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	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
2	<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
3	Rapidly Progressing to ESRD in an Individual with Coexisting ADPKD and Masked Klinefelter and Gitelman Syndromes. Genes, 2022, 13, 394.	2.4	1
4	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	4.1	14
5	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
6	Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. Frontiers in Genetics, 2022, 13, 652454.	2.3	27
7	Description of Two New Cases of AQP1 Related Pulmonary Arterial Hypertension and Review of the Literature. Genes, 2022, 13, 927.	2.4	5
8	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
9	Novel genes and sex differences in COVID-19 severity. Human Molecular Genetics, 2022, 31, 3789-3806.	2.9	38
10	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
11	Molecular and histologic insights on early onset cardiomyopathy in Danon disease females. Clinical Genetics, 2021, 99, 481-483.	2.0	3
12	Analysis of complex structural variants in the DMD gene in one family. Neuromuscular Disorders, 2021, 31, 253-263.	0.6	4
13	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
14	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
15	<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
16	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
17	A Possible Association Between Zika Virus Infection and CDK5RAP2 Mutation. Frontiers in Genetics, 2021, 12, 530028.	2.3	1
18	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

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19	Novel TNIP2 and TRAF2 Variants Are Implicated in the Pathogenesis of Pulmonary Arterial Hypertension. Frontiers in Medicine, 2021, 8, 625763.	2.6	13
20	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
21	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
22	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
23	Schuursâ€“Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	2.4	13
24	Novel Genetic and Molecular Pathways in Pulmonary Arterial Hypertension Associated with Connective Tissue Disease. Cells, 2021, 10, 1488.	4.1	17
25	Tenorio syndrome: Description of 14 novel cases and review of the clinical and molecular features. Clinical Genetics, 2021, 100, 405-411.	2.0	2
26	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
27	Expanding the Evidence of a Semi-Dominant Inheritance in GDF2 Associated with Pulmonary Arterial Hypertension. Cells, 2021, 10, 3178.	4.1	12
28	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
29	Heterozygous pathogenic variants inGLI1are a common finding in isolated postaxial polydactyly A/B. Human Mutation, 2020, 41, 265-276.	2.5	6
30	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
31	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
32	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
33	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
34	A six-attribute classification of geneticmosaicism. Genetics in Medicine, 2020, 22, 1743-1757.	2.4	34
35	Molecular characterization of Spanish patients with MECP2 duplication syndrome. Clinical Genetics, 2020, 97, 610-620.	2.0	16
36	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

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37	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
38	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
39	Characterization of rare ABCC8 variants identified in Spanish pulmonary arterial hypertension patients. Scientific Reports, 2020, 10, 15135.	3.3	19
40	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
41	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
42	Can we identify individuals with an ALPL variant in adults with persistent hypophosphatasaemia?. Orphanet Journal of Rare Diseases, 2020, 15, 51.	2.7	10
43	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
44	New mutations associated with Hirschsprung disease. Anales De PediatrÃa (English Edition), 2020, 93, 222-227.	0.2	1
45	Clinical heterogeneity of Pulmonary Arterial Hypertension associated with variants in TBX4. PLoS ONE, 2020, 15, e0232216.	2.5	21
46	Customized Massive Parallel Sequencing Panel for Diagnosis of Pulmonary Arterial Hypertension. Genes, 2020, 11, 1158.	2.4	21
47	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
48	Biomarkers in Vestibular Schwannomaâ€“Associated Hearing Loss. Frontiers in Neurology, 2019, 10, 978.	2.4	26
49	MRX93 syndrome (BRWD3 gene): five new patients with novel mutations. Clinical Genetics, 2019, 95, 726-731.	2.0	13
50	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
51	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
52	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
53	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
54	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

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55	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	12.8	24
56	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
57	Somatic activating mutations in PIK3CA cause generalized lymphatic anomaly. Journal of Experimental Medicine, 2019, 216, 407-418.	8.5	96
58	Constitutional mosaicism in RASA1â€related capillary malformationâ€arteriovenous malformation. Clinical Genetics, 2019, 95, 516-519.	2.0	10
59	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
60	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
61	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
62	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
63	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
64	FAM46A mutations are responsible for autosomal recessive osteogenesis imperfecta. Journal of Medical Genetics, 2018, 55, 278-284.	3.2	55
65	Noncoding copy-number variations are associated with congenital limb malformation. Genetics in Medicine, 2018, 20, 599-607.	2.4	42
66	mTOR mutations in Smithâ€Kingsmore syndrome: Four additional patients and a review. Clinical Genetics, 2018, 93, 762-775.	2.0	36
67	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
68	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
69	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
70	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform. Epigenomics, 2018, 10, 941-954.	2.1	31
71	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
72	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	42

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73	Abnormal bone turnover in individuals with low serum alkaline phosphatase. Osteoporosis International, 2018, 29, 2147-2150.	3.1	30
74	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
75	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
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	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
78	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	1.2	36
79	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
80	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
81	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. Scientific Reports, 2017, 7, 12288.	3.3	23
82	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellisâ€“van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571.	2.9	50
83	P2X receptors up-regulate the cell-surface expression of the neuronal glycine transporter GlyT2. Neuropharmacology, 2017, 125, 99-116.	4.1	16
84	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
85	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48
86	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
87	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
88	TP53 and CDKN1A mutation analysis in families with Liâ€“Fraumeni and Liâ€“Fraumeni like syndromes. Familial Cancer, 2017, 16, 243-248.	1.9	14
89	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
90	Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH. PLoS ONE, 2017, 12, e0172363.	2.5	26

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91	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
92	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
93	46,<scp>XX</scp> ovotesticular <scp>DSD</scp> associated with a <i><scp>SOX</scp>3</i> gene duplication in a <i><scp>SRY</scp></i>â€negative boy. Clinical Endocrinology, 2016, 85, 673-675.	2.4	28
94	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
95	The role of CDKN2A/B deletions in pediatric acute lymphoblastic leukemia. Pediatric Hematology and Oncology, 2016, 33, 415-422.	0.8	26
96	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. Trends in Genetics, 2016, 32, 444-455.	6.7	81
97	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
98	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
99	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
100	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
101	Cover Image, Volume 170A, Number 4, April 2016. American Journal of Medical Genetics, Part A, 2016, 170, i.	1.2	0
102	Two novel <i>POC1A</i> 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
103	A view on clinical genetics and genomics in Spain: of challenges and opportunities. Molecular Genetics & Genomic Medicine, 2016, 4, 376-391.	1.2	8
104	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
105	Clinical, biochemical and genetic spectrum of low alkaline phosphatase levels in adults. European Journal of Internal Medicine, 2016, 29, 40-45.	2.2	57
106	Spectrum of mutations and genotypeâ€“phenotype analysis in Noonan syndrome patients with RIT1 mutations. Human Genetics, 2016, 135, 209-222.	3.8	75
107	Rothmundâ€“Thomson Syndrome: novel pathogenic mutations and frequencies of variants in the <i><scp>RECQL</scp>4</i> and <i><scp>USB</scp>1 (C16orf57)</i> gene. Molecular Genetics & Genomic Medicine, 2016, 4, 359-366.	1.2	19
108	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

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109	Array CGH Analysis of Paired Blood and Tumor Samples from Patients with Sporadic Wilms Tumor. PLoS ONE, 2015, 10, e0136812.	2.5	8
110	A founder EIF2AK4 mutation causes an aggressive form of pulmonary arterial hypertension in Iberian Gypsies. Clinical Genetics, 2015, 88, 579-583.	2.0	57
111	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
112	Identification of a premature stop codon mutation in the PHGDH gene in severe Neuâ€Laxova syndromeâ€"evidence for phenotypic variability. American Journal of Medical Genetics, Part A, 2015, 167, 1323-1329.	1.2	22
113	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
114	Analysis of invdupdel(8p) rearrangement: Clinical, cytogenetic and molecular characterization. American Journal of Medical Genetics, Part A, 2015, 167, 1018-1025.	1.2	22
115	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
116	New microdeletion and microduplication syndromes: a comprehensive review. Genetics and Molecular Biology, 2014, 37, 210-219.	1.3	84
117	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
118	OSX/SP7 Mutations and Osteogenesis Imperfecta. , 2014, , 173-179.		1
119	Simpson-Golabi-Behmel syndrome types I and II. Orphanet Journal of Rare Diseases, 2014, 9, 138.	2.7	75
120	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
121	CDKN1C mutations: two sides of the same coin. Trends in Molecular Medicine, 2014, 20, 614-622.	6.7	89
122	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta. , 2014, , 181-186.		1
123	Two mutations in IFITM5 causing distinct forms of osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1136-1142.	1.2	37
124	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
125	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
126	Familial imbalance in 16p13.11 leads to a dosage compensation rearrangement in an unaffected carrier. BMC Medical Genetics, 2014, 15, 116.	2.1	3

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127	Clinical utility gene card for: Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	2.8	50
128	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
129	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
130	A New Overgrowth Syndrome is due to Mutations inRNF125. Human Mutation, 2014, 35, 1436-1441.	2.5	33
131	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
132	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	8.2	153
133	Clinical, biochemical, and molecular studies in pyridoxineâ€dependent epilepsy. Antisense therapy as possible new therapeutic option. Epilepsia, 2013, 54, 239-248.	5.1	43
134	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
135	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
136	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
137	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
138	Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574.	6.2	240
139	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
140	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
141	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
142	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
143	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
144	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. Scientific Reports, 2013, 3, 1346.	3.3	39

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145	Detection of Hypomethylation Syndrome among Patients with Epigenetic Alterations at theGNASLocus. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1060-E1067.	3.6	37
146	NOTCH2mutations in Alagille syndrome. Journal of Medical Genetics, 2012, 49, 138-144.	3.2	197
147	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
148	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
149	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
150	Stratification of Wilms tumor by genetic and epigenetic analysis. Oncotarget, 2012, 3, 327-335.	1.8	101
151	Constitutional and somatic methylation status of DMRH19 and KvDMR in Wilms tumor patients. Genetics and Molecular Biology, 2012, 35, 714-724.	1.3	11
152	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
153	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. Human Mutation, 2012, 33, 343-350.	2.5	178
154	Clinical utility of chromosomal microarray analysis in invasive prenatal diagnosis. Human Genetics, 2012, 131, 513-523.	3.8	82
155	Methylation screening of reciprocal genome-wide UPDs identifies novel human-specific imprinted genesâ€. Human Molecular Genetics, 2011, 20, 3188-3197.	2.9	55
156	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
157	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
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