Maria Rita Passos-Bueno

List of Publications by Year
in descending order

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257 papers	13,107 citations	36303 51 h-index	31849 101 g-index
272 all docs	272 docs citations	272 times ranked	15492 citing authors

#	Article	IF	Citations
1	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
2	Mutations in the proteolytic enzyme calpain 3 cause limb-girdle muscular dystrophy type 2A. Cell, 1995, 81, 27-40.	28.9	922
3	A gene related to Caenorhabditis elegans spermatogenesis factor fer-1 is mutated in limb-girdle muscular dystrophy type 2B. Nature Genetics, 1998, 20, 37-42.	21.4	626
4	Autosomal recessive limbgirdle muscular dystrophy, LGMD2F, is caused by a mutation in the Î´â€“sarcoglycan gene. Nature Genetics, 1996, 14, 195-198.	21.4	417
5	Limb-girdle muscular dystrophy type 2G is caused by mutations in the gene encoding the sarcomeric protein telethonin. Nature Genetics, 2000, 24, 163-166.	21.4	312
6	Clinical spectrum of fibroblast growth factor receptor mutations. Human Mutation, 1999, 14, 115-125.	2.5	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). Human Molecular Genetics, 2000, 9, 2051-2058.	2.9	259
8	RAB23 Mutations in Carpenter Syndrome Imply an Unexpected Role for Hedgehog Signaling in Cranial-Suture Development and Obesity. American Journal of Human Genetics, 2007, 80, 1162-1170.	6.2	229
9	Caveolin-3 in muscular dystrophy. Human Molecular Genetics, 1998, 7, 871-877.	2.9	200
10	Rare variants inSOS2andLZTR1are associated with Noonan syndrome. Journal of Medical Genetics, 2015, 52, 413-421.	3.2	187
11	Reconstruction of Large Cranial Defects in Nonimmunosuppressed Experimental Design With Human Dental Pulp Stem Cells. Journal of Craniofacial Surgery, 2008, 19, 204-210.	0.7	185
12	Exomic variants of an elderly cohort of Brazilians in the ABraOM database. Human Mutation, 2017, 38, 751-763.	2.5	181
13	Modeling non-syndromic autism and the impact of TRPC6 disruption in human neurons. Molecular Psychiatry, 2015, 20, 1350-1365.	7.9	175
14	The sarcoglycan complex in the six autosomal recessive limb-girdle muscular dystrophies. Human Molecular Genetics, 1996, 5, 1963-1969.	2.9	167
15	Stem cell proliferation under low intensity laser irradiation: A preliminary study. Lasers in Surgery and Medicine, 2008, 40, 433-438.	2.1	155
16	High serum endostatin levels in Down syndrome: implications for improved treatment and prevention of solid tumours. European Journal of Human Genetics, 2001, 9, 811-814.	2.8	145
17	Serum creatine-kinase (CK) and pyruvate-kinase (PK) activities in Duchenne (DMD) as compared with Becker (BMD) muscular dystrophy. Journal of the Neurological Sciences, 1991, 102, 190-196.	0.6	136
18	The Seventh Form of Autosomal Recessive Limb-Girdle Muscular Dystrophy Is Mapped to 17q11-12. American Journal of Human Genetics, 1997, 61, 151-159.	6.2	136

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19	Genetics of Craniosynostosis: Genes, Syndromes, Mutations and Genotype-Phenotype Correlations. Frontiers of Oral Biology, 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. American Journal of Human Genetics, 2002, 71, 1320-1329.	6.2	128
21	The facioscapulohumeral muscular dystrophy (FSHD1) gene affects males more severely and more frequently than females. American Journal of Medical Genetics Part A, 1998, 77, 155-161.	2.4	123
22	Asymptomatic carriers and gender differences in facioscapulohumeral muscular dystrophy (FSHD). Neuromuscular Disorders, 2004, 14, 33-38.	0.6	114
23	Fat Grafts Supplemented with Adipose-Derived Stromal Cells in the Rehabilitation of Patients with Craniofacial Microsomia. Plastic and Reconstructive Surgery, 2013, 132, 141-152.	1.4	114
24	High mutation detection rate inTCOF1 among Treacher Collins syndrome patients reveals clustering of mutations and 16 novel pathogenic changes. Human Mutation, 2000, 16, 315-322.	2.5	112
25	Genomic screening for beta-sarcoglycan gene mutations: missense mutations may cause severe limb-girdle muscular dystrophy type 2E (LGMD 2E). Human Molecular Genetics, 1996, 5, 1953-1961.	2.9	111
26	Familial spongiform encephalopathy associated with a novel prion protein gene mutation. Annals of Neurology, 1997, 42, 138-146.	5.3	110
27	Whole-genome array-CGH screening in undiagnosed syndromic patients: old syndromes revisited and new alterations. Cytogenetic and Genome Research, 2006, 115, 254-261.	1.1	103
28	A Noncoding Expansion in EIF4A3 Causes Richieri-Costa-Pereira Syndrome, a Craniofacial Disorder Associated with Limb Defects. American Journal of Human Genetics, 2014, 94, 120-128.	6.2	99
29	Limb-girdle muscular dystrophy: one gene with different phenotypes, one phenotype with different genes. Current Opinion in Neurology, 2000, 13, 511-517.	3.6	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. Human Molecular Genetics, 1996, 5, 815-820.	2.9	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. Nature Communications, 2018, 9, 475.	12.8	86
33	Syndromes of the first and second pharyngeal arches: A review. American Journal of Medical Genetics, Part A, 2009, 149A, 1853-1859.	1.2	85
34	A review of craniofacial disorders caused by spliceosomal defects. Clinical Genetics, 2015, 88, 405-415.	2.0	85
35	Clinical variability in calpainopathy: What makes the difference?. European Journal of Human Genetics, 2002, 10, 825-832.	2.8	84
36	Sarcoglycanopathies are responsible for 68% of severe autosomal recessive limb-girdle muscular dystrophy in the Brazilian population. Journal of the Neurological Sciences, 1999, 164, 44-49.	0.6	81

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37	Heterozygous Mutations of FREM1 Are Associated with an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. PLoS Genetics, 2011, 7, e1002278.	3.5	80
38	Analysis of the CTG repeat in skeletal muscle of young and adult myotonic dystrophy patients: when does the expansion occur?. Human Molecular Genetics, 1995, 4, 401-406.	2.9	79
39	A gene which causes severe ocular alterations and occipital encephalocele (Knobloch syndrome) is mapped to 21q22.3. Human Molecular Genetics, 1996, 5, 843-847.	2.9	76
40	New Source of Muscle-Derived Stem Cells with Potential for Alveolar Bone Reconstruction in Cleft Lip and/or Palate Patients. Tissue Engineering - Part A, 2009, 15, 427-435.	3.1	71
41	Gastric Bypass and Sleeve Gastrectomy: the Same Impact on IL-6 and TNF-Î±. Prospective Clinical Trial. Obesity Surgery, 2013, 23, 1252-1261.	2.1	69
42	Dysferlin Protein Analysis in Limb-Girdle Muscular Dystrophies. Journal of Molecular Neuroscience, 2001, 17, 71-80.	2.3	67
43	Mesenchymal Stem Cells Derived From Canine Umbilical Cord Veinâ€"A Novel Source for Cell Therapy Studies. Stem Cells and Development, 2010, 19, 395-402.	2.1	67
44	Apparent association of mental retardation and specific patterns of deletions screened with probes cf56a and cf23a in Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 1991, 39, 437-441.	2.4	66
45	Cellular interference in craniofrontonasal syndrome: males mosaic for mutations in the X-linked EFNB1 gene are more severely affected than true hemizygotes. Human Molecular Genetics, 2013, 22, 1654-1662.	2.9	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. Molecular Psychiatry, 1998, 3, 438-441.	7.9	65
47	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	2.4	63
48	A new form of autosomal dominant limb-girdle muscular dystrophy (LGMD1G) with progressive fingers and toes flexion limitation maps to chromosome 4p21. European Journal of Human Genetics, 2004, 12, 1033-1040.	2.8	61
49	A Novel Autosomal Recessive GJA1 Missense Mutation Linked to Craniometaphyseal Dysplasia. PLoS ONE, 2013, 8, e73576.	2.5	61
50	Decreased cellular uptake and metabolism in Allan-Herndon-Dudley syndrome (AHDS) due to a novel mutation in the MCT8 thyroid hormone transporter. Journal of Medical Genetics, 2005, 43, 457-460.	3.2	59
51	Differential methylation is associated with non-syndromic cleft lip and palate and contributes to penetrance effects. Scientific Reports, 2017, 7, 2441.	3.3	59
52	Screening of TCOF1 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. Journal of Medical Genetics, 2002, 39, 493-495.	3.2	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. Journal of Medical Genetics, 2008, 45, 539-543.	3.2	53
54	Further evidence of the importance of RIT1 in Noonan syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2952-2957.	1.2	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. Journal of the Neurological Sciences, 1991, 101, 141-147.	0.6	52
56	Genetic heterogeneity for Duchenne-like muscular dystrophy (DLMD) based on linkage and 50 DAG analysis. Human Molecular Genetics, 1993, 2, 1945-1947.	2.9	50
57	Mutations in collagen 18A1 (COL18A1) and their relevance to the human phenotype. Anais Da Academia Brasileira De Ciencias, 2006, 78, 123-131.	0.8	50
58	Telethonin protein expression in neuromuscular disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 33-40.	3.8	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. European Journal of Human Genetics, 2004, 12, 521-526.	2.8	49
60	Deficiency of Merosin (Laminin M or Î±2) in Congenital Muscular Dystrophy Associated with Cerebral White Matter Alterations. Neuropediatrics, 1995, 26, 293-297.	0.6	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.. Journal of Medical Genetics, 1998, 35, 951-953.	3.2	48
62	Molecular screening for microdeletions at 9p22â€p24 and 11q23â€q24 in a large cohort of patients with trigonocephaly. Clinical Genetics, 2005, 67, 503-510.	2.0	48
63	Understanding the basis of auriculocondylar syndrome: Insights from human, mouse and zebrafish genetic studies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 306-317.	1.6	48
64	Deletion of the Basement Membrane Heparan Sulfate Proteoglycan Type XVIII Collagen Causes Hypertriglyceridemia in Mice and Humans. PLoS ONE, 2010, 5, e13919.	2.5	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.. Journal of Medical Genetics, 1996, 33, 97-102.	3.2	45
66	Rare Variants in the Epithelial Cadherin Gene Underlying the Genetic Etiology of Nonsyndromic Cleft Lip with or without Cleft Palate. Human Mutation, 2015, 36, 1029-1033.	2.5	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. European Journal of Medical Genetics, 2011, 54, e425-e432.	1.3	44
68	Genetics and genomics in Brazil: a promising future. Molecular Genetics & Genomic Medicine, 2014, 2, 280-291.	1.2	44
69	Transcriptome of iPSC-derived neuronal cells reveals a module of co-expressed genes consistently associated with autism spectrum disorder. Molecular Psychiatry, 2021, 26, 1589-1605.	7.9	44
70	Estimate of the proportion of Duchenne muscular dystrophy with autosomal recessive inheritance. American Journal of Medical Genetics Part A, 1989, 32, 407-410.	2.4	42
71	The phenotype of chromosome 2p-linked limb-girdle muscular dystrophy. Neuromuscular Disorders, 1996, 6, 483-490.	0.6	42
72	EIF4A3 deficient human iPSCs and mouse models demonstrate neural crest defects that underlie Richieri-Costa-Pereira syndrome. Human Molecular Genetics, 2017, 26, 2177-2191.	2.9	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. Genomics, 1995, 27, 192-195.	2.9	41
74	Absence of calpain 3 in a form of limb-girdle muscular dystrophy (LGMD2A). Journal of the Neurological Sciences, 1997, 146, 173-178.	0.6	41
75	Description of a new mutation and characterization ofFGFR1, FGFR2, andFGFR3 mutations among Brazilian patients with syndromic craniosynostoses. American Journal of Medical Genetics Part A, 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. American Journal of Medical Genetics Part A, 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. Stem Cells International, 2012, 2012, 1-7.	2.5	40
78	Screening of deletions in the dystrophin gene with the cDNA probes Cf23a, Cf56a, and Cf115.. Journal of Medical Genetics, 1990, 27, 145-150.	3.2	39
79	Mutations in PCYT1A Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 94, 113-119.	6.2	39
80	Dystrophin immunostaining in muscles from patients with different types of muscular dystrophy: a Brazilian study. Journal of the Neurological Sciences, 1990, 98, 221-233.	0.6	38
81	A deletion including the brain promoter of the Duchenne muscular dystrophy gene is not associated with mental retardation. Neuromuscular Disorders, 1992, 2, 117-120.	0.6	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. Diabetes Care, 2007, 30, 275-279.	8.6	38
83	Molecular Analysis in Brazilian Cystic Fibrosis Patients Reveals Five Novel Mutations. Genetic Testing and Molecular Biomarkers, 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. Journal of Neurodevelopmental Disorders, 2019, 11, 13.	3.1	37
85	Half the dystrophin gene is apparently enough for a mild clinical course: confirmation of its potential use for gene therapy. Human Molecular Genetics, 1994, 3, 919-922.	2.9	36
86	TCOF1 mutation database: Novel mutation in the alternatively spliced exon 6A and update in mutation nomenclature. Human Mutation, 2005, 25, 429-434.	2.5	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. Journal of Medical Genetics, 2008, 45, 447-450.	3.2	36
88	Region 8q24 is a susceptibility locus for nonsyndromic oral clefting in Brazil. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 464-468.	1.6	36
89	Susceptibility to DNA Damage as a Molecular Mechanism for Non-Syndromic Cleft Lip and Palate. PLoS ONE, 2013, 8, e65677.	2.5	35
90	Whole-genome sequencing of 1,171 elderly admixed individuals from Brazil. Nature Communications, 2022, 13, 1004.	12.8	35

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91	CNS malformations in Knobloch syndrome with splice mutation inCOL18A1 gene. American Journal of Medical Genetics, Part A, 2007, 143A, 1514-1518.	1.2	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.	6.2	33
93	Parental origin of mutations in sporadic cases of Treacher Collins syndrome. European Journal of Human Genetics, 2003, 11, 718-722.	2.8	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 maintainance of the C-terminus domain of dystrophin enough to ensure a milder Becker muscular dystrophy phenotype?. Human Molecular Genetics, 1993, 2, 39-42.	2.9	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 FG syndrome. American Journal of Medical Genetics, Part A, 2005, 139A, 221-226.	1.2	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.	1.2	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.	1.2	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 nonâ€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.	1.2	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.	2.8	31
103	Validating GWAS Variants from Microglial Genes Implicated in Alzheimerâ€™s Disease. Journal of Molecular Neuroscience, 2017, 62, 215-221.	2.3	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.	3.2	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.	7.9	30
106	A functional SNP in the promoter region of TCOF1 is associated with reduced gene expression and YY1 DNAâ€“protein interaction. Gene, 2005, 359, 44-52.	2.2	29
107	Auriculo-condylar syndrome: mapping of a first locus and evidence for genetic heterogeneity. European Journal of Human Genetics, 2008, 16, 145-152.	2.8	29
108	Actin cytoskeleton dynamics in stem cells from autistic individuals. Scientific Reports, 2018, 8, 11138.	3.3	29

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109	Point mutation in a Becker muscular dystrophy patient. Human Molecular Genetics, 1993, 2, 75-77.	2.9	28
110	Apert p.Ser252Trp Mutation in FGFR2 Alters Osteogenic Potential and Gene Expression of Cranial Periosteal Cells. Molecular Medicine, 2007, 13, 422-442.	4.4	28
111	RareRELNvariants affect Reelin-DAB1 signal transduction in autism spectrum disorder. Human Mutation, 2018, 39, 1372-1383.	2.5	28
112	Pfeiffer mutation in an apert patient: How wide is the spectrum of variability due to mutations in the FGFR2 gene?. American Journal of Medical Genetics Part A, 1997, 71, 243-245.	2.4	27
113	HTR1B and HTR2C in autism spectrum disorders in Brazilian families. Brain Research, 2009, 1250, 14-19.	2.2	27
114	Cnbp ameliorates Treacher Collins Syndrome craniofacial anomalies through a pathway that involves redox-responsive genes. Cell Death and Disease, 2016, 7, e2397-e2397.	6.3	27
115	Partial ?-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. Arquivos De Neuro-Psiquiatria, 2001, 59, 161-164.	0.8	26
117	Effects of antipsychotics with different weight gain liabilities on human in vitro models of adipose tissue differentiation and metabolism. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2011, 35, 1884-1890.	4.8	26
118	Is bone transplantation the gold standard for repair of alveolar bone defects?. Journal of Tissue Engineering, 2014, 5, 204173141351935.	5.5	26
119	An 11q11â€“q13.3 duplication, including FGF3 and FGF4 genes, in a patient with syndromic multiple craniosynostoses. American Journal of Medical Genetics, Part A, 2007, 143A, 1912-1918.	1.2	25
120	Schinzelâ€“Giedion syndrome in two Brazilian patients: Report of a novel mutation in SETBP1 and literature review of the clinical features. American Journal of Medical Genetics, Part A, 2015, 167, 1039-1046.	1.2	25
121	Posttranscriptional Interaction Between miRâ€450aâ€5p and miRâ€28â€5p and STAT1 mRNA Triggers Osteoblastic Differentiation of Human Mesenchymal Stem Cells. Journal of Cellular Biochemistry, 2017, 118, 4045-4062.	2.6	25
122	Importance of Zinc Transporter 8 Autoantibody in the Diagnosis of Type 1 Diabetes in Latin Americans. Scientific Reports, 2017, 7, 207.	3.3	25
123	Metaâ€Analyses Support Previous and Novel Autism Candidate Genes: Outcomes of an Unexplored Brazilian Cohort. Autism Research, 2020, 13, 199-206.	3.8	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?. American Journal of Medical Genetics Part A, 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?. Journal of Medical Genetics, 1993, 30, 131-134.	3.2	24
126	Craniosynostosis in pycnodysostosis: Broadening the spectrum of the cranial flat bone abnormalities. American Journal of Medical Genetics, Part A, 2010, 152A, 2599-2603.	1.2	24

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127	Vertical transmission of a frontonasal phenotype caused by a novel ALX4 mutation. American Journal of Medical Genetics, Part A, 2013, 161, 600-604.	1.2	24
128	Improvement ofIn VitroOsteogenic 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.	2.5	24
129	Apert and Crouzon syndromesâ€"Cognitive development, brain abnormalities, and molecular aspects. American Journal of Medical Genetics, Part A, 2016, 170, 1532-1537.	1.2	24
130	The recurrent PPP1CB mutation p.Pro49Arg in an additional Noonanâ€like syndrome individual: Broadening the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 824-828.	1.2	24
131	The influence of population stratification on genetic markers associated with type 1 diabetes. Scientific Reports, 2017, 7, 43513.	3.3	24
132	Impact of rare variants in <scp>ARHGAP29</scp> to the etiology of oral clefts: role of lossâ€ofâ€function vs missense variants. Clinical Genetics, 2017, 91, 683-689.	2.0	24
133	No evidence of genetic heterogeneity in Brazilian facioscapulohumeral muscular dystrophy familes (FSHD) with 4q markers. Human Molecular Genetics, 1993, 2, 557-562.	2.9	23
134	Deficiency of Î±-Actinin-3 (ACTN3) Occurs in Different Forms of Muscular Dystrophy. Neuropediatrics, 1997, 28, 223-228.	0.6	23
135	Evidence thatBCL3plays a role in the etiology of nonsyndromic oral clefts in Brazilian families. Genetic Epidemiology, 2002, 23, 364-374.	1.3	23
136	Clinical evaluation and COL2A1 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.	1.3	23
137	Zebrafish sp7 mutants show tooth cycling independent of attachment, eruption and poor differentiation of teeth. Developmental Biology, 2018, 435, 176-184.	2.0	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.	0.6	22
139	Further evidence for the organisation of the four sarcoglycans proteins within the dystrophinâ€"glycoprotein complex. European Journal of Human Genetics, 1999, 7, 251-254.	2.8	22
140	Genetics and Management of the Patient with Orofacial Cleft. Plastic Surgery International, 2012, 2012, 1-11.	0.7	22
141	Novel mutations inIRF6in nonsyndromic cleft lip with or without cleft palate: When shouldIRF6mutational screening be done?. American Journal of Medical Genetics, Part A, 2009, 149A, 1319-1322.	1.2	21
142	Reduced transcription of TCOF1 in adult cells of Treacher Collins syndrome patients. BMC Medical Genetics, 2009, 10, 136.	2.1	21
143	Novel variants in GNAI3 associated with auriculocondylar syndrome strengthen a common dominant negative effect. European Journal of Human Genetics, 2015, 23, 481-485.	2.8	21
144	COL18A1 is highly expressed during human adipocyte differentiation and the SNP c.1136C > T in its "frizzled" motif is associated with obesity in diabetes type 2 patients. Anais Da Academia Brasileira De Ciencias, 2008, 80, 167-177.	0.8	21

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145	Dystrophin immunofluorescence pattern in manifesting and asymptomatic carriers of Duchenne's and Becker muscular dystrophies of different ages. Neuromuscular Disorders, 1991, 1, 177-183.	0.6	20
146	Becker and limb-girdle muscular dystrophies: A psychiatric and intellectual level comparative study. American Journal of Medical Genetics Part A, 1995, 60, 33-38.	2.4	19
147	Does the P172H mutation at theTM4SF2gene cause X-linked mental retardation?. , 2004, 124A, 413-415.		19
148	Novel point mutations in the dystrophin gene. Human Mutation, 1997, 10, 217-222.	2.5	18
149	CD105 is regulated by hsa-miR-1287 and its expression is inversely correlated with osteopotential in SHED. Bone, 2018, 106, 112-120.	2.9	18
150	Assessment of the 50-kDa dystrophin-associated glycoprotein in Brazilian patients with severe childhood autosomal recessive muscular dystrophy. Journal of the Neurological Sciences, 1994, 123, 122-128.	0.6	17
151	TCOF1 mutations excluded from a role in other first and second branchial arch-related disorders. American Journal of Medical Genetics Part A, 2002, 111, 324-327.	2.4	17
152	Transforming Growth Factor-Î± and Nonsyndromic Cleft Lip with or without Palate in Brazilian Patients: Results of a Large Case-Control Study. Cleft Palate-Craniofacial Journal, 2004, 41, 387-391.	0.9	17
153	Functionally conserved cis-regulatory elements of COL18A1 identified through zebrafish transgenesis. Developmental Biology, 2010, 337, 496-505.	2.0	17
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155	Altered mTORC1 signaling in multipotent stem cells from nearly 25% of patients with nonsyndromic autism spectrum disorders. Molecular Psychiatry, 2015, 20, 551-552.	7.9	17
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