INGEMM - INSTITUTO DE GENÉTICA MÉDICA Y MOLECULAR

Altmuller, F; Lissewski, C; Bertola, D; Flex, E; Stark, Z; Spranger, S; Baynam, G; Buscarilli, M; Dyack, S; Gillis, J; Yntema, HG; Pantaleoni, F; van Loon, RLE; MacKay, S; Mina, K; Schanze, I; Tan, TY; Walsh, M; White, SM; Niewisch, MR; Garcia-Minaur, S; Plaza, D; Ahmadian, MR; Cave, H; Tartaglia, M; Zenker, M. Genotype and phenotype spectrum of NRAS germline variants. EUROPEAN JOURNAL OF HUMAN GENETICS. 2017; 25(7):823-831.Article. IF -4,287

Andrade, RC; dos Santos, ACE; Neto, JCD; Nevado, J; Lapunzina, P; Vargas, FR. TP53 and CDKN1A mutation analysis in families with Li-Fraumeni and Li-Fraumeni like syndromes. FAMILIAL CANCER. 2017; 16(2):243-248.Article. IF -1,66

Barraza-Garcia, J; Rivera-Pedroza, CI; Hisado-Oliva, A; Belinchon-Martinez, A; Sentchordi-Montane, L; Duncan, EL; Clark, GR; del Pozo, A; Ibanez-Garikano, K; Offiah, A; Prieto-Matos, P; Cormier-Daire, V; Heath, KE. Broadening the phenotypic spectrum of POP1-skeletal dysplasias: identification of POP1 mutations in a mild and severe skeletal dysplasia. CLINICAL GENETICS. 2017; 92(1):91-98.Article. IF -3,326

Benito-Sanz, S; Belinchon-Martinez, A; Aza-Carmona, M; de la Torre, C; Huber, C; Gonzalez-Casado, I; Ross, JL; Thomas, NS; Zinn, AR; Cormier-Daire, V; Heath, KE. Identification of 15 novel partial SHOX deletions and 13 partial duplications, and a review of the literature reveals intron 3 to be a hotspot region. JOURNAL OF HUMAN GENETICS. 2017; 62(2):229-234.Review. IF -2,471

Blanco-Kelly, F; Palomares, M; Vallespin, E; Villaverde, C; Martin-Arenas, R; Velez-Monsalve, C; Lorda-Sanchez, I; Nevado, J; Trujillo-Tiebas, MJ; Lapunzina, P; Ayuso, C; Corton, M. Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH. PLOS ONE. 2017; 12(2):-e0172363.Article. IF -2,806

Blanco-Lago, R; Malaga-Dieguez, I; Granizo-Martinez, JJ; Carrera-Garcia, L; Barruz-Galian, P; Lapunzina, P; Nevado-Blanco, J. Wolf-Hirschhorn syndrome. Description of a Spanish cohort of 51 cases and a literature review. REVISTA DE NEUROLOGIA. 2017; 64(9):393-400.Review. IF -0,743

Borobia, Alberto M; Dapia, Irene; Tong, Hoi Y; Arias, Pedro; Munoz, Mario; Tenorio, Jair; Hernandez, Rafael; Garcia Garcia, Irene; Gordo, Gema; Ramirez, Elena; Frias, Jesus; Lapunzina, Pablo; Carcas, Antonio J. Clinical Implementation of Pharmacogenetic

Testing in a Hospital of the Spanish National Health System: Strategy and Experience Over 3 Years.. Clinical and translational science. 2017; ():-.Article. IF -0,86

Caparros-Martin, JA; Aglan, MS; Temtamy, S; Otaify, GA; Valencia, M; Nevado, J; Vallespin, E; Del Pozo, A; de Castro, CP; Calatrava-Ferreras, L; Gutierrez, P; Bueno, AM; Sagastizabal, B; Guillen-Navarro, E; Ballesta-Martinez, M; Gonzalez, V; Basaran, SY; Buyukoglan, R; Sarikepe, B; Espinoza-Valdez, C; Cammarata-Scalisi, F; Martinez-Glez, V; Heath, KE; Lapunzina, P; Ruiz-Perez, VL. Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. MOLECULAR GENETICS & GENOMIC MEDICINE. 2017; 5(1):28-39.Article. IF -NO TIENE

Corcuera, BL; Jimenez, E; Bartolome-Martin, D; Zafra, F; Lapunzina, P; Aragon, C; Villarejo-Lopez, L. Regulation of the neuronal glycine transporter GLYT2 by P2X purinergic receptors. JOURNAL OF NEUROCHEMISTRY. 2017; 142():190-190.Meeting Abstract. IF -4,083

Del Rey, G; Gryngarten, MG; Barruz, P; Alvarez, MAM; Venara, MC; Azrak, SR; Arcari, AJ; Casali, B; Boywitt, A; De Bellis, R; Escobar, ME; Lapunzina, P; Nevado, J. DELETION UPSTREAM OF SOX9 IN A 46 XY FEMALE WITHOUT CAMPOMELIC DYSPLASIA, AND IDENTIFIED IN HER MOTHER WITH PREMATURE OVARIAN FAILURE. HORMONE RESEARCH IN PAEDIATRICS. 2017; 88():555-556.Meeting Abstract. IF -1,844

Escudero, A; Carrasco, P; Nevado, J; Palomares, M; Vallespin, E; Lapunzina, P; Fernandez, L; Vela, M; Gonzalo, I; Gonzalez, B; Bueno, D; del Pozo, A; Perez-Martinez, A. HIGH RESOLUTION TECHNOLOGIES IN B-CELL ACUTE LYMPHOBLASTIC LEUKEMIA. HAEMATOLOGICA. 2017; 102():659-659.Meeting Abstract. IF -7,702

Fernandez, Luis; Tenorio, Jair; Polo-Vaquero, Coral; Vallespin, Elena; Palomares-Bralo, Maria; Garcia-Minaur, Sixto; Santos-Simarro, Fernando; Arias, Pedro; Carnicer, Hernan; Giannivelli, Silvina; Medina, Juan; Perez-Piaya, Rosa; Solis, Jorge; Rodriguez, Monica; Villagra, Alexandra; Rodriguez, Laura; Nevado, Julian; Martinez-Glez, Victor; Heath, Karen E; Lapunzina, Pablo. In-frame Variants in FLNA Proximal Rod 1 Domain Associate With a Predominant Cardiac Valvular Phenotype.. Revista espanola de cardiologia (English ed.). 2017; ():-.Article. IF -4,485

Flottmann, Ricarda; Kragesteen, Bjort K; Geuer, Sinje; Socha, Magdalena; Allou, Lila; Sowinska-Seidler, Anna; Bosquillon de Jarcy, Laure; Wagner, Johannes; Jamsheer, Aleksander; Oehl-Jaschkowitz, Barbara; Wittler, Lars; de Silva, Deepthi; Kurth, Ingo; Maya, Idit; Santos-Simarro, Fernando; Hulsemann, Wiebke; Klopocki, Eva; Mountford, Roger; Fryer, Alan; Borck, Guntram; Horn, Denise; Lapunzina, Pablo; Wilson, Meredith; Mascrez, Benedicte; Duboule, Denis; Mundlos, Stefan; Spielmann, Malte. Noncoding copy-number variations are associated with congenital limb malformation.. Genetics in medicine : official journal of the American College of Medical Genetics. 2017; ():-.Article. IF -8,229

Galvez, E; Sebastian, E; Madero, L; Catala, A; Belendez, C; de Heredia, CD; Galera, A; Plaza, D; Badell, I; Vallespin, E; Lapunzina, P; Perona, R; Surralles, J; Bueren, J; Sevilla, J. NEXT GENERATION SEQUENCING IN BONE MARROW FAILURE SYNDROMES. HAEMATOLOGICA. 2017; 102():225-225.Meeting Abstract. IF -7,702

Garcia, M; Barrio, R; Garcia-Lavandeira, M; Garcia-Rendueles, AR; Escudero, A; Diaz-Rodriguez, E; Del Blanco, DG; Fernandez, A; de Rijke, YB; Vallespin, E; Nevado, J; Lapunzina, P; Matre, V; Hinkle, PM; Hokken-Koelega, ACS; de Miguel, MP; Cameselle-Teijeiro, JM; Nistal, M; Alvarez, CV; Moreno, JC. The syndrome of central hypothyroidism and macroorchidism: IGSF1 controls TRHR and FSHB expression by differential modulation of pituitary TGF beta and Activin pathways. SCIENTIFIC REPORTS. 2017; 7():-42937.Article. IF -4,259

Garcia-Morato, MB; Garcia-Minaur, S; Garicano, JM; Simarro, FS; Molina, LDP; Lopez-Granados, E; Cerdan, AF; Pena, RR. Mutations in PIK3R1 can lead to APDS2, SHORT syndrome or a combination of the two. CLINICAL IMMUNOLOGY. 2017; 179():77-80.Article. IF -3,99

Garcia-Morato, MB; Nevado, J; Gonzalez-Granado, LI; Urgelles, AS; Pena, RR; Cerdan, AF. Chronic granulomatous disease caused by maternal uniparental isodisomy of chromosome 16. JOURNAL OF ALLERGY AND CLINICAL IMMUNOLOGY-IN PRACTICE. 2017; 5(4):1146-1148.Letter. IF -5,317

Gkourogianni, A; Andrew, M; Tyzinski, L; Crocker, M; Douglas, J; Dunbar, N; Fairchild, J; Funari, MFA; Heath, KE; Jorge, AAL; Kurtzman, T; LaFranchi, S; Lalani, S; Lebl, J; Lin, YZ; Los, E; Newbern, D; Nowak, C; Olson, M; Popovic, J; Pruhova, S; Elblova, L; Quintos, JB; Segerlund, E; Sentchordi, L; Shinawi, M; Stattin, EL; Swartz, J; del Angel, AG; Cuellar, SD; Hosono, H; Sanchez-Lara, PA; Hwa, V; Baron, J; Nilsson, O; Dauber, A. Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM. 2017; 102(2):460-469.Article. IF -5,455

Gomez-Gonzalez, C; Esteban-Rodriguez, MI; Ruano, Y; Vallespin, E; Lapunzina, P; Martinez, P; Pascual, SI; Molano, J; Prior, C. Molecular Diagnosis of Limb-girdle Muscular Dystrophy Type 2A by Next-generation Sequencing. ANNALS OF INDIAN ACADEMY OF NEUROLOGY. 2017; 20(2):164-165.Letter. IF -0,95

Gordo, G; Tenorio, J; Arias, P; Santos-Simarro, F; Garcia-Minaur, S; Moreno, J C; Nevado, J; Vallespin, E; Rodriguez-Laguna, L; de Mena, R; Dapia, I; Palomares, M; Del Pozo, A; Ibanez, K; Silla, J C; Barroso, E; Ruiz Perez, V L; Martinez-Glez, V; Lapunzina, P. mTOR mutations in Smith-Kingsmore syndrome: four additional patients and a review.. Clinical genetics. 2017; ():-.Article. IF -3,326

Ibarra-Ramirez, M; Campos-Acevedo, LD; Lugo-Trampe, J; Martinez-Garza, LE; Martinez-Glez, V; Valencia-Benitez, M; Lapunzina, P; Ruiz-Perez, V. Phenotypic Variation in Patients with Homozygous c.1678G > T Mutation in EVC Gene: Report of Two Mexican Families with Ellis-van Creveld Syndrome. AMERICAN JOURNAL OF CASE REPORTS. 2017; 18():-.Article. IF -NO TIENE

Jimenez-Rolando, B; Noval, S; Rosa-Perez, I; Mata Diaz, E; Del Pozo, A; Ibanez, C; Silla, J C; Montano, V E F; Martin-Arenas, R; Vallespin, E. Next generation sequencing in the diagnosis of Stargardt's disease.. Archivos de la Sociedad Espanola de Oftalmologia. 2017; ():-.Article. IF -NO TIENE

Kalish, JM; Biesecker, LG; Brioude, F; Deardorff, MA; Di Cesare-Merlone, A; Druley, T; Ferrero, GB; Lapunzina, P; Larizza, L; Maas, S; Macchiaiolo, M; Maher, ER; Maitz, S; Martinez-Agosto, JA; Mussa, A; Robinson, P; Russo, S; Selicorni, A; Hennekam, RC. Nomenclature and definition in asymmetric regional body overgrowth. AMERICAN JOURNAL OF MEDICAL GENETICS PART A. 2017; 173(7):1735-1738.Article. IF -2,259

Laorden, NR; Mejorada, RL; Rodriguez, JMP; Vallespin, E; Montesa, A; Estelles, DL; Villaguzman, JC; Estelles, DL; Villaguzman, JC; Ibanez, K; Gonzalez-Billalabeitia, E; Magraner, L; Garde, J; Polo, SH; Arija, JA; Villatoro, R; Valderrama, BP; Lapunzina, P; Marcos, EC; Hidalgo, DO. Prevalence and baseline clinico-pathological associations of germline deleterious mutations in DNA repair genes (gmDDR) in a metastatic castration resistant prostate cancer (mCRPC) prospective spanish cohort (PROREPAIR-B study). ANNALS OF ONCOLOGY. 2017; 28():-.Meeting Abstract. IF -11,855

Leiter, SM; Parker, VER; Welters, A; Knox, R; Rocha, N; Clark, G; Payne, F; Lotta, L; Harris, J; Guerrero-Fernandez, J; Gonzalez-Casado, I; Garcia-Minaur, S; Gordo, G; Wareham, N; Martinez-Glez, V; Allison, M; O'Rahilly, S; Barroso, I; Meissner, T; Davies, S; Hussain, K; Temple, K; Barreda-Bonis, AC; Kummer, S; Semple, RK. Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. EUROPEAN JOURNAL OF ENDOCRINOLOGY. 2017; 177(2):175-186.Article. IF -4,101

Marcos, EC; Laorden, NR; Rodriguez, JMP; del Pozo, A; Saez, MI; Colmenero, AM; Puente, J; Silla-Castro, JC; Mejorada, RL; Garcia-Carbonero, I; Rivera, L; Vidal, MJM; Barrera, RM; Parra, EMF; Florez, YC; Borrega, P; Baamonde, MAGD; Pritchard, C; Lapunzina, P; Hidalgo, DO. PROREPAIR-B: A prospective cohort study of DNA repair defects in metastatic castration resistant prostate cancer (mCRPC). ANNALS OF ONCOLOGY. 2017; 28():-.Meeting Abstract. IF -11,855

Marquez-Rodas, I; Lobo, M; Flores-Sanchez, C; Sanz, M; Luque, S; Lizarraga, S; Gonzalez-Asanza, C; Pajares, JA; Peligros, MI; Bueno, O; Mata, C; Lopez, C; Lopez-Tarruella, S; Jerez, Y; Munoz-Martin, A; Blanco, M; Die-Trill, M; Justel, JP; Solera, J; Martin, M. Five Years of Multidisciplinary Care in Hereditary Cancer: Our Experience in a Spanish University Hospital. ONCOLOGY. 2017; 92(2):68-74.Article. IF -2,262

Mediero, S; Mardero, OD; de los Bueis, AB; Martin, SN; Garcia-Minaur, S. Keratoconus associated with Williams-Beuren syndrome: a new case report. INTERNATIONAL JOURNAL OF OPHTHALMOLOGY. 2017; 10(4):658-660.Letter. IF -1,177

Meerschaut, I; Rochefort, D; Revencu, N; Petre, J; Corsello, C; Rouleau, GA; Hamdan, FF; Michaud, JL; Morton, J; Radley, J; Ragge, N; Garcia-Minaur, S; Lapunzina, P; Bralo, MP; Mori, MA; Moortgat, S; Benoit, V; Mary, S; Bockaert, N; Oostra, A; Vanakker, O; Velinov, M; de Ravel, TJL; Mekahli, D; Sebat, J; Vaux, KK; DiDonato, N; Hanson-Kahn, AK; Hudgins, L; Dallapiccola, B; Novelli, A; Tarani, L; Andrieux, J; Parker, MJ; Neas, K; Ceulemans, B; Schoonjans, AS; Prchalova, D; Havlovicova, M; Hancarova, M; Budisteanu, M; Dheedene, A; Menten, B; Dion, PA; Lederer, D; Callewaert, B. FOXP1-related intellectual disability syndrome: a recognisable entity. JOURNAL OF MEDICAL GENETICS. 2017; 54(9):613-623.Article. IF -5,451

Munoz-Cabello, Patricia; Garcia-Minaur, Sixto; Espinel-Vallejo, Manuel Eliecer; Fernandez-Franco, Lorenzo; Stephens, Alexandra; Santos-Simarro, Fernando; Lapunzina-Badia, Pablo; McAllister, Marion. Translation and Cross-Cultural Adaptation with Preliminary Validation of GCOS-24 for Use in Spain.. Journal of genetic counseling. 2017; ():-.Article. IF -1,938

Palencia-Campos, A; Ullah, A; Nevado, J; Yildirim, R; Unal, E; Ciorraga, M; Barruz, P; Chico, L; Piceci-Sparascio, F; Guida, V; De Luca, A; Kayserili, HL; Ullah, I; Burmeister, M; Lapunzina, P; Ahmad, W; Morales, AV; Ruiz-Perez, VL. GLI1 inactivation is associated with developmental phenotypes overlapping with Ellis-van Creveld syndrome. HUMAN MOLECULAR GENETICS. 2017; 26(23):4556-4571.Article. IF -5,34

Palomares-Bralo, M; Vallespin, E; del Pozo, A; Ibanez, K; Silla, JC; Galan, E; Gordo, G; Martinez-Glez, V; Alba-Valdivia, LI; Heath, KE; Garcia-Minaur, S; Lapunzina, P; Santos-Simarro, F. Pitfalls of trio-based exome sequencing: imprinted genes and parental mosaicism-MAGEL2 as an example. GENETICS IN MEDICINE. 2017; 19(11):1283-1285.Letter. IF -8,229

Peces, R; Afonso, S; Peces, C; Nevado, J; Selgas, R. Living kidney transplantation between brothers with unrecognized renal amyloidosis as the first manifestation of familial Mediterranean fever: a case report. BMC MEDICAL GENETICS. 2017; 18():-97.Article. IF -2,198

Rivera-Pedroza, CI; Barraza-Garcia, J; Paumard-Hernandez, B; Nevado, J; Orbea-Gallardo, C; del Pozo, JS; Heath, KE. Chromosome 1p31.1p31.3 Deletion in a Patient with Craniosynostosis, Central Nervous System and Renal Malformation: Case Report and Review of the Literature. MOLECULAR SYNDROMOLOGY. 2017; 8(1):30-35.Article. IF -NO TIENE

Rodriguez, F; Vallejos, C; Giraudo, F; Unanue, N; Hernandez, MI; Godoy, P; Celis, S; Martin-Arenas, R; Palomares-Bralo, M; Heath, KE; Lopez, MT; Cassorla, F. Copy number variants of Ras/MAPK pathway genes in patients with isolated cryptorchidism. ANDROLOGY. 2017; 5(5):923-930.Article. IF -2,427

Rodriguez, FA; Vallejos, C; Unanue, N; Hernandez, MI; Celis, S; Martin-Arenas, R; Bralo, MP; Heath, KE; Lopez, MT; Cassorla, F. MOLECULAR STUDY OF RAS/ MAPK PATHWAY GENES IN PATIENTS WITH CRYPTORCHIDISM. HORMONE RESEARCH IN PAEDIATRICS. 2017; 88():12-12.Meeting Abstract. IF -1,844

Rodriguez-Jimenez, C; Santos-Simarro, F; Campos-Barros, A; Camarena, C; Lledin, D; Vallespin, E; del Pozo, A; Mena, R; Lapunzina, P; Rodriguez-Novoa, S. A new variant in PHKA2 is associated with glycogen storage disease type IXa. MOLECULAR GENETICS AND METABOLISM REPORTS. 2017; 10():52-55.Article. IF -NO TIENE

Rodriguez-Zabala, M; Aza-Carmona, M; Rivera-Pedroza, CI; Belinchon, A; Guerrero-Zapata, I; Barraza-Garcia, J; Vallespin, E; Lu, M; del Pozo, A; Glucksman, MJ; Santos-Simarro, F; Heath, KE. FGF9 mutation causes craniosynostosis along with multiple synostoses. HUMAN MUTATION. 2017; 38(11):1471-1476.Article. IF -4,601

Salas, PC; Lapunzina, P; Perez-Martinez, A. Genetic predisposition to childhood cancer. ANALES DE PEDIATRIA. 2017; 87(3):125-127.Editorial Material. IF -1,14

Sanchez-Montenegro, Carlos; Vilanova-Sanchez, Alejandra; Barrena-Delfa, Saturnino; Tenorio, Jair; Santos-Simarro, Fernando; Garcia-Minaur, Sixto; Lapunzina, Pablo; Martinez-Martinez, Leopoldo. Costello Syndrome and Umbilical Ligament Rhabdomyosarcoma in Two Pediatric Patients: Case Reports and Review of the Literature.. Case reports in genetics. 2017; 2017():1587610-.Article. IF -NO TIENE

Santos-Simarro, F; Vallespin, E; del Pozo, A; Ibanez, K; Silla, JC; Fernandez, L; Nevado, J; Gonzalez-Pecellin, H; Montano, VEF; Martin, R; Valdivia, LA; Garcia-Minaur, S; Lapunzina, P; Palomares-Bralo, M. Eye coloboma and complex cardiac malformations belong to the clinical spectrum of PUF60 variants. CLINICAL GENETICS. 2017; 92(3):350-351.Letter. IF -3,326

Tardivo, A; Masotto, B; Espeche, L; Solari, AP; Nevado, J; Rozental, S. 16p11.2 Microdeletion: first report in Argentina. ARCHIVOS ARGENTINOS DE PEDIATRIA. 2017; 115(6):E449-E453.Article. IF -0,403

Tenorio, J; Alvarez, I; Riancho-Zarrabeitia, L; Martos-Moreno, GA; Mandrile, G; Crespo, MD; Sukchev, M; Sherif, M; Kramer, I; Darnaude-Ortiz, MT; Arias, P; Gordo, G; Dapia, I; Martinez-Villanueva, J; Gomez, R; Iturzaeta, JM; Otaify, G; Garcia-Unzueta, M; Rubinacci, A; Riancho, JA; Aglan, M; Temtamy, S; Hamid, MA; Argente, J; Ruiz-Perez, VL; Heath, KE; Lapunzina, P. Molecular and clinical analysis of ALPL in a cohort of

patients with suspicion of Hypophosphatasia. AMERICAN JOURNAL OF MEDICAL GENETICS PART A. 2017; 173(3):601-610.Article. IF -2,259

Teresa, MAG; Abal, RP; Torres, SR; Urabayen, DG; Martinez, SG; Trang, H; Barros, AC. Spanish patients with central hypoventilation syndrome included in the European Registry. The 2015 data. ANALES DE PEDIATRIA. 2017; 86(5):255-263.Article. IF -1,14

Tornero, C; Aguado, P; Garcia, S; ATenorio, J; Lapunzina, P; Heath, K; Buno, A; Iturzaeta, JM; Monjo, I; Plasencia, C; Balsa, A. Low Alkaline Phosphatase Levels: Could it be Hypophosphatasia?.. JOURNAL OF BONE AND MINERAL RESEARCH. 2017; 32():S181-S182.Meeting Abstract. IF -6,284

Tornero, C; Aguado, P; Garcia, S; Tenorio, J; Lapunzina, P; Buno, A; Iturzaeta, JM; Plasencia, C; Monjo, I; Balsa, A. LOW ALKALINE PHOSPHATASE LEVELS: COULD IT BE HYPOPHOSPHATASIA?. ANNALS OF THE RHEUMATIC DISEASES. 2017; 76():416-416.Meeting Abstract. IF -12,811

Tornero, C; Aguado, P; Garcia-Carazo, S; Tenorio, JA; Lapunzina, P; Heath, K; Buno, A; Iturzaeta, JM; Monjo, I; Plasencia, C; Balsa, A. Low Alkaline Phosphatase Levels: Could It be Hypophosphatasia?. ARTHRITIS & RHEUMATOLOGY. 2017; 69():-.Meeting Abstract. IF -6,918

Vasques, GA; Hisado-Oliva, A; Funari, MFA; Lerario, AM; Quedas, EPS; Solberg, P; Heath, KE; Jorge, AAL. Long-term response to growth hormone therapy in a patient with short stature caused by a novel heterozygous mutation in NPR2. JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM. 2017; 30(1):111-116.Article. IF -1,233

Villafuerte, B; Gonzalez, A; Moya, C; Garzon, L; Herranz, A; Natera-De Benito, D; Gallego, M; Palomares, M; Nevado, J; Moreno, JC. NKX2-1 GENE DEFECTS IN A PEDIATRIC COHORT WITH SUSPECTED BRAIN-LUNG-THYROID SYNDROME. HORMONE RESEARCH IN PAEDIATRICS. 2017; 88():376-377.Meeting Abstract. IF -1,844

Villarejo-Lopez, L; Jimenez, E; Bartolome-Martin, D; Zafra, F; Lapunzina, P; Aragon, C; Lopez-Corcuera, B. P2X receptors up-regulate the cell-surface expression of the neuronal glycine transporter GlyT2. NEUROPHARMACOLOGY. 2017; 125():99-116.Article. IF -5,012

Writzl, K; Maver, A; Kovacic, L; Martinez-Valero, P; Contreras, L; Satrustegui, J; Castori, M; Faivre, L; Lapunzina, P; van Kuilenburg, ABP; Radovic, S; Thauvin-Robinet, C; Peterlin, B; del Arco, A; Hennekam, RC. De Novo Mutations in SLC25A24 Cause a Disorder Characterized by Early Aging, Bone Dysplasia, Characteristic Face, and Early Demise. AMERICAN JOURNAL OF HUMAN GENETICS. 2017; 101(5):844-855.Article. IF -9,025

Nº Documentos	Tipo de documento	FI	1º Cuartil	1º Decil
32	Artículos	92,332	10	3
5	Cartas	18,999	2	1
1	Editoriales	1,14	0	0
11	Meeting Abstract	74,742	7	6
2	Revisiones	3,214	0	0
	FI Originales	**92,332**		
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