

Maria Rita Passos-Bueno

List of Publications by Year in descending order

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257
papers

13,107
citations

36203

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docs citations

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times ranked

15492
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#	ARTICLE	IF	CITATIONS
1	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	13.5	1,422
2	Mutations in the proteolytic enzyme calpain 3 cause limb-girdle muscular dystrophy type 2A. <i>Cell</i> , 1995, 81, 27-40.	13.5	922
3	A gene related to <i>Caenorhabditis elegans</i> spermatogenesis factor <i>fer-1</i> is mutated in limb-girdle muscular dystrophy type 2B. <i>Nature Genetics</i> , 1998, 20, 37-42.	9.4	626
4	Autosomal recessive limb-girdle muscular dystrophy, LGMD2F, is caused by a mutation in the "sarcoglycan gene. <i>Nature Genetics</i> , 1996, 14, 195-198.	9.4	417
5	Limb-girdle muscular dystrophy type 2G is caused by mutations in the gene encoding the sarcomeric protein telethonin. <i>Nature Genetics</i> , 2000, 24, 163-166.	9.4	312
6	Clinical spectrum of fibroblast growth factor receptor mutations. <i>Human Mutation</i> , 1999, 14, 115-125.	1.1	284
7	Collagen XVIII, containing an endogenous inhibitor of angiogenesis and tumor growth, plays a critical role in the maintenance of retinal structure and in neural tube closure (Knobloch syndrome). <i>Human Molecular Genetics</i> , 2000, 9, 2051-2058.	1.4	259
8	RAB23 Mutations in Carpenter Syndrome Imply an Unexpected Role for Hedgehog Signaling in Cranial-Suture Development and Obesity. <i>American Journal of Human Genetics</i> , 2007, 80, 1162-1170.	2.6	229
9	Caveolin-3 in muscular dystrophy. <i>Human Molecular Genetics</i> , 1998, 7, 871-877.	1.4	200
10	Rare variants in <i>SOS2</i> and <i>LZTR1</i> are associated with Noonan syndrome. <i>Journal of Medical Genetics</i> , 2015, 52, 413-421.	1.5	187
11	Reconstruction of Large Cranial Defects in Nonimmunosuppressed Experimental Design With Human Dental Pulp Stem Cells. <i>Journal of Craniofacial Surgery</i> , 2008, 19, 204-210.	0.3	185
12	Exomic variants of an elderly cohort of Brazilians in the ABraOM database. <i>Human Mutation</i> , 2017, 38, 751-763.	1.1	181
13	Modeling non-syndromic autism and the impact of TRPC6 disruption in human neurons. <i>Molecular Psychiatry</i> , 2015, 20, 1350-1365.	4.1	175
14	The sarcoglycan complex in the six autosomal recessive limb-girdle muscular dystrophies. <i>Human Molecular Genetics</i> , 1996, 5, 1963-1969.	1.4	167
15	Stem cell proliferation under low intensity laser irradiation: A preliminary study. <i>Lasers in Surgery and Medicine</i> , 2008, 40, 433-438.	1.1	155
16	High serum endostatin levels in Down syndrome: implications for improved treatment and prevention of solid tumours. <i>European Journal of Human Genetics</i> , 2001, 9, 811-814.	1.4	145
17	Serum creatine-kinase (CK) and pyruvate-kinase (PK) activities in Duchenne (DMD) as compared with Becker (BMD) muscular dystrophy. <i>Journal of the Neurological Sciences</i> , 1991, 102, 190-196.	0.3	136
18	The Seventh Form of Autosomal Recessive Limb-Girdle Muscular Dystrophy Is Mapped to 17q11-12. <i>American Journal of Human Genetics</i> , 1997, 61, 151-159.	2.6	136

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19	Genetics of Craniosynostosis: Genes, Syndromes, Mutations and Genotype-Phenotype Correlations. <i>Frontiers of Oral Biology</i> , 2008, 12, 107-143.	1.5	134
20	Molecular Analysis of Collagen XVIII Reveals Novel Mutations, Presence of a Third Isoform, and Possible Genetic Heterogeneity in Knobloch Syndrome. <i>American Journal of Human Genetics</i> , 2002, 71, 1320-1329.	2.6	128
21	The facioscapulohumeral muscular dystrophy (FSHD1) gene affects males more severely and more frequently than females. <i>American Journal of Medical Genetics Part A</i> , 1998, 77, 155-161.	2.4	123
22	Asymptomatic carriers and gender differences in facioscapulohumeral muscular dystrophy (FSHD). <i>Neuromuscular Disorders</i> , 2004, 14, 33-38.	0.3	114
23	Fat Grafts Supplemented with Adipose-Derived Stromal Cells in the Rehabilitation of Patients with Craniofacial Microsomia. <i>Plastic and Reconstructive Surgery</i> , 2013, 132, 141-152.	0.7	114
24	High mutation detection rate inTCOF1 among Treacher Collins syndrome patients reveals clustering of mutations and 16 novel pathogenic changes. <i>Human Mutation</i> , 2000, 16, 315-322.	1.1	112
25	Genomic screening for beta-sarcoglycan gene mutations: missense mutations may cause severe limb-girdle muscular dystrophy type 2E (LGMD 2E). <i>Human Molecular Genetics</i> , 1996, 5, 1953-1961.	1.4	111
26	Familial spongiform encephalopathy associated with a novel prion protein gene mutation. <i>Annals of Neurology</i> , 1997, 42, 138-146.	2.8	110
27	Whole-genome array-CGH screening in undiagnosed syndromic patients: old syndromes revisited and new alterations. <i>Cytogenetic and Genome Research</i> , 2006, 115, 254-261.	0.6	103
28	A Noncoding Expansion in EIF4A3 Causes Richieri-Costa-Pereira Syndrome, a Craniofacial Disorder Associated with Limb Defects. <i>American Journal of Human Genetics</i> , 2014, 94, 120-128.	2.6	99
29	Limb-girdle muscular dystrophy: one gene with different phenotypes, one phenotype with different genes. <i>Current Opinion in Neurology</i> , 2000, 13, 511-517.	1.8	93
30	Linkage analysis in autosomal recessive limb-girdle muscular dystrophy (AR LGMD) maps a sixth form to 5q33-34 (LGMD2F) and indicates that there is at least one more subtype of AR LGMD. <i>Human Molecular Genetics</i> , 1996, 5, 815-820.	1.4	92
31	Seven autosomal recessive limb-girdle muscular dystrophies in the Brazilian population: from LGMD2A to LGMD2G. , 1999, 82, 392-398.		90
32	Discordant congenital Zika syndrome twins show differential in vitro viral susceptibility of neural progenitor cells. <i>Nature Communications</i> , 2018, 9, 475.	5.8	86
33	Syndromes of the first and second pharyngeal arches: A review. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1853-1859.	0.7	85
34	A review of craniofacial disorders caused by spliceosomal defects. <i>Clinical Genetics</i> , 2015, 88, 405-415.	1.0	85
35	Clinical variability in calpainopathy: What makes the difference?. <i>European Journal of Human Genetics</i> , 2002, 10, 825-832.	1.4	84
36	Sarcoglycanopathies are responsible for 68% of severe autosomal recessive limb-girdle muscular dystrophy in the Brazilian population. <i>Journal of the Neurological Sciences</i> , 1999, 164, 44-49.	0.3	81

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37	Heterozygous Mutations of <i>FREM1</i> Are Associated with an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. <i>PLoS Genetics</i> , 2011, 7, e1002278.	1.5	80
38	Analysis of the CTG repeat in skeletal muscle of young and adult myotonic dystrophy patients: when does the expansion occur?. <i>Human Molecular Genetics</i> , 1995, 4, 401-406.	1.4	79
39	A gene which causes severe ocular alterations and occipital encephalocele (Knobloch syndrome) is mapped to 21q22.3. <i>Human Molecular Genetics</i> , 1996, 5, 843-847.	1.4	76
40	New Source of Muscle-Derived Stem Cells with Potential for Alveolar Bone Reconstruction in Cleft Lip and/or Palate Patients. <i>Tissue Engineering - Part A</i> , 2009, 15, 427-435.	1.6	71
41	Gastric Bypass and Sleeve Gastrectomy: the Same Impact on IL-6 and TNF- α . <i>Prospective Clinical Trial. Obesity Surgery</i> , 2013, 23, 1252-1261.	1.1	69
42	Dysferlin Protein Analysis in Limb-Girdle Muscular Dystrophies. <i>Journal of Molecular Neuroscience</i> , 2001, 17, 71-80.	1.1	67
43	Mesenchymal Stem Cells Derived From Canine Umbilical Cord Vein – A Novel Source for Cell Therapy Studies. <i>Stem Cells and Development</i> , 2010, 19, 395-402.	1.1	67
44	Apparent association of mental retardation and specific patterns of deletions screened with probes <i>cf56a</i> and <i>cf23a</i> in Duchenne muscular dystrophy. <i>American Journal of Medical Genetics Part A</i> , 1991, 39, 437-441.	2.4	66
45	Cellular interference in craniofrontonasal syndrome: males mosaic for mutations in the X-linked <i>EFNB1</i> gene are more severely affected than true hemizygotes. <i>Human Molecular Genetics</i> , 2013, 22, 1654-1662.	1.4	66
46	The short variant of the polymorphism within the promoter region of the serotonin transporter gene is a risk factor for late onset Alzheimer's disease. <i>Molecular Psychiatry</i> , 1998, 3, 438-441.	4.1	65
47	Autosomal recessive <i>POLR1D</i> mutation with decrease of <i>TCOF1</i> mRNA is responsible for Treacher Collins syndrome. <i>Genetics in Medicine</i> , 2014, 16, 720-724.	1.1	63
48	A new form of autosomal dominant limb-girdle muscular dystrophy (LGMD1G) with progressive fingers and toes flexion limitation maps to chromosome 4p21. <i>European Journal of Human Genetics</i> , 2004, 12, 1033-1040.	1.4	61
49	A Novel Autosomal Recessive <i>GJA1</i> Missense Mutation Linked to Craniometaphyseal Dysplasia. <i>PLoS ONE</i> , 2013, 8, e73576.	1.1	61
50	Decreased cellular uptake and metabolism in Allan-Herndon-Dudley syndrome (AHDS) due to a novel mutation in the <i>MCT8</i> thyroid hormone transporter. <i>Journal of Medical Genetics</i> , 2005, 43, 457-460.	1.5	59
51	Differential methylation is associated with non-syndromic cleft lip and palate and contributes to penetrance effects. <i>Scientific Reports</i> , 2017, 7, 2441.	1.6	59
52	Screening of <i>TCOF1</i> in patients from different populations: confirmation of mutational hot spots and identification of a novel missense mutation that suggests an important functional domain in the protein treacle. <i>Journal of Medical Genetics</i> , 2002, 39, 493-495.	1.5	55
53	New SMS mutation leads to a striking reduction in spermine synthase protein function and a severe form of Snyder-Robinson X-linked recessive mental retardation syndrome. <i>Journal of Medical Genetics</i> , 2008, 45, 539-543.	1.5	53
54	Further evidence of the importance of <i>RIT1</i> in Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2952-2957.	0.7	53

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55	Immunofluorescence dystrophin study in Duchenne dystrophy through the concomitant use of two antibodies directed against the carboxy-terminal and the amino-terminal region of the protein. <i>Journal of the Neurological Sciences</i> , 1991, 101, 141-147.	0.3	52
56	Genetic heterogeneity for Duchenne-like muscular dystrophy (DLMD) based on linkage and 50 DAG analysis. <i>Human Molecular Genetics</i> , 1993, 2, 1945-1947.	1.4	50
57	Mutations in collagen 18A1 (COL18A1) and their relevance to the human phenotype. <i>Anais Da Academia Brasileira De Ciencias</i> , 2006, 78, 123-131.	0.3	50
58	Telethonin protein expression in neuromuscular disorders. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2002, 1588, 33-40.	1.8	49
59	Maternal MTHFR interacts with the offspring's BCL3 genotypes, but not with TGFA, in increasing risk to nonsyndromic cleft lip with or without cleft palate. <i>European Journal of Human Genetics</i> , 2004, 12, 521-526.	1.4	49
60	Deficiency of Merosin (Laminin M or $\alpha 2$) in Congenital Muscular Dystrophy Associated with Cerebral White Matter Alterations. <i>Neuropediatrics</i> , 1995, 26, 293-297.	0.3	48
61	A first missense mutation in the delta sarcoglycan gene associated with a severe phenotype and frequency of limb-girdle muscular dystrophy type 2F (LGMD2F) in Brazilian sarcoglycanopathies.. <i>Journal of Medical Genetics</i> , 1998, 35, 951-953.	1.5	48
62	Molecular screening for microdeletions at 9p22-p24 and 11q23-q24 in a large cohort of patients with trigonocephaly. <i>Clinical Genetics</i> , 2005, 67, 503-510.	1.0	48
63	Understanding the basis of auriculocondylar syndrome: Insights from human, mouse and zebrafish genetic studies. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2013, 163, 306-317.	0.7	48
64	Deletion of the Basement Membrane Heparan Sulfate Proteoglycan Type XVIII Collagen Causes Hypertriglyceridemia in Mice and Humans. <i>PLoS ONE</i> , 2010, 5, e13919.	1.1	46
65	Main clinical features of the three mapped autosomal recessive limb-girdle muscular dystrophies and estimated proportion of each form in 13 Brazilian families.. <i>Journal of Medical Genetics</i> , 1996, 33, 97-102.	1.5	45
66	Rare Variants in the Epithelial Cadherin Gene Underlying the Genetic Etiology of Nonsyndromic Cleft Lip with or without Cleft Palate. <i>Human Mutation</i> , 2015, 36, 1029-1033.	1.1	45
67	Using a combination of MLPA kits to detect chromosomal imbalances in patients with multiple congenital anomalies and mental retardation is a valuable choice for developing countries. <i>European Journal of Medical Genetics</i> , 2011, 54, e425-e432.	0.7	44
68	Genetics and genomics in Brazil: a promising future. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 280-291.	0.6	44
69	Transcriptome of iPSC-derived neuronal cells reveals a module of co-expressed genes consistently associated with autism spectrum disorder. <i>Molecular Psychiatry</i> , 2021, 26, 1589-1605.	4.1	44
70	Estimate of the proportion of Duchenne muscular dystrophy with autosomal recessive inheritance. <i>American Journal of Medical Genetics Part A</i> , 1989, 32, 407-410.	2.4	42
71	The phenotype of chromosome 2p-linked limb-girdle muscular dystrophy. <i>Neuromuscular Disorders</i> , 1996, 6, 483-490.	0.3	42
72	EIF4A3 deficient human iPSCs and mouse models demonstrate neural crest defects that underlie Richieri-Costa-Pereira syndrome. <i>Human Molecular Genetics</i> , 2017, 26, 2177-2191.	1.4	42

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73	Confirmation of the 2p Locus for the Mild Autosomal Recessive Limb-Girdle Muscular Dystrophy Gene (LGMD2B) in Three Families Allows Refinement of the Candidate Region. <i>Genomics</i> , 1995, 27, 192-195.	1.3	41
74	Absence of calpain 3 in a form of limb-girdle muscular dystrophy (LGMD2A). <i>Journal of the Neurological Sciences</i> , 1997, 146, 173-178.	0.3	41
75	Description of a new mutation and characterization of FGFR1, FGFR2, and FGFR3 mutations among Brazilian patients with syndromic craniosynostoses. <i>American Journal of Medical Genetics Part A</i> , 1998, 78, 237-241.	2.4	41
76	Mapping of the autosomal recessive (AR) craniometaphyseal dysplasia locus to chromosome region 6q21-22 and confirmation of genetic heterogeneity for mild AR spondylocostal dysplasia. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 482-491.	2.4	41
77	Optimization of Parameters for a More Efficient Use of Adipose-Derived Stem Cells in Regenerative Medicine Therapies. <i>Stem Cells International</i> , 2012, 2012, 1-7.	1.2	40
78	Screening of deletions in the dystrophin gene with the cDNA probes Cf23a, Cf56a, and Cf115.. <i>Journal of Medical Genetics</i> , 1990, 27, 145-150.	1.5	39
79	Mutations in PCYT1A Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 94, 113-119.	2.6	39
80	Dystrophin immunostaining in muscles from patients with different types of muscular dystrophy: a Brazilian study. <i>Journal of the Neurological Sciences</i> , 1990, 98, 221-233.	0.3	38
81	A deletion including the brain promoter of the Duchenne muscular dystrophy gene is not associated with mental retardation. <i>Neuromuscular Disorders</i> , 1992, 2, 117-120.	0.3	38
82	Functional Vascular Endothelial Growth Factor -634G>C SNP Is Associated With Proliferative Diabetic Retinopathy: A case-control study in a Brazilian population of European ancestry. <i>Diabetes Care</i> , 2007, 30, 275-279.	4.3	38
83	Molecular Analysis in Brazilian Cystic Fibrosis Patients Reveals Five Novel Mutations. <i>Genetic Testing and Molecular Biomarkers</i> , 2000, 4, 69-74.	1.7	37
84	A Brazilian cohort of individuals with Phelan-McDermid syndrome: genotype-phenotype correlation and identification of an atypical case. <i>Journal of Neurodevelopmental Disorders</i> , 2019, 11, 13.	1.5	37
85	Half the dystrophin gene is apparently enough for a mild clinical course: confirmation of its potential use for gene therapy. <i>Human Molecular Genetics</i> , 1994, 3, 919-922.	1.4	36
86	TCOF1 mutation database: Novel mutation in the alternatively spliced exon 6A and update in mutation nomenclature. <i>Human Mutation</i> , 2005, 25, 429-434.	1.1	36
87	High frequency of submicroscopic chromosomal imbalances in patients with syndromic craniosynostosis detected by a combined approach of microsatellite segregation analysis, multiplex ligation-dependent probe amplification and array-based comparative genome hybridisation. <i>Journal of Medical Genetics</i> , 2008, 45, 447-450.	1.5	36
88	Region 8q24 is a susceptibility locus for nonsyndromic oral clefting in Brazil. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 464-468.	1.6	36
89	Susceptibility to DNA Damage as a Molecular Mechanism for Non-Syndromic Cleft Lip and Palate. <i>PLoS ONE</i> , 2013, 8, e65677.	1.1	35
90	Whole-genome sequencing of 1,171 elderly admixed individuals from Brazil. <i>Nature Communications</i> , 2022, 13, 1004.	5.8	35

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91	CNS malformations in Knobloch syndrome with splice mutation in COL18A1 gene. American Journal of Medical Genetics, Part A, 2007, 143A, 1514-1518.	0.7	34
92	Linkage Analysis in a Large Brazilian Family with van der Woude Syndrome Suggests the Existence of a Susceptibility Locus for Cleft Palate at 17p11.2-11.1. American Journal of Human Genetics, 1999, 65, 433-440.	2.6	33
93	Parental origin of mutations in sporadic cases of Treacher Collins syndrome. European Journal of Human Genetics, 2003, 11, 718-722.	1.4	33
94	Human Stem Cell Cultures from Cleft Lip/Palate Patients Show Enrichment of Transcripts Involved in Extracellular Matrix Modeling By Comparison to Controls. Stem Cell Reviews and Reports, 2011, 7, 446-457.	5.6	33
95	Is the maintenance of the C-terminus domain of dystrophin enough to ensure a milder Becker muscular dystrophy phenotype?. Human Molecular Genetics, 1993, 2, 39-42.	1.4	32
96	Absence of correlation between utrophin localization and quantity and the clinical severity in Duchenne/Becker dystrophies. American Journal of Medical Genetics Part A, 1995, 58, 305-309.	2.4	32
97	An Xq22.3 duplication detected by comparative genomic hybridization microarray (Array-CGH) defines a new locus (FGS5) for FC syndrome. American Journal of Medical Genetics, Part A, 2005, 139A, 221-226.	0.7	32
98	IRF6 is a risk factor for nonsyndromic cleft lip in the Brazilian population. American Journal of Medical Genetics, Part A, 2012, 158A, 2170-2175.	0.7	32
99	Polymorphisms at Regions 1p22.1 (rs560426) and 8q24 (rs1530300) Are Risk Markers for Nonsyndromic Cleft Lip and/or Palate in the Brazilian Population. American Journal of Medical Genetics, Part A, 2013, 161, 1177-1180.	0.7	32
100	Absence of correlation between skewed X inactivation in blood and serum creatine-kinase levels in Duchenne/Becker female carriers. , 1998, 80, 356-361.		31
101	Genetic contribution for non-syndromic cleft lip with or without cleft palate (NS CL/P) in different regions of Brazil and implications for association studies. American Journal of Medical Genetics, Part A, 2011, 155, 1581-1587.	0.7	31
102	Collybistin binds and inhibits mTORC1 signaling: a potential novel mechanism contributing to intellectual disability and autism. European Journal of Human Genetics, 2016, 24, 59-65.	1.4	31
103	Validating GWAS Variants from Microglial Genes Implicated in Alzheimer's Disease. Journal of Molecular Neuroscience, 2017, 62, 215-221.	1.1	31
104	Evidence of genetic heterogeneity in the autosomal recessive adult forms of limb-girdle muscular dystrophy following linkage analysis with 15q probes in Brazilian families.. Journal of Medical Genetics, 1993, 30, 385-387.	1.5	30
105	Analysis of the serotonin transporter polymorphism (5-HTTLPR) in Brazilian patients affected by dysthymia, major depression and bipolar disorder. Molecular Psychiatry, 2000, 5, 348-349.	4.1	30
106	A functional SNP in the promoter region of TCOF1 is associated with reduced gene expression and YY1 DNA-protein interaction. Gene, 2005, 359, 44-52.	1.0	29
107	Auriculo-condylar syndrome: mapping of a first locus and evidence for genetic heterogeneity. European Journal of Human Genetics, 2008, 16, 145-152.	1.4	29
108	Actin cytoskeleton dynamics in stem cells from autistic individuals. Scientific Reports, 2018, 8, 11138.	1.6	29

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109	Point mutation in a Becker muscular dystrophy patient. <i>Human Molecular Genetics</i> , 1993, 2, 75-77.	1.4	28
110	Apert p.Ser252Trp Mutation in FGFR2 Alters Osteogenic Potential and Gene Expression of Cranial Periosteal Cells. <i>Molecular Medicine</i> , 2007, 13, 422-442.	1.9	28
111	Rare <i>RELN</i> variants affect Reelin-DAB1 signal transduction in autism spectrum disorder. <i>Human Mutation</i> , 2018, 39, 1372-1383.	1.1	28
112	Pfeiffer mutation in an apert patient: How wide is the spectrum of variability due to mutations in the FGFR2 gene?. <i>American Journal of Medical Genetics Part A</i> , 1997, 71, 243-245.	2.4	27
113	HTR1B and HTR2C in autism spectrum disorders in Brazilian families. <i>Brain Research</i> , 2009, 1250, 14-19.	1.1	27
114	Cnbp ameliorates Treacher Collins Syndrome craniofacial anomalies through a pathway that involves redox-responsive genes. <i>Cell Death and Disease</i> , 2016, 7, e2397-e2397.	2.7	27
115	Partial β -sarcoglycan deficiency with retention of the dystrophin-glycoprotein complex in a LGMD2D family. , 2000, 23, 984-988.		26
116	Prion disease resembling frontotemporal dementia and parkinsonism linked to chromosome 17. <i>Arquivos De Neuro-Psiquiatria</i> , 2001, 59, 161-164.	0.3	26
117	Effects of antipsychotics with different weight gain liabilities on human in vitro models of adipose tissue differentiation and metabolism. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , 2011, 35, 1884-1890.	2.5	26
118	Is bone transplantation the gold standard for repair of alveolar bone defects?. <i>Journal of Tissue Engineering</i> , 2014, 5, 204173141351935.	2.3	26
119	An 11q11-q13.3 duplication, including <i>FGF3</i> and <i>FGF4</i> genes, in a patient with syndromic multiple craniosynostoses. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1912-1918.	0.7	25
120	Schinzel-Giedion syndrome in two Brazilian patients: Report of a novel mutation in <i>SETBP1</i> and literature review of the clinical features. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1039-1046.	0.7	25
121	Posttranscriptional Interaction Between miR-450a-5p and miR-28-5p and STAT1 mRNA Triggers Osteoblastic Differentiation of Human Mesenchymal Stem Cells. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 4045-4062.	1.2	25
122	Importance of Zinc Transporter 8 Autoantibody in the Diagnosis of Type 1 Diabetes in Latin Americans. <i>Scientific Reports</i> , 2017, 7, 207.	1.6	25
123	Meta-Analyses Support Previous and Novel Autism Candidate Genes: Outcomes of an Unexplored Brazilian Cohort. <i>Autism Research</i> , 2020, 13, 199-206.	2.1	25
124	Severe nonspecific X-linked mental retardation caused by a proximally Xp located gene: Intragenic heterogeneity or a new form of X-linked mental retardation?. <i>American Journal of Medical Genetics Part A</i> , 1993, 46, 172-175.	2.4	24
125	Cosegregation of schizophrenia with Becker muscular dystrophy: susceptibility locus for schizophrenia at Xp21 or an effect of the dystrophin gene in the brain?. <i>Journal of Medical Genetics</i> , 1993, 30, 131-134.	1.5	24
126	Craniosynostosis in pycnodysostosis: Broadening the spectrum of the cranial flat bone abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2599-2603.	0.7	24

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127	Vertical transmission of a frontonasal phenotype caused by a novel <i>ALX4</i> mutation. American Journal of Medical Genetics, Part A, 2013, 161, 600-604.	0.7	24
128	Improvement of <i>In Vitro</i> Osteogenic Potential through Differentiation of Induced Pluripotent Stem Cells from Human Exfoliated Dental Tissue towards Mesenchymal-Like Stem Cells. Stem Cells International, 2015, 2015, 1-9.	1.2	24
129	Apert and Crouzon syndromes—Cognitive development, brain abnormalities, and molecular aspects. American Journal of Medical Genetics, Part A, 2016, 170, 1532-1537.	0.7	24
130	The recurrent <i>PPP1CB</i> mutation p.Pro49Arg in an additional Noonan-like syndrome individual: Broadening the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 824-828.	0.7	24
131	The influence of population stratification on genetic markers associated with type 1 diabetes. Scientific Reports, 2017, 7, 43513.	1.6	24
132	Impact of rare variants in <i>ARHGAP29</i> to the etiology of oral clefts: role of loss of function vs missense variants. Clinical Genetics, 2017, 91, 683-689.	1.0	24
133	No evidence of genetic heterogeneity in Brazilian facioscapulohumeral muscular dystrophy families (FSHD) with 4q markers. Human Molecular Genetics, 1993, 2, 557-562.	1.4	23
134	Deficiency of β -Actinin-3 (ACTN3) Occurs in Different Forms of Muscular Dystrophy. Neuropediatrics, 1997, 28, 223-228.	0.3	23
135	Evidence that <i>BCL3</i> plays a role in the etiology of nonsyndromic oral clefts in Brazilian families. Genetic Epidemiology, 2002, 23, 364-374.	0.6	23
136	Clinical evaluation and <i>COL2A1</i> gene analysis in 21 Brazilian families with Stickler syndrome: Identification of novel mutations, further genotype/phenotype correlation, and its implications for the diagnosis. European Journal of Medical Genetics, 2008, 51, 183-196.	0.7	23
137	Zebrafish <i>sp7</i> mutants show tooth cycling independent of attachment, eruption and poor differentiation of teeth. Developmental Biology, 2018, 435, 176-184.	0.9	23
138	Intrafamilial variability in dystrophin abundance correlated with difference in the severity of the phenotype. Journal of the Neurological Sciences, 1993, 119, 38-42.	0.3	22
139	Further evidence for the organisation of the four sarcoglycans proteins within the dystrophin-glycoprotein complex. European Journal of Human Genetics, 1999, 7, 251-254.	1.4	22
140	Genetics and Management of the Patient with Orofacial Cleft. Plastic Surgery International, 2012, 2012, 1-11.	0.7	22
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