Maria Rita Passos-Bueno

List of Publications by Year
in descending order

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257 papers	13,107 citations	36203 51 h-index	31759 101 g-index
272 all docs	272 docs citations	272 times ranked	15492 citing authors

#	Article	IF	Citations
1	Copy number variations in a Brazilian cohort with autism spectrum disorders highlight the contribution of cell adhesion genes. Clinical Genetics, 2022, 101, 134-141.	1.0	13
2	New locus underlying auriculocondylar syndrome (ARCND): 430 kb duplication involvingTWIST1regulatory elements. Journal of Medical Genetics, 2022, 59, 895-905.	1.5	4
3	<scp>FMR1</scp> premutation in children with autism spectrum disorders: Should additional diagnostic tests be performed?. American Journal of Medical Genetics, Part A, 2022, 188, 1334-1337.	0.7	0
4	Recurrence of COVID-19 associated with reduced T-cell responses in a monozygotic twin pair. Open Biology, 2022, 12, 210240.	1.5	5
5	Interleukin-17a Induces Neuronal Differentiation of Induced-Pluripotent Stem Cell-Derived Neural Progenitors From Autistic and Control Subjects. Frontiers in Neuroscience, 2022, 16, 828646.	1.4	5
6	Dystrophin genetic variants and autism. Discover Mental Health, 2022, 2, 1.	1.0	0
7	Whole-genome sequencing of 1,171 elderly admixed individuals from Brazil. Nature Communications, 2022, 13, 1004.	5.8	35
8	Mutations in trpÎ³, the homologue of TRPC6 autism candidate gene, causes autism-like behavioral deficits in Drosophila. Molecular Psychiatry, 2022, 27, 3328-3342.	4.1	6
9	Rare CACNA1H and RELN variants interact through mTORC1 pathway in oligogenic autism spectrum disorder. Translational Psychiatry, 2022, 12, .	2.4	3
10	Reply to Lombardo, 2020: An additional route of investigation: what are the mechanisms controlling ribosomal protein genes dysregulation in autistic neuronal cells?. Molecular Psychiatry, 2021, 26, 1436-1437.	4.1	2
11	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.	4.1	44
12	Extreme phenotypes approach to investigate host genetics and COVID-19 outcomes. Genetics and Molecular Biology, 2021, 44, e20200302.	0.6	6
13	Admixture/fine-mapping in Brazilians reveals a West African associated potential regulatory variant (rs114066381) with a strong female-specific effect on body mass and fat mass indexes. International Journal of Obesity, 2021, 45, 1017-1029.	1.6	4
14	TCF7L2 rs7903146 polymorphism association with diabetes and obesity in an elderly cohort from Brazil. PeerJ, 2021, 9, e11349.	0.9	5
15	Biased pathogenic assertions of loss of function variants challenge molecular diagnosis of admixed individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 357-363.	0.7	4
16	Complement C4 Is Reduced in iPSC-Derived Astrocytes of Autism Spectrum Disorder Subjects. International Journal of Molecular Sciences, 2021, 22, 7579.	1.8	8
17	Congenital limb deficiency: Genetic investigation of 44 individuals presenting mainly longitudinal defects in isolated or syndromic forms. Clinical Genetics, 2021, 100, 615-623.	1.0	4
18	Neuroprogenitor Cells From Patients With TBCK Encephalopathy Suggest Deregulation of Early Secretory Vesicle Transport. Frontiers in Cellular Neuroscience, 2021, 15, 803302.	1.8	2

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19	Metaâ€Analyses Support Previous and Novel Autism Candidate Genes: Outcomes of an Unexplored Brazilian Cohort. Autism Research, 2020, 13, 199-206.	2.1	25
20	Recapitulation of Neural Crest Specification and EMT via Induction from Neural Plate Border-like Cells. Stem Cell Reports, 2020, 15, 776-788.	2.3	11
21	Human levator veli palatini muscle: a novel source of mesenchymal stromal cells for use in the rehabilitation of patients with congenital craniofacial malformations. Stem Cell Research and Therapy, 2020, 11, 501.	2.4	3
22	Phenotypeâ€“genotype analysis of 242 individuals with <scp>RASopathies</scp>: 18â€year experience of a tertiary center in Brazil. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 896-911.	0.7	10
23	Structural variation of the malaria-associated human glycophorin A-B-E region. BMC Genomics, 2020, 21, 446.	1.2	7
24	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
25	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.	1.5	37
26	A fast degrading PLLA composite with a high content of functionalized octacalcium phosphate mineral phase induces stem cells differentiation. Journal of the Mechanical Behavior of Biomedical Materials, 2019, 93, 93-104.	1.5	15
27	Zebrafish sp7 mutants show tooth cycling independent of attachment, eruption and poor differentiation of teeth. Developmental Biology, 2018, 435, 176-184.	0.9	23
28	Discordant congenital Zika syndrome twins show differential in vitro viral susceptibility of neural progenitor cells. Nature Communications, 2018, 9, 475.	5.8	86
29	Genetics of Cleft Lip and Cleft Palate: Perspectives in Surgery Management and Outcome. , 2018, , 25-35.		3
30	CD105 is regulated by hsa-miR-1287 and its expression is inversely correlated with osteopotential in SHED. Bone, 2018, 106, 112-120.	1.4	18
31	MRPL53, a New Candidate Gene for Orofacial Clefting, Identified Using an eQTL Approach. Journal of Dental Research, 2018, 97, 33-40.	2.5	8
32	Richieriâ€Costaâ€Pereira syndrome: Expanding its phenotypic and genotypic spectrum. Clinical Genetics, 2018, 93, 800-811.	1.0	15
33	RareRELNvariants affect Reelin-DAB1 signal transduction in autism spectrum disorder. Human Mutation, 2018, 39, 1372-1383.	1.1	28
34	Development of a comprehensive noninvasive prenatal test. Genetics and Molecular Biology, 2018, 41, 545-554.	0.6	8
35	Actin cytoskeleton dynamics in stem cells from autistic individuals. Scientific Reports, 2018, 8, 11138.	1.6	29
36	Complexity of the 5â€² Untranslated Region of EIF4A3, a Critical Factor for Craniofacial and Neural Development. Frontiers in Genetics, 2018, 9, 149.	1.1	6

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37	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.	0.7	24
38	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.	1.2	25
39	EIF4A3 deficient human iPSCs and mouse models demonstrate neural crest defects that underlie Richieri-Costa-Pereira syndrome. Human Molecular Genetics, 2017, 26, 2177-2191.	1.4	42
40	Validating GWAS Variants from Microglial Genes Implicated in Alzheimerâ€™s Disease. Journal of Molecular Neuroscience, 2017, 62, 215-221.	1.1	31
41	Differential methylation is associated with non-syndromic cleft lip and palate and contributes to penetrance effects. Scientific Reports, 2017, 7, 2441.	1.6	59
42	Targeted molecular investigation in patients within the clinical spectrum of Auriculocondylar syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 938-945.	0.7	11
43	Exomic variants of an elderly cohort of Brazilians in the ABraOM database. Human Mutation, 2017, 38, 751-763.	1.1	181
44	Importance of Zinc Transporter 8 Autoantibody in the Diagnosis of Type 1 Diabetes in Latin Americans. Scientific Reports, 2017, 7, 207.	1.6	25
45	The influence of population stratification on genetic markers associated with type 1 diabetes. Scientific Reports, 2017, 7, 43513.	1.6	24
46	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.	1.0	24
47	Integrative Variation Analysis Reveals that a Complex Genotype May Specify Phenotype in Siblings with Syndromic Autism Spectrum Disorder. PLoS ONE, 2017, 12, e0170386.	1.1	2
48	Neuromuscular disorders: genes, genetic counseling and therapeutic trials. Genetics and Molecular Biology, 2016, 39, 339-348.	0.6	16
49	Apert and Crouzon syndromesâ€”Cognitive development, brain abnormalities, and molecular aspects. American Journal of Medical Genetics, Part A, 2016, 170, 1532-1537.	0.7	24
50	Cnbp ameliorates Treacher Collins Syndrome craniofacial anomalies through a pathway that involves redox-responsive genes. Cell Death and Disease, 2016, 7, e2397-e2397.	2.7	27
51	Cell Type-Dependent Nonspecific Fibroblast Growth Factor Signaling in Apert Syndrome. Stem Cells and Development, 2016, 25, 1249-1260.	1.1	3
52	Craniosynostosis in 10q26 deletion patients: A consequence of brain underdevelopment or altered suture biology?. American Journal of Medical Genetics, Part A, 2016, 170, 403-409.	0.7	8
53	Detection of small copy number variations (CNVs) in autism spectrum disorder (ASD) by custom array comparative genomic hybridization (aCGH). Research in Autism Spectrum Disorders, 2016, 23, 145-151.	0.8	7
54	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

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55	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.	1.1	45
56	Recurrence of frontometaphyseal dysplasia in two sisters with a mutation in FLNA and an atypical paternal phenotype: Insights into genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2015, 167, 1161-1164.	0.7	2
57	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.	1.2	24
58	Novel variants in GNAI3 associated with auriculocondylar syndrome strengthen a common dominant negative effect. European Journal of Human Genetics, 2015, 23, 481-485.	1.4	21
59	Stem Cells to Understand the Pathophysiology of Autism Spectrum Disorders. Pancreatic Islet Biology, 2015, , 121-142.	0.1	0
60	Altered mTORC1 signaling in multipotent stem cells from nearly 25% of patients with nonsyndromic autism spectrum disorders. Molecular Psychiatry, 2015, 20, 551-552.	4.1	17
61	Intragenic Deletion in the <i>LIFR</i> Gene in a Long-Term Survivor with StÃ¼ve-Wiedemann Syndrome. Molecular Syndromology, 2015, 6, 87-90.	0.3	6
62	A review of craniofacial disorders caused by spliceosomal defects. Clinical Genetics, 2015, 88, 405-415.	1.0	85
63	Increased In Vitro Osteopotential in SHED Associated with Higher IGF2 Expression When Compared with hASCs. Stem Cell Reviews and Reports, 2015, 11, 635-644.	5.6	14
64	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.	0.7	25
65	Rare variants inSOS2andLZTR1are associated with Noonan syndrome. Journal of Medical Genetics, 2015, 52, 413-421.	1.5	187
66	Modeling non-syndromic autism and the impact of TRPC6 disruption in human neurons. Molecular Psychiatry, 2015, 20, 1350-1365.	4.1	175
67	Investigation of 15q11-q13, 16p11.2 and 22q13 CNVs in Autism Spectrum Disorder Brazilian Individuals with and without Epilepsy. PLoS ONE, 2014, 9, e107705.	1.1	17
68	Genetics and genomics in Brazil: a promising future. Molecular Genetics & Genomic Medicine, 2014, 2, 280-291.	0.6	44
69	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	1.1	63
70	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.	2.6	99
71	MTHFR rs2274976 polymorphism is a risk marker for nonsyndromic cleft lip with or without cleft palate in the Brazilian population. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 30-35.	1.6	16
72	Further evidence of the importance of RIT1 in Noonan syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2952-2957.	0.7	53

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73	Mutations in PCYT1A Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 94, 113-119.	2.6	39
74	Is bone transplantation the gold standard for repair of alveolar bone defects?. Journal of Tissue Engineering, 2014, 5, 204173141351935.	2.3	26
75	Challenges in the Orthodontic Treatment of a Patient with Pycnodysostosis. Cleft Palate-Craniofacial Journal, 2014, 51, 735-739.	0.5	15
76	Gastric Bypass and Sleeve Gastrectomy: the Same Impact on IL-6 and TNF-Î±. Prospective Clinical Trial. Obesity Surgery, 2013, 23, 1252-1261.	1.1	69
77	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
78	A microduplication of 5p15.33 reveals CLPTM1L as a candidate gene for cleft lip and palate. European Journal of Medical Genetics, 2013, 56, 222-225.	0.7	11
79	Vertical transmission of a frontonasal phenotype caused by a novel ALX4 mutation. American Journal of Medical Genetics, Part A, 2013, 161, 600-604.	0.7	24
80	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.	0.7	48
81	Susceptibility to DNA Damage as a Molecular Mechanism for Non-Syndromic Cleft Lip and Palate. PLoS ONE, 2013, 8, e65677.	1.1	35
82	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.	1.4	66
83	Stem Cells as a Good Tool to Investigate Dysregulated Biological Systems in Autism Spectrum Disorders. Autism Research, 2013, 6, 354-361.	2.1	12
84	Fat Grafts Supplemented with Adipose-Derived Stromal Cells in the Rehabilitation of Patients with Craniofacial Microsomia. Plastic and Reconstructive Surgery, 2013, 132, 141-152.	0.7	114
85	Contribution of polymorphisms in genes associated with craniofacial development to the risk of nonsyndromic cleft lip and/or palate in the Brazilian population. Medicina Oral, Patologia Oral Y Cirugia Bucal, 2013, 18, e414-e420.	0.7	15
86	Novel Molecular Pathways Elicited by Mutant FGFR2 May Account for Brain Abnormalities in Apert Syndrome. PLoS ONE, 2013, 8, e60439.	1.1	12
87	A Novel Autosomal Recessive GJA1 Missense Mutation Linked to Craniometaphyseal Dysplasia. PLoS ONE, 2013, 8, e73576.	1.1	61
88	Genetics and Management of the Patient with Orofacial Cleft. Plastic Surgery International, 2012, 2012, 1-11.	0.7	22
89	Optimization of Parameters for a More Efficient Use of Adipose-Derived Stem Cells in Regenerative Medicine Therapies. Stem Cells International, 2012, 2012, 1-7.	1.2	40
90	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

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91	FGFR2 Mutation Confers a Less Drastic Gain of Function in Mesenchymal Stem Cells Than in Fibroblasts. Stem Cell Reviews and Reports, 2012, 8, 685-695.	5.6	11
92	Saethreâ€“Chotzen phenotype with learning disability and hyper IgE phenotype in a patient due to complex chromosomal rearrangement involving chromosomes 3 and 7. American Journal of Medical Genetics, Part A, 2012, 158A, 1680-1685.	0.7	9
93	A complex chromosomal rearrangement involving chromosomes 2, 5, and X in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 529-536.	1.1	10
94	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
95	Auriculoâ€condylar syndrome. Confronting a diagnostic challenge. American Journal of Medical Genetics, Part A, 2012, 158A, 59-65.	0.7	16
96	Efeitos de diferentes pressÃµes de aspiraÃ§Ã£o do tecido adiposo na obtenÃ§Ã£o de cÃ©lulas-tronco mesenquimais. Revista Brasileira De Cirurgia Plastica, 2012, 27, 509-513.	0.0	14
97	Centro de Estudos do Genoma Humano:. Revista Neurociencias, 2012, 20, 194-199.	0.0	1
98	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.	0.7	44
99	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.	2.5	26
100	Histological and radiological changes in cranial bone in the presence of bone wax. Acta Cirurgica Brasileira, 2011, 26, 274-278.	0.3	6
101	Heterozygous Mutations of FREM1 Are Associated with an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. PLoS Genetics, 2011, 7, e1002278.	1.5	80
102	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
103	Craniometaphyseal dysplasia with severe craniofacial involvement shows homozygosity at 6q21â€22.1 locus. American Journal of Medical Genetics, Part A, 2011, 155, 1106-1108.	0.7	8
104	The Richieriâ€Costa and Pereira syndrome: Report of two Brazilian siblings and review of literature. American Journal of Medical Genetics, Part A, 2011, 155, 1173-1177.	0.7	11
105	Obesity in pycnodysostosis due to UPD1: Possible effect of an imprinted gene on chromosome 1. American Journal of Medical Genetics, Part A, 2011, 155, 1483-1486.	0.7	5
106	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.	0.7	31
107	Saethre-Chotzen Syndrome, Pro136His TWIST Mutation, Hearing Loss, and External and Middle Ear Structural Anomalies: Report on a Brazilian Family. Cleft Palate-Craniofacial Journal, 2010, 47, 548-552.	0.5	7
108	Collybistin and gephyrin are novel components of the eukaryotic translation initiation factor 3 complex. BMC Research Notes, 2010, 3, 242.	0.6	9

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109	SOX17 Mutations Implicated in Urinary Tract Abnormalities. Human Mutation, 2010, 31, V-V.	1.1	1
110	Craniosynostosis in pycnodysostosis: Broadening the spectrum of the cranial flat bone abnormalities. American Journal of Medical Genetics, Part A, 2010, 152A, 2599-2603.	0.7	24
111	Deletion of the Basement Membrane Heparan Sulfate Proteoglycan Type XVIII Collagen Causes Hypertriglyceridemia in Mice and Humans. PLoS ONE, 2010, 5, e13919.	1.1	46
112	Effects of uterine cervix constriction on Wistar rats. Acta Cirurgica Brasileira, 2010, 25, 469-474.	0.3	1
113	An experimental model for the study of craniofacial deformities. Acta Cirurgica Brasileira, 2010, 25, 264-268.	0.3	8
114	Functionally conserved cis-regulatory elements of COL18A1 identified through zebrafish transgenesis. Developmental Biology, 2010, 337, 496-505.	0.9	17
115	Mesenchymal Stem Cells Derived From Canine Umbilical Cord Veinâ€"A Novel Source for Cell Therapy Studies. Stem Cells and Development, 2010, 19, 395-402.	1.1	67
116	Alveolar osseous defect in rat for cell therapy: preliminary report. Acta Cirurgica Brasileira, 2010, 25, 313-317.	0.3	12
117	HTR1B and HTR2C in autism spectrum disorders in Brazilian families. Brain Research, 2009, 1250, 14-19.	1.1	27
118	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.	0.7	21
119	Syndromes of the first and second pharyngeal arches: A review. American Journal of Medical Genetics, Part A, 2009, 149A, 1853-1859.	0.7	85
120	Reduced transcription of TCOF1 in adult cells of Treacher Collins syndrome patients. BMC Medical Genetics, 2009, 10, 136.	2.1	21
121	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.	1.6	71
122	Stem cell proliferation under low intensity laser irradiation: A preliminary study. Lasers in Surgery and Medicine, 2008, 40, 433-438.	1.1	155
123	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
124	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.	0.7	23
125	Genetics of Craniosynostosis: Genes, Syndromes, Mutations and Genotype-Phenotype Correlations. Frontiers of Oral Biology, 2008, 12, 107-143.	1.5	134
126	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.	1.5	53

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127	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.	1.5	36
128	Reconstruction of Large Cranial Defects in Nonimmunosuppressed Experimental Design With Human Dental Pulp Stem Cells. Journal of Craniofacial Surgery, 2008, 19, 204-210.	0.3	185
129	Unusual phenotype in a female patient with a Gly25Ala substitution in the signal peptide region of the COL2A1 gene. Clinical Dysmorphology, 2008, 17, 225-226.	0.1	3
130	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.	0.3	21
131	Hydrocephalus and moderate mental retardation in a boy with Van der Woude phenotype and IRF6 gene mutation. Clinical Dysmorphology, 2007, 16, 163-166.	0.1	5
132	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.	2.6	229
133	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.	4.3	38
134	Apert p.Ser252Trp Mutation in FGFR2 Alters Osteogenic Potential and Gene Expression of Cranial Periosteal Cells. Molecular Medicine, 2007, 13, 422-442.	1.9	28
135	CNS malformations in Knobloch syndrome with splice mutation inCOL18A1 gene. American Journal of Medical Genetics, Part A, 2007, 143A, 1514-1518.	0.7	34
136	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.	0.7	25
137	Whole-genome array-CGH screening in undiagnosed syndromic patients: old syndromes revisited and new alterations. Cytogenetic and Genome Research, 2006, 115, 254-261.	0.6	103
138	Mutations in collagen 18A1 (COL18A1) and their relevance to the human phenotype. Anais Da Academia Brasileira De Ciencias, 2006, 78, 123-131.	0.3	50
139	Mutational Screening of FGFR1, CER1, and CDON in a Large Cohort of Trigonocephalic Patients. Cleft Palate-Craniofacial Journal, 2006, 43, 148-151.	0.5	15
140	Further evidence of association between mutations inFGFR2 and syndromic craniosynostosis with sacrococcygeal eversion. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 629-633.	1.6	15
141	Molecular screening for microdeletions at 9p22-p24 and 11q23-q24 in a large cohort of patients with trigonocephaly. Clinical Genetics, 2005, 67, 503-510.	1.0	48
142	How pathogenic is the p.D104N/endostatin polymorphic allele ofCOL18A1in Knobloch syndrome?. Human Mutation, 2005, 25, 314-315.	1.1	5
143	TCOF1 mutation database: Novel mutation in the alternatively spliced exon 6A and update in mutation nomenclature. Human Mutation, 2005, 25, 429-434.	1.1	36
144	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.	0.7	32

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145	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.	1.5	59
146	Characterization of human Collagen XVIII promoter 2: Interaction of Sp1, Sp3 and YY1 with the regulatory region and a SNP that increases transcription in hepatocytes. Matrix Biology, 2005, 24, 550-559.	1.5	12
147	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.	1.0	29
148	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.	1.4	49
149	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.	1.4	61
150	Does the P172H mutation at theTM4SF2gene cause X-linked mental retardation?. , 2004, 124A, 413-415.		19
151	Fine mapping and clinical reevaluation of a Brazilian pedigree with a severe form of X-linked mental retardation associated with other neurological dysfunction. , 2004, 127A, 321-323.		8
152	Asymptomatic carriers and gender differences in facioscapulohumeral muscular dystrophy (FSHD). Neuromuscular Disorders, 2004, 14, 33-38.	0.3	114
153	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.	0.5	17
154	Parental origin of mutations in sporadic cases of Treacher Collins syndrome. European Journal of Human Genetics, 2003, 11, 718-722.	1.4	33
155	Immunological Methods for the Analysis of Protein Expression in Neuromuscular Diseases. , 2003, 217, 355-378.		7
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