

# Research and Diagnosis of Inherited Metabolic Diseases

## PUBLICATIONS

- \* Aldámiz-Echevarría L, Llarena M, Bueno MA, Dalmau J, Vitoria I, Fernández-Marmiesse A, Andrade F, Blasco J, Alcalde C, Gil D, García MC, González-Lamuno D, Ruiz M, Ruiz MA, Peña-Quintana L, González D, Sánchez-Valverde F, Desviat LR, Pérez B, Couce ML. Molecular epidemiology, genotype-phenotype correlation and BH4 responsiveness in Spanish patients with phenylketonuria. *J Hum Genet.* 2016;61(8):731-44. Article. IF: 2.471; Q3
- \* Gallego-Villar L, Rivera-Barahona A, Cuevas-Martín C, Guenzel A, Pérez B, Barry MA, Murphy MP, Logan A, González-Quintana A, Martín MA, Medina S, Gil-Izquierdo A, Cuezva JM, Richard E, Desviat LR. In vivo evidence of mitochondrial dysfunction and altered redox homeostasis in a genetic mouse model of propionic acidemia: Implications for the pathophysiology of this disorder. *Free Radical Bio Med.* 2016;96:1-12. Article. IF: 5.606; Q1
- \* Jansen JC, Cirak S, van Scherpenzeel M, Timal S, Reunert J, Rust S, Pérez B, Vicogne D, Krawitz P, Wada Y, Ashikov A, Pérez-Cerdá C, Medrano C, Arnoldy A, Hoischen A, Huijben K, Steenbergen G, Quelhas D, Diogo L, Rymen D, Jaeken J, Guffon N, Cheillan D, van den Heuvel LP, Maeda Y, Kaiser O, Schara U, Gerner P, van den Boogert MAW, Holleboom AG, Nassogne MC, Sokal E, Salomon J, van den Bogaart G, Drenth JPH, Huynen MA, Veltman JA, Wevers RA, Morava E, Matthijs G, Foulquier F, Marquardt T, Lefeber DJ. CCDC115 deficiency causes a disorder of golgi homeostasis with abnormal protein glycosylation. *Am J Hum Genet.* 2016;98(2):310-21. Article. IF: 9.025; D1
- \* Martínez-Pizarro A, Desviat LR, Ugarte M, Pérez B, Richard E. Endoplasmic reticulum stress and autophagy in homocystinuria patients with remethylation defects. *Plos One.* 2016;11(3):e0150357. Article. IF: 2.806; Q1
- \* Ortigoza-Escobar JD, Molero-Luis M, Arias A, Marti-Sánchez L, Rodríguez-Pombo P, Artuch R, Pérez-Dueñas B. Treatment of genetic defects of thiamine transport and metabolism. *Expert Rev Neurother.* 2016;16(7):755-63. Review. IF: 3.149; Q2
- \* Ortigoza-Escobar JD, Molero-Luis M, Arias A, Oyarzábal A, Darín N, Serrano M, García-Cazorla A, Tondo M, Hernández M, García-Villoria J, Casado M, Gort L, Mayr JA, Rodríguez-Pombo P, Ribes A, Artuch R, Pérez-Dueñas B. Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. *Brain.* 2016;139(1):31-8. Article. IF: 10.292; D1
- \* Ortigoza-Escobar JD, Oyarzábal A, Montero R, Artuch R, Jou C, Jiménez C, Gort L, Briones P, Muchart J, López-Gallardo E, Emperador S, Pesini ER, Montoya J, Pérez B, Rodríguez-Pombo P, Pérez-Dueñas B. Ndufs4 related Leigh syndrome: A case report and review of the literature. *Mitochondrion.* 2016;28:73-8. Review. IF: 3.704; Q2
- \* Oyarzabal A, Bravo-Alonso I, Sánchez-Arago M, Rejas MT, Merinero B, García-Cazorla A, Artuch R, Ugarte M, Rodríguez-Pombo P. Mitochondrial response to the BCKDK-deficiency: Some clues to understand the positive dietary response in this form of autism. *Bba-Mol Basis Dis.* 2016;1862(4):592-600. Article. IF: 5.476; Q1
- \* Sala PR, Ruijter G, Acquaviva C, Chabli A, de Sain-van der Velden MGM, García-Villoria J, Heiner-Fokkema MR, Jeannesson-Thivisol E, Leckstrom K, Franzson L, Lynes G, Olesen J, Onkenhout W, Petrou P, Drousiotou A, Ribes A, Vianey-Saban C, Merinero B. Pilot

experience with an external quality assurance scheme for acylcarnitines in plasma/serum. *JIMD Rep.* 2016;30:23-31. Article. IF: Not Indexed

- \* Stojiljkovic M, Klaassen K, Djordjevic M, Sarajlija A, Brasil S, Kecman B, Grkovic S, Kostic J, Rodríguez-Pombo P, Desviat LR, Pavlovic S, Pérez B. Molecular and phenotypic characteristics of seven novel mutations causing branched-chain organic acidurias. *Clin Genet.* 2016;90(3):252. Article. IF: 3.326; Q2
- \* Vega AI, Medrano C, Navarrete R, Desviat LR, Merinero B, Rodríguez-Pombo P, Vitoria I, Ugarte M, Pérez-Cerdá C, Pérez B. Molecular diagnosis of glycogen storage disease and disorders with overlapping clinical symptoms by massive parallel sequencing. *Genet Med.* 2016;18(10):1037-43. Article. IF: 8.229; D1