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Life & Medical Sciences

Dr Aurora Pujol received her MD from the Autonomous University of Barcelona in 1993 and her PhD in Cellular and Molecular Biology from the University of Heidelberg/ German Cancer Research Center in 1998. She trained in Human Genetics with Prof Jean Louis Mandel at the IGBMC, Strasbourg, generating and characterizing mouse models for a rare neurometabolic disorder, X-linked adrenoleukodystrophy (X-ALD). In 2002, she obtained a position as clinician in Medical Genetics at the Louis Pasteur University Hospital, and combined genetic diagnosis with fundamental molecular genetics research as Junior Group Leader at the IGBMC. In 2005, she moved back to Barcelona as an ICREA Research Professor and Director of the Neurometabolic Diseases Laboratory at IDIBELL. In 2017 she obtained the certification in Clinical Genomics by the ABMGG, at the NIH, USA. Her lab is of international reference for genomic diagnostics and translational research for the leukodystrophies.

Research interests

Our mission is to improve disease management of rare brain disorders through precision medicine. This spans the whole spectrum from genomic diagnosis to disease-modifying treatments and biomarker identification. A first research line revolves around adrenoleukodystrophy (ALD), a rare neurometabolic disease made popular by the movie Lorenzo's oil. We are applying strategies for integration of –omic approaches to gain insights into pathomechanisms. These involve redox and metabolic homeostasis, mitochondria dynamics, proteostasis and cellular stress responses. Tailored preclinical tests have yielded four licensed patents on reprofilled drugs and three phase II/III clinical trials for ALD. A second research line aims at gene discovery through clinical genomics. We are identifying novel disease-causing genes and modeling disease using iPS-derived organoid cultures and zebrafish. Results are advancing scientific knowledge while serving the undiagnosed community.

Selected publications

– Fourcade S, Morató L, Parameswaran J, Ruiz M, Ruiz-Cortés T, Jové M, Naudí A, Martínez-Redondo A, Dierssen M, Ferrer I, Villarroya F, Pamplona R, Vaquero A, Portero-Otín M, **Pujol A*** 2017, 'Loss of SIRT2 leads to axonal degeneration and locomotor disability associated with redox and energy imbalance', *Aging Cell*, 16(6):1404-141.

– Launay N, Ruiz M, Grau L, Ortega FJ, Ilieva EV, Martínez JJ, Galea E, Ferrer I, Knecht E, Fourcade S. & **Pujol A***. 2017, 'Tauroursodeoxycholic bile acid arrests axonal degeneration by inhibiting the unfolded protein response in X-linked adrenoleukodystrophy'. *Acta Neuropathol.* 133(2):283-301.

– 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, **Pujol A***. Oxidative Stress and Mitochondrial Dynamics Malfunction are linked in Pelizaeus-Merzbacher Disease. Brain Pathol. 2017 Dec 26.

– Falkenberg KD, Braverman NE, Moser AB, Steinberg SJ, Klouwer FCC, Schlüter A, Ruiz M, **Pujol A**, Engvall M, Naess K, Körver-Keularts I, Ferdinandusse S, Wanders RJA, Waterham HR. Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. Am J Hum Genet. 2017 Dec 7;101(6):965-976.

– Mattioli F, Schaefer E, Magee A, Mark P, Mancini GM, Dieterich K, Von Allmen G, Alders M, Coutton C, Vieville G, Engelen M, Cobben JM, Juusola J, **Pujol A**, Mandel JL, Piton A 2017, 'Mutations in Histone Acetylase Modifier BRPF1 Cause an Autosomal-Dominant Form of Intellectual Disability with Associated Ptosis', *Am J Hum Genet.*, 100(1):105-116.

– Kury S, van Woerden GM, Besnard T, Onori MP, Latypova X, Towne MC, Cho MT, Prescott TE, Ploeg MA, Sanders S, Stessman HAF, **Pujol, A** et al; , 'De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability', *Am J Human Genet.* 2017, 101, 5, 768 – 788.

Selected research activities

Diagnosis of over 200 cases of rare neurogenetic disorders using genomics.