

## RESEARCH AND DIAGNOSIS OF INHERITED METABOLIC DISEASES GROUP

 **Publications: 13**

 **Impact Factor: 41.87**

 **Q1: 6**



### PUBLICATIONS

- Alcaide P, Krijt J, Ruiz-Sala P, Jesina P, Ugarte M, Kozich V, Merinero B. Enzymatic diagnosis of homocystinuria by determination of cystathionine-beta-synthase activity in plasma using LC-MS/MS. *Clin Chim Acta*. 2015;438:261-5. Article. IF: 2.799; Q1.
- Brasil S, Richard E, Jorge-Finnigan A, Leal F, Merinero B, Banerjee R, Desviat LR, Ugarte M, Pérez B. Methylmalonic aciduria cblB type: characterization of two novel mutations and mitochondrial dysfunction studies. *Clin Genet*. 2015;87(6):576-81. Article. IF: 3.892; Q2.
- Huemer M, Kozich V, Rinaldo P, Baumgartner MR, Merinero B, Pasquini E, Ribes A, Blom HJ. Newborn screening for homocystinurias and methylation disorders: systematic review and proposed guidelines. *J Inherit Metab Dis*. 2015;38(6):1007-19. Review. IF: 3.541; Q2.NO
- Matos L, Goncalves V, Pinto E, Laranjeira F, Prata MJ, Jordán P, Desviat LR, Pérez B, Alves S. Functional analysis of splicing mutations in the IDS gene and the use of antisense oligonucleotides to exploit an alternative therapy for MPS II. *Bba-Mol Basis Dis*. 2015;1852(12):2712-21. Article. IF: 5.158; Q1.
- Richard E, Pérez B, Pérez-Cerdá C, Desviat LR. Understanding molecular mechanisms in propionic acidemia and investigated therapeutic strategies. *Expert Opin Orphan D*. 2015;3(12):1427-38. Review. IF: 0.464; Q4.
- Rivera-Barahona A, Sánchez-Alcudia R, Viecelli HM, Rufenacht V, Pérez B, Ugarte M, Haberle J, Thony B, Desviat LR. Functional characterization of the SPF/ASH splicing variation in otc deficiency of mice and man. *Plos One*. 2015;10(4):e0122966. Article. IF: 3.057; Q1.
- Serrano M, de Diego V, Muchart J, Cuadras D, Felipe A, Macaya A, Velázquez R, Poo MP, Fons C, O'Callaghan MM, García-Cazorla A, Boix C, Robles B, Carratala F, Giros M, Briones P, Gort L, Artuch R, Pérez-Cerdá C, Jaeken J, Pérez B, Pérez-Dueñas B. Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. *Orphanet J Rare Dis*. 2015;10:138. Article. IF: 3.29; Q2.
- Stojiljkovic M, Klaassen K, Djordjevic M, Sarajlija A, Kecman B, Ugrin M, Zukic B, Desviat LR, Pavlovic S, Pérez B. Tetrahydrobiop-  
 terin deficiency among Serbian patients presenting with hyperphenylalaninemia. *J Pediatr Endocr Met*. 2015;28(42463):477-80. Article. IF: 0.912; Q4.
- Vallejo-Torres L, Castilla I, Couce ML, Pérez-Cerdá C, Martín-Hernández E, Pineda M, Campistol J, Arrospide A, Morris S, Serrano-Aguilar P. Cost-effectiveness analysis of a national newborn screening program for biotinidase deficiency. *Pediatrics*. 2015;136(2):E424-32. Article. IF: 5.196; D1.
- Vitoria I, Martín-Hernández E, Peña-Quintana L, Bueno M, Quijada-Fraile P, Dalmau J, Molina-Marrero S, Pérez B, Merinero B. Carnitine-acylcarnitine translocase deficiency: experience with four cases in Spain and review of the literature. *JIMD Rep*. 2015;20:11-20. Article. Not Indexed



### 3.5 Cancer and Human Molecular Genetics Area

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- Yuste-Checa P, Gámez A, Brasil S, Desviat LR, Ugarte M, Pérez-Cerdá C, Pérez B. The effects of PMM2-CDG-causing mutations on the folding, activity, and stability of the PMM2 protein. *Hum Mutat.* 2015;36(9):851-60. Article. IF: 5.089; Q1.
- Yuste-Checa P, Medrano C, Gámez A, Desviat LR, Matthijs G, Ugarte M, Pérez-Cerdá C, Pérez B. Antisense-mediated therapeutic pseudoexon skipping in TMEM165-CDG. *Clin Genet.* 2015;87(1):42-8. Article. IF: 3.892; Q2.

