

Pablo Lapunzina

List of Publications by Year in descending order

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222
papers

9,345
citations

47409

49
h-index

62345

84
g-index

238
all docs

238
docs citations

238
times ranked

14715
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. <i>Nature Reviews Endocrinology</i> , 2018, 14, 229-249.	4.3	388
2	Diversity and Function of Mutations in P450 Oxidoreductase in Patients with Antley-Bixler Syndrome and Disordered Steroidogenesis. <i>American Journal of Human Genetics</i> , 2005, 76, 729-749.	2.6	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. <i>Genome Research</i> , 2014, 24, 554-569.	2.4	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. <i>Journal of Clinical Oncology</i> , 2019, 37, 490-503.	0.8	255
5	Identification of a Frameshift Mutation in Osterix in a Patient with Recessive Osteogenesis Imperfecta. <i>American Journal of Human Genetics</i> , 2010, 87, 110-114.	2.6	246
6	Mutations in WNT1 Cause Different Forms of Bone Fragility. <i>American Journal of Human Genetics</i> , 2013, 92, 565-574.	2.6	240
7	Risk of tumorigenesis in overgrowth syndromes: A comprehensive review. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2005, 137C, 53-71.	0.7	237
8	Extending the phenotype of recurrent rearrangements of 16p11.2: Deletions in mentally retarded patients without autism and in normal individuals. <i>European Journal of Medical Genetics</i> , 2009, 52, 77-87.	0.7	226
9	Mutations in Two Nonhomologous Genes in a Head-to-Head Configuration Cause Ellis-van Creveld Syndrome. <i>American Journal of Human Genetics</i> , 2003, 72, 728-732.	2.6	200
10	<i>NOTCH2</i> mutations in Alagille syndrome. <i>Journal of Medical Genetics</i> , 2012, 49, 138-144.	1.5	197
11	Epigenetic inactivation of the Sotos overgrowth syndrome gene histone methyltransferase NSD1 in human neuroblastoma and glioma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 21830-21835.	3.3	190
12	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. <i>Human Mutation</i> , 2012, 33, 343-350.	1.1	178
13	HRAS mutation analysis in Costello syndrome: Genotype and phenotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1-7.	0.7	164
14	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. <i>Journal of Clinical Investigation</i> , 2014, 124, 4693-4708.	3.9	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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, 1876-1888.	1.8	127
16	Deletions Involving Long-Range Conserved Nongenic Sequences Upstream and Downstream of FOXL2 as a Novel Disease-Causing Mechanism in Blepharophimosis Syndrome. <i>American Journal of Human Genetics</i> , 2005, 77, 205-218.	2.6	116
17	Reduced hepatic expression of farnesoid X receptor in hereditary cholestasis associated to mutation in ATP8B1. <i>Human Molecular Genetics</i> , 2004, 13, 2451-2460.	1.4	107
18	Stratification of Wilms tumor by genetic and epigenetic analysis. <i>Oncotarget</i> , 2012, 3, 327-335.	0.8	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. <i>Human Molecular Genetics</i> , 2013, 22, 124-139.	1.4	97
20	Molecular and clinical characterization of cardio-facio-cutaneous (CFC) syndrome: Overlapping clinical manifestations with Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 799-807.	0.7	96
21	Molecular and clinical analysis of <i>RAF1</i> in Noonan syndrome and related disorders: dephosphorylation of serine 259 as the essential mechanism for mutant activation. <i>Human Mutation</i> , 2010, 31, 284-294.	1.1	96
22	Genome-Wide Allelic Methylation Analysis Reveals Disease-Specific Susceptibility to Multiple Methylation Defects in Imprinting Syndromes. <i>Human Mutation</i> , 2013, 34, n/a-n/a.	1.1	96
23	Somatic activating mutations in <i>PIK3CA</i> cause generalized lymphatic anomaly. <i>Journal of Experimental Medicine</i> , 2019, 216, 407-418.	4.2	96
24	Epigenetic biomarkers: Current strategies and future challenges for their use in the clinical laboratory. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2017, 54, 529-550.	2.7	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. <i>Trends in Molecular Medicine</i> , 2014, 20, 614-622.	3.5	89
27	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. <i>Human Genetics</i> , 2016, 135, 569-586.	1.8	85
28	New microdeletion and microduplication syndromes: a comprehensive review. <i>Genetics and Molecular Biology</i> , 2014, 37, 210-219.	0.6	84
29	Clinical utility of chromosomal microarray analysis in invasive prenatal diagnosis. <i>Human Genetics</i> , 2012, 131, 513-523.	1.8	82
30	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. <i>Trends in Genetics</i> , 2016, 32, 444-455.	2.9	81
31	Comparative study of three diagnostic approaches (FISH, STRs and MLPA) in 30 patients with 22q11.2 deletion syndrome. <i>Clinical Genetics</i> , 2005, 68, 373-378.	1.0	79
32	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome-Osteogenesis imperfecta phenotypic spectrum. <i>Human Mutation</i> , 2012, 33, 1444-1449.	1.1	77
33	Simpson-Golabi-Behmel syndrome types I and II. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 138.	1.2	75
34	Spectrum of mutations and genotype-phenotype analysis in Noonan syndrome patients with RIT1 mutations. <i>Human Genetics</i> , 2016, 135, 209-222.	1.8	75
35	Cancer Genes Hypermethylated in Human Embryonic Stem Cells. <i>PLoS ONE</i> , 2008, 3, e3294.	1.1	75
36	Microhomology-Mediated Mechanisms Underlie Non-Recurrent Disease-Causing Microdeletions of the FOXL2 Gene or Its Regulatory Domain. <i>PLoS Genetics</i> , 2013, 9, e1003358.	1.5	72

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37	The consequences of uniparental disomy and copy number neutral loss-of-heterozygosity during human development and cancer. <i>Biology of the Cell</i> , 2011, 103, 303-317.	0.7	68
38	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silver-Russell and Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1377-1387.	1.4	68
39	Clinical and Molecular Evaluation of SHOX/PAR1 Duplications in Léri-Weill Dyschondrosteosis (LWD) and Idiopathic Short Stature (ISS). <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E404-E412.	1.8	60
40	Macrocephaly-capillary malformation: Analysis of 13 patients and review of the diagnostic criteria. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 3101-3106.	0.7	57
41	A founder <i>EIF2AK4</i> mutation causes an aggressive form of pulmonary arterial hypertension in Iberian Gypsies. <i>Clinical Genetics</i> , 2015, 88, 579-583.	1.0	57
42	Clinical, biochemical and genetic spectrum of low alkaline phosphatase levels in adults. <i>European Journal of Internal Medicine</i> , 2016, 29, 40-45.	1.0	57
43	Long interspersed nuclear element-1 (LINE1)-mediated deletion of <i>EVC</i> , <i>EVC2</i> , <i>C4orf6</i> , and <i>STK32B</i> in Ellis-van Creveld syndrome with borderline intelligence. <i>Human Mutation</i> , 2008, 29, 931-938.	1.1	55
44	Methylation screening of reciprocal genome-wide UPDs identifies novel human-specific imprinted genes. <i>Human Molecular Genetics</i> , 2011, 20, 3188-3197.	1.4	55
45	<i>FAM46A</i> mutations are responsible for autosomal recessive osteogenesis imperfecta. <i>Journal of Medical Genetics</i> , 2018, 55, 278-284.	1.5	55
46	Widening the mutation spectrum of <i>EVC</i> and <i>EVC2</i> : ectopic expression of Weyer variants in NIH 3T3 fibroblasts disrupts hedgehog signaling. <i>Human Mutation</i> , 2009, 30, 1667-1675.	1.1	54
47	A deletion and a duplication in distal 22q11.2 deletion syndrome region. Clinical implications and review. <i>BMC Medical Genetics</i> , 2009, 10, 48.	2.1	54
48	Expanding the mutation spectrum in 182 Spanish probands with craniosynostosis: identification and characterization of novel <i>TCF12</i> variants. <i>European Journal of Human Genetics</i> , 2015, 23, 907-914.	1.4	53
49	CLAPO syndrome: identification of somatic activating <i>PIK3CA</i> mutations and delineation of the natural history and phenotype. <i>Genetics in Medicine</i> , 2018, 20, 882-889.	1.1	52
50	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. <i>EBioMedicine</i> , 2021, 65, 103246.	2.7	52
51	De Novo Mutations in <i>SLC25A24</i> Cause a Disorder Characterized by Early Aging, Bone Dysplasia, Characteristic Face, and Early Demise. <i>American Journal of Human Genetics</i> , 2017, 101, 844-855.	2.6	51
52	<i>CDKN1C</i> (<i>p57</i> (<i>Kip2</i>)) analysis in Beckwith-Wiedemann syndrome (BWS) patients: Genotype-phenotype correlations, novel mutations, and polymorphisms. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1390-1397.	0.7	50
53	Clinical utility gene card for: Beckwith-Wiedemann Syndrome. <i>European Journal of Human Genetics</i> , 2014, 22, 435-435.	1.4	50
54	<i>GLI1</i> inactivation is associated with developmental phenotypes overlapping with Ellis-van Creveld syndrome. <i>Human Molecular Genetics</i> , 2017, 26, 4556-4571.	1.4	50

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55	<i>FOXP1</i>-related intellectual disability syndrome: a recognisable entity. <i>Journal of Medical Genetics</i> , 2017, 54, 613-623.	1.5	48
56	Refining the phenotype associated with <i>GNB1</i> mutations: Clinical data on 18 newly identified patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2259-2275.	0.7	47
57	Beckwithâ€“Wiedemann syndrome and uniparental disomy 11p: fine mapping of the recombination breakpoints and evaluation of several techniques. <i>European Journal of Human Genetics</i> , 2011, 19, 416-421.	1.4	44
58	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. <i>European Journal of Human Genetics</i> , 2016, 24, 784-793.	1.4	44
59	Alagille Syndrome: Cutaneous Manifestations in 38 Children. <i>Pediatric Dermatology</i> , 2005, 22, 11-14.	0.5	43
60	PAR1 deletions downstream ofSHOX are the most frequent defect in a Spanish cohort of LÃ©ri-Weill dyschondrosteosis (LWD) probands. <i>Human Mutation</i> , 2006, 27, 1062-1062.	1.1	43
61	CDKN1C Mutations in HELLP/Preeclamptic Mothers of Beckwithâ€“Wiedemann Syndrome (BWS) Patients. <i>Placenta</i> , 2009, 30, 551-554.	0.7	43
62	Clinical, biochemical, and molecular studies in pyridoxineâ€“dependent epilepsy. Antisense therapy as possible new therapeutic option. <i>Epilepsia</i> , 2013, 54, 239-248.	2.6	43
63	A patient with Simpson-Golabi-Behmel syndrome and hepatocellular carcinoma.. <i>Journal of Medical Genetics</i> , 1998, 35, 153-156.	1.5	42
64	A Novel Dominant Hyperekplexia Mutation Y705C Alters Trafficking and Biochemical Properties of the Presynaptic Glycine Transporter GlyT2. <i>Journal of Biological Chemistry</i> , 2012, 287, 28986-29002.	1.6	42
65	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotypeâ€“phenotype correlations. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1354-1369.	0.7	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. <i>Human Molecular Genetics</i> , 2015, 24, 4126-4137.	1.4	42
67	Noncoding copy-number variations are associated with congenital limb malformation. <i>Genetics in Medicine</i> , 2018, 20, 599-607.	1.1	42
68	Further delineation of Malan syndrome. <i>Human Mutation</i> , 2018, 39, 1226-1237.	1.1	42
69	Familial Occurrence of the IMAGE Association: Additional Clinical Variants and a Proposed Mode of Inheritance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005, 90, 3186-3190.	1.8	39
70	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. <i>Scientific Reports</i> , 2013, 3, 1346.	1.6	39
71	Novel genes and sex differences in COVID-19 severity. <i>Human Molecular Genetics</i> , 2022, 31, 3789-3806.	1.4	38
72	Multiple Giant Pilomatricoma in Familial Sotos Syndrome. <i>Pediatric Dermatology</i> , 2008, 25, 122-125.	0.5	37

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73	Characterization of a 8q21.11 Microdeletion Syndrome Associated with Intellectual Disability and a Recognizable Phenotype. <i>American Journal of Human Genetics</i> , 2011, 89, 295-301.	2.6	37
74	Detection of Hypomethylation Syndrome among Patients with Epigenetic Alterations at the <i>GNAS</i> Locus. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E1060-E1067.	1.8	37
75	Two mutations in <i>IFITM5</i> causing distinct forms of osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1136-1142.	0.7	37
76	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 28-39.	0.6	37
77	Clinical Implementation of Pharmacogenetic Testing in a Hospital of the Spanish National Health System: Strategy and Experience Over 3 Years. <i>Clinical and Translational Science</i> , 2018, 11, 189-199.	1.5	37
78	Molecular and clinical analysis of <i>ALPL</i> in a cohort of patients with suspicion of Hypophosphatasia. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 601-610.	0.7	36
79	Nomenclature and definition in asymmetric regional body overgrowth. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1735-1738.	0.7	36
80	<i>mTOR</i> mutations in Smith-Kingsmore syndrome: Four additional patients and a review. <i>Clinical Genetics</i> , 2018, 93, 762-775.	1.0	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 <i>KDEL2</i> and <i>HSP47</i> as a Key Determinant in Osteogenesis Imperfecta Caused by Bi-allelic Variants in <i>KDEL2</i> . <i>American Journal of Human Genetics</i> , 2020, 107, 989-999.	2.6	35
83	Variants in members of the cadherin-catenin complex, <i>CDH1</i> and <i>CTNND1</i> , cause blepharocheilodontic syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 210-219.	1.4	34
84	A six-attribute classification of genetic mosaicism. <i>Genetics in Medicine</i> , 2020, 22, 1743-1757.	1.1	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. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2583-2588.	0.7	33
86	Adults with Sotos syndrome: Review of 21 adults with molecularly confirmed <i>NSD1</i> alterations, including a detailed case report of the oldest person. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2105-2111.	0.7	33
87	A New Overgrowth Syndrome is due to Mutations in <i>RNF125</i> . <i>Human Mutation</i> , 2014, 35, 1436-1441.	1.1	33
88	The syndrome of central hypothyroidism and macroorchidism: <i>IGSF1</i> controls <i>TRHR</i> and <i>FSHB</i> expression by differential modulation of pituitary <i>TGFβ2</i> and Activin pathways. <i>Scientific Reports</i> , 2017, 7, 42937.	1.6	33
89	Germline and Mosaic Variants in <i>PRKACA</i> and <i>PRKACB</i> Cause a Multiple Congenital Malformation Syndrome. <i>American Journal of Human Genetics</i> , 2020, 107, 977-988.	2.6	33
90	Constitutional mosaic genome-wide uniparental disomy due to diploidisation: an unusual cancer-predisposing mechanism. <i>Journal of Medical Genetics</i> , 2011, 48, 212-216.	1.5	32

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91	Customized high resolution CGH array for clinical diagnosis reveals additional genomic imbalances in previously well-defined pathological samples. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1950-1960.	0.7	32
92	The role of ZFP57 and additional KRAB-zinc finger proteins in the maintenance of human imprinted methylation and multi-locus imprinting disturbances. <i>Nucleic Acids Research</i> , 2020, 48, 11394-11407.	6.5	32
93	MLPA vs multiprobe FISH: comparison of two methods for the screening of subtelomeric rearrangements in 50 patients with idiopathic mental retardation. <i>Clinical Genetics</i> , 2006, 69, 228-233.	1.0	31
94	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform. <i>Epigenomics</i> , 2018, 10, 941-954.	1.0	31
95	Clinical and molecular analyses of Beckwith-Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2740-2749.	0.7	30
96	Abnormal bone turnover in individuals with low serum alkaline phosphatase. <i>Osteoporosis International</i> , 2018, 29, 2147-2150.	1.3	30
97	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1615-1626.	1.4	29
98	46,XX ovotesticular DSD associated with a SOX3 gene duplication in a SRY-negative boy. <i>Clinical Endocrinology</i> , 2016, 85, 673-675.	1.2	28
99	Oculocerebrocutaneous (Delleman) Syndrome: Report of Two Cases. <i>Neuropediatrics</i> , 2005, 36, 50-54.	0.3	27
100	Report of a newly identified patient with mutations in BMP1 and underlying pathogenetic aspects. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1143-1150.	0.7	27
101	The most recurrent monogenic disorders that overlap with the phenotype of Rett syndrome. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 609-620.	0.7	27
102	Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. <i>Frontiers in Genetics</i> , 2022, 13, 652454.	1.1	27
103	The role of CDKN2A/B deletions in pediatric acute lymphoblastic leukemia. <i>Pediatric Hematology and Oncology</i> , 2016, 33, 415-422.	0.3	26
104	Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH. <i>PLoS ONE</i> , 2017, 12, e0172363.	1.1	26
105	Biomarkers in Vestibular Schwannoma-Associated Hearing Loss. <i>Frontiers in Neurology</i> , 2019, 10, 978.	1.1	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. <i>Revista Espanola De Cardiologia (English Ed)</i> , 2016, 69, 1011-1019.	0.4	25
107	Follow-up and Risk of Tumors in Overgrowth Syndromes. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2005, 18, 1227-35.	0.4	24
108	Síndrome LEOPARD: una variante del síndrome de Noonan con fuerte asociación a miocardiopatía hipertrófica. <i>Revista Espanola De Cardiologia</i> , 2013, 66, 350-356.	0.6	24

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109	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. <i>Nature Communications</i> , 2019, 10, 797.	5.8	24
110	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. <i>Scientific Reports</i> , 2017, 7, 12288.	1.6	23
111	Risks of congenital anomalies in large for gestational age infants. <i>Journal of Pediatrics</i> , 2002, 140, 200-204.	0.9	22
112	Clinical and molecular studies on two further families with Simpson-Golabi-Behmel syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 272-277.	0.7	22
113	Functional Characterization of NIPBL Physiological Splice Variants and Eight Splicing Mutations in Patients with Cornelia de Lange Syndrome. <i>International Journal of Molecular Sciences</i> , 2014, 15, 10350-10364.	1.8	22
114	Identification of a premature stop codon mutation in the <i>PHGDH</i> gene in severe Neu-Laxova syndrome—evidence for phenotypic variability. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1323-1329.	0.7	22
115	Analysis of invdupdel(8p) rearrangement: Clinical, cytogenetic and molecular characterization. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1018-1025.	0.7	22
116	Genetic analyses of aplastic anemia and idiopathic pulmonary fibrosis patients with short telomeres, possible implication of DNA-repair genes. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 82.	1.2	21
117	Clinical heterogeneity of Pulmonary Arterial Hypertension associated with variants in TBX4. <i>PLoS ONE</i> , 2020, 15, e0232216.	1.1	21
118	Customized Massive Parallel Sequencing Panel for Diagnosis of Pulmonary Arterial Hypertension. <i>Genes</i> , 2020, 11, 1158.	1.0	21
119	Ultraviolet light-related neural tube defects?. , 1996, 67, 106-106.		20
120	A novel insertion in the <i>FGFR2</i> gene in a patient with Crouzon phenotype and sacroccygeal tail. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2005, 73, 61-64.	1.6	20
121	<i>FOXL2</i> copy number changes in the molecular pathogenesis of BPES: unique cohort of 17 deletions. <i>Human Mutation</i> , 2010, 31, n/a-n/a.	1.1	20
122	Higher frequency of uncommon 1.5-2 Mb deletions found in familial cases of 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 71-75.	0.7	19
123	Rothmund—Thomson Syndrome: novel pathogenic mutations and frequencies of variants in the <i>RECQL4</i> and <i>USB1</i> (C16orf57) gene. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 359-366.	0.6	19
124	Characterization of rare <i>ABCC8</i> variants identified in Spanish pulmonary arterial hypertension patients. <i>Scientific Reports</i> , 2020, 10, 15135.	1.6	19
125	Eye coloboma and complex cardiac malformations belong to the clinical spectrum of <i>PUF60</i> variants. <i>Clinical Genetics</i> , 2017, 92, 350-351.	1.0	18
126	Sotos syndrome is associated with leukemia/lymphoma. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1244-1245.	0.7	17

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127	LEOPARD Syndrome: A Variant of Noonan Syndrome Strongly Associated With Hypertrophic Cardiomyopathy. <i>Revista Espanola De Cardiologia (English Ed)</i> , 2013, 66, 350-356.	0.4	17
128	Prediction models for voriconazole pharmacokinetics based on pharmacogenetics: AN exploratory study in a Spanish population. <i>International Journal of Antimicrobial Agents</i> , 2019, 54, 463-470.	1.1	17
129	Novel Genetic and Molecular Pathways in Pulmonary Arterial Hypertension Associated with Connective Tissue Disease. <i>Cells</i> , 2021, 10, 1488.	1.8	17
130	Molecular diagnosis of Beckwith-Wiedemann Syndrome using quantitative methylation-sensitive polymerase chain reaction. <i>Genetics in Medicine</i> , 2006, 8, 628-634.	1.1	16
131	P2X receptors up-regulate the cell-surface expression of the neuronal glycine transporter GlyT2. <i>Neuropharmacology</i> , 2017, 125, 99-116.	2.0	16
132	Further delineation of neuropsychiatric findings in Tatton-Brown-Rahman syndrome due to disease-causing variants in DNMT3A: seven new patients. <i>European Journal of Human Genetics</i> , 2020, 28, 469-479.	1.4	16
133	Molecular characterization of Spanish patients with <i>MECP2</i> duplication syndrome. <i>Clinical Genetics</i> , 2020, 97, 610-620.	1.0	16
134	Hyperekplexia (Startle Disease). <i>Molecular Diagnosis and Therapy</i> , 2003, 7, 125-128.	1.3	16
135	Beckwith-Wiedemann syndrome due to 11p15.5 paternal duplication associated with Klinefelter syndrome and a <i>de novo</i> pericentric inversion of chromosome Y. <i>European Journal of Medical Genetics</i> , 2005, 48, 159-166.	0.7	15
136	Unrelated chromosomal anomalies found in patients with suspected 22q11.2 deletion. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1134-1141.	0.7	15
137	A parallel study of different array-CGH platforms in a set of Spanish patients with developmental delay and intellectual disability. <i>Gene</i> , 2013, 521, 82-86.	1.0	15
138	Semilobar holoprosencephaly, coronal craniosynostosis, and multiple congenital anomalies: A severe expression of the Genoa syndrome or a newly recognized syndrome?. <i>American Journal of Medical Genetics Part A</i> , 2001, 102, 258-260.	2.4	14
139	Craniofacial dyssynostosis: Description of the first four Spanish cases and review. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 41-48.	0.7	14
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