

# Research and Diagnosis of Inherited Metabolic Diseases Group



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## Research Activity

### DOCTORAL THESES

Medrano Rodríguez C. Bases Moleculares de los defectos congénitos de glicosilación[dissertation]. Madrid: UAM: 2017(01/03/2017).

Directors: Pérez González B, Pérez Cerdá C.

### BOOKS AND BOOK CHAPTERS

**Cornejo V, Raimann B, Pérez González B, Ruiz Desviat L, Arias C.** Bases moleculares y cromosómicas de las enfermedades genéticas. Hiperfenilalaninemias. In: Colombo M, Cornejo V, Raimann E, (Eds). Errores Innatos en el Metabolismo del Niño. Chile: Editorial universitaria, 2017. Capítulo 3

**Pérez González B, Ruiz Desviat L, Martínez M.** Bases moleculares y cromosómicas de las enfermedades genéticas. Deficiencia de Pterinas. In: Colombo M, Cornejo V, Raimann E, (Eds). Errores Innatos en el Metabolismo del Niño. Chile: Editorial universitaria, 2017. Capítulo 3

**Pérez González B, Ruiz Desviat L.** Bases moleculares y cromosómicas de las enfermedades genéticas. Genética molecular de enfermedades metabólicas hereditarias y herramientas diagnósticas más usadas en genética molecular. In: Colombo M, Cornejo V, Raimann E, (Eds). Errores Innatos en el Metabolismo del Niño. Chile: Editorial universitaria, 2017. Capítulo 1

### PUBLICATIONS

Alonso-Barroso E, Brasil S, Briso-Montiano A, Navarrete R, Pérez-Cerdá C, Ugarte M, Pérez B, Desviat LR, Richard E. Generation and characterization of a human iPSC line from

patient with propionic acidemia due to defects in the PCCA gene. Stem Cell Res. 2017; 23: 173-7. Editorial Material. IF: 1.829; Q3

- Bellusci M, Quijada-Fraile P, Barrio-Carreras D, Martín-Hernández E, García-Silva M, Merinero B, Pérez B, Hernández-Lain A. Carnitine palmitoyltransferase IA deficiency: abnormal muscle biopsy findings in a child presenting with Reye's syndrome. J Inher Metab Dis. 2017; 40(5): 751-2. Editorial Material. IF: 4.092; Q1
- Bravo-Alonso I, Navarrete R, Arribas-Carreira L, Perona A, Abia D, Couce ML, García-Cazorla A, Morais A, Domingo R, Ramos MA, Swanson MA, Van Hove JLG, Ugarte M, Pérez B, Pérez-Cerdá C, Rodríguez-Pombo P. Nonketotic hyperglycinemia: Functional assessment of missense variants in GLDC to understand phenotypes of the disease. Hum Mutat. 2017; 38(6): 678-91. Article. IF: 5.359; Q1
- Cabezas OR, Flanagan SE, Stanescu H, García-Martínez E, Caswell R, Lango-Allen H, Antán-Gamero M, Argente J, Busse AM, Brandli A, Cheshire C, Crowne E, Dumitriu S, Drynda R, Hamilton-Shield JP, Hayes W, Hofherr A, Iancu D, Issler N, Jefferies C, Jones P, Johnson M, Kesselheim A, Klootwijk E, Koettgen M, Lewis W, Martos JM, Mozere M, Norman J, Patel V, Parrish A, Pérez-Cerdá C, Pozo J, Rahman SA, Sebire N, Tekman M, Turnpenny PD, van't Hoff W, Viering DHHM, Weedon MN, Wilson P, Guay-Woodford L, Kleta R, Hussain K, Ellard S, Bockenhauer D. Polycystic kidney disease with hyperinsulinemic hypoglycemia caused by a promoter mutation in phosphomannomutase 2. J Am Soc Nephrol. 2017; 28(8): 2529-39. Article. IF: 8.655; D1
- Casado M, Ferrer-López I, Ruiz-Sala P, Pérez-Cerdá C, Artuch R. Urine oligosaccharide tests for the diagnosis of oligosaccharidoses. Rev Anal Chem. 2017; 36(3): 20160019. Article. IF: 2.111; Q2
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- Coughlin CR, Swanson MA, Kronquist K, Acquaviva C, Hutchin T, Rodríguez-Pombo P, Vaisanen ML, Spector E, Creadon-Swindell



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  - Rivera-Barahona A, Fulgencio-Covián A, Pérez-Cerdá C, Ramos R, Barry MA, Ugarte M, Pérez B, Richard E, Desviat LR. Dysregulated miRNAs and their pathogenic implications for the neurometabolic disease propionic acidemia. *SCI Rep-Uk*. 2017; 7: 5727. Article. IF: 4.122; Q1
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## PUBLIC PROJECTS

**Pérez González MB.** Enfermedades neurometabólicas hereditarias: avances en el diagnóstico, la patofisiología y desarrollo de terapias dirigidas a estabilizar proteínas. (PI16/00573). ISCIII. 2017-2019.

Management centre: UAM

**Ruiz Desviat MI.** Propionic acidemia: from models and mechanisms to biomarkers and therapies. (SAF2016-76004-R). MINECO. 2017-2019.

Management centre: UAM

## PRIVATE PROJECTS

**Pérez González MB.** Bases moleculares de las enfermedades neurometabólicas y desarrollo de terapias específicas de mutación. Fundación Isabel Gemio. 2016-2020.

Management centre: Fundación Severo Ochoa

**Pérez González MB.** Bases moleculares del síndrome de deficiencia Glut1 (Glut1ds) e investigación en terapias. CIBERER. 2017-Ongoing.

Management centre: CIBERER

**Ruiz Desviat L.** Análisis genómicos y transcriciómicos para identificar defectos de Splicing y evaluación in vivo de la terapia antisentido. Fundación Ramón Areces. 2015-2018.

Management centre: Fundación Severo Ochoa

## INTERNATIONAL PROJECTS

**Pérez González MB.** European research network directed towards improving diagnosis and treatment of inborn error of glycosylation (E-rare-3 JTC2015). European Union. 2016-2018.

Management centre: UAM

**Pérez González MB.** Strengthening the research potential of image through reinforcement of biomedical science of rare diseases in Serbia – en route for innovation (Serbordisinn) (FP7-REGPOT). European Union. 2013-2016.

Management centre: UAM

**Ruiz Desviat MI.** Networking towards clinical application of antisense-mediated exon skipping. European cooperation in science and technology (COST BM1207). European Union. 2013-2017.

Management centre: UAM

## PATENTS AND TRADEMARKS

**Pérez González B, Ruiz Desviat L, Jorge Finnigan A, Ugarte Pérez M, Martínez Ruiz A, Underhaug J, Barnejee R,** inventors; UAM, Universidad de Bergen, Universidad de Michigan, assignees; Compounds useful for the treatment of methylmalonic aciduria. P201330171; 2013 February 11.

**Pérez González B, Gámez Abascal A, Yuste Checa P, Arduim Brasil S, Ugarte Pérez M, Martínez Ruiz A, Underhaug J,** inventors; UAM, Universidad de Bergen, assignees; Compounds for treating congenital disorders of glycosylation. EPI6382373.5; 2016.