

Dislipemias de Origen Genético y Enfermedades Metabólicas

Publicaciones: 6

Q1: 1



ACTIVIDAD DE INVESTIGACIÓN

Publicaciones

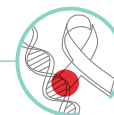
- Gómez-González C, Rosas-Alonso R, Rodríguez-Antolín C, García-Guede A, Ibáñez de Cáceres I, Sanguino J, Pascual SI, Esteban I, del Pozo A, Mori MA, Torres RJ, Prior C. Symptomatic heterozygous X-Linked myotubular myopathy female patient with a large deletion at Xq28 and decrea-

se expression of normal allele. Eur J Med Genet. 2021; 64(4): 104170. Article. IF: 2.465; Q3

- Mazo MA, Sanguino J, Martín-Gullón I, Rubio J. Formation of carbon nanofibers with Ni catalyst supported on a micro-mesoporous glass. Micropor Mesopor Mat. 2021; 323: 111168. Article. IF: 5.876; Q1
- Rodríguez-Antolín C, Rosas-Alonso R, Cruz

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- Rosas-Alonso R, Queiruga J, Arias P, del Monte A, Yuste F, Rodríguez-Antolín C, Losantos-García I, Borobia AM, Rodríguez-Novoa S. Analytical validation of a laboratory-development multi-gene pharmacogenetic assay. Pharmacogenet Genom. 2021; 31(8): 177-84. Article. IF: 2; Q4
- Santos-Simarro F, Pacio M, Cueto-González AM, Mansilla E, Valenzuela-Palafoff MI, López-Grondo-



na F, Lledin MD, Schuffelmann C, del Pozo A, Solís M, Vallcorba P, Lapunzina P, Suso JJM, Siccha SM, Montejo JM, Mena R, Jiménez-Rodríguez C, García-Miñaur S, Palomares-Bralo M. Mosaic variegated aneuploidy syndrome 2 caused by biallelic variants in CEP57, two new cases and review of the phenotype. *Eur J Med Genet.* 2021; 64(11): 104338. Review. IF: 2.465; Q3

- Siccha SM, Cueto AM, Parrón-Pajares M, González-Morán G, Pacio-Míguez M, del Pozo A, Solís M, Rodríguez-Jiménez C, Caino S, Fano V, Heath KE, García-Miñaur S, Palomares-Bralo M, Santos-Simarro F. Delineation of the clinical and radiological features of Stuve-Wiedemann syndrome childhood survivors, four new cases and review of the literature. *Am J Med Genet A.* 2021; 185(3): 856-65. Review. IF: 2.578; Q3

■ Proyectos de investigación

Rodríguez Novoa SM. Diagnóstico genético de la hipercolesterolemia familiar mediante secuenciación masiva. Estudio funcional de nuevas variantes y detección de mosaïcismo. Estudio de miRNAs (PI18/00917). ISCIII. 2019-2021.

Centro de Gestión: FIBHULP

Rodríguez Novoa SM. Renal tubular and markers of bone turnover in hbv mono-infected patients during long term treatment with entecavir or tenofovir. Bristol-Myers Squibb International Corporation. 2011-Ongoing.

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■ Patentes y marcas

Rodríguez Novoa SM, del Monte Vergara A, Rosas Alonso R, Queiruga Parada J, Yuste González F, authors; FIBHULP, assignee. Trademark name: Pharma Genfinder; CM18332183; 2020 November 05.



Ibáñez de Cáceres I, de Castro Carpeño J, Jiménez Hernández J, Rodríguez Antolín C, Rodríguez Jiménez C, Rosas Alonso R, Cruz Castellanos P, Burdiel Herencia M, Pernía Arias O, Diestro Tejada MD, Esteban Rodríguez MI, inventors; FIBHULP, assignee. miR-151A-3p as an universal endogenous control for exosome cargo normalization. EP19382252.5 (EP3719144), PCT/EP2020/059774 PCT Direct, EP20719957.1 (EP3947733), US17/601,657; 2019 April 05.