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 decrease 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 P, Higuera O, Sánchez-Cabrero D, Esteban-Rodríguez I, Peláez-García A, Montano VEF, Rodríguez-Jiménez C, de Cáceres II, de Castro J. Novel SLC12A2-ROS1 fusion in non-small cell lung cancer with a significant response to crizotinib: the importance of choosing the appropriate next-generation sequencing assay. Oncologist. 2021; 26(6): E908-12. Article. IF: 5.837; Q2
- 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 multigene pharmacogenetic assay. Pharmacogenet Genom. 2021; 31(8): 177-84. Article. IF: 2; Q4
- Santos-Simarro F, Pacio M, Cueto-González AM, Mansilla E, Valenzuela-Palafoll MI, López-Grondo-

na F, Lledin MD, Schuffelmann C, del Pozo A, Solis 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, Solis 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 mosaicismo. Estudio de miRNAs (PI18/00917). ISCIII. 2019-2021.

Centro de Gestión: FIBHULP

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

Centro de Gestión: FIBHULP

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.

PHARMA GENFINDER

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.