

INGEMM - INSTITUTO DE GENETICA MEDICA Y MOLECULAR (INSTITUTE OF MEDICAL AND MOLECULAR GENETICS) GROUP



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QI: **14**



PUBLICATIONS

- Abascal F, Ezkurdia I, Rodríguez-Rivas J, Rodríguez JM, del Pozo A, Vázquez J, Valencia A, Tress ML. Alternatively spliced homologous exons have ancient origins and are highly expressed at the protein level. *Plos Comput Biol.* 2015;11(6):e1004325. Article. IF: 4.587; Q1.
- Barroso E, Berges-Soria J, Benito-Sanz S, Rivera-Pedroza CI, Ballesta-Martínez MJ, López-González V, Guillén-Navarro E, Heath KE. Identification of the fourth duplication of upstream ihh regulatory elements, in a family with craniosynostosis philadelphia type, helps to define the phenotypic characterization of these regulatory elements. *Am J Med Genet A.* 2015;167(4):902-6. Letter. IF: 2.082; Q3.
- Caparros-Martin JA, de Luca A, Cartault F, Aglan M, Temtamy S, Otaify GA, Mehrez M, Valencia M, Vázquez L, Alessandri JL, Nevado J, Rueda-Arenas I, Heath KE, Digilio MC, Dallapiccola B, Goodship JA, Mill P, Lapunzina P, Ruiz-Pérez VL. Specific variants in WDR35 cause a distinctive form of Ellis-van Creveld syndrome by disrupting the recruitment of the EvC complex and SMO into the cilium. *Hum Mol Genet.* 2015;24(14):4126-37. Article. IF: 5.985; DI.
- Carcavilla A, García-Miñaur S, Pérez-Aytes A, Vendrell T, Pinto I, Guillén-Navarro E, González-Meneses A, Aoki Y, Grinberg D, Ezquieta B. Cardiofaciocutaneous syndrome, a Noonan syndrome related disorder: Clinical and molecular findings in 11 patients. *Med Clin.* 2015;144(2):67-72. Article. IF: 1.267; Q2.
- Cardoso LCD, Rodríguez-Laguna L, Crespo MD, Vallespín E, Palomares-Bralo M, Martín-Arenas R, Rueda-Arenas I, de Faria PAS, García-Miguel P, Lapunzina P, Vargas FR, Seuanez HN, Martínez-Glez V. Array CGH Analysis of paired blood and tumor samples from patients with sporadic wilms tumor. *Plos One.* 2015;10(8):e0136812. Article. IF: 3.057; Q1.
- Ezkurdia I, Calvo E, del Pozo A, Vázquez J, Valencia A, Tress ML. The potential clinical impact of the release of two drafts of the human proteome. *Expert Rev Proteomic.* 2015;12(6):579-93. Review. IF: 3.465; Q2.
- Ferreira S, Ortiz A, Germain DP, Viana-Baptista M, Caldeira-Gomes A, Camprecios M, Fenollar-Cortes M, Gallegos-Villalobos A, García D, García-Robles JA, Egido J, Gutiérrez-Rivas E, Herrero JA, Mas S, Oancea R, Peres P, Salazar-Martin LM, Solera-García J, Alves H, Garman SC, Oliveira JP. The alpha-galactosidase A p.Arg118Cys variant does not cause a Fabry disease phenotype: Data from individual patients and family studies. *Mol Genet Metab.* 2015;114(2):248-58. Article. IF: 3.093; Q2.
- Fung WLA, Butcher NJ, Costain G, Andrade DM, Boot E, Chow EWC, Chung B, Cytrynbaum C, Faghfouri H, Fishman L, García-Miñaur S, George S, Lang AE, Repetto G, Shugar A, Silversides C, Swillen A, van Amelsvoort T, McDonald-McGinn DM, Bassett AS. Practical guidelines for managing adults with 22q11.2 deletion syndrome. *Genet Med.* 2015;17(8):599-609. Review. IF: 7.71; DI.
- García-Santiago FA, Martínez-Glez V, Santos F, García-Miñaur S, Mansilla E, Meneses AG, Rosell J, Granero AP, Vallespín E, Fernández L, Sierra B, Oliver-Bonet M, Palomares M, de Torres ML, Mori MA, Nevado J, Heath KE, Delicado A, Lapunzina P. Analysis of Invdupdel(8p) rearrangement: clinical, cytogenetic and molecular characterization. *Am J Med Genet A.* 2015;167(5):1018-25. Article. IF: 2.082; Q3.



3.5 Cancer and Human Molecular Genetics Area

- Gordo-Gilart R, Andueza S, Hierro L, Martínez-Fernández P, D'Agostino D, Jara P, Álvarez L. Functional analysis of ABCB4 mutations relates clinical outcomes of progressive familial intrahepatic cholestasis type 3 to the degree of MDR3 floppase activity. *Gut*. 2015;64(1):147-55. Article. IF: 14.921; DI.
- Gutiérrez-Repiso C, Colomo N, Rojo-Martínez G, Valdés S, Tapia MJ, Esteva I, de Adana MSR, Rubio-Martín E, Lago-Sampedro A, Santiago P, Velasco I, García-Fuentes E, Moreno JC, Soriguer F. Evolution of urinary iodine excretion over eleven years in an adult population. *Clin Nutr*. 2015;34(4):712-8. Article. IF: 4.487; Q1.
- Hisado-Oliva A, Garre-Vázquez Al, Santaolalla-Caballero F, Belinchón A, Barreda-Bonis AC, Vasques GA, Ramírez J, Luzuriaga C, Carlone G, González-Casado I, Benito-Sanz S, Jorge AA, Campos-Barros A, Heath KE. Heterozygous NPR2 mutations cause disproportionate short stature, similar to Leri-Weill Dyschondrosteosis. *J Clin Endocr Metab*. 2015;100(8):E1133-42. Article. IF: 5.531; Q1.
- Kamieniak MM, Rico D, Milne RL, Muñoz-Repeta I, Ibáñez K, Grillo MA, Domingo S, Borrego S, Cazorla A, García-Bueno JM, Hernando S, García-Donas J, Hernández-Agudo E, Cajal TRY, Robles-Díaz L, Márquez-Rodas I, Cusido M, Sáez R, Lacambra-Calvet C, Osorio A, Urioste M, Cigudosa JC, Paz-Ares L, Palacios J, Benítez J, García MJ. Deletion at 6q24.2-26 predicts longer survival of high-grade serous epithelial ovarian cancer patients. *Mol Oncol*. 2015;9(2):422-36. Article. IF: 5.367; Q1.
- Ley-Martos M, Salado-Reyes MJ, Espinosa-Rosso R, Solera-García J, Jiménez-Jiménez L. Variability in the clinical presentation of Pompe disease: development following enzyme replacement therapy. *Rev Neurologia*. 2015;61(9):416-20. Article. IF: 0.684; Q4.
- Lupiáñez DG, Kraft K, Heinrich V, Krawitz P, Brancati F, Kloppock E, Hom D, Kayserili H, Opitz JM, Laxova R, Santos-Simarro F, Gilbert-Dussardier B, Wittler L, Borschwiwer M, Haas SA, Osterwalder M, Franke M, Timmermann B, Hecht J, Spielmann M, Visel A, Mundlos S. Disruptions of topological chromatin domains cause pathogenic rewiring of gene-enhancer interactions. *Cell*. 2015;161(5):1012-1025. Article. IF: 28.71; DI.
- Mattos EP, da Silva AA, Magalhaes JAA, Leite JCL, Leistner-Segal S, Gus-Kessler R, Pérez JA, Vedolín LM, Torreblanca-Zanca A, Lapunzina P, Ruiz-Pérez VL, Sanseverino MTV. Identification of a premature stop codon mutation in the PHGDH gene in severe Neu-Laxova syndrome evidence for phenotypic variability. *Am J Med Genet A*. 2015;167(6):1323-29. Article. IF: 2.082; Q3.
- Moccia E, Guillén-Ponce C, Earl J, Márquez M, Solera J, Salazar-López MT, Calcedo-Arnaiz C, Vázquez-Sequeiros E, Montans J, Muñoz-Beltran M, Vicente-Bartulos A, González-Gordaliza C, Sanjuanbenito A, Guerrero C, Mendaña E, Lisa E, Lobo E, Martínez JC, Real FX, Malats N, Carrato A. PanGen-Fam: Spanish registry of hereditary pancreatic cancer. *Eur J Cancer*. 2015;51(14):1911-7. Article. IF: 6.163; Q1.
- Nevado J, Rosenfeld JA, Mena R, Palomares-Bralo M, Vallespin E, Mori MA, Tenorio JA, Gripp KW, Denenberg E, del Campo M, Plaja A, Martín-Arenas R, Santos-Simarro F, Armengol L, Gowans G, Orera M, Sánchez-Hombre MC, Corbacho-Fernández E, Fernández-Jaen A, Haldeman-Englert C, Saitta S, Dubbs H, Benedicte DB, Li X, Devaney L, Dinulos MB, Vallee S, Crespo MC, Fernández B, Fernández-Montano VE, Rueda-Arenas I, de Torres ML, Ellison JW, Raskin S, Venegas-Vega CA, Fernández-Ramárez F, Delicado A, García-Miñaur S, Lapunzina P. PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. *Eur J Hum Genet*. 2015;23(12):1615-26. Article. IF: 4.58; Q1.
- Paumard-Hernández B, Berges-Soria J, Barroso E, Rivera-Pedroza CI, Pérez-Carrizosa V, Benito-Sanz S, López-Messa E, Santos F, García-Recuero II, Romance A, Ballesta-Martínez MJ, López-González V, Campos-Barros A, Cruz J, Guillen-Navarro E, del Pozo JS, Lapunzina P, García-Miñaur S, Heath KE. Expanding the mutation spectrum in 182 Spanish probands with craniosynostosis: identification and characterization of novel TCF12 variants. *Eur J Hum Genet*. 2015;23(7):907-14. Article. IF: 4.58; Q1.
- Rojnueangnit K, Xie J, Gomes A, Sharp A, Callens T, Chen YJ, Liu Y, Cochran M, Abbott MA, Atkin J, Babovic-Vuksanovic D, Barnett CP, Crenshaw M, Bartholomew DW, Basel L, Bellus G, Ben-Shachar S, Bialer MG, Bick D, Blumberg B, Cortes F, David KL, Destree A, Duat-Rodríguez A, Earl D, Escobar L, Eswara M, Ezquieta B, Frayling IM, Frydman M, Gardner K, Gripp KW, Hernandez-Chico C, Heyrman K, Ibrahim J, Janssens S, Keena BA, Llano-Rivas I, Leppig K, McDonald M, Misra VK, Mulbury J, Narayanan V, Orenstein N, Galvin-Parton P, Pedro H, Pivnick EK, Powell CM, Randolph L, Raskin S, Rosell J, Rubin K, Seashore M, Schaaf CP, Scheuerle A, Schultz M, Schorry E, Schnur R, Siqueland E, Tkachuk A, Tonsgard J, Upadhyaya M, Verma IC, Wallace S, Williams C, Zackai E, Zonana J, Lazaro C, Claes K, Korf B, Martin Y, Legius E, Messiaen L. High Incidence of Noonan Syndrome Features including short stature and pulmonic stenosis in patients carrying *nfl* missense mutations affecting p.ARG1809: Genotype-phenotype correlation. *Hum Mutat*. 2015;36(11):1052-63. Article. IF: 5.089; Q1.
- Sánchez TB, Rodríguez AD, Extremera VC, Lapunzina P, Bralo MP, Blanco JN. Clinical phenotype of a patient with FOXP1 deletion. *An Pediatr*. 2015;82(4):280-1. Letter. IF: 0.773; Q4.
- Tenorio J, Navas P, Barrios E, Fernández L, Nevado J, Quezada CA, López-Meseguer M, Arias P, Mena R, Lobo JL, Álvarez C, Heath K, Escribano-Subias P, Lapunzina P. A founder EIF2AK4 mutation causes an aggressive form of pulmonary arterial hypertension in Iberian Gypsies. *Clin Genet*. 2015;88(6):579-83. Article. IF: 3.892; Q2.
- Valencia M, Tabet L, Yazbeck N, Araújo A, Ruiz-Pérez VL, Charaffedine K, Fares F, Badra R, Farra C. Ellis-van Creveld Syndrome: mutations uncovered in lebanese families. *Case Rep Genet*. 2015;2015:528481. Article. Not Indexed
- Veriñ H, Fernández-Miñano A, Benito-Sanz S, Janssens S, Callewaert B, de Waele K, de Schepper J, Francois I, Menten B, Heath KE, Gómez-Skarmeta JL, de Baere E. Profiling of conserved non-coding elements upstream of SHOX and functional characterisation of the SHOX cis-regulatory landscape. *Sci Rep-Uk*. 2015;5: 17667. Article. IF: 5.228; Q1.