: Aurora Pujol. Co-investigator: Carlos Casasnovas, Neurology Department, Bellvitge Hospital 2020-2021.
Registered EUDRACT Number: 2019-004733-17. Sponsor Protocol Number: XAMNPEAP2019

5- Randomized placebo-controlled phase IIb/III study for clinical and molecular study of dimethyl fumarate action.
Principal Investigator: Aurora Pujol. Co-investigator: Carlos Casasnovas, Neurology Department, Bellvitge Hospital 2022-2025.

Patents (Active)

1. Combination of **N-acetylcysteine and lipoic acid** for the treatment of a disease with axonal damage and concomitant oxidative lesions
Inventors: Aurora Pujol.
Request number: P201030728
Priority country and date: Spain, May 17th, 2010
Extended to countries: US
Entity owner: IDIBELL PCT ES11/070195/ Minoryx Therapeutics, In exploitation

2. **Pioglitazone** for use in the treatment of adrenoleukodystropy
Inventors: Aurora Pujol.
Request number: P8278EP00.
Priority country and date: Spain, March 23th,2012
Extended to countries: US
Licensed to Minoryx Therapeutics Inc, in exploitation

3. Specific **mTOR inhibitors** in the treatment of X-linked Adrenoleukodystrophy.
Inventors: Aurora Pujol, Erwin Knecht.
Request number: EP14382353.2
Priority country and date: Spain, September 19th, 2014
PCT (PCT/EP2015071563).
Entity owner: IDIBELL/ICREA/Principe Felipe Research Center, Spain
Extended to countries: EU, US
Licensed to SOMBiotech, April 12th, 2017

4. Methods and compositions for the **Diagnosis and for the Treatment** of adrenoleukodystrophy
Inventors: Aurora Pujol, Adolfo López de Munain, Manuel Portero, Reinald Pamplona.

Request number: EP15382366
Priority country and date: Spain, July 14th, 2015
Extended to countries: EU, US
Entity owner: IDIBELL/ ICREA/ Biodonostia/ Universitat de Lleida
Licensed to SOMBiotech, April 12th, 2017

5. **NRF2 activation** for the prevention and/or treatment of axonal degeneration.
Inventors: Aurora Pujol, Stephane Fourcade.
Priority country and date: USA
Extended to countries: US15/957,601.
Entity owner: IDIBELL/ ICREA

Orphan Drug Designations

1- **Pioglitazone** for the treatment of adrenoleukodystrophy.
European Medicines Agency EU/3/14/1245, on 19/02/2014

2- **Temsirolimus** for the treatment of Adrenoleukodystrophy.
European Medicines Agency EU/3/16/1669, on 15/06/ 2016.

3- **Dimethyl fumarate** for the treatment of Adrenoleukodystrophy
European Medicines Agency EU/3/19/2236, on 09/02/2020

4- **Fingolimod** for the treatment of Adrenoleukodystrophy
European Medicines Agency EU/3/21/2520 on 12/11/ 2020

Selected Peer-Reviewed Publications

Citation Metrics

Total Articles in Publication List:	**150**
Total Articles as Senior Author:	**63**
Total Impact Factor	**675**
Sum of the Times Cited:	**>20.000**
Average Citation per Article:	**102**
H index:	**49**
I10 index:	**116**

ORCID ID: https://orcid.org/0000-0002-9606-0600

Google Scholar: https://scholar.google.com/citations?user=RN4lgoYAAAAJ

***last or corresponding author, & featured**

1. Isolation of a fully infectious variant of parvovirus H-1 supplanting the standard strain in human cells.
H Faisst S, Faisst SR, Dupressoir T, Plaza S, **Pujol A**, Jauniaux JC, Rhode S, Rommelaere J.
J Virol.1995;69: 4538-4543. IF: 5.8 (D1 Virology 2/25).

2. Sequencing analysis of a 40.2 kb fragment of yeast chromosome X reveals 19 open reading frames including URA2 (5'end), TRK1, PBS2, SPT10, GCD14, RPE1, PHO86, NCA3, ASF1, CCT7, GZF3, two tRNA genes, three remnant delta elements and a Ty4 transposon.
Cziepluch C, Kordes E, **Pujol A**, Jauniaux JC.
Yeast.1996; 12: 1471-1474. IF: 2.2 (Q1 Biochemistry & Applied Microbiology 21/123).

3. Complete nucleotide sequence of Saccharomyces cerevisiae chromosome X.
Galibert F, Alexandraki D, Baur A, Boles E, Chalwatzis N, Chuat JC, Coster F, Cziepluch C, De Haan M, Domdey H, Durand P, Entian KD, Gatius M, Goffeau A, Grivell LA, Hennemann A, Herbert CJ, Heumann K, Hilger F, Hollenberg CP, Huang ME, Jacq C, Jauniaux JC, Katsoulou C, Karpfinger-Hartl L, **Pujol A** and 32 other coauthors.
EMBO J. 1996; 15: 2031-2049. IF: 13.17 (D1 Biochemistry and molecular biology 9/295).

4. The nucleotide sequence of Saccharomyces cerevisiae chromosome XV.
Dujon B, K. Albermann, M. Aldea, D. Alexandraki, W. Ansorge, J. Arino, V. Benes, C. Bohn, M. Bolotin-Fukuhara, R. Bordonné, **Pujol A** [.....], G. Volckaert, H. Voss, R. Wambutt, H. Wedler, S. Wiemann, B. Winsor, K. H. Wolfe, A. Zollner, E. Zumstein, K. Kleine.
Nature 1997; 387(6632 Suppl):98-102. IF: 42.35 (D1 Biochemistry and molecular biology 9/295).

5. Sequence and analysis of a 36.2 kb fragment from the right arm of yeast chromosome XV reveals 19 open reading frames including SNF2 (5' end), CPA1, SLY41, a putative transport ATPase, a putative ribosomal protein and an SNF2 homologue.
Poirey R, Cziepluch C, Tobiasch E, **Pujol A**, Kordes E, Jauniaux JC.
Yeast.1997; 13: 479-482. IF: 2.2 (Biochemistry & Applied Microbiology 21/123).

6. Inhibition of parvovirus minute virus of mice replication by a peptide involved in the oligomerization of nonstructural protein NS1.
Pujol A, Deleu L, Nüesch JPF, Jauniaux JC, Rommelaere J.
J Virol.1997; 71: 7393-7403. IF: 5.8 (Virology 2/25).

7. Activation of promoter P4 of the autonomous parvovirus minute virus of mice at early S phase is required for productive infection.
Deleu L, **Pujol A**, Faisst S, Rommelaere J.
J Virol. 1999; 73:3877-85. IF: 5.9. (Virology 3/26).

8. Characterization of the Adrenoleukodystrophy-Related (ABCD2) gene promoter: Inductibility by Retinoic acid and Forskolin.
Pujol A, Troffer-Charlier N, Metzger E, Chimini G, Mandel JL.
Genomics. 2000; 70:131-139. IF: 3.4 (Genetics and heredity 29/114).

9. Inhibition of Transcription-regulating Properties of Nonstructural Protein 1 (NS1) of Parvovirus Minute Virus of Mice by a dominant-negative mutant form of NS1.
Pujol A, Deleu L, Nüesch JPF, Rommelaere J. (First and second author contributed equally).
J Gen Virol. 2001; 82: 1917-1927. IF: 3.2. (Biotechnology and applied microbiology 18/131).

10. ABCD1 Mutations and the X-linked Adrenoleukodystrophy Mutation Database: Role in Diagnosis and clinical correlation.
Kemp S, **Pujol A**, Waterham HR, van Geel BM, Boehm CD, Raymond GV, Cutting GR, Wanders RJA and Moser HW. Review.
Human Mutation. 2001;18: 499-515. IF: 6.1 (Genetics and heredity 12/113).

11. Late onset neurological phenotype of the X-ALD gene inactivation in mice: a mouse model for adrenomyeloneuropathy.
Pujol A*, Hindelang C, Callizot N, Bartsch U, Schachner M, Mandel JL.
Hum Mol Genet. 2002; 1:11(5):499-505. IF: 8.7 (Biochemistry and molecular biology 17/266). Cited by 189

12. Mouse liver PMP70 and ALDP: homomeric interactions prevail in vivo.
Guimaraes CP, Domingues P, Aubourg P, Fouquet F, **Pujol A**, Jimenez-Sanchez G, Sa-Miranda C, Azevedo JE**.**
Biochim Biophys Acta. 2004; 689:235-43. IF: 3.04 (Biochemistry and molecular biology 97/261).

13. Functional overlap between ABCD1 (ALD) and ABCD2 (ALDR) transporters: a therapeutic target for X-adrenoleukodystrophy.
Pujol A*, Ferrer I, Camps C, Metzger E, Hindelang C, Callizot N, Ruiz M, Pampols T, Giros M, Mandel JL.
Hum Mol Gen. 2004;13:2997-3006. IF: 7.8 (Biochemistry and molecular biology 23/261).

14. Inactivation of the peroxisomal ABCD2 transporter in the mouse leads to late-onset ataxia involving mitochondria, Golgi and endoplasmic reticulum damage.
Ferrer I, Kapfhammer JP, Hindelang C, Kemp S, Troffer-Charlier N, Broccoli V, Callyzot N, Mooyer P, Selhorst J, Vreken P, Wanders RJ, Mandel JL, **Pujol A***.
Hum Mol Genet. 2005; 14:3565-77. IF: 7.7 (Biochemistry and molecular biology 22/261).

15. The evolutionary origin of peroxisomes: an ER-peroxisome connection.
Schluter A, Fourcade S, Ripp R, Mandel JL, Poch O, **Pujol A***.
Mol Biol Evol. 2006; 23:838-45. IF: 7.9 (Biochemistry and molecular biology 25/262).

16. PeroxisomeDB: a database for the peroxisomal proteome, functional genomics and disease.
Schluter A, Fourcade S, Domènech-Estévez E, Gabaldón T, Huerta-Cepas J, Berthommier G, Ripp R, Wanders RJ, Poch O, **Pujol A***.
Nuc Acids Res. 2007;35:D815-22. IF: 6.9 (Biochemistry and molecular biology 29/263).

17. The role of peroxisomal ABC transporters in the mouse adrenal gland: the loss of Abcd2 (ALDR), Not Abcd1 (ALD), causes oxidative damage.
Lu JF, Barron-Casella E, Deering R, Heinzer AK, Moser AB, deMesy Bentley KL, Wand GS, C McGuinness M, Pei Z, Watkins PA, **Pujol A**, Smith KD, Powers JM.
Lab Invest. 2007; 87:261-72. IF: 4.4 (D1, Pathology 5/66).

18. Steroid hormones control circadian Elovl3 expression in mouse liver.
Brolinson A, Fourcade S, Jakobsson A, **Pujol A**, Jacobsson A.
Endocrinology. 2008; 149:3158-66. IF: 4.9 (Endocrinology and metabolism 20/93).

19. Early oxidative damage underlying neurodegeneration in X-adrenoleukodystrophy.
Fourcade S, López-Erauskin J, Galino J, Duval C, Naudi A, Jove M, Kemp S, Villarroya F, Ferrer I, Pamplona R, Portero-Otin M, **Pujol A*.**
Hum Mol Genet. 2008; 17:1762-73. IF: 7.2 (Genetics and heredity 13/138).
&Featured by Faculty of 1000 (Professor George Perry, Dean of Texas University).

20. A key role for the peroxisomal ABCD2 transporter in fatty acid homeostasis.
Fourcade S, Ruiz M, Camps C, Schlüter A, Houten SM, Mooyer PA, Pàmpols T, Dacremont G, Wanders RJ, Giròs M, **Pujol A***.
Am J Physiol Endocrinol Metab. 2009;296:E211-21. IF: 4.3 (Endocrinology and metabolism 22/105).

21. Insulin-like growth factor-1 and neurotrophin-3 gene therapy prevents motor decline in an X-linked adrenoleukodystrophy mouse model.
Mastroeni R, Bensadoun JC, Charvin D, Aebischer P, **Pujol A**, Raoul C.
Ann Neurol. 2009;66:117-22. IF: 9.317 (Clinical Neurology 3/167).

22. ABCD2 is abundant in adipose tissue and opposes the accumulation of dietary erucic acid (C22:1) in fat.
Liu J, Sabeva NS, Bhatnagar S, Li X, **Pujol A**, Graf GA.
J Lipid Res. 2010;51:162-8. IF: 6.115 (Q1 Biochemistry and molecular biology 42/286).

23. PeroxisomeDB 2.0: an integrative view of the global peroxisomal metabolome.
Schlüter A, Real-Chicharro A, Gabaldón T, Sánchez-Jiménez F**, Pujol A***.
Nucleic Acids Res. 2010;38:D800-5. IF: 7.836 (Q1, Biochemistry and molecular biology 30/286).

24. Valproic acid induces pharmacological antioxidant effects in X-adrenoleukodystrophy.
Fourcade S, Ruiz M, Guilera C, Hahnen E, Brichta L, Naudi A, Portero-Otín M, Dacremont G, Cartier N, Wanders RJ, Kemp S, Mandel JL, Wirth B, Pamplona R, Aubourg P and **Pujol A*.**
Hum Mol Genet. 2010 ;19:2005-14. IF: 8.058 (D1, Genetics and heredity 13/156).

25. Current and future pharmacological treatment strategies in X-linked Adrenoleukodystrophy.
Berger J, **Pujol A**, Aubourg P, Forss-Petter S.
Brain Pathol. 2010;20:845-56. Review. IF: 4.741 (D1, Pathology: 6/76).

26. Pathomechanisms underlying X-Adrenoleukodystrophy: a three-hit hypothesis.
Singh I, **Pujol A***.
Brain Pathol. 2010;20:838-44. Review. IF: 4.741 (D1, Pathology: 6/76).

27. General Aspects and neuropathology of X-linked Adrenoleukodystrophy.
Ferrer I, Aubourg P, **Pujol A*.**
Brain Pathol. 2010;20:817-30. Review. IF: 4.741 (D1, Pathology: 6/76).

28. Schilder's disease a heterogenous group of disorders known as X-linked adrenoleukodystrophy.

Ferrer I, **Pujol A*.**
Brain Pathol. 2010;20:815-16. Review. IF: 4.741 (D1, Pathology: 6/76).

29. Invariant NKT cells in adrenoleukodystrophy patients and mice.
Gautron AS, Giquel B, Beaudoin L, Autrusseau E, Speak A, Platt F, Kemp S, **Pujol A**, Aubourg P, Lehuen A.
J Neuroimmunol. 2010; 229:204-11. IF: 2.901 (Q2 Inmunology 65/134).

30. Staging Anti-Inflammatory Therapy in Alzheimer's Disease.
Lichtenstein MP, Carriba P, Masgrau R, **Pujol A**, Galea E.
Front. Ag. Neurosci. 2010; Oct 25;2:142.

31. Antioxidants halt axonal degeneration in a mouse model of X-linked adrenoleukodystrophy.
López-Erauskin J, Fourcade S, Galino J, Ruiz M, Schlüter A, Martínez JJ, Pamplona R, Naudi A, Portero-Otin M, Ferrer I and **Pujol A***.
Ann Neurol 2011; 70:84-92. IF: 11.089 (D1 Clinical Neurology 3/192).

32. Phylogenomic evidence for a Myxococcal contribution to the mitochondrial fatty acid beta-oxidation. Schlüter A, Ruiz-Trillo I, **Pujol A***.
PloS ONE 2011; 6(7):e21989. IF: 4.092 (Q1, biology:12/85).

33. Oxidative damage compromises energy metabolism in the axonal degeneration mouse model of X-adrenoleukodystrophy.
Galino J, Ruiz M, Fourcade S, Schlüter A, López-Erauskin J, Guilera C, Jove M, Naudi A, García-Arumi E, Andreu AL, Starkov A, Pamplona R, Ferrer I, Porter-Otin M, **Pujol A***.
Antioxid Redox Signal 2011 15:2095-2107. IF: 8.456. (D1, Endocrinology and metabolism 5/122).

34. Amyloid generation and dysfunctional immunoproteasome activation with disease progression in animal model of familial Alzheimer Disease.
Aso E, Lomonio S, Joda L, Lopez I, Carmona M, Fernandez N, Moreno J, 21ueves S**, Pujol A,** Pamplona R, Portero-Otín M, Martín V, Diaz M, Ferrer I**.**
Brain Pathol, 2012 Sep;22(5):636-53. IF: 4.739 (Q1, Pathology: 9/77).

35. JNK/ERK/FAK mediate pro-migratory actions of basic fibroblast growth factor in astrocytes via MCP1 and COX.
Lichtenstein MP, Madrigal JM, **Pujol A** and Galea E.
Neurosignals, 2012;20(2):86-102. IF: 2.561 (Q3 Neurosciences 146/252).

36. Oxidative stress underlying axonal degeneration in adrenoleukodystrophy: A paradigm for multifactorial neurodegenerative diseases?
Galea E, Launay N, Portero-Otin M, Ruiz M, Pamplona R, Aubourg P, Ferrer I and **Pujol A***.
Biochim Biophys Acta-Mol Basis Dis. 2012 Sep;1822(9):1475-88. Invited Review IF: 4.910 (Q1, Biochemistry and Molecular Biology 58/290). Cited by 90
&Featured by Faculty of 1000 (Professor Mike Johnston, Kennedy Krieger Institute and John Hopkins University Hospital)

37. Functional genomic analysis unravels a metabolic-inflammatory interplay in adrenoleukodystrophy.
Schlüter A, Espinosa L, Fourcade S, Galino J, López E, Ilieva E, Morató L, Asheuer M, Cook T, McLaren A, Reid J, Kelly F, Bates S, Aubourg P, Galea E, **Pujol A***.
Hum Mol Genet. 2012 Mar 1;21(5):1062-77. IF: 8.058 (D1, Biochemistry and Molecular Biology 28/290).

38. Bezafibrate lowers very long-chain fatty acids in X-linked adrenoleukodystrophy fibroblasts by inhibiting fatty acid elongation.
Engelen M, Schackmann MJ, Ofman R, Sanders RJ, Dijkstra IM, Houten SM, Fourcade S, **Pujol A**, Poll-The BT, Wanders RJ, Kemp S.
J Inherit Metab Dis. 2012 Nov;35(6):1137-45. IF: 4.070 (Q1, Genetics & Heredity 40/161).

39. The absence of ABCD2 reveals a novel role for peroxisomes in metabolic responses to dietary lipids.
Liu J, Liang S, Liu X, Brown A, Newman KE, Bhatnagar S, Li X. **Pujol A**, Graf G.
J Lipid Res 2012 Jun;53(6):1071-9. IF: 4.386 (Q1, Biochemistry and molecular biology 69/290).

40. Neurochemistry and the non-motor aspects of Parkinson Disease.
Ferrer I, López-González I, Carmona M, Dalfó E, **Pujol A** and Martínez A.
Neurobiol Dis 2012, Jun;45 (3) 508-26. Invited Review. IF: 5.624 (Q1, Neurosciences 34/252).

41. ATP and noradrenaline activate CREB in astrocytes via non-canonical Ca2+ and cyclic AMP independent pathways.
Carriba P, Pardo L, Parra-Damas A, Lichtenstein MP, Saura CA, **Pujol A**, Masgrau R, Galea E.
Glia 2012, Sep;60(9):1330-44. IF: 5.066 (Q1 Neurosciences 43/252).

42. Bezafibrate administration improves behavioral deficits and tau pathology in P301S mice.
Dumont M, Stack C, Elipenahli C, Jainuddin S, Gerges M, Starkova N, Calingasan NY, Yang L, Tampellini D, Starkov A A, Chan RB, Di Paolo G, **Pujol A,** Beal F.
Hum Mol Genet 2012 Dec 1;21(23):5091-105. IF: 8.058 (D1, Biochemistry and Molecular Biology 28/290).

43. Oxidative stress modulates mitochondrial failure and cyclophilin D function in X-linked adrenoleukodystrophy.
López-Erauskin J, Galino J, Bianchi P, Fourcade S, Andreu AL, Ferrer I, Muñoz-Pinedo C, and **Pujol A*.**
Brain, 2012 Dec;135 (Pt12):3584-98. IF: 10.870 (D1 Neurosciences 10/252).
& Scientific commentary by Marc Fransen, Brain 2012 Dec;135 (Pt12):3584-98.
&Comment by Paolo Bernardi on EMBO Reports, EMBO Rep. 2013 Jan;14(1):7-9

44. Oxidative stress regulates UPS and immunoproteasome functioning in a mouse model of X-adrenoleukodystrophy.
Launay N, Ruiz M, Fourcade S, Schlüter A, Guilera C, Ferrer I, Knecht E and **Pujol A*.**
Brain, 2013 Mar;136(Pt 3):891-904. IF: 10.870 (D1 Neurosciences 10/252).

45. Cyclophilin D as a potential target for antioxidants in neurodegeneration: the X-ALD case.
López-Erauskin J, Ferrer I, Galea E, **Pujol A*.**
Biol Chem, 2013 May; 394(5):621-629. Review.
IF: 2.683 (Q3 Biochemistry and Molecular Biology 158/290).

46. Glutathione imbalance in patients with X-linked adrenoleukodystrophy.
Petrillo S, Piemonte F, Pastore A, Tozzi G, Aiello C, **Pujol A**, Cappa M, Bertini E.
Mol Genet Metab 2013 Aug;109(4):366-70. IF: 2.834 (Q2 Genetics & Heredity 72/161).

47. Functional genomics unravels dysregulation of cortical olfactory receptors in Parkinson disease: novel putative chemoreceptors in the human brain.
Garcia-Esparcia P, Schlüter A, Carmona M, Moreno J, Ansoleaga B, Gustincich S, Ferrer I, and **Pujol A.**
J Neuropathol Exp Neurol, 2013 Jun ;72(6) :524-539. IF: 4.345 (Q1 Pathology 12/77; Neurosciences 60/252).

48. Impaired mitochondria oxidative phosphorylation in the peroxisomal disease X-linked adrenoleukodystrophy.
López-Erauskin J, Galino J, Ruiz M, Cuezva JM, Fabregat I, Pamplona R, Ferrer I, Portero-Otín M, Fourcade S, and **Pujol A*.**
Human Mol Genet, 2013 Aug 15;22(16):3296-305. IF: 7.692 (D1, Biochemistry and Molecular Biology 28/290).
& Issue Cover August 15th. & Featured in MDLinx http://www.mdlinx.com/

49. Pioglitazone halts axonal degeneration in a mouse model of X-adrenoleukodystrophy.
Morató L, Galino J, Ruiz M, Calingasan N, Starkov AA, Dumont M, Naudí A, Martinez JJ, Aubourg P, Portero-Otín M, Pamplona R, Galea E, Beal F, Ferrer I, Fourcade S and **Pujol A*.**
Brain, 2013 Aug;136(Pt 8):2432-43. IF: 9.915 (D1 Neurosciences 10/252).
& Research Highlights in Nature Rev Neurology, July 16 2013 doi:10.1038/nrneurol.2013.141
& Scientific Commentary by Carlos Moraes, Brain. 2013 Aug;136(Pt 8):2339-41
& Press release of the European Human Genetics Conference on 08 June 2013, Paris France

50. Histone deacetylase inhibitor upregulates peroxisomal fatty acid oxidation and inhibits apoptotic cell death in abcd1-deficient glial cells.
Singh J, Khan M, **Pujol A**, Baarine M, Singh I.
PloS One. 2013 Jul 26;8(7):e70712. IF: 3.730 (Q1 Multidisciplinary Sciences 7/56).

51. Mitochondrial dysfunction and oxidative damage cooperatively fuel axonal degeneration in X-linked adrenoleukodystrophy.
Fourcade S, López-Erauskin J, Ruiz M, Ferrer I, **Pujol A*.**
Biochimie. 2014 Mar;98:143-9. Invited Review. IF: 3.142 (Q2 Biochemistry and Molecular Biology 119/290).

52. PGC-1° overexpression exacerbates b-amyloid and tau deposition in a transgenic mouse model of Alzheimer's disease.

Dumont M, Stack C, Elipenahli C, Jainuddin S, Launay N, Gerges M, Starkova N, Starkov AA, Calingasan NY, Tampellini D, **Pujol A**, Beal MF.
FASEB J. 2014 Apr 28(4):1745-55. IF: 5.704 (Q1 Biochemistry & Molecular Biology 44/290).

53. PDR-1/hParkin negatively regulates the phagocytosis of apoptotic cell corpses in C.elegans.
Cabello J, Sämann J, Gómez-Orte E, Erazo T, Coppa A, **Pujol A**, Büssing I, Schulze B, Lizcano J, Ferrer I, Baumeister R, Dalfo E.
Cell Death Dis 2014 Mar 13;5:e1120. IF: 6.044 (Q1 Cell Biology 35/184).

54. Methylene blue upregulates Nrf2/ARE genes and prevents tau-related neurotoxicity.
Stack C, Jainuddin S, Elipenahli C, Gerges M, Starkova N,. Starkov AA, Portero-Otin M, Launay N, **Pujol A,** Kaidery, N; Thomas B; Beal MF, Dumont M.
Hum Mol Genet 2014 Jul 15;23(14):3716-32. IF: 7.692 (D1, Biochemistry and Molecular Biology 28/290).

55. Mitochondrial dysfunction in white matter disorders.
Morató L, Bertini E, Verrigni G, Ardissone A, Ruiz M, Ferrer I, Uziel G and **Pujol A*.**
Glia. 2014 Nov;62(11):1878-94. IF: 5.066 (Q1 Neurosciences 43/252).

56. Pharmacological inhibition of Poly(ADP-Ribose) Polymerases improves fitness and mitochondrial Function in Skeletal Muscle.
Pirinen E, Canto C, Jo YS, Morató L, Zhang H, Menzies K, Williams EG, Mouchiroud L, Moullan N, Hagberg C, Li W, Timmers S, Imhof R, Verbeek J, **Pujol A**, van Loon B, Viscomi C, Zeviani M, Schrauwen P, Sauve A, Schoonjans K, Auwerx J.
Cell Metab. 2014 Jun 3;19(6):1034-41. IF: 16.747 (D1 Cell Biology 8/185).

57. ABCD2 alters peroxisome proliferator-activated receptor α signaling in vitro, but does not impair responses to fenofibrate therapy in a mouse model of diet-induced obesity.
Liu X, Liu J, Liang S, Schlüter A, Fourcade S, Aslibekyan S, **Pujol A**, Graf GA. .
Mol Pharmacol. 2014 Nov;86(5):505-13. IF: 4.120 (Q1 Pharmacology & Pharmacy 34/256).

58. Improvement of the Rett syndrome phenotype in a MeCP2 mouse model upon treatment with levodopa and a dopa-decarboxylase inhibitor.
Szczesna K, de la Caridad O, Petazzi P, Soler M, Roa L, Saez MA, Fourcade S, **Pujol A**, Artuch-Iriberri R, Molero-Luis M, Vidal A, Huertas D, Esteller M.
Neuropsychopharmacology. 2014. Nov;39(12):2846-56. IF: 8.678. (D1 Neurosciences 17/252).

59. Autophagy induction halts axonal degeneration in a mouse model of X-adrenoleukodystrophy.
Launay N, Aguado C, Fourcade S, Ruiz M, Grau L, Riera J, Guilera C, Giròs M, Ferrer I, Knecht E, **Pujol A*.**
Acta Neuropathol. 2015 Mar;129(3):399-415. IF: 11.360 (D1 Neurosciences 10/256).
& XIV National Research Prize of the College of Physicians of Cordoba, November 2016

60. Mitochondrial Disease Sequence Data Resource (MseqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities.

Falk MJ, Shen L, Gonzalez M, Leipzig J, Lott MT, Stassen AP, Diroma MA, Navarro-Gomez D, Yeske P, Bai R, Boles RG, Brilhante V, Ralph D, DaRe JT, Shelton R, Terry SF, Zhang Z, Copeland WC, van Oven M, Prokisch H, Wallace DC, Attimonelli M, Krotoski D, Zuchner S, Gai X;. **Pujol A** and other 40 co-authors.

Mol Genet Metab. 2015 Mar;114(3):388-396. IF: 3.093 (Q2 Genetics & Heredity 64/166).

61. Deregulation of purine metabolism in Alzheimer's disease.

Ansoleaga B, Jové M, Schlüter A, Garcia-Esparcia P, Moreno J, **Pujol A**, Pamplona R, Portero-Otín M, Ferrer I.

Neurobiol Aging. 2015 Jan;36(1):68-80. IF: 5.153 (Q1 Neurosciences 36/256).

62. Astrocytes and mitochondria from adrenoleukodystrophy protein (ABCD1)-deficient mice reveal that the adrenoleukodystrophy-associated very long-chain fatty acids target several cellular energy-dependent functions.

Kruska N, Schönfeld P, **Pujol A,** Reiser G.

Biochim Biophys Acta. 2015 Jan 10;1852(5):925-936. IF: 5.089 (Q1 Biochemistry & Molecular Biology 54/291).

63. Neuroinflammatory Signals in Alzheimer Disease and APP/PS1 Transgenic Mice:Correlations With Plaques, Tangles, and Oligomeric Species.

López-González I, Schlüter A, Aso E, Garcia-Esparcia P, Ansoleaga B, Llorens F, Carmona M, Moreno J, Fuso A, Portero-Otin M, Pamplona R, **Pujol A,** Ferrer I.

J Neuropathol Exp Neurol. 2015 Apr;74(4):319-44. IF: 3.432 (Q1 Clinical Neurology 28/194).

64. Activation of Sirtuin 1 as therapy for the peroxisomal disease adrenoleukodystrophy.

Morató L, Ruiz M, Boada J, Calingasan NY, Galino J, Guilera C, Jové M, Naudí A, Ferrer I, Pamplona R, Portero-Otín M, Serrano M, Beal MF , Fourcade S and **Pujol A*.**

Cell Death Differ. 2015 Mar 27. IF: 8.218 (D1 Biochemistry & Molecular Biology 23/289).

65. Oxidative stress, mitochondrial and proteostasis malfunction in adrenoleukodystrophy: a paradigm for axonal degeneration.

Fourcade S, Ferrer I, **Pujol A*.**

Free Radic Biol Med. 2015 Jun 11. Pii: S0891-5849(15)00264-6 Invited Review. IF: 5.784 (Q1 Biochemistry & Molecular Biology 37/289).

66. Altered glycolipid and glycerophospholipid signaling drive inflammatory cascades in adrenomyeloneuropathy.

Ruiz M, Jové M, Schlüter A, Casasnovas C, Villarroya F, Guilera C, Ortega FJ, Naudí A, Pamplona R, Gimeno R, Fourcade S, Portero-Otín M, **Pujol A*.**

Hum Mol Genet. 2015 Dec 15;24(24):6861-76. IF: 5.986 (D1 Genetics & Heredity 16/164).

67. Novel Therapeutic Targets and Drug Candidates for Modifying Disease Progression in Adrenoleukodystrophy.

Pujol A.

Endocr Dev. 2016;30:147-60.

68. Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition).
Klionsky DJ, Abdelmohsen K, Abe A, Abedin MJ, Abeliovich H, Acevedo Arozena A, Adachi H, Adams CM, Adams PD, Adeli K, Adhihetty PJ, Adler SG, Agam G, Agarwal R, **Pujol A** and 94 coauthors. **Autophagy**. 2016;12(1):1-222. IF: 8.593 (Q1 Cell Biology 22/190).

69. Targeted activation of CREB in reactive astrocytes is neuroprotective in focal acute cortical injury.
Pardo L, Schlüter A, Valor LM, Barco A, Giralt M, Golbano A, Hidalgo J, Jia P, Zhao Z, Jové M, Portero-Otin M, Ruiz M, Giménez-Llort L, Masgrau R, **Pujol A,** Galea E.
Glia. 2016 May;64(5):853-74. IF: 6.200 (D1 Neurosciences 23/259).

70. Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H/SPG35 in 4 families.
Soehn AS, Rattay TW, Beck-Wödl S, Schäferhoff K, Monk D, Döbler-Neumann M, Hörtnagel K, Schlüter A, Ruiz M, **Pujol A**, Züchner S, Riess O, Schüle R, Bauer P, Schöls L.
Neurology. 2016 Jul 12;87(2):186-91. IF: 8.320 (D1 Clinical Neurology 9/194).

71. A view on clinical genetics and genomics in Spain: of challenges and opportunities.
Pàmpols T, Ramos FJ, Lapunzina P, Gozalo-Salellas I, Pérez-Jurado LA, **Pujol A*.**
Mol Genet Genomic Med. 2016 Jul 18;4(4):376-91.

72. De novo loss of function mutations in KIAA2022 are associated with epilepsy and neurodevelopmental delay in females.
Webster R, Cho MT, Retterer K, Millan F, Nowak C, Douglas J, Ahmad A, Raymond GV, Johnson MR, **Pujol A**, Begtrup A, McKnight D, Devinsky O, Chung WK.
Clin Genet. 2017 May 91 (5):765-763. IF: 3.512 (Q2 Genetics & Heredity 58/171).

73. Mutations in Histone Acetylase Modifier BRPF1 Cause an Autosomal-Dominant Form of Intellectual Disability with Associated Ptosis.
Mattioli F, Schaefer E, Magee A, Mark P, Mancini GM, Dieterich K, Von Allmen G, Alders M, Coutton C, van Slegtenhorst M, Vieville G, Engelen M, Cobben JM, Juusola J, **Pujol A**, Mandel JL, Piton A.
Am J Hum Genet. 2017 Jan 5;100(1):105-116. IF: 8.855 (D1 Genetics & Heredity 12/171).

74. Tauroursodeoxycholic bile acid arrests axonal degeneration by inhibiting the unfolded protein response in X-linked adrenoleukodystrophy.
Launay N, Ruiz M, Grau L, Ortega FJ, Ilieva EV, Martínez JJ, Galea E, Ferrer I, Knecht E, Fourcade S, **Pujol A*.**
Acta Neuropathol. 2017 Feb;133(2):283-301. IF: 15.876 (D1 Neurosciences 9/258).
& International Patent

75. De novo mutations in protein kinase genes CAMK2A and CAMK2B cause intellectual disability.
Küry S, Van Woerden GM, Besnard T, Proietti Onori M, Latypova X, Towne MC , Cho MT, Prescott TE, Ploeg MA, Sanders S, Stessman HAF, **Pujol A** and 82 coauthors.
Am J of Hum Genet, 2017 Nov 2;101(5):768-788. IF: 8.855 (D1 Genetics & Heredity 12/171).

76. Loss of SIRT2 leads to axonal degeneration and locomotor disability associated with redox and energy imbalance.

Fourcade S, Ruiz M, Villarroya F, Portero-Otin M, Pamplona R, Naudi A, Jove M, Ferrer I, Vaquero A, **Pujol A*.**

Aging Cell. 2017, Dec;16(6):1404-1413. IF: 7.627 (D1 Geriatrics/Gerontology 2/49).
& Editorial in Aging

77. Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder.

Falkenberg KD, Braverman NE, Moser AB, Steinberg SJ, Klouwer FC, Schlüter A, Ruiz M, **Pujol A,** Engvall M, Naess K, van Spronsen FJ, Körver-Keularts I, Rubio-Gozalbo ME, Ferdinandusse S. Wanders RJA, Waterham HR.

Am J of Hum Genet, 2017 Dec 7;101(6):965-976. IF: 8.855 (D1 Genetics & Heredity 12/171).

78. Oxidative Stress and Mitochondrial Dynamics Malfunction are interlinked in Pelizaeus-Merzbacher Disease.

Ruiz M, Bégou M, Launay N, Ranea-Robles P, Bianchi P, López-Erauskin J, Morató L, Guilera C, Petit B, Vaurs-Barriere C, Guéret-Gonthier C, Bonnet-Dupeyron MN, Fourcade S, Auwerx J, Boespflug-Tanguy O, and **Pujol A***.

Brain Pathology, 2018 Sep;28(5):611-630. PMID: 29027761 IF: 6.352 (D1 Neurosciences 26/267).
& Issue Cover

79. Genetic variants in HSD17B3, SMAD3, and IPO11 impact circulating lipids in response to fenofibrate in individuals with type 2 diabetes.

Shah HS, Gao H, Morieri ML, Doria A, Mychaleckyi JC, McLeod HL, Buse JB, Wagner MJ, Motsinger-Reif AA; **Pujol A** ACCORD/ACCORDion Investigators.

Clin Pharmacol Ther 2018 Apr;103(4):712-721. PMID: 28736931 IF: 6.336 (D1 Pharmacology & Pharmacy 15/267).

80. Evaluation of afferent pain pathways in adrenomyeloneuropathic patients.

Yagüe S, Veciana M, Casasnovas C, Ruiz M, Pedro J, Valls-Solé J, **Pujol A*.**

Clinical Neurophysiol. 2018 Mar;129(3):507-515. PMID: 29367165 IF: 3.675 (Q2 Clinical Neurology 51/199).

81. Epigenomic signature of adrenoleukodystrophy predicts compromised oligodendrocyte differentiation.

Schlüter A, Sandoval J, Fourcade S, Díaz-Lagares A, Ruiz M, Casaccia P, Esteller M, and **Pujol A*.**

Brain Pathol, 2018 28(6):902-919. PMID: 29476661 IF: 6.352 (D1 Neurosciences 26/267)**.**
& Top20 Most Read Article in Brain Pathology 2017-2018

82. Dual molecular effects of dominant RORA mutations cause two variants of syndromic intellectual disability with either autistic features or cerebellar ataxia.

Guissart C, Latypova X, Rollier P, Khan TN, Stamberger H, McWalter K, Cho M, Kjaergaard S, **Pujol A** and 37 additional coauthors.

Am J of Hum Genet, 2018 May 3;102(5):744-759 PMID: 29656859 IF: 9.924 (D1 Genetics & Heredity 9/174).

83. SIRT2 in age-related neurodegenerative disorders.
Fourcade S, Outeiro-TF and **Pujol A*.**
Editorial, Aging, 2018 Mar 3;10(3):295-296**.** PMID: 29514133 IF: 5.515 (D1 Geriatrics & Gerontology 3/53).

84. Inhibition of Gsk3b Reduces Nfkb1 Signaling and Rescues Synaptic Activity to Improve the Rett Syndrome Phenotype in Mecp2-Knockout Mice.
Jorge-Torres OC, Szczesna K, Roa L, Casal C, Gonzalez-Somermeyer L, Soler M, Velasco CD, Martínez-San Segundo P, Petazzi P, Sáez MA, Delgado-Morales R, Fourcade S, **Pujol A,** Huertas D, Llobet A, Guil S, Esteller M.
Cell Rep. 2018 May 8;23(6):1665-1677. PMID: 29742424 IF: 7.815 (Q1 Cell Biology 29/193).

85. Aberrant regulation of the GSK-3β/NRF2 axis unveils a novel therapy for adrenoleukodystrophy.
Ranea-Robles P, Launay N, Ruiz M, Calingasan NY, Dumont M, Naudí A, Portero-Otín M, Pamplona R, Ferrer I, Beal F, Fourcade S and **Pujol A*.**
EMBO Mol Med, 2018 Aug;10(8). PMID: 29997171 IF: 10.624 (D1 Medicine, Research & Experimental 6/136).
& International Patent,
& Orphan Drug Designation
& Phase II clinical trial granted by Spanish NIH (890.000) 2022-2026

86. Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy.
Pant D, Dorboz I, Schluter A, Fourcade S, Launay N,(35 coauthors). Gleeson JG, Pujades C, Fatemi A, Boespflug-Tanguy-O and **Pujol A*.**
J Clin Invest 2019 Mar 1;129(3):1240-1256. PMID: 30620337 IF: 11.864 (D1 Medicine, Research & Experimental 3/139).
& Orphan Drug Designation for Fingolimod to treat DEGS1-leukodystrophy (on progress)
& https://scienceunderthesurface.wordpress.com/?s=degs1
& Local TV: https://www.ccma.cat/tv3/marato/una-investigacio-financada-per-la-marato-descobreix-una-nova-leucodistrofia-infantil-i-la-cura-potencial/noticia/2897153/
& National Press: https://www.abc.es/espana/catalunya/abci-cientificos-espanoles-descubren-nuevo-tipo-leucodistrofia-infantil-y-potencial-cura-201901101344_noticia.html

& Best Talk Award, SSIEM Congress Rotterdam 3-6th September 2019

87. A novel mutation in the GFAP gene expands the phenotype of Alexander Disease.
Casasnovas C, Verdura E, Vélez-Santamaria V, Schlüter A, Pons-Escoda A, Homedes C, Ruiz M, Fourcade S, Launay N and **Pujol A*.**
J Med Genet, 2019, Dec;56(12):846-849. Pii: jmedgenet-2018-105959. Doi: 10.1136/jmedgenet-2018-105959. PMID: 31004048 IF: 4.943 (D1 Genetics & Heredity 30/178).
& Issue Cover December 2019

88. Biomarker identification, safety and efficacy of high-dose antioxidants for adrenomyeloneuropathy: a phase II pilot study.
Casasnovas C, Ruiz M, Schlüter A, Naudí A, Fourcade S, Veciana M, Castañer S, Albertí A, Bargalló N, Johnson M, Raymond GV, Fatemi A, Moser AB, Villarroya F, Portero-Otín M, Artuch R, Pamplona R, and **Pujol A***.
Neurotherapeutics, 2019 May 10. 16(4) SI:1167-1182. Doi: 10.1007/s13311-019-00735-2. PMID: 31077039 IF: 6.035 (D1 Clinical Neurology 21/204).

89. FAHN/SPG35: a narrow phenotypic spectrum across disease classifications.
Rattay T, Lindig T, Baets J, Smets K, Deconinck T, Söhn A, Hörtnagel K, Eckstein K, Wiethoff S, Reichbauer J, Krageloh-Mann I, Plecko B, Munchau A, Wilken B, Giese AK, Ortibus E, López de Munain A, **Pujol A,** Bassi M, D'Angelo MG, De Jonghe P, Zuchner S, Schoels L, Schüle R.
Brain 2019 Jun 1;142(6):1561-1572. PMID: 31135052 IF: 11.387 (D1 Clinical Neurology 6/204).

90. Clinical presentation and proteomic signature of patients with TANGO2 mutations.
Mingirulli N, Pyle A, Hathazi D, Alston CL, Kohlschmidt N, O'Grady G, Waddell L, Evesson F, Cooper SBT, Turner C, Duff J, Topf A, Yubero D, Jou C, Nascimento A, Ortez C, García-Cazorla A, Gross C, O'Callaghan M, Santra S, Preece MA, Champion M, Korenev S, Chronopoulou E, Anirban M, Pierre G, McArthur D, Thompson K, Navas P, Ribes A, Tort F, Schlüter A, **Pujol A**, Montero R, Sarquella G, Lochmüller H, Jiménez-Mallebrera C, Taylor RW, Artuch R, Grünert SC Roos A, Horvath R.
J Inherit Metab Dis. 2019, Jul 24. 43(2):297-308. Doi: 10.1002/jimd.12156. PMID: 31339582 IF: 4.750 (Q1 Genetics & Heredity 44/175).

91. Parkinsonism and Spastic Paraplegia type 7: expanding the spectrum of mitochondrial parkinsonism.
De la Casa-Fages B, Fernández-Eulate G, Gamez J, Barahona-Hernando R, Morís G, García-Barcina M, Infante J, Zulaica M, Fernández-Pelayo U, Muñoz-Oreja M, Urtasun M, Olaskoaga A, Zelaya V, Jericó I, Saez-Villaverde R, Catalina I, Sola E, Martínez-Sáez E, **Pujol A**, Ruiz M, Schlüter A, Spinazzola A, Muñoz-Blanco JL, Grandas F, Holt I, Álvarez V, López de Munaín A.
Mov Disord, 2019, Oct;34(10):1547-1561. PMID: 31433872 IF: 8.22 (D1 Clinical Neurology 12/199).

92. Complete loss of KCNA1 activity causes neonatal epileptic encephalopathy and dyskinesia.
Verdura E, Fons C, Schlüter A, Ruiz M, Fourcade S, Casasnovas. C, Castellano A, **Pujol A***.
J Med Genet, 2019, Oct 5. 57(2):132-137. PMID: 31586945 IF: 4.943 (Q1 Genetics & Heredity 18/174).

93. Truncating variants in UBAP1 associated with childhood-onset nonsyndromic hereditary spastic paraplegia.
S Gu, C Chen, J Mokry, H Cope, N Launay, K Flanigan, M Waldrop, R Schrader, J Juusola, O Goker-Alpan, A Milunsky, A Schlüter, M Troncoso, **A Pujol**, K Tan, C P Schaff, L Meng,
Hum Mutat, 2019 Nov 7. 41(3):632-640. PMID: 31696996 IF: 4.124 (Q1 Genetics & Heredity 45/178).

94. Reply to: "Mitochondrial Parkinsonism due to SPG7/Paraplegin variants with secondary mtDNA depletion".
De la Casa-Fages B, Fernández-Eulate G, Gamez J, Barahona-Hernando R, Morís G, García-Barcina M, Infante J, Zulaica M, Fernández-Pelayo U, Muñoz-Oreja M, Urtasun M, Olaskoaga A, Zelaya V, Jericó I, Saez-Villaverde R, Catalina I, Sola E, Martínez-Sáez E, **Pujol A**, Ruiz M, Schlüter A, Spinazzola A, Muñoz-Blanco JL, Grandas F, Holt I, Álvarez V, López de Munaín A.
Mov Disord. 2019 Dec;34(12):1932-1933. PMID: 31845766 IF: 8.22 (D1 Clinical Neurology 12/199) .

95. A deep intronic splice variant advises reexamination of presumably dominant SPG7 cases.
E Verdura; A Schlüter; G Fernández-Eulate; R Ramos-Martín; M Zulaica; M Ruiz; S Fourcade; C Casasnovas; A López de Munain, **A Pujol*.**
Ann Clin Transl Neurol, 2020 Jan;7(1):105-111. PMID: 31854126 IF:4.511 (Q2 Clinical Neurology 59/208).

96. POLR3A variants with striatal involvement and extrapyramidal movement disorder.
I Harting, M Al-Saady, I Krägeloh-Mann, A Bley, M Hempel, T Bierhals, S Karch, U Moog, G Bernard, R Huntsman, R M.L. van Spaendonk , M Vreeburg , A Rodríguez Palmero, **A Pujol**, M S. Van der Knaap, P J.W. Pouwels , N I. Wolf.
Neurogenetics, 2020 Jan 15. 21(2):121-133. PMID: 31940116 IF 2.660 (Q2 Clinical Neurology 140/208) .

97. The peroxisomal fatty acid transporter ABCD1/pmp-4 is required in the C. elegans hypodermis for axonal maintenance: a worm model for adrenoleukodystrophy.
A Coppa, S Guha, S Fourcade, J Parameswaran, M Ruiz, A B. Moser, A Schlüter, M P. Murphy, JM Lizcano, A Miranda-Vizuete, E Dalfó, **A Pujol***.
Free Radic Biol Med, 2020 May 20;152:797-809. PMID: 32017990 IF: 7.376 (Q1 Biochemistry & Molecular Biology 44/295) .

98. Phenotypic correlations in a large single center cohort of patients with BSCL2 nerve disorders: a clinical, neurophysiological and muscle MRI study.
Fernández-Eulate G, Fernández-Torrón R, Guisasola A, Iglesias Gaspar MT, Diaz-Manera J, Maneiro M, Zulaica M, Olasagasti V, Formica AF, Espinal JB, Ruiz M, Schlüter A, **Pujol A**, Poza JJ, López de Munain A.
Eur J Neurol. 2020 Aug;27(8):1364-1373. PMID: 32320108 IF : 6.089 (Q1 Clinical Neurology 30/208).

99. HNRNPH1-related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome.
Reichert SC, Li R, Turner S, van Jaarsveld RH, Massink MPG, van den Boogaard MH, Del Toro M, Rodríguez-Palmero A, Fourcade S, Schlüter A, Planas-Serra L, **Pujol A**, Iascone M, Maitz S, Loong L, Stewart H, De Franco E, Ellard S, Frank J, Lewandowski R.
Clin Genet. 2020 Jul;98(1):91-98. PMID: 32335897 IF:4.438 (Q1 Genetics & Heredity 54/176).

100. A global effort to define the human genetics of protective immunity to SARS-CoV-2 infection.

Casanova JL, Su H, in behalf of Covid Human Genetics Effort. **Pujol A** and 38 coauthors
Cell. 2020 Jun 11;181(6):1194-1199. PMID: 32405102 IF: 41.584 (D1 Biochemistry & Molecular Biology 2/295).

101. High-dose biotin restores redox balance, energy and lipid homeostasis, and axonal health in a model of adrenoleukodystrophy.
Fourcade S, Goicoechea L, Parameswaran J, Schlüter A, Launay N, Ruiz M, Seyer A, Colsch B, Calingasan NY, Ferrer I, Beal MF, Sedel F, **Pujol A***.
Brain Pathol. 2020 Sep;30(5):945-963. PMID: 32511826 IF: 6.508 (Q1 Clinical Neurology 27/208).

102. Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity.
Matalonga L, Laurie S, Papakonstantinou A, Piscia D, Mereu E, Bullich G, Thompson R, Horvath R, Pérez-Jurado L, Riess O, Gut I, van Ommen GJ, Lochmüller H, Beltran S; RD–Connect Genome-Phenome Analysis Platform and URD-Cat Data Contributors.
J Mol Diagn. 2020 Sep;22(9):1205-1215. PMID: 32619640 IF: 5.568 (Q1 Pathology 13/77)

103. Expanding the Clinical and Genetic Spectrum of PCYT2-related Disorders
Vélez-Santamaría V, Verdura V, Macmurdo C, Planas-Serra L, Schlüter A, Casas J, Martínez J, Casasnovas C1, Si Y, Thompson SS, Maroofian R, **Pujol A*.**
Brain, 2020, Sep 1;143(9): e76. PMID: 32889549 IF: 13.501 (D1 Clinical Neurology 6/208).

104. Ceramide signaling in inherited and multifactorial brain metabolic diseases.
Pant D, Aguilera-Albesa S, **Pujol A***.
Neurobiology of Disease, Invited Review.2020 Sep;143:105014. PMID: 32653675 IF: 5.996 (Q1 Neurosciences 54/273).

105. A novel hypomorphic splice variant in EIF2B5 gene is associated with mild ovarioleukodystrophy
Rodríguez-Palmero A, Schlüter A, Verdura E, Ruiz M, Martínez JJ, Gourlaouen I, Ka C, Lobato R, Casasnovas C, Le Gac G, Fourcade S, **Pujol A***
Ann Clin Transl Neurol, 2020 Sep;7(9):1574-1579. PMID: 33173540 IF:4.511 (Q2 Clinical Neurology 59/208). IF: 3.66)

106. Case Report: Benign Infantile Seizures Temporally Associated With COVID-19
García-Howard M, Herranz-Aguirre M, Moreno-Galarraga L, Urretavizcaya-Martínez M, Alegría-Echauri J, Gorría-Redondo N, Planas-Serra L, Schlüter A, Gut M, **Pujol A*** and Aguilera-Albesa S
Frontiers in Pediatrics, 2020 Aug 6;8:507. PMID: 32850563 IF: 3.418 (Q1 Pediatrics 24/129)

107. Auto-antibodies against type I IFNs in patients with life-threatening COVID-19.
Bastard P, Rosen LB, Zhang Q, [56 additional coauthors]Covid Human Genetic Effort, Casanova JL
Science. 2020 Oct 23;370(6515):eabd4585. PMID: 32972996 IF: 47.728 (D1 Multidisciplinary Sciences 2/72)

& Extensive coverage on Catalan and national TV (TV3, Tele5, La Sexta, Antena 3, Canarias7, Euskal Telebista); national radio (Cadena Ser, Onda Regional de Murcia along with major Spanish newspapers (El Periódico, La Vanguardia, Heraldo, La Voz de Galicia, Diario 16, 20 minutos, El Norte de Castilla).

&Extensive international coverage
&TOP10 discoveries of 2020 after Nature 2020 Dec;588(7839):596-598

108. Inborn errors of type I IFN Immunity in patients with severe COVID-19
Zhang Q, Jing H, Bastard P, [64 additional coauthors] **Pujol A**, Covid Human Genetic Effort, Casanova JL, and Su HC
Science, 2020 Oct 23;370(6515):eabd4570. PMID: 32972995 IF: 47.728 (D1 Multidisciplinary Sciences 2/72)

& Extensive coverage on Catalan and national TV (TV3, Tele5, La Sexta, Antena 3, Canarias7, Euskal Telebista); national radio (Cadena Ser, Onda Regional de Murcia along with major Spanish newspapers (El Periódico, La Vanguardia, Heraldo, La Voz de Galicia, Diario 16, 20 minutos, El Norte de Castilla).

&Extensive international coverage
& TOP10 discoveries of 2020 highlighted in Nature 2020 Dec;588(7839):596-598

109. Impairment of the Mitochondrial One-Carbon Metabolism Enzyme SHMT1 causes a novel brain and heart developmental syndrome
García-Cazorla A, Verdura E, Juliá-Palacios N, Anderson EN, Goicoechea L, Tsogtbaatar E, Dsouza NR, Schlüter A, Urreizti R, Planas-Serra L, Tarnowski JM, Gavrilova RH, SHMT2 Working Group, Ruiz M, Rodríguez-Palmero A, Fourcade S, Cogné B, Besnard T, Vincent M, Bézieau S, Folmes CD, Zimmermann MT, Klee EW, Pandey U, Artuch R, Cousin MA, **Pujol A***
Acta Neuropathol, 2020 Dec;140(6):971-975. PMID: 33015733 IF: 17.088 (D1 Clinical Neurology 5/208)

&https://www.lavanguardia.com/vida/20201005/483870086261/descubren-una-nueva-enfermedad-genetica-rara-del-sistema-nervioso-y-cardiaco.html

&https://www.ccma.cat/324/descobreixen-una-nova-malaltia-en-nens-amb-problemes-cognitius-motors-i-cardiacs/noticia/3050915/

& Best Talk Award, SSIEM Virtual Congress December 3rd 2020

110. The Value of Mouse Models of Rare Diseases: A Spanish Experience.
Murillo-Cuesta S, Artuch R, Asensio F, de la Villa P, Dierssen M, Enríquez JA, Fillat C, Fourcade S, Ibáñez B, Montoliu L, Oliver E, **Pujol A**, Salido E, Vallejo M, Varela-Nieto I.
Front Genet. 2020 Oct 14;11:583932. Doi: 10.3389/fgene.2020.583932. eCollection 2020. PMID: 33173540 IF:4.599 (Q2 Genetics & Heredity 48/176)

111. Lipid alterations in human frontal cortex in ALS-FTLD-TDP43 proteinopathy spectrum are partly related to peroxisome impairment.
Andrés-Benito P, Gelpi E, Jové M, Mota-Martorell N, Obis È, Portero-Otin M, Povedano M, **Pujol A**, Pamplona R, Ferrer I.

Neuropathol Appl Neurobiol, 2021 Jun;47(4):544-563. PMID: 33332650 IF:6.25 (D1 Clinical Neurology 35/212)

112. DLG4-related synaptopathy: A new rare brain disorder
Rodriguez-Palmero A, Boerrigter MM, Gómez-Andrés D, (78 coauthors), **Pujol A*** and Tumer Z.
Genetics in Medicine, 2021 May;23(5):888-899. PMID: 33597769 IF:8.964 (D1 Genetics & Heredity 13/175)

113. Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in males.
Baldassarri M, Picchiotti N, Fava F, (22 co-authors), Planas-Serra L; Schluter A, Spanish Covid HGE; **Pujol A**;; GEN-COVID Multicenter Study; Renieri A, MD; Frullanti E; Mari F.
Ebiomedicine, 2021 Mar;65:103246. PMID: 33647767 IF: 11.205 (Q1 Medecine, Research & Experimental 14/139)

114. Epigenome-Wide Association Study of COVID-19 Severity with Respiratory Failure.
Castro de Moura M; Davalos V; Planas-Serra L; Alvarez-Errico D; Arribas C; Ruiz M; Aguilera-Albesa S; Troya J; Valencia-Ramos J; Vélez-Santamaria V; Rodriguez-Palmero A; Villar-Garcia J; Horcajada JP; Albu S; Casasnovas C; Rull A, Reverte L; Dietl B; Dalmau D; Arranz M; Llucià-Carol L; Planas-A; Pérez-Tur J; Fernandez-Cadenas I; Villares P; Tenorio J; Colobran R; Martin-Nalda A; Soler-Palacin P; Vidal F; **Pujol A***; Esteller M.
Ebiomedicine, 2021.Apr; 66:103339 . PMID: 33867313 IF: 11.205 (Q1 Medecine, Research & Experimental 14/139)

115. Neutralizing Autoantibodies to Type I IFNs in >10% of Patients with Severe COVID-19 Pneumonia Hospitalized in Madrid, Spain.
Troya J; Bastard P; Planas-Serra L; Ryan P; Ruiz M; de Carranza M; Torres J; Martínez A; Abel L; Casanova JL; **Pujol A***.
J Clin Immunol 2021 Apr 13;1-9. PMID: 33851338 IF: 8.542 (Q1 Immunology 36/162)

116. Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder.
Kour S, Rajan D, Fortuna T, Anderson E, Ward C, Lee Y, Lee S, Beom Shin Y, Chae JH, Choi M, Verdura E, **Pujol A**, McCurry H, Landers J, Agnihotri A, E E, Moody S, Phornphutkul C, Guillen M, Begtrup A, Houlden H, Kirschner J, Schorling D, Rudnik-Schöneborn S, Strom T, Leiz S, Juliette K, Wagner M, Issa M, Elbendary H, Stanley V, Maroofian R, Gleeson J, Zaki M, Senderek J, and Pandey U.
Nature Communications 2021 May 7;12(1):2558. PMID: 33963192 IF:17.694 (D1 6/74 Multidisciplinary Sciences).

117. CSVS, a crowdsourcing database of the Spanish population genetic variability.
Peña-Chilet M, Roldán G, Perez-Florido J, Ortuño FM, Carmona R, Aquino V, Lopez-Lopez D, Loucera C, Fernandez-Rueda JL, Gallego A, García-Garcia F, González-Neira A, Pita G, Núñez-Torres R, Santoyo-López J, Ayuso C, Minguez P, Avila-Fernandez A, Corton M, Moreno-Pelayo MÁ, Morin M, Gallego-Martinez A, Lopez-Escamez JA, Borrego S, Antiñolo G, Amigo J, Salgado-Garrido

J, Pasalodos-Sanchez S, Morte B; Spanish Exome Crowdsourcing Consortium, Carracedo Á, Alonso Á, Dopazo J.

Nucleic Acids Res. 2021 Jan 8;49(D1):D1130-D1137. PMID: 32990755 IF: 19.160 (D1 8/297 Biochemistry & Molecular Biology).

118. Anti-oxidant MitoQ rescue of AWB chemosensory neuron impairment in a C. Elegans model of X-linked Adrenoleukodystrophy.
Guha S, **Pujol A**, Dalfo E.
MicroPubl Biol. 2021 Jan 14;2021. PMID: 33474532

119. SARS-CoV-2-related MIS-C: A key to the viral and genetic causes of Kawasaki disease?.
Sancho-Shimizu V, Brodin P, Cobat A, Biggs CM, Toubiana J, Lucas CL, Henrickson SE, Belot A; MIS-C@CHGE, Tangye SG, Milner JD, Levin M, Abel L, Bogunovic D, Casanova JL, Zhang SY.
J Exp Med. 2021 Jun 7;218(6):e20210446.. PMID: 33904890 IF: 17.579 (D1 11/162 Immunology).

120. Modulation of mitochondrial and inflammatory homeostasis through RIP140 is neuroprotective in an adrenoleukodystrophy mouse model.
Ranea-Robles P, Galino J, Espinosa L, Schlüter A, Ruiz M, Calingasan NY, Villarroya F, Naudí A, Pamplona R, Ferrer I, Beal MF, Portero-Otín M, Fourcade S, **Pujol A*.**
Neuropathol Appl Neurobiol. 48(1):e12747.2021. PMID: 34237158 IF 6.250 (D1 Clinical Neurology 35/212).

121. Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN-β.
Vinh DC, Abel L, Bastard P, Cheng MP, Condino-Neto A, Gregersen PK, Haerynck F, Cicalese MP, Hagin D, Soler-Palacín P, Planas AM, **Pujol A**, Notarangelo LD, Zhang Q, Su HC, Casanova JL, Meyts I; COVID Human Genetic Effort.
J Clin Immunol. 2021 Jun 8:1-18. PMID: 34101091 IF: 8.542 (Q1 Immunology 36/162)

122. X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19.
Asano T, Boisson B, Onodi F, Matuozzo D, Moncada-Velez M, Maglorius Renkilaraj MRL, Zhang P, Meertens L, Bolze A, Materna M, Korniotis S, Gervais A, Talouarn E, Bigio B, Seeleuthner Y, Bilguvar K, Zhang Y, Neehus AL, Planas-Serra L, Schlüter A, **Pujol A,** (85 additional co-authors), Casanova JL.
Sci Immunol. 2021 Aug 19;6(62):eabl4348. PMID: 34413140 IF: 30.658 (D1 Immunology 5/162)

123. Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths.
Bastard P, Gervais A, Le Voyer T, Rosain J, Philippot Q, Manry J, Michailidis E, Hoffmann HH, Eto S, Garcia-Prat M, Bizien L, Parra-Martínez A, Yang R, Haljasmägi L, Migaud M, Särekannu K, Maslovskaja J, de Prost N, Tandjaoui-Lambiotte Y, Luyt CE, Amador-Borrero B, Gaudet A, Poissy J, Morel P, Richard P, Cognasse F, **Pujol A.**, (105 addiontal co-authors), Casanova JL.
Sci Immunol. 2021 Aug 19;6(62):eabl4340. PMID: 34413139 IF: 30.658 (D1 Immunology 5/162)

124. Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy.
Verdura E, Rodríguez-Palmero A, Vélez-Santamaria V, Planas-Serra L, de la Calle I, Raspall-Chaure M, Roubertie A, Benkirane M, Saettini F, Pavinato L, Mandrile G, O'Leary M, O'Heir E, Barredo E, Chacón A, Michaud V, Goizet C, Ruiz M, Schlüter A, Rouvet I, Sala-Coromina J, Fossati C, Iascone M, Canonico F, Marcé-Grau A, de Souza P, Adams DR, Casasnovas C, Rehm HL, Mefford HC, González Gutierrez-Solana L, Brusco A, Koenig M, Macaya A, **Pujol A*.**
Brain. 2021 Aug 20:awab124. PMID: 34415322 IF: 15.255 (D1 Clinical Neurology 6/212)

125.From Your Nose to Your Toes: A Review of Severe Acute Respiratory Syndrome Coronavirus 2 Pandemic–Associated Pernio.
Arkin LM, Moon JJ, Tran JM, Asgari S, O'Farrelly C, Casanova JL, Cowen EW, Mays JW, Singh AM, Drolet BA, COVID Human Genetic Effort.
J Invest Dermatol. 2021. 141(12):2791-2796. PMID: 34561087 IF: 7.590 (D1 Dermatology 5/69)

126. The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call.
Bellusci M, Paredes-Fuentes AJ, Ruiz-Pesini E, Gómez B; MITOSPAIN Working Group, Martín MA, Montoya J, Artuch R.
Genes (Basel). 2021 Oct 9;12(10):1590 PMID: 34680984 IF: 4.141 (Q2 Genetics and Heredity 72/175)

127 Biallelic Variants in the Mitochondrial Rnase P Subunit PRORP cause mitochondrial tRNA processingdefects and pleiotropic multixystem presentations.
Hochberg I, Demain LAM, Richer J, Thompson K, Urquhart JE, Rea A, Pagarkar W, Rodríguez-Palmero A, Schlüter A, Verdura E, **Pujol A**, Quijada-Fraile P, Amberger A, Deutschmann AJ, Demetz S, Gillespie M, Belyantseva IA, McMillan HJ, Barzik M, Beaman GM, Motha R, Ying NG K, O´Sullivan J, Williams SG, Bhaskar SS, Lawrence IR, Jenkinson EM, Zambonin JL, Blumenfeld Z, Yalonetsky S, Oerum S, Rossmanith W, Genomics England Research.
American Journal of Human Genetics, 2021 Oct 22;144(9):2659-2669. PMID: 35325049 IF: 11.043 (D1 Genetics Heredity 10/175)

128. Epigenetic profiling linked to multisystem inflammatory syndrome in children (MIS-C): A multicenter, retrospective study.
Davalos V, García-Prieto CA, Ferrer G, Aguilera-Albesa S, Valencia-Ramos J, Rodríguez-Palmero A, Ruiz M, Planas-Serra L, Jordan I, Alegría I, Flores-Pérez P, Cantarín V, Fumadó V, Viadero MT, Rodrigo C, Méndez-Hernández M, López-Granados E, Colobran R, Rivière JG, Soler-Palacín P, **Pujol A**, Esteller M.
EClinicalMedicine. 2022 Jun 25;50:101515. PMID: 35770252 IF: 17.033 (D1 Medicine, General & Internal 12/172)

129. A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection.
Andreakos E, Abel L, Vinh DC, Kaja E, Drolet BA, Zhang Q, O'Farrelly C, Novelli G, Rodríguez-Gallego C, Haerynck F, Prando C, **Pujol A,** COVID Human Genetic Effort*, Su HC, Casanova JL, Spaan AN.

Nature Immunology 2022 23(2):159-164, 2022. PMID: 35012964 IF:31.250 (D1 Immunology 4/162)

Extensive press releases:
&https://www.elmundo.es/ciencia-y-salud/salud/2022/03/05/62222e60e4d4d855558b457a.html
&https://www.20minutos.es/noticia/4919130/0/estudio-genes-personas-superresistentes-covid-no-contagiados-contactos-estrechos-idibell/
&https://www.cope.es/programas/fin-de-semana/noticias/terminator-del-covid-asi-son-los-pacientes-super-resistentes-por-que-son-clave-para-acabar-con-virus-20211211_1670599

TV: https://cnnespanol.cnn.com/video/covid-genes-virus-celulas-investigacion-dra-aurora-pujol-vls/

130. Diagnosis of genetic white matter disorders by singleton whole-exome and genome sequencing using interactome-driven prioritization.
Schlüter A, Rodríguez Palmero A, Verdura E, Vélez Santamaría V, Ruiz M, Fourcade S, Planas Serra L, Martínez JJ, Guilera C, Girós M, Artuch R, Yoldi ME, O´Callaghan M, García Cazorla A, Armstrong J, Redin C, Mandel JL, Conejo D, Sierra-Córcoles C, Beltran S, Gut M, Vázquez E, del Toro M, Troncoso M, Pérez-Jurado LA, Gutiérrez Solana LG, López de Munain A, Casanovas C, Aguilera Albesa S, Macaya A, **Pujol A***
Neurology 1;98(9):e912-e923, 2022. PMID: 35012964 IF: 12.258 (D1 Clinical Neurology 9/212)
Extensive coverage in written press: La Vanguardia, TV3,
Broadcast in main TV2 program: El Cazador de Cerebros : "Rare Disease" May 9th 2022

131. Biallelic variants in SLC35B2 cause a novel chondrodysplasia with hypomyelinating leukodystrophy.
Guasto A, Dubail J, Aguilera-Albesa S, Paganini C, Vanhulle C, Haouari W, Gorría-Redondo N, Aznal-Sainz E, Boddaert N, Planas-Serra L, Schlüter A, Verdura E, Bruneel A, Rossi A, Huber C, **Pujol A***, Cormier-Daire V.
Brain. 2022 Oct 21;145(10):3711-3722. PMID: 35325049 IF:15.255 (D1 Clinical Neurology 6/212)

132. Low Lymphocytes and IFN-Neutralizing Autoantibodies as Biomarkers of COVID-19 Mortality.
Troya J, Bastard P, Casanova JL, Abel L, **Pujol A***.
J Clin Immunol 3:1-4. Online ahead of print, 2022. PMID: 35243564 IF: 8.542 (Q1 Immunology 36/162)

133. Human genetic and immunological determinants of critical COVID-19 pneumonia.
Zhang Q, Bastard P; COVID Human Genetic Effort, Cobat A, Casanova JL.
Nature 603 (7902):587-598, 2022. PMID: 35090163 IF: 69.504 (D1 1/74 Multidisciplinary Sciences)

134. Novel genes and sex differences in COVID-19 severity.
Cruz R, Diz-de Almeida S, López de Heredia M, Quintela I, Ceballos FC, Pita G, Lorenzo-Salazar,JM, **Pujol A** (and 46 coauthors) Flores C, Lapunzina P, Carracedo A.

Hum Mol Genet. 2022 Nov 10;31(22):3789-3806. doi: 10.1093/hmg/ddac132. PMID: 35708486 IF: 5.121 (Q1 36/175 Genetics & Heredity)

135. Accelerated biological aging in COVID-19 patients.
Cao X, Li W, Wang T, Ran D, Davalos V, Planas-Serra L, **Pujol A**, Esteller M, Wang X, Yu H.
Nat Commun. Apr 19;13(1):2135, 2022. PMID:35440567 IF: 17.694 (D1 6/74 Multidisciplinary Sciences)

136. Studying severe long COVID to understand post-infectious disorders beyond COVID-19.
Brodin P, Casari G, Townsend L, O'Farrelly C, Tancevski I, Löffler-Ragg J, Mogensen TH, Casanova JL; COVID Human Genetic Effort.
Nat Med. Apr 5. 28(5):879-882, 2022. PMID: 35383311 IF: 87.244 (D1 1/297 Biochemistry & Molecular Biology).

137. CIBERER: Spanish national network for research on rare diseases: A highly productive collaborative initiative.
Luque J, Mendes I, Gómez B, Morte B, López de Heredia M, Herreras E, Corrochano V, Bueren J, Gallano P, Artuch R, Fillat C, Pérez-Jurado LA, Montoliu L, Carracedo Á, Millán JM, Webb SM, Palau F; CIBERER Network, Lapunzina P.
Clin Genet. 101(5-6):481-493, 2022. PMID: 35060122 IF: 4.296 (Q2 63/175 Genetics & Heredity).

138. Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases.
Bullich G, Matalonga L, Pujadas M, Papakonstantinou A, Piscia D, Tonda R, Artuch R, Gallano P, Garrabou G, González JR, Grinberg D, Guitart M, Laurie S, Lázaro C, Luengo C, Martí R, Milà M, Ovelleiro D, Parra G, **Pujol A**, Tizzano E, Macaya A, Palau F, Ribes A, Pérez-Jurado LA, Beltran S; Undiagnosed Rare Disease Program of Catalonia (URD-Cat) Consortium.
J. Mol Diagn. 24(5):529-542, 2022. PMID: 35569879 IF: 5.341 (Q1 18/77 Pathology).

139. The risk of COVID-19 death is much greater and age-dependent with type I IFN autoantibodies.
Manry J, Bastard P, Gervais A, Voyer TL, Rosain J, Philippot Q, Michailidis E, Hoffmann HH, Eto S, Garcia-Prat M, Bizien L, Parra-Martínez A, Troya J, Trouillet-Assant S, Belot A, Saker K, Garçon P, Rivière JG. **Pujol A** (43 additional coauthors), Zhang Q, Casanova JL, Abel L, Cobat A.
Proc Natl Acad Sci USA. 119(21):e2200413119, 2022. PMID: 35576468 IF: 11.205 (Q1 8/72 Multidisciplinary Sciences).

140. Loss of of seryl-tRNA synthetase (SARS1) causes complex spastic paraplegia and cellular senescence
E Verdura, B Senger, M Raspall-Chaure, A Schlüter, N Launay, M Ruiz, **Pujol A***
J Medical Genetics 2022. 59 (12), 1227-1233 PMID: 36041817 IF: 5.945 (Q1 Genetics & Heredity 28/175)

141. Activating cannabinoid receptor 2 preserves axonal health through GSK-3β/NRF2 axis in adrenoleukodystrophy.

Parameswaran J, Goicoechea L, Planas-Serra L, Pastor A, Ruiz M, Calingasan NY, Guilera C, Aso E, Boada J, Pamplona R, Portero-Otín M, de la Torre R, Ferrer I, Casasnovas C, Fourcade S. **Pujol A***

Acta Neuropathol. 2022 Aug;144(2):241-258 PMID: 35778568 IF: 15.887 (D1 Clinical Neurology 5/212)

142. Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency.
Campbell TM, Liu Z, Zhang Q, Moncada-Velez M, (45 additional authors) COVID Human Genetic Effort, Bergman P, Abel L, Cobat A, Casanova JL, Meyts I, Bryceson YT.
J Exp Med. 2022 Jul 4;219(7):e20220202. PMID: 35670811 IF: 17.579 (D1 Inmunology 11/162)

143. Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia.
Zhang Q, Matuozzo D, Le Pen J, Lee D, Moens L, (38 additional authors), Covid Human Genetic Effort, Bastard P, Su HC, Boisson-Dupuis S, Abel L, Rice CM, Zhang SY, Cobat A, Casanova JL.
J Exp Med. 2022 Aug 1;219(8):e20220131. PMID: 35708626 IF: 17.579 (D1 Inmunology 11/162)

144. Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs.
Bastard P, Vazquez S, Liu J, Laurie MT, Wang CY, Gervais A, Le Voyer T, Bizien L, Zamecnik C, Philippot Q, Rosain J, Catherinot E, Willmore A, Mitchell AM, Bair R, Garçon P, Kenney H, Fekkar A, **Pujol A** (42 additional coauthors), Zhang Q, Anderson MS, Casanova JL, DeRisi JL.
Sci Immunol. 2022 Jun 14:eabp8966. PMID: 35857576 IF: 30.658 (D1 Inmunology 5/162)

145. Autoantibodies against type I IFNs in patients with critical influenza pneumonia.
Zhang Q, Pizzorno A, Miorin L, Bastard P, COVID Human Genetic Effort; (68 coauthors)Jouanguy E, Abel L, Cobat A, Trouillet-Assant S, García-Sastre A, Casanova JL
J Exp Med. 2022 Nov 7;219(11):e20220514. PMID: 36112363 IF: 17.579 (D1 Inmunology 11/162)

146. Decoding the Human Genetic and Immunological Basis of COVID-19 mRNA Vaccine-Induced Myocarditis.
Bolze A, Mogensen TH, Zhang SY, Abel L, Andreakos E, Arkin LM, Borghesi A, Brodin P, Hagin D, Novelli G, Okada S, Peter J, Renia L, Severe K, Tiberghien P, Vinh DC; COVID human genetic effort, Cirulli ET, Casanova JL, Hsieh EWY.
J Clin Immunol. 2022 Oct;42(7):1354-1359. PMID: 36207567 IF: 8.542 (Q1 Inmunology 36/162)

147. Rare predicted loss-of-function variants of type I IFN immunity genes are associated with life-threatening COVID-19.
Matuozzo D, Talouarn E, Marchal A, Manry J, Seeleuthner Y, Zhang Y, Bolze A, Chaldebas M, Milisavljevic B, Zhang P, Gervais A, Bastard P, Asano T, Bizien L, Barzaghi F, Abolhassani H, Tayoun AA, Aiuti A, Darazam IA, Allende LM, Alonso-Arias R, **Pujol A** (49 additional collaborators),; COVID Human Genetic Effort -Dupuis S, Su HC, Boisson B, Jouanguy E, Casanova JL, Zhang Q, Abel L, Cobat A
medRxiv. 2022 Oct 25:2022.10.22.22281221. PMID: 36324795

148. Epigenetic activation of antiviral sensors and effectors of interferon response pathways during SARS-CoV-2 infection.

Bińkowski J, Taryma-Leśniak O, Łuczkowska K, Niedzwiedź A, Lechowicz K, Strapagiel D, Jarczak J, Davalos V, **Pujol A**, Esteller M, Kotfis K, Machaliński B, Parczewski M, Wojdacz TK.

Biomed Pharmacother. 2022 Sep;153:113396..PMID: 36076479. IF: 6.529 (Q1 12/77 Pathology)

149. Human type I IFN deficiency does not impair B cell response to SARS-CoV-2 mRNA vaccination.

Sokal A, Bastard P, Chappert P, Barba-Spaeth G, Fourati S, Vanderberghe A, Lagouge-Roussey P, Meyts I, Gervais A, Bouvier-Alias M, Azzaoui I, Fernández I, de la Selle A, Zhang Q, Bizien L, Pellier I, **Pujol A**, (21 additional coauthors), Abel L, Rey FA, Weill JC, Reynaud CA, Tangye SG, Casanova JL, Mahévas M.

J Exp Med. 2023 Jan 2;220(1):e20220258. PMID: 36342455 IF: 17.579 (D1 Inmunology 11/162)

150. Inborn errors of OAS–RNase L in SARS-CoV-2–related multisystem inflammatory syndrome in children.

D Lee, JL Pen, A Yatim, B Dong, YAquino, M Ogishi, R Pescarmona, E Talouarn, AM Planas, I Meyts, **Pujol A**, (65 additional coauthors) L Alsina, S Keles, E Haddad, L Abel, A Belot, L Quintana-Murci, CM Rice, R H. Silverman, SY Zhang, JL Casanova

Science 2022 Dec 20;eabo3627. PMID: 36538032 IF: 63.832 (D1 Multidisciplinary Sciences 2/74)

Book Chapters

Pujol A. Of a mouse model for X-linked Adrenoleukodystrophy and beyond In: X-linked adrenoleukodystrophy. June 2005, eds Berger J., Stöckler S and Köhler W, Sps Verlagsgesellschaft Heilbronn.

Pujol A and Aubourg P**.** Chapter 12: The ABCD subfamily: peroxisomal transporters in health and disease. In: The Genetics and biochemistry of ATP Binding Cassette transporters. May 2011; eds. Linton KJ and Holland ISBN 9814280062, World Scientific Publishing.

Pujol A. Novel therapeutic targets and drug candidates for modifying disease progression in adrenoleukodystrophy. 'Advanced Therapies in Paediatric Endocrinology and Diabetology'. 2015, in press. NLM ID: 101667831. A volume in the series 'Endocrine Development', edited by Marco Cappa, Rome (Italy) for S. Karger AG. Basel (Switzerland).

Oral Presentations at International venues

Talks at International Congress

1. **Late-onset phenotype of the X-ALD gene inactivation in mice: a mouse model for X-Adrenomyeloneuropathy.** 39th SSIMD, Prague, September 2001
2. **Mouse models of X-Linked adrenoleukodystrophy and phenotypic correction by transgenic overexpression of the ALDR gene: functional redundancy at the peroxisomal membrane?** 52nd ASHG. Baltimore, October 2002
3. **Functional redundancy between ABCD1 and ABCD2 transporters in the mouse**: Ixth IEM. Brisbane. September 2003.
4. **Overlapping functions of ABCD1 and ABCD2 transporters: implications for therapy.** 53rd ASHG. Los Angeles, November 2003.
5. **Functional overlap between ABCD1 (ALD) and ABCD2 (ALDR) transporters: a therapeutic target for X-linked adrenoleukodystrophy**. 36th EHSG, Munich, June 2004**. Young Investigator Award.**
6. **Valproic acid stimulates ABCD2 gene expression: a novel potential therapy for X-adrenoleukodystrophy**.38th EHSG, European Society for Human Genetics, Amsterdam, May 2006
7. **Neurodegeneration in X-ALD: dangerous liaisons between mitochondria and peroxisomes?** 17th ESN, European Society for Neurochemistry, Salamanca, Spain. May 2007
8. **Mitochondria-peroxisome cross-talk orchestrated through RIP140 and fatty acids**. XXXI Congress of the Spanish Society for Biochemistry and Molecular Biology. Bilbao, September 2008.
9. **Mouse models for X-adrenoleukodystrophy: opportunities for gene therapy**. Invited speaker. EPFL Gene Therapy Workshop. Lausanne June 2008.
10. **Valproic acid as a therapeutic strategy in X-linked adrenoleukodystrophy.** Invited speaker. Second AMN Day International Symposium. London March 2009.
11. **Therapeutic challenge in white matter disease.** Invited speaker**.** Chairman in the 2nd ELA Research Foundation Congress, Luxembourg, June 2009.
12. **Oxidative stress underlying neurodegeneration in X-linked adrenoleukodystrophy.** Invited speaker**.** Third AMN Day International Symposium. London, 9th May 2010.
13. **A cocktail of antioxidants halts axonal damage and disability in a mouse model of X-adrenoleukodystrophy.** Invited speaker**.** United Leukodystrophy Foundation Annual Conference. DeKalb, Chicago, 8-9th July 2010.
14. **Towards a therapeutic clinical trial for AMN.** Invited speaker**.** European Leukodystrophy Association, Paris, France, Colloque Familles/Chercheurs" 16-17 April 2011.
15. **Clinical trials with antioxidants in adrenoleukodystrophy**. Invited speaker. United Leukodystrophy Association, ULF Conference De Kalb, Chicago, USA,11th of July 2011.
16. **Metabolic failure and oxidative stress intertwinning due to fatty acid excess: rationale for a clinical trial for adrenoleukodystrophy**, Mérida, México, Cell Signaling Networks Conference, International Society for Biochemistry and Molecular Biology, 22-26 October 2011
17. **Antioxidants halt axonal degeneration and disability in X- adrenoleukodystrophy (X-ALD) mouse model: towards a clinical trial**, Invited speaker. Porto, Portugal, VIII International

Symposium of the Portuguese Society for Metabolic Disease (SPDM) Inborn Errors of Metabolism and Neurodegeneration, 3-4 November 2011

18. **Mitochondria failure underlying axonal degeneration in adrenoleukodystrophy, a peroxisomal disease .** Invited speaker. Freiburg, Germany, 17th European Bioenergetics Conference, 15-20 September 2012
19. **Pioglitazone halts axonal degeneration in a mouse model of Adrenoleukodystrophy: Translation into Clinical Trials.** Invited speaker. Cancun, Mexico, Satellite Meeting 11th Biennial ISN-ASN (International Society for Neurochemistry-American Society for Neurochemistry). Myelin: From Basic to Translational Research. April 16th-20th 2013.
20. **Pioglitazone prevents mitochondria dysfunction and halts axonal degeneration in a mouse model of X-adrenoleukodystrophy.** Oral Communication. Paris, France, European Human Genetics Conference 2013. June 8-11th 2013. Press release of the European Human Genetics Conference on 08 June 2013, Paris France
21. **Redox and Metabolic Dysfunction in Adrenoleukodystrophy: Translation into Clinical Trials.** Oral communication. XI European Meeting on Glial Cells in Health and Disease. Berlin Germany, July 2-6th 2013
22. **Mitochondria failure and rescue by pioglitazone: a clinical trial for adrenomyeloneuropathy.** Invited speaker. United Leukodystrophy Foundation annual meeting. De Kalb, Chicago USA, July 18-20th 2013
23. **Targeting oxidative stress and mitochondrial dysfunction to halt axonal degeneration.** Invited speaker. 2nd International Symposium on Multiple Sclerosis and Myelin Disorders. Leipzig, Germany, October 11-13th 2013
24. **Fighting oxidative stress and mitochondrial dysfunction halts axonal degeneration: the adrenoleukodystrophy case.** Invited Speaker. "The core of neuronal communication: axonal biology, degeneration and regeneration."International Summer *School, University of Trento, Centre for Integrative Biology, September 30th-October 4th, 2013*
25. **Blocking oxidative stress as therapy for adrenoleukodystrophy.** Invited Speaker. "Advanced Therapies in Paediatric Endocrinology and Diabetology". Bambino Gesù Childrens Hospital. Rome 27-28 October 2014.
26. **Identification of drug targets in X-ALD: Translation to the clinics**. Invited Speaker. 3th European Leukodystrophy Association Scientific Congress. Paris, June 3-6th, 2015.
27. **Integrative–omics analysis uncovers lipid-mediated inflammation in adrenoleukodystrophy.** Invited Speaker. XII European Meeting on Glial Cells in Health and Disease (EuroGlia). Bilbao, Spain, July 15 – 18, 2015.
28. **Metabolomics and Oxidative Stress Biomarkers as Outcome Measures in X-ALD.** Invited Speaker. Emerging and Translational Biology of Leukodystrophies. Baltimore, US, May 12-14th 2016
29. **A CB2 Agonist Halts Axonal Degeneration in a Mouse Model of Adrenoleukodystrophy.** Invited Speaker. Gordon Research Conference. Myelin Meeting. Lucca, Italy, May 15th-20th 2016.
30. **A combination of antioxidants as disease modifying treatment in adrenomyeloneuropathy: A phase II clinical trial**
    Invited Speaker. 3rd International Symposium on Multiple Sclerosis and Myelin Disorders. Berlin, Germany, October 7th-9th 2016
31. **The Diagnosis and Novel Gene Identification for Inherited Brain White Matter Disorders in Spain**

Speaker. Fifth International Conference on Rare and Undiagnosed Diseases. Karolinska Institute, Stockholm Sweden, August 30-31st, 2017

32. **Concerted inflammatory and redox dyshomeostasis in adrenoleukodystrophy.**
    Invited Speaker. 2nd Symposium ELA-European Leukodystrophy Foundation. Luxembourg 4-5th December, 2017
33. **Biallelic mutations of the dihydroceramide desaturase DEGS1 gene cause a novel hypomyelinating leukodystrophy with a therapeutic hope.**
    Late Breaking Talk, Plenary. European Society of Human Genetics, Milan, Italy, June 17-19th 2018.
34. **Degs1, a novel gene causing leukodystrophy**.
    Undiagnosed Disease Network International Congress, Naples, Italy, June 20th, 2018.
35. **Biotin for adrenoleukodystrophy: How does it work?**
    Invited Speaker. 1st Vitamins for Neurodegeneration Symposium, Recordati Rare Diseases, Paris, France, June 26-29thh, 2018.
36. **Precision Medicine for the Leukodystrophies: from diagnostics to therapy in one go.**
    Invited Speaker. 4th International Symposium on Multiple Sclerosis and Myelin Disorders. Berlin, Germany, October 12th-14th 2018.
37. **DEGS1-Leukodystrophy, a novel disorder of sphingolipid metabolism impacts mitochondria dynamics.**
    Late Breaking Talk, Plenary. SSIEM Society for Inherited Errors of Metabolism, Rotterdam, The Netherlands, September 3th- 6th, 2019
    PRIZE OF THE JURY to the BEST TALK
38. **Biallelic Variants in SVBP cause centrosome defects and complex hereditary spastic paraplegia and intellectual disability**
    Invited Speaker. Eight International Conference on Rare and Undiagnosed Diseases, Nijmegen, Holland, February 7th-8th 2020
39. **The lipid transporter ABCD1 causing adrenoleukodystrophy orchestrates organelle cross-talk.**
    Invited Plenary Speaker. 8th FEBS Special Meeting: ABC Proteins – from Genetic Disease to Multidrug Resistance". Innsbruck, Austria, March 1-7th, 2020
40. **Personalized Medicine for adrenoleukodystrophy: identifying biomarkers through multiomics integration**
    Invited Plenary Speaker. United Leukodystrophy Foundation Symposium. Dekalb, Illinois, June 23th-25th, 2020. Virtual Conference.
41. **A novel brain and heart developmental syndrome caused by impairment of the mitochondrial one-carbon metabolism enzyme SHMT2**
    Late Breaking Talk, Plenary. SSIEM Society for Inherited Errors of Metabolism 2020, December 3rd 2020.
    PRIZE OF THE JURY to BEST TALK
42. **Biallelic variants in PIK4A cause hypomyelinating leukodystrophy**
    9th International Conference on Rare and Undiagnosed Diseases, Mayo Clinic Rochester. April 10-12th 2021
43. **Novel metabolic genes causing spastic paraplegias: the SHMT2 and PIA4KA cases**
    Invited Speaker 20th Tom Wahlig Symposium, Essen, Germany Nov 26, 2021

44. **Loss of function of the phosphoinositide kinase PI4KA causes hypomyelinating leukodystrophy**
    14th International Congress of Inborn Errors of Metabolism. ICIEM 2021, Sydney November 21 – 24th 2021
45. **Novel players in ceramide metabolism: DEGS1 loss causes a novel leukodystrophy with therapeutic options.**
    Invited Plenary Speaker. Gordon Research Conference. A Holistic Approach to Understanding Simple and Complex Sphingolipids. Il Cioco, Berga, Italy, March 27 – April 1st, 2022

Other Invited International presentations (Selection)

1. **Potential role of ABCD2/ALDR as therapeutic target for X-ALD treatment in the mouse model**.1st Symposium on Therapy for X-linked adrenoleukodystrophy. Kennedy Krieger Institute, Baltimore, USA, 5th March 2003
2. **Antioxidants halt axonal degeneration and bioenergetic failure in adrenoleukodystrophy**, Department of Neurology and Neurosciences, Weill Cornell Medical College, New York, USA. 7th of December 2010
3. **The loss of Abcd1 impacts bioenergetic and redox homeostasis leading to axonal degeneration,** Department of Pharmacological Sciences, University of Kentucky, Lexington, USA, 9th of December 2010
4. **Oxidative stress and metabolic failure intertwinning underlying axonal degeneration in adrenoleukodystrophy: Novel therapeutic approaches**, Kennedy Krieger Institute, Johns Hopkins University, Baltimore, USA, 17th of November 2011
5. **Intertwining of oxidant stress and bioenergetic failure in adrenoleukodystrophy: Therapeutic implications**, Burke Medical Research Institute, White Plains, New York, USA. 20th of March 2012
6. **Oxidative stress and mitochondria dysfunction in X-adrenoleukodystrophy:** Biogen Inc, Boston, 1st of May 2012**.**
7. **Mitochondrial dysfunction underlying axonal degeneration: therapeutic implications for X-ALD.** Massachusetts General Institute for Neurodegenerative diseases (MIND). Massachusetts General Hospital. Boston, USA, 2nd of May 2012.
8. **Mitochondria dysfunction and bioenergetic failure underlying adrenoleukodystrophy: translation into clinical trials**. Grand Rounds of the Neurology Department, Weill Cornell Medical College, New York, USA, 5th of December 2012
9. **Bioenergetics and Redox dyshomeostasis underlying axonal degeneration in Adrenoleukodystrophy: Translation into Clinical Trials**
   HJKRI- Hunter James Kelly Research Institute University at Buffalo, May 15th, 2013
10. **Adrenoleukodystrophy: Pathology and Therapeutics** Darby Children's Research Institute, Medical University of South Carolina, Charleston, May 22nd, 2013
11. **Redox control of bioenergetics and proteostasis (and viceversa): the adrenoleukodystrophy case.** Center for Mitochondrial and Epigenomic Medicine. Children's Hospital of Philadelphia (CHOP), U Penn, Philadelphia, February 12th, 2015
12. **Precision medicine for the Leukodystrophies: novel genes, mechanisms and therapies.**

Grand Rounds at the Personalized Medicine Department. Mayo Clinic, Minnesota, August 8th, 2018

13. **Design of rational pharmacological approaches for adrenoleukodystrophy**
    Journées d'actualités physiopathologiques de Strasbourg, Faculté de Medecine, Strasbourg, France, December 6th, 2019
14. **Understanding and Treating Metabolic Leukodystrophies: From Bench to Bedside.**
    Invited Speaker. Translational Sessions. Sanofi Framingham, Massachusetts, January 27th-28th, 2020
15. **Implementing Genomic Medicine for Neurological Disorders: Diagnosis, Gene Discovery and Therapy**
    Grand Rounds at the Children's Hospital of Pittsburgh of University of Pittsburg Medical Center. June 30th, 2022