Pablo Lapunzina

List of Publications by Year
in descending order

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222 papers	9,345 citations	41344 49 h-index	54911 84 g-index
238 all docs	238 docs citations	238 times ranked	13704 citing authors

#	Article	IF	Citations
1	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	9.6	388
2	Diversity and Function of Mutations in P450 Oxidoreductase in Patients with Antley-Bixler Syndrome and Disordered Steroidogenesis. American Journal of Human Genetics, 2005, 76, 729-749.	6.2	321
3	Genome-wide parent-of-origin DNA methylation analysis reveals the intricacies of human imprinting and suggests a germline methylation-independent mechanism of establishment. Genome Research, 2014, 24, 554-569.	5.5	311
4	PROREPAIR-B: A Prospective Cohort Study of the Impact of Germline DNA Repair Mutations on the Outcomes of Patients With Metastatic Castration-Resistant Prostate Cancer. Journal of Clinical Oncology, 2019, 37, 490-503.	1.6	255
5	Identification of a Frameshift Mutation in Osterix in a Patient with Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 87, 110-114.	6.2	246
6	Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574.	6.2	240
7	Risk of tumorigenesis in overgrowth syndromes: A comprehensive review. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 53-71.	1.6	237
8	Extending the phenotype of recurrent rearrangements of 16p11.2: Deletions in mentally retarded patients without autism and in normal individuals. European Journal of Medical Genetics, 2009, 52, 77-87.	1.3	226
9	Mutations in Two Nonhomologous Genes in a Head-to-Head Configuration Cause Ellis-van Creveld Syndrome. American Journal of Human Genetics, 2003, 72, 728-732.	6.2	200
10	NOTCH2mutations in Alagille syndrome. Journal of Medical Genetics, 2012, 49, 138-144.	3.2	197
11	Epigenetic inactivation of the Sotos overgrowth syndrome gene histone methyltransferase NSD1 in human neuroblastoma and glioma. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 21830-21835.	7.1	190
12	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. Human Mutation, 2012, 33, 343-350.	2.5	178
13	HRAS mutation analysis in Costello syndrome: Genotype and phenotype correlation. American Journal of Medical Genetics, Part A, 2006, 140A, 1-7.	1.2	164
14	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	8.2	153
15	Novel (60%) and Recurrent (40%) Androgen Receptor Gene Mutations in a Series of 59 Patients with a 46,XY Disorder of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 1876-1888.	3.6	127
16	Deletions Involving Long-Range Conserved Nongenic Sequences Upstream and Downstream of FOXL2 as a Novel Disease-Causing Mechanism in Blepharophimosis Syndrome. American Journal of Human Genetics, 2005, 77, 205-218.	6.2	116
17	Reduced hepatic expression of farnesoid X receptor in hereditary cholestasis associated to mutation in ATP8B1. Human Molecular Genetics, 2004, 13, 2451-2460.	2.9	107
18	Stratification of Wilms tumor by genetic and epigenetic analysis. Oncotarget, 2012, 3, 327-335.	1.8	101

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19	The ciliary Evc/Evc2 complex interacts with Smo and controls Hedgehog pathway activity in chondrocytes by regulating Sufu/Gli3 dissociation and Gli3 trafficking in primary cilia. Human Molecular Genetics, 2013, 22, 124-139.	2.9	97
20	Molecular and clinical characterization of cardioâ€facioâ€cutaneous (CFC) syndrome: Overlapping clinical manifestations with Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 799-807.	1.2	96
21	Molecular and clinical analysis ofRAF1in Noonan syndrome and related disorders: dephosphorylation of serine 259 as the essential mechanism for mutant activation. Human Mutation, 2010, 31, 284-294.	2.5	96
22	Genome-Wide Allelic Methylation Analysis Reveals Disease-Specific Susceptibility to Multiple Methylation Defects in Imprinting Syndromes. Human Mutation, 2013, 34, n/a-n/a.	2.5	96
23	Somatic activating mutations in PIK3CA cause generalized lymphatic anomaly. Journal of Experimental Medicine, 2019, 216, 407-418.	8.5	96
24	Epigenetic biomarkers: Current strategies and future challenges for their use in the clinical laboratory. Critical Reviews in Clinical Laboratory Sciences, 2017, 54, 529-550.	6.1	92
25	Macrocephaly-cutis marmorata telangiectatica congenita: Report of six new patients and a review. , 2004, 130A, 45-51.		90
26	CDKN1C mutations: two sides of the same coin. Trends in Molecular Medicine, 2014, 20, 614-622.	6.7	89
27	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
28	New microdeletion and microduplication syndromes: a comprehensive review. Genetics and Molecular Biology, 2014, 37, 210-219.	1.3	84
29	Clinical utility of chromosomal microarray analysis in invasive prenatal diagnosis. Human Genetics, 2012, 131, 513-523.	3.8	82
30	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. Trends in Genetics, 2016, 32, 444-455.	6.7	81
31	Comparative study of three diagnostic approaches (FISH, STRs and MLPA) in 30 patients with 22q11.2 deletion syndrome. Clinical Genetics, 2005, 68, 373-378.	2.0	79
32	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome-Osteogenesis imperfecta phenotypic spectrum. Human Mutation, 2012, 33, 1444-1449.	2.5	77
33	Simpson-Golabi-Behmel syndrome types I and II. Orphanet Journal of Rare Diseases, 2014, 9, 138.	2.7	75
34	Spectrum of mutations and genotypeâ€“phenotype analysis in Noonan syndrome patients with RIT1 mutations. Human Genetics, 2016, 135, 209-222.	3.8	75
35	Cancer Genes Hypermethylated in Human Embryonic Stem Cells. PLoS ONE, 2008, 3, e3294.	2.5	75
36	Microhomology-Mediated Mechanisms Underlie Non-Recurrent Disease-Causing Microdeletions of the FOXL2 Gene or Its Regulatory Domain. PLoS Genetics, 2013, 9, e1003358.	3.5	72

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37	The consequences of uniparental disomy and copy number neutral loss-of-heterozygosity during human development and cancer. Biology of the Cell, 2011, 103, 303-317.	2.0	68
38	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387.	2.8	68
39	Clinical and Molecular Evaluation of SHOX/PAR1 Duplications in LÃ©ri-Weill Dyschondrosteosis (LWD) and Idiopathic Short Stature (ISS). Journal of Clinical Endocrinology and Metabolism, 2011, 96, E404-E412.	3.6	60
40	Macrocephalyâ€“capillary malformation: Analysis of 13 patients and review of the diagnostic criteria. American Journal of Medical Genetics, Part A, 2010, 152A, 3101-3106.	1.2	57
41	A founder EIF2AK4 mutation causes an aggressive form of pulmonary arterial hypertension in Iberian Gypsies. Clinical Genetics, 2015, 88, 579-583.	2.0	57
42	Clinical, biochemical and genetic spectrum of low alkaline phosphatase levels in adults. European Journal of Internal Medicine, 2016, 29, 40-45.	2.2	57
43	Long interspersed nuclear element-1 (LINE1)-mediated deletion ofEVC,EVC2,C4orf6, andSTK32B in Ellisâ€“van Creveld syndrome with borderline intelligence. Human Mutation, 2008, 29, 931-938.	2.5	55
44	Methylation screening of reciprocal genome-wide UPDs identifies novel human-specific imprinted genesâ€. Human Molecular Genetics, 2011, 20, 3188-3197.	2.9	55
45	FAM46A mutations are responsible for autosomal recessive osteogenesis imperfecta. Journal of Medical Genetics, 2018, 55, 278-284.	3.2	55
46	Widening the mutation spectrum ofEVCandEVC2: ectopic expression of Weyer variants in NIH 3T3 fibroblasts disrupts hedgehog signaling. Human Mutation, 2009, 30, 1667-1675.	2.5	54
47	A deletion and a duplication in distal 22q11.2 deletion syndrome region. Clinical implications and review. BMC Medical Genetics, 2009, 10, 48.	2.1	54
48	Expanding the mutation spectrum in 182 Spanish probands with craniosynostosis: identification and characterization of novel TCF12 variants. European Journal of Human Genetics, 2015, 23, 907-914.	2.8	53
49	CLAPO syndrome: identification of somatic activating PIK3CA mutations and delineation of the natural history and phenotype. Genetics in Medicine, 2018, 20, 882-889.	2.4	52
50	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
51	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, 844-855.	6.2	51
52	CDKN1C (p57^{Kip2}) analysis in Beckwithâ€“Wiedemann syndrome (BWS) patients: Genotypeâ€“phenotype correlations, novel mutations, and polymorphisms. American Journal of Medical Genetics, Part A, 2010, 152A, 1390-1397.	1.2	50
53	Clinical utility gene card for: Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	2.8	50
54	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellisâ€“van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571.	2.9	50

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55	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48
56	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
57	Beckwithâ€“Wiedemann syndrome and uniparental disomy 11p: fine mapping of the recombination breakpoints and evaluation of several techniques. European Journal of Human Genetics, 2011, 19, 416-421.	2.8	44
58	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	2.8	44
59	Alagille Syndrome: Cutaneous Manifestations in 38 Children. Pediatric Dermatology, 2005, 22, 11-14.	0.9	43
60	PAR1 deletions downstream ofSHOX are the most frequent defect in a Spanish cohort of LÃ©ri-Weill dyschondrosteosis (LWD) probands. Human Mutation, 2006, 27, 1062-1062.	2.5	43
61	CDKN1C Mutations in HELLP/Preeclamptic Mothers of Beckwithâ€“Wiedemann Syndrome (BWS) Patients. Placenta, 2009, 30, 551-554.	1.5	43
62	Clinical, biochemical, and molecular studies in pyridoxineâ€dependent epilepsy. Antisense therapy as possible new therapeutic option. Epilepsia, 2013, 54, 239-248.	5.1	43
63	A patient with Simpson-Golabi-Behmel syndrome and hepatocellular carcinoma.. Journal of Medical Genetics, 1998, 35, 153-156.	3.2	42
64	A Novel Dominant Hyperekplexia Mutation Y705C Alters Trafficking and Biochemical Properties of the Presynaptic Glycine Transporter GlyT2. Journal of Biological Chemistry, 2012, 287, 28986-29002.	3.4	42
65	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2013, 161, 1354-1369.	1.2	42
66	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. Human Molecular Genetics, 2015, 24, 4126-4137.	2.9	42
67	Noncoding copy-number variations are associated with congenital limb malformation. Genetics in Medicine, 2018, 20, 599-607.	2.4	42
68	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	42
69	Familial Occurrence of the IMAGe Association: Additional Clinical Variants and a Proposed Mode of Inheritance. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 3186-3190.	3.6	39
70	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. Scientific Reports, 2013, 3, 1346.	3.3	39
71	Novel genes and sex differences in COVID-19 severity. Human Molecular Genetics, 2022, 31, 3789-3806.	2.9	38
72	Multiple Giant Pilomatricoma in Familial Sotos Syndrome. Pediatric Dermatology, 2008, 25, 122-125.	0.9	37

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73	Characterization of a 8q21.11 Microdeletion Syndrome Associated with Intellectual Disability and a Recognizable Phenotype. American Journal of Human Genetics, 2011, 89, 295-301.	6.2	37
74	Detection of Hypomethylation Syndrome among Patients with Epigenetic Alterations at theGNASLocus. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1060-E1067.	3.6	37
75	Two mutations in IFITM5 causing distinct forms of osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1136-1142.	1.2	37
76	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. Molecular Genetics & Genomic Medicine, 2017, 5, 28-39.	1.2	37
77	Clinical Implementation of Pharmacogenetic Testing in a Hospital of the Spanish National Health System: Strategy and Experience Over 3 Years. Clinical and Translational Science, 2018, 11, 189-199.	3.1	37
78	Molecular and clinical analysis of ALPL in a cohort of patients with suspicion of Hypophosphatasia. American Journal of Medical Genetics, Part A, 2017, 173, 601-610.	1.2	36
79	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	1.2	36
80	mTOR mutations in Smithâ€Kingsmore syndrome: Four additional patients and a review. Clinical Genetics, 2018, 93, 762-775.	2.0	36
81	A prenatally diagnosed patient with full monosomy 21: Ultrasound, cytogenetic, clinical, molecular, and necropsy findings. , 2004, 127A, 69-73.		35
82	Interaction between KDELR2 and HSP47 as a Key Determinant in Osteogenesis Imperfecta Caused by Bi-allelic Variants in KDELR2. American Journal of Human Genetics, 2020, 107, 989-999.	6.2	35
83	Variants in members of the cadherinâ€“catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. European Journal of Human Genetics, 2018, 26, 210-219.	2.8	34
84	A six-attribute classification of geneticmosaicism. Genetics in Medicine, 2020, 22, 1743-1757.	2.4	34
85	Capillary malformation of the lower lip, lymphatic malformation of the face and neck, asymmetry and partial/generalized overgrowth (CLAPO): Report of six cases of a new syndrome/association. American Journal of Medical Genetics, Part A, 2008, 146A, 2583-2588.	1.2	33
86	Adults with Sotos syndrome: Review of 21 adults with molecularly confirmed NSD1 alterations, including a detailed case report of the oldest person. American Journal of Medical Genetics, Part A, 2011, 155, 2105-2111.	1.2	33
87	A New Overgrowth Syndrome is due to Mutations inRNF125. Human Mutation, 2014, 35, 1436-1441.	2.5	33
88	The syndrome of central hypothyroidism and macroorchidism: IGSF1 controls TRHR and FSHB expression by differential modulation of pituitary TGFÎ² and Activin pathways. Scientific Reports, 2017, 7, 42937.	3.3	33
89	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	6.2	33
90	Constitutional mosaic genome-wide uniparental disomy due to diploidisation: an unusual cancer-predisposing mechanism. Journal of Medical Genetics, 2011, 48, 212-216.	3.2	32

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91	Customized high resolution CGHâ€array for clinical diagnosis reveals additional genomic imbalances in previous wellâ€defined pathological samples. American Journal of Medical Genetics, Part A, 2013, 161, 1950-1960.	1.2	32
92	The role of ZFP57 and additional KRAB-zinc finger proteins in the maintenance of human imprinted methylation and multi-locus imprinting disturbances. Nucleic Acids Research, 2020, 48, 11394-11407.	14.5	32
93	MLPA vs multiprobe FISH: comparison of two methods for the screening of subtelomeric rearrangements in 50 patients with idiopathic mental retardation. Clinical Genetics, 2006, 69, 228-233.	2.0	31
94	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform. Epigenomics, 2018, 10, 941-954.	2.1	31
95	Clinical and molecular analyses of Beckwithâ€“Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. American Journal of Medical Genetics, Part A, 2016, 170, 2740-2749.	1.2	30
96	Abnormal bone turnover in individuals with low serum alkaline phosphatase. Osteoporosis International, 2018, 29, 2147-2150.	3.1	30
97	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. European Journal of Human Genetics, 2015, 23, 1615-1626.	2.8	29
98	46,<scp>XX</scp> ovotesticular <scp>DSD</scp> associated with a <scp>SOX</scp>3 gene duplication in a <scp>SRY</scp>â€negative boy. Clinical Endocrinology, 2016, 85, 673-675.	2.4	28
99	Oculocerebrocutaneous (Delleman) Syndrome: Report of Two Cases. Neuropediatrics, 2005, 36, 50-54.	0.6	27
100	Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. American Journal of Medical Genetics, Part A, 2014, 164, 1143-1150.	1.2	27
101	The most recurrent monogenic disorders that overlap with the phenotype of Rett syndrome. European Journal of Paediatric Neurology, 2019, 23, 609-620.	1.6	27
102	Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. Frontiers in Genetics, 2022, 13, 652454.	2.3	27
103	The role of CDKN2A/B deletions in pediatric acute lymphoblastic leukemia. Pediatric Hematology and Oncology, 2016, 33, 415-422.	0.8	26
104	Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH. PLoS ONE, 2017, 12, e0172363.	2.5	26
105	Biomarkers in Vestibular Schwannomaâ€“Associated Hearing Loss. Frontiers in Neurology, 2019, 10, 978.	2.4	26
106	Molecular Analysis of BMPR2 , TBX4 , and KCNK3 and Genotype-Phenotype Correlations in Spanish Patients and Families With Idiopathic and Hereditary Pulmonary Arterial Hypertension. Revista Espanola De Cardiologia (English Ed), 2016, 69, 1011-1019.	0.6	25
107	Follow-up and Risk of Tumors in Overgrowth Syndromes. Journal of Pediatric Endocrinology and Metabolism, 2005, 18, 1227-35.	0.9	24
108	SÃndrome LEOPARD: una variante del sÃndrome de Noonan con fuerte asociaciÃ³n a miocardiopatÃa hipertrÃ³fica. Revista Espanola De Cardiologia, 2013, 66, 350-356.	1.2	24

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109	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	12.8	24
110	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. Scientific Reports, 2017, 7, 12288.	3.3	23
111	Risks of congenital anomalies in large for gestational age infants. Journal of Pediatrics, 2002, 140, 200-204.	1.8	22
112	Clinical and molecular studies on two further families with Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics, Part A, 2005, 138A, 272-277.	1.2	22
113	Functional Characterization of NIPBL Physiological Splice Variants and Eight Splicing Mutations in Patients with Cornelia de Lange Syndrome. International Journal of Molecular Sciences, 2014, 15, 10350-10364.	4.1	22
114	Identification of a premature stop codon mutation in the PHGDH gene in severe Neuâ€Łaxova syndromeâ€”evidence for phenotypic variability. American Journal of Medical Genetics, Part A, 2015, 167, 1323-1329.	1.2	22
115	Analysis of invdupdel(8p) rearrangement: Clinical, cytogenetic and molecular characterization. American Journal of Medical Genetics, Part A, 2015, 167, 1018-1025.	1.2	22
116	Genetic analyses of aplastic anemia and idiopathic pulmonary fibrosis patients with short telomeres, possible implication of DNA-repair genes. Orphanet Journal of Rare Diseases, 2019, 14, 82.	2.7	21
117	Clinical heterogeneity of Pulmonary Arterial Hypertension associated with variants in TBX4. PLoS ONE, 2020, 15, e0232216.	2.5	21
118	Customized Massive Parallel Sequencing Panel for Diagnosis of Pulmonary Arterial Hypertension. Genes, 2020, 11, 1158.	2.4	21
119	Ultraviolet light-related neural tube defects?. , 1996, 67, 106-106.		20
120	A novel insertion in theFGFR2gene in a patient with Crouzon phenotype and sacrococcygeal tail. Birth Defects Research Part A: Clinical and Molecular Teratology, 2005, 73, 61-64.	1.6	20
121	FOXL2copy number changes in the molecular pathogenesis of BPES: unique cohort of 17 deletions. Human Mutation, 2010, 31, n/a-n/a.	2.5	20
122	Higher frequency of uncommon 1.5-2 Mb deletions found in familial cases of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2005, 136A, 71-75.	1.2	19
123	Rothmundâ€“Thomson Syndrome: novel pathogenic mutations and frequencies of variants in the <scp>RECQL</scp>4 and <scp>USB</scp>1 (C16orf57) gene. Molecular Genetics & Genomic Medicine, 2016, 4, 359-366.	1.2	19
124	Characterization of rare ABCC8 variants identified in Spanish pulmonary arterial hypertension patients. Scientific Reports, 2020, 10, 15135.	3.3	19
125	Eye coloboma and complex cardiac malformations belong to the clinical spectrum of <scp>PUF60</scp> variants. Clinical Genetics, 2017, 92, 350-351.	2.0	18
126	Sotos syndrome is associated with leukemia/lymphoma. American Journal of Medical Genetics, Part A, 2007, 143A, 1244-1245.	1.2	17

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127	LEOPARD Syndrome: A Variant of Noonan Syndrome Strongly Associated With Hypertrophic Cardiomyopathy. Revista Espanola De Cardiologia (English Ed), 2013, 66, 350-356.	0.6	17
128	Prediction models for voriconazole pharmacokinetics based on pharmacogenetics: AN exploratory study in a Spanish population. International Journal of Antimicrobial Agents, 2019, 54, 463-470.	2.5	17
129	Novel Genetic and Molecular Pathways in Pulmonary Arterial Hypertension Associated with Connective Tissue Disease. Cells, 2021, 10, 1488.	4.1	17
130	Molecular diagnosis of Beckwith-Wiedemann Syndrome using quantitative methylation-sensitive polymerase chain reaction. Genetics in Medicine, 2006, 8, 628-634.	2.4	16
131	P2X receptors up-regulate the cell-surface expression of the neuronal glycine transporter GlyT2. Neuropharmacology, 2017, 125, 99-116.	4.1	16
132	Further delineation of neuropsychiatric findings in Tatton-Brown-Rahman syndrome due to disease-causing variants in DNMT3A: seven new patients. European Journal of Human Genetics, 2020, 28, 469-479.	2.8	16
133	Molecular characterization of Spanish patients with MECP2 duplication syndrome. Clinical Genetics, 2020, 97, 610-620.	2.0	16
134	Hyperekplexia (Startle Disease). Molecular Diagnosis and Therapy, 2003, 7, 125-128.	1.1	16
135	Beckwithâ€“Wiedemann syndrome due to 11p15.5 paternal duplication associated with Klinefelter syndrome and a â€œde novoâ€•pericentric inversion of chromosome Y. European Journal of Medical Genetics, 2005, 48, 159-166.	1.3	15
136	Unrelated chromosomal anomalies found in patients with suspected 22q11.2 deletion. American Journal of Medical Genetics, Part A, 2008, 146A, 1134-1141.	1.2	15
137	A parallel study of different array-CGH platforms in a set of Spanish patients with developmental delay and intellectual disability. Gene, 2013, 521, 82-86.	2.2	15
138	Semilobar holoprosencephaly, coronal craniosynostosis, and multiple congenital anomalies: A severe expression of the Genoa syndrome or a newly recognized syndrome?. American Journal of Medical Genetics Part A, 2001, 102, 258-260.	2.4	14
139	Craniofacial dyssynostosis: Description of the first four Spanish cases and review. American Journal of Medical Genetics, Part A, 2005, 132A, 41-48.	1.2	14
140	Two novel POC1A mutations in the primordial dwarfism, SOFT syndrome: Clinical homogeneity but also unreported malformations. American Journal of Medical Genetics, Part A, 2016, 170, 210-216.	1.2	14
141	TP53 and CDKN1A mutation analysis in families with Liâ€“Fraumeni and Liâ€“Fraumeni like syndromes. Familial Cancer, 2017, 16, 243-248.	1.9	14
142	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	4.1	14
143	Germinal mosaicism in Simpsonâ€Golabiâ€Behmel syndrome. Clinical Genetics, 2007, 72, 384-386.	2.0	13
144	Direct tandem duplication in chromosome 19q characterized by array CGH. European Journal of Medical Genetics, 2008, 51, 257-263.	1.3	13

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145	MRX93 syndrome (BRWD3 gene): five new patients with novel mutations. Clinical Genetics, 2019, 95, 726-731.	2.0	13
146	Development, behaviour and sensory processing in Marshallâ€“Smith syndrome and Malan syndrome: phenotype comparison in two related syndromes. Journal of Intellectual Disability Research, 2020, 64, 956-969.	2.0	13
147	Novel TNIP2 and TRAF2 Variants Are Implicated in the Pathogenesis of Pulmonary Arterial Hypertension. Frontiers in Medicine, 2021, 8, 625763.	2.6	13
148	Schuursâ€“Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	2.4	13
149	Autosomal recessive hydrocephalus due to aqueduct stenosis: report of a further family and implications for genetic counselling. Journal of Maternal-Fetal and Neonatal Medicine, 2002, 12, 64-66.	1.5	12
150	Hyperekplexia (Startle Disease). Molecular Diagnosis and Therapy, 2003, 7, 125-128.	1.1	12
151	Impact of NGS in the medical sciences: genetic syndromes with an increased risk of developing cancer as an example of the use of new technologies. Genetics and Molecular Biology, 2014, 37, 241-249.	1.3	12
152	Expanding the Evidence of a Semi-Dominant Inheritance in GDF2 Associated with Pulmonary Arterial Hypertension. Cells, 2021, 10, 3178.	4.1	12
153	Segmental uniparental disomy leading to homozygosity for a pathogenic mutation in three recessive metabolic diseases. Molecular Genetics and Metabolism, 2012, 105, 270-271.	1.1	11
154	Constitutional and somatic methylation status of DMRH19 and KvDMR in Wilms tumor patients. Genetics and Molecular Biology, 2012, 35, 714-724.	1.3	11
155	A new variant in PHKA2 is associated with glycogen storage disease type IXa. Molecular Genetics and Metabolism Reports, 2017, 10, 52-55.	1.1	11
156	The G397A (E133K) change in theAGGF1 (VG5Q) gene is a single nucleotide polymorphism in the Spanish population. American Journal of Medical Genetics, Part A, 2006, 140A, 2832-2833.	1.2	10
157	Otopalatodigital syndrome type 2 in two siblings with a novel filamin A 629G>T mutation: Clinical, pathological, and molecular findings. American Journal of Medical Genetics, Part A, 2007, 143A, 1120-1125.	1.2	10
158	Umbilical Cord Stricture is Not a Genetic Anomaly: A Study in Twins. Pediatric and Developmental Pathology, 2008, 11, 363-369.	1.0	10
159	A novel mutation in CDKN1C in sibs with Beckwithâ€“Wiedemann syndrome and cleft palate, sensorineural hearing loss, and supernumerary flexion creases. American Journal of Medical Genetics, Part A, 2013, 161, 192-197.	1.2	10
160	Pitfalls of trio-based exome sequencing: imprinted genes and parental mosaicismâ€”MAGEL2 as an example. Genetics in Medicine, 2017, 19, 1283-1285.	2.4	10
161	The Brain-Lung-Thyroid syndrome (BLTS): A novel deletion in chromosome 14q13.2-q21.1 expands the phenotype to humoral immunodeficiency. European Journal of Medical Genetics, 2018, 61, 393-398.	1.3	10
162	Constitutional mosaicism in RASA1 â€related capillary malformationâ€arteriovenous malformation. Clinical Genetics, 2019, 95, 516-519.	2.0	10

#	Article	IF	Citations
163	Can we identify individuals with an ALPL variant in adults with persistent hypophosphatasaemia?. Orphanet Journal of Rare Diseases, 2020, 15, 51.	2.7	10
164	Laurinâ€“Sandrow syndrome: Review and redefinition. American Journal of Medical Genetics, Part A, 2008, 146A, 2557-2565.	1.2	9
165	<scp>CIBERER</scp>: Spanish national network for research on rare diseases: A highly productive collaborative initiative. Clinical Genetics, 2022, 101, 481-493.	2.0	9
166	Euchromatic variant 16p+. Implications in prenatal diagnosis. Prenatal Diagnosis, 2006, 26, 535-538.	2.3	8
167	Segmental uniparental isodisomy of chromosome 6 causing transient diabetes mellitus and merosinâ€deficient congenital muscular dystrophy. American Journal of Medical Genetics, Part A, 2014, 164, 2908-2913.	1.2	8
168	Array CGH Analysis of Paired Blood and Tumor Samples from Patients with Sporadic Wilms Tumor. PLoS ONE, 2015, 10, e0136812.	2.5	8
169	A view on clinical genetics and genomics in Spain: of challenges and opportunities. Molecular Genetics & Genomic Medicine, 2016, 4, 376-391.	1.2	8
170	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, 1325-1329.	0.8	8
171	Phenotype-loci associations in networks of patients with rare disorders: application to assist in the diagnosis of novel clinical cases. European Journal of Human Genetics, 2018, 26, 1451-1461.	2.8	8
172	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. European Journal of Human Genetics, 2019, 27, 1379-1388.	2.8	8
173	Further definition of the proximal 19p13.3 microdeletion/microduplication syndrome and implication of <scp>PIAS4</scp> as the major contributor. Clinical Genetics, 2020, 97, 467-476.	2.0	8
174	Severe congenital nephrogenic diabetes insipidus in a compound heterozygote with a new large deletion of the AQP2 gene. A case report. Molecular Genetics & Genomic Medicine, 2019, 7, e00568.	1.2	7
175	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellisâ€van Creveld syndrome caused by hypomorphic mutations in the EVC gene. Human Mutation, 2020, 41, 2087-2093.	2.5	7
176	Biallelic truncating variants in MAPKAPK5 cause a new developmental disorder involving neurological, cardiac, and facial anomalies combined with synpolydactyly. Genetics in Medicine, 2021, 23, 679-688.	2.4	7
177	<scp>TBL1XR1</scp> associated intellectual disability, a new missense variant with dysmorphic features plus autism: Expanding the phenotypic spectrum. Clinical Genetics, 2021, 99, 812-817.	2.0	7
178	Segmental undergrowth is associated with pathogenic variants in vascular malformation genes: A retrospective caseâ€series study. Clinical Genetics, 2022, 101, 296-306.	2.0	7
179	Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwithâ€“Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. Clinical Epigenetics, 2022, 14, .	4.1	7
180	Cutaneous and ophthalmic signs as a clue to early diagnosis of severe neurofibromatosis type 2: report of a novel mutation that predicts this poor prognosis. Clinical and Experimental Dermatology, 2014, 39, 557-559.	1.3	6

#	Article	IF	Citations
181	Costello Syndrome and Umbilical Ligament Rhabdomyosarcoma in Two Pediatric Patients: Case Reports and Review of the Literature. Case Reports in Genetics, 2017, 2017, 1-13.	0.2	6
182	Heterozygous pathogenic variants inGLI1are a common finding in isolated postaxial polydactyly A/B. Human Mutation, 2020, 41, 265-276.	2.5	6
183	Pathogenic variants in <scp>KPTN</scp>, a rare cause of macrocephaly and intellectual disability. American Journal of Medical Genetics, Part A, 2020, 182, 2222-2225.	1.2	6
184	Radiological Findings in Multidetector Computed Tomography (MDCT) of Hereditary and Sporadic Pulmonary Veno-Occlusive Disease: Certainties and Uncertainties. Diagnostics, 2021, 11, 141.	2.6	6
185	A novel SMARCAL1 missense mutation that affects splicing in a severely affected Schimke immunoosseous dysplasia patient. European Journal of Medical Genetics, 2016, 59, 363-366.	1.3	5
186	Coâ€occurrence of neurofibromatosis type 1 and optic nerve gliomas with autosomal dominant polycystic kidney disease type 2. Molecular Genetics & Genomic Medicine, 2020, 8, e1321.	1.2	5
187	Preimplantation genetic testing for a chr14q32 microdeletion in a family with Kagami-Ogata syndrome and Temple syndrome. Journal of Medical Genetics, 2022, 59, 253-261.	3.2	5
188	Mosaic Variegated Aneuploidy syndrome 2 caused by biallelic variants in CEP57, two new cases and review of the phenotype. European Journal of Medical Genetics, 2021, 64, 104338.	1.3	5
189	Description of Two New Cases of AQP1 Related Pulmonary Arterial Hypertension and Review of the Literature. Genes, 2022, 13, 927.	2.4	5
190	Additional case of an uncommon 22q11.2 reciprocal rearrangement in a phenotypically normal mother of children with 22q11.2 deletion and 22q11.2 duplication syndromes. American Journal of Medical Genetics, Part A, 2012, 158A, 2963-2968.	1.2	4
191	Multiple copy number variants in a pediatric patient with Hb H disease and intellectual disability. American Journal of Medical Genetics, Part A, 2016, 170, 986-991.	1.2	4
192	Analysis of complex structural variants in the DMD gene in one family. Neuromuscular Disorders, 2021, 31, 253-263.	0.6	4
193	Other tumors in Sotos syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 228-228.	1.2	3
194	Mietensâ€“Weber syndrome: two new patients and a review. Clinical Dysmorphology, 2006, 15, 175-177.	0.3	3
195	Familial imbalance in 16p13.11 leads to a dosage compensation rearrangement in an unaffected carrier. BMC Medical Genetics, 2014, 15, 116.	2.1	3
196	Variantes que mantienen el marco de lectura en el dominio Rod 1 proximal del gen FLNA se asocian con un predominio del fenotipo valvular. Revista Espanola De Cardiologia, 2018, 71, 545-552.	1.2	3
197	Coexistence of autosomal dominant polycystic kidney disease type 1 and hereditary renal hypouricemia type 2: A model of earlyâ€onset and fast cyst progression. Clinical Genetics, 2020, 97, 857-868.	2.0	3
198	Molecular and histologic insights on early onset cardiomyopathy in Danon disease females. Clinical Genetics, 2021, 99, 481-483.	2.0	3

#	Article	IF	Citations
199	Unusual fourâ€generation chromosomeâ€22 rearrangement: When â€œnormalityâ€•masks abnormality. American Journal of Medical Genetics, Part A, 2009, 149A, 1561-1564.	1.2	2
200	Recurrence of Hirschsprung disease due to maternal mosaicism of a novel <scp>RET</scp> gene mutation. Clinical Genetics, 2014, 85, 401-402.	2.0	2
201	Implementation of chromosomal microarrays in a cohort of patients with intellectual disability at the Argentinean public health system. Molecular Biology Reports, 2020, 47, 6863-6878.	2.3	2
202	Prenatal ultrasound findings in Koolenâ€de Vries foetuses: Central nervous system anomalies are frequent markers of this syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1649.	1.2	2
203	Tenorio syndrome: Description of 14 novel cases and review of the clinical and molecular features. Clinical Genetics, 2021, 100, 405-411.	2.0	2
204	Diastrophic dysplasia diagnosed in a case published 100 years ago. , 1998, 77, 334-336.		1
205	OSX/SP7 Mutations and Osteogenesis Imperfecta. , 2014, , 173-179.		1
206	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta. , 2014, , 181-186.		1
207	In-frame Variants in FLNA Proximal Rod 1 Domain Associate With a Predominant Cardiac Valvular Phenotype. Revista Espanola De Cardiologia (English Ed), 2018, 71, 545-552.	0.6	1
208	First report case with negative genetic study (array CGH, exome sequencing) in patients with vertical transmission of Zika virus infection and associated brain abnormalities . The Application of Clinical Genetics, 2019, Volume 12, 141-150.	3.0	1
209	New mutations associated with Hirschsprung disease. Anales De PediatrÃa (English Edition), 2020, 93, 222-227.	0.2	1
210	A Possible Association Between Zika Virus Infection and CDK5RAP2 Mutation. Frontiers in Genetics, 2021, 12, 530028.	2.3	1
211	Birth of two healthy girls following preimplantation genetic diagnosis and gestational surrogacy in a rapidly progressive autosomal dominant polycystic kidney diseaseÂcase using tolvaptan. CKJ: Clinical Kidney Journal, 2021, 14, 1987-1989.	2.9	1
212	The portrayal of dwarfism without skeletal dysplasia in art: Proportionate short stature due to growth hormone deficiency and other disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 186-191.	1.6	1
213	Macrocephaly-Cutis Marmorata Telangiectatica Congenita (Macrocephaly-Capillary Malformation). , 2008, , 323-332.		1
214	Rapidly Progressing to ESRD in an Individual with Coexisting ADPKD and Masked Klinefelter and Gitelman Syndromes. Genes, 2022, 13, 394.	2.4	1
215	Abstract CT225: A randomized trial of integrated genomics, organoids and avatar mouse models for personalized treatment of pancreatic cancer. Cancer Research, 2022, 82, CT225-CT225.	0.9	1
216	Research in two large pediatric residencies in Argentina. Academic Medicine, 1998, 73, 803-5.	1.6	0

#	Article	IF	Citations
217	Reply to correspondence from Martï¿½nez-Frï¿½as et al.??Nowadays it is preceptive to perform chromosomal studies with high resolution G-bands and FISH techniques when necessary?. American Journal of Medical Genetics Part A, 2002, 108, 255-255.	2.4	0
218	New findings in craniofacial dyssynostosis. , 2005, 134A, 344-345.		0
219	Cover Image, Volume 170A, Number 4, April 2016. American Journal of Medical Genetics, Part A, 2016, 170, i.	1.2	0
220	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
221	Historical and geographical distribution of the founder mutation c.610G>A; p.Ala204Thr in the CLCNKB gene linked to Bartter syndrome type III in Spain. CKJ: Clinical Kidney Journal, 2021, 14, 1990-1993.	2.9	0
222	Molecular diagnosis of limb-girdle muscular dystrophy type 2A by next-generation sequencing. Annals of Indian Academy of Neurology, 2017, 20, 164.	0.5	0