Francesco Brancati

List of Publications by Year
in descending order

185	12,879	36303 51	29157 104
papers	citations	h-index	g-index

190	190	190	15657
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	RIPK4 regulates cellâ€“cell adhesion in epidermal development and homeostasis. Human Molecular Genetics, 2022, , .	2.9	1
2	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	4.1	41
3	De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. Human Genetics and Genomics Advances, 2022, 3, 100111.	1.7	7
4	HDAC9structural variants disruptingTWIST1transcriptional regulation lead to craniofacial and limb malformations. Genome Research, 2022, 32, 1242-1253.	5.5	5
5	Expanding the PURA syndrome phenotype: A child with the recurrent PURA p.(Phe233del) pathogenic variant showing similarities with cutis laxa. Molecular Genetics & Genomic Medicine, 2021, 9, e1562.	1.2	8
6	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). International Journal of Molecular Sciences, 2021, 22, 1190.	4.1	16
7	Craniosynostosis is a feature of <scp>CHD7</scp>â€related <scp>CHARGE</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2160-2163.	1.2	2
8	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	3.8	4
9	Biological insights in the pathogenesis of hypermobile Ehlers-Danlos syndrome from proteome profiling of patients' dermal myofibroblasts. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166051.	3.8	12
10	Copy number variation analysis implicates novel pathways in patients with oculoâ€auriculoâ€vertebralâ€spectrum and congenital heart defects. Clinical Genetics, 2021, 100, 268-279.	2.0	9
11	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.1	11
12	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	3.3	10
13	Targeted Next-Generation Sequencing Indicates a Frequent Oligogenic Involvement in Primary Ovarian Insufficiency Onset. Frontiers in Endocrinology, 2021, 12, 664645.	3.5	5
14	Matrix Metalloproteinases Inhibition by Doxycycline Rescues Extracellular Matrix Organization and Partly Reverts Myofibroblast Differentiation in Hypermobile Ehlers-Danlos Syndrome Dermal Fibroblasts: A Potential Therapeutic Target?. Cells, 2021, 10, 3236.	4.1	5
15	A recurrent, de novo pathogenic variant in ARPC4 disrupts actin filament formation and causes a neurodevelopmental disorder with microcephaly and speech delay. Human Genetics and Genomics Advances, 2021, 3, 100072.	1.7	4
16	Thickness mapping of individual retinal layers and sectors by Spectralis SpectralÂDomainâ€optical Coherence Tomography in Autosomal Dominant Optic Atrophy. Acta Ophthalmologica, 2020, 98, e390.	1.1	0
17	Mutation analysis of the FBN1 gene in a cohort of patients with Marfan Syndrome: A 10-year single center experience. Clinica Chimica Acta, 2020, 501, 154-164.	1.1	13
18	Multisystemic manifestations in a cohort of 75 classical Ehlers-Danlos syndrome patients: natural history and nosological perspectives. Orphanet Journal of Rare Diseases, 2020, 15, 197.	2.7	20

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19	Craniosynostosisâ€microphthalmia syndrome belongs to the spectrum of <scp>BCOR</scp>â€related disorders. Clinical Genetics, 2020, 98, 413-415.	2.0	0
20	Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. Italian Journal of Pediatrics, 2020, 46, 130.	2.6	14
21	Tremor is a major feature of 9p13 deletion syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2694-2698.	1.2	1
22	Molecular Genetics and Pathogenesis of Ehlersâ€“Danlos Syndrome and Related Connective Tissue Disorders. Genes, 2020, 11, 547.	2.4	29
23	Molecular Genetics of Niemannâ€“Pick Type C Disease in Italy: An Update on 105 Patients and Description of 18 NPC1 Novel Variants. Journal of Clinical Medicine, 2020, 9, 679.	2.4	21
24	Fundamental role of BMP15 in human ovarian folliculogenesis revealed by null and missense mutations associated with primary ovarian insufficiency. Human Mutation, 2020, 41, 983-997.	2.5	20
25	Measles skin rash: Infection of lymphoid and myeloid cells in the dermis precedes viral dissemination to the epidermis. PLoS Pathogens, 2020, 16, e1008253.	4.7	13
26	Cellular and Molecular Mechanisms in the Pathogenesis of Classical, Vascular, and Hypermobile Ehlersâ€'Danlos Syndromes. Genes, 2019, 10, 609.	2.4	46
27	TAB2 c.1398dup variant leads to haploinsufficiency and impairs extracellular matrix homeostasis. Human Mutation, 2019, 40, 1886-1898.	2.5	5
28	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
29	Genotypic Categorization of Loeys-Dietz Syndrome Based on 24 Novel Families and Literature Data. Genes, 2019, 10, 764.	2.4	20
30	H2AFY promoter deletion causes PITX1 endoactivation and Liebenberg syndrome. Journal of Medical Genetics, 2019, 56, 246-251.	3.2	20
31	Clinical and molecular characterization of an 18â€monthâ€old infant with autosomal recessive cutis laxa type 1C due to a novel LTBP4 pathogenic variant, and literature review. Molecular Genetics & Genomic Medicine, 2019, 7, e00735.	1.2	12
32	Delineation of MidXq28â€duplication syndrome distal to MECP2 and proximal to RAB39B genes. Clinical Genetics, 2019, 96, 246-253.	2.0	6
33	Mutational spectrum and clinical signatures in 114 families with hereditary multiple osteochondromas: insights into molecular properties of selected exostosin variants. Human Molecular Genetics, 2019, 28, 2133-2142.	2.9	12
34	Expanding the Clinical and Mutational Spectrum of Recessive AEBP1-Related Classical-Like Ehlers-Danlos Syndrome. Genes, 2019, 10, 135.	2.4	23
35	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
36	Clinical and Molecular Characterization of Classical-Like Ehlers-Danlos Syndrome Due to a Novel TNXB Variant. Genes, 2019, 10, 843.	2.4	16

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37	Chronic mucocutaneous candidiasis and connective tissue disorder in humans with impaired JNK1-dependent responses to IL-17A/F and TGF-Î². Science Immunology, 2019, 4, .	11.9	45
38	Italian validation of the functional difficulties questionnaire (FDQâ€9) and its correlation with major determinants of quality of life in adults with hypermobile Ehlersâ€“Danlos syndrome/hypermobility spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 25-34.	1.7	11
39	Recessive mutations in the neuronal isoforms of DST , encoding dystonin, lead to abnormal actin cytoskeleton organization and HSAN type VI. Human Mutation, 2019, 40, 106-114.	2.5	30
40	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. Journal of Clinical Investigation, 2019, 129, 5374-5380.	8.2	27
41	Keratoderma-Deafness-Mucocutaneous Syndrome Associated with Phe142Leu in the GJB2 Gene. Acta Dermato-Venereologica, 2019, 99, 1192-1194.	1.3	1
42	Dermal fibroblast-to-myofibroblast transition sustained by Î±vÃŸ3 integrin-ILK-Snail1/Slug signaling is a common feature for hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 1010-1023.	3.8	34
43	A novel mutation in CDH11, encoding cadherinâ€11, cause Branchioskeletogenital (Elsahyâ€Waters) syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2028-2033.	1.2	13
44	Exploring relationships between joint hypermobility and neurodevelopment in children (4â€“13 years) with hereditary connective tissue disorders and developmental coordination disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 546-556.	1.7	19
45	Differential Enzymatic Activity of Rat ADAR2 Splicing Variants Is Due to Altered Capability to Interact with RNA in the Deaminase Domain. Genes, 2018, 9, 79.	2.4	9
46	Multifaced Roles of the Î±vÎ²3 Integrin in Ehlersâ€“Danlos and Arterial Tortuosity Syndromesâ€™ Dermal Fibroblasts. International Journal of Molecular Sciences, 2018, 19, 982.	4.1	24
47	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. European Journal of Human Genetics, 2018, 26, 1266-1271.	2.8	12
48	Homozygous Recessive Versican Missense Variation Is Associated With Early Teeth Loss in a Pakistani Family. Frontiers in Genetics, 2018, 9, 723.	2.3	4
49	Transcriptome analysis of skin fibroblasts with dominant negative COL3A1 mutations provides molecular insights into the etiopathology of vascular Ehlers-Danlos syndrome. PLoS ONE, 2018, 13, e0191220.	2.5	31
50	Ehlersâ€“Danlos syndrome, classical type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 27-39.	1.6	116
51	Barber-Say Syndrome and Ablepharon-Macrostomia Syndrome: A Patient's View. Molecular Syndromology, 2017, 8, 172-178.	0.8	6
52	Clinical and molecular characterization of a 13-year-old Indian boy with cutis laxa type 2B: Identification of two novel PYCR1 mutations by amplicon-based semiconductor exome sequencing. Journal of Dermatological Science, 2017, 88, 141-143.	1.9	6
53	The Glu331del mutation in the CYP17A1 gene causes atypical congenital adrenal hyperplasia in a 46,XX female. Gynecological Endocrinology, 2017, 33, 918-922.	1.7	1
54	The 2017 international classification of the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	1.6	1,163

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55	A Small Supernumerary Marker Derived from the Pericentromeric Region of Chromosome 5: Case Report and Delineation of Partial Trisomy 5p Phenotype. Cytogenetic and Genome Research, 2017, 153, 22-28.	1.1	3
56	Identification and characterization of 5â€² CCG interruptions in complex DMPK expanded alleles. European Journal of Human Genetics, 2017, 25, 257-261.	2.8	38
57	Uniparental disomy of chromosome 1 unmasks recessive mutations of PPT1 in a boy with neuronal ceroid lipofuscinosis type 1. Brain and Development, 2017, 39, 182-183.	1.1	5
58	GLUT10â€"Lacking in Arterial Tortuosity Syndromeâ€"Is Localized to the Endoplasmic Reticulum of Human Fibroblasts. International Journal of Molecular Sciences, 2017, 18, 1820.	4.1	15
59	Assessment of the retinal posterior pole in dominant optic atrophy by spectral-domain optical coherence tomography and microperimetry. PLoS ONE, 2017, 12, e0174560.	2.5	17
60	Glucose transporter type 10â€"lacking in arterial tortuosity syndromeâ€"facilitates dehydroascorbic acid transport. FEBS Letters, 2016, 590, 1630-1640.	2.8	25
61	Formation of new chromatin domains determines pathogenicity of genomic duplications. Nature, 2016, 538, 265-269.	27.8	582
62	Two unique TUBB3 mutations cause both CFEOM3 and malformations of cortical development. American Journal of Medical Genetics, Part A, 2016, 170, 297-305.	1.2	51
63	An experimental analysis of bed load transport in gravel-bed braided rivers with high grain Reynolds numbers. Advances in Water Resources, 2016, 94, 160-173.	3.8	10
64	Small fiber neuropathy is a common feature of Ehlers-Danlos syndromes. Neurology, 2016, 87, 155-159.	1.1	90
65	Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3Î²,5Î±,6Î²-Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to NPC1 and SMPD1 Mutations. Clinica Chimica Acta, 2016, 455, 39-45.	1.1	42
66	Characterization of endocrine features and genotypeâ€"phenotypes correlations in blepharophimosisâ€"ptosisâ€"epicanthus inversus syndrome type 1. Journal of Endocrinological Investigation, 2016, 39, 227-233.	3.3	19
67	Transcriptome-Wide Expression Profiling in Skin Fibroblasts of Patients with Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility Type. PLoS ONE, 2016, 11, e0161347.	2.5	40
68	GLUT10 deficiency leads to oxidative stress and non-canonical Î±vÎ²3 integrin-mediated TGFÎ² signalling associated with extracellular matrix disarray in arterial tortuosity syndrome skin fibroblasts. Human Molecular Genetics, 2015, 24, 6769-6787.	2.9	42
69	Bruchâ€™s membrane abnormalities in PRDM5-related brittle cornea syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 145.	2.7	19
70	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€"Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
71	Insights in the etiopathology of galactosyltransferase II (GalT-II) deficiency from transcriptome-wide expression profiling of skin fibroblasts of two sisters with compound heterozygosity for two novel B3GALT6 mutations. Molecular Genetics and Metabolism Reports, 2015, 2, 1-15.	1.1	27
72	Mutations in the NHEJ Component XRCC4 Cause Primordial Dwarfism. American Journal of Human Genetics, 2015, 96, 412-424.	6.2	71

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73	Quantitative ultrasound at the phalanges in a cohort of monozygotic twins of different ages. Radiologia Medica, 2015, 120, 277-282.	7.7	6
74	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	6.2	61
75	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025.	28.9	1,725
76	Neurodevelopmental attributes of joint hypermobility syndrome/Ehlersâ€“Danlos syndrome, hypermobility type: Update and perspectives. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 107-116.	1.6	45
77	Differential diagnosis and diagnostic flow chart of joint hypermobility syndrome/ehlersâ€danlos syndrome hypermobility type compared to other heritable connective tissue disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 6-22.	1.6	91
78	Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. Acta Neuropathologica Communications, 2015, 3, 44.	5.2	45
79	Generation of Human Induced Pluripotent Stem Cells from Extraembryonic Tissues of Fetuses Affected by Monogenic Diseases. Cellular Reprogramming, 2015, 17, 275-287.	0.9	18
80	Cerebral cavernous malformations associated to meningioma: High penetrance in a novel family mutated in thePDCD10gene. Neuroradiology Journal, 2015, 28, 289-293.	1.2	11
81	Characterization of ANKRD11 mutations in humans and mice related to KBG syndrome. Human Genetics, 2015, 134, 181-190.	3.8	52
82	p63â€dependent and independent mechanisms of nectinâ€1 and nectinâ€4 regulation in the epidermis. Experimental Dermatology, 2015, 24, 114-119.	2.9	25
83	Membranous Nectin-4 expression is a risk factor for distant relapse of T1-T2, N0 luminal-A early breast cancer. Oncogenesis, 2014, 3, e118-e118.	4.9	35
84	Unbiased next generation sequencing analysis confirms the existence of autosomal dominant Alport syndrome in a relevant fraction of cases. Clinical Genetics, 2014, 86, 252-257.	2.0	121
85	A Novel LIPE Nonsense Mutation Found Using Exome Sequencing in Siblings With Late-Onset Familial PartialÂLipodystrophy. Canadian Journal of Cardiology, 2014, 30, 1649-1654.	1.7	58
86	Nectin-4 Mutations Causing Ectodermal Dysplasia with Syndactyly Perturb the Rac1 Pathway and the Kinetics of Adherens Junction Formation. Journal of Investigative Dermatology, 2014, 134, 2146-2153.	0.7	33
87	Partial lipodystrophy associated with muscular dystrophy of unknown genetic origin. Muscle and Nerve, 2014, 49, 928-930.	2.2	13
88	De Novo 13q13.3-21.31 deletion involving RB1 gene in a patient with hemangioendothelioma of the liver. Italian Journal of Pediatrics, 2014, 40, 5.	2.6	6
89	Late diagnosis of lateral meningocele syndrome in a 55â€yearâ€old woman with symptoms of joint instability and chronic musculoskeletal pain. American Journal of Medical Genetics, Part A, 2014, 164, 528-534.	1.2	16
90	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. American Journal of Human Genetics, 2014, 95, 622-632.	6.2	34

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91	Nosology and inheritance pattern(s) of joint hypermobility syndrome and Ehlersâ€Danlos syndrome, hypermobility type: A study of intrafamilial and interfamilial variability in 23 Italian pedigrees. American Journal of Medical Genetics, Part A, 2014, 164, 3010-3020.	1.2	70
92	Further delineation of Loeys-Dietz syndrome type 4 in a family with mild vascular involvement and a TGFB2 splicing mutation. BMC Medical Genetics, 2014, 15, 91.	2.1	29
93	Clinical and molecular characterization of 40 patients with classic Ehlersâ€“Danlos syndrome: identification of 18 COL5A1 and 2 COL5A2 novel mutations. Orphanet Journal of Rare Diseases, 2013, 8, 58.	2.7	101
94	Thoracic Aortic Aneurysm in Infancy in Aneurysmsâ€“<scp>O</scp>steoarthritis Syndrome Due to a Novel <scp>SMAD</scp>3 Mutation: Further Delineation of the Phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 1028-1035.	1.2	58
95	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	1.1	57
96	Neurobehavioral phenotype observed in KBG syndrome caused by ANKRD11 mutations. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 17-23.	1.7	39
97	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. Orphanet Journal of Rare Diseases, 2013, 8, 63.	2.7	60
98	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
99	Clinical utility gene card for: Joubert Syndrome - update 2013. European Journal of Human Genetics, 2013, 21, 1187-1187.	2.8	20
100	A Mathematical Model for the Flow Resistance and the Related Hydrodynamic Dispersion Induced by River Dunes. Journal of Applied Mathematics, 2013, 2013, 1-9.	0.9	5
101	Ablepharon macrostomia syndrome: A distinct genetic entity clinically related to the group of FRASâ€“FREM complex disorders. American Journal of Medical Genetics, Part A, 2013, 161, 3012-3017.	1.2	7
102	The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803.	3.9	25
103	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
104	Understanding pyrrolineâ€5â€carboxylate synthetase deficiency: clinical, molecular, functional, and expression studies, structureâ€based analysis, and novel therapy with arginine. Journal of Inherited Metabolic Disease, 2012, 35, 761-776.	3.6	44
105	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	21.4	157
106	Type III and V collagens modulate the expression and assembly of EDA+ fibronectin in the extracellular matrix of defective Ehlersâ€“Danlos syndrome fibroblasts. Biochimica Et Biophysica Acta - General Subjects, 2012, 1820, 1576-1587.	2.4	18
107	Homeotic Arm-to-Leg Transformation Associated with Genomic Rearrangements at the PITX1 Locus. American Journal of Human Genetics, 2012, 91, 629-635.	6.2	111
108	Kinematic and Diffusion Tensor Imaging Definition of Familial Marcus Gunn Jaw-Winking Synkinesis. PLoS ONE, 2012, 7, e51749.	2.5	18

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109	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	2.5	74
110	De Barsy Syndrome: A genetically heterogeneous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. American Journal of Medical Genetics, Part A, 2012, 158A, 927-931.	1.2	37
111	Management of pain and fatigue in the joint hypermobility syndrome (a.k.a. Ehlersâ€“Danlos syndrome,) Tj ETQq1 1 0.784314 rgBT /Ov Medical Genetics, Part A, 2012, 158A, 2055-2070.	1.2	124
112	Mutations in FKBP14 Cause a Variant of Ehlers-Danlos Syndrome with Progressive Kyphoscoliosis, Myopathy, and Hearing Loss. American Journal of Human Genetics, 2012, 90, 201-216.	6.2	136
113	Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. Orphanet Journal of Rare Diseases, 2012, 7, 4.	2.7	64
114	Two families confirm SchÃ¶pf-Schulz-Passarge syndrome as a discrete entity within the WNT10A phenotypic spectrum. Clinical Genetics, 2011, 79, 92-95.	2.0	20
115	Clinical utility gene card for: Joubert syndrome. European Journal of Human Genetics, 2011, 19, 1017-1017.	2.8	0
116	Primary hypertrophic osteoarthropathy: A new family supporting genetic heterogeneity. Joint Bone Spine, 2011, 78, 218-219.	1.6	4
117	Copy-Number Variations Involving the IHH Locus Are Associated with Syndactyly and Craniosynostosis. American Journal of Human Genetics, 2011, 88, 70-75.	6.2	89
118	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. American Journal of Human Genetics, 2011, 88, 767-777.	6.2	106
119	Mutations in ANKRD11 Cause KBG Syndrome, Characterized by Intellectual Disability, Skeletal Malformations, and Macrodontia. American Journal of Human Genetics, 2011, 89, 289-294.	6.2	205
120	Putaminal, but not nigral alterations, characterize hemiparkinsonism-hemiatrophy syndrome: A case report. Movement Disorders, 2011, 26, 352-354.	3.9	5
121	Primary Focal Hyperhidrosis in a New Family Not Linked to Known Loci. Dermatology, 2011, 223, 335-342.	2.1	12
122	Two Novel Mutations Affecting Splicing in the IRF6 Gene Associated With van der Woude Syndrome. Journal of Craniofacial Surgery, 2010, 21, 1654-1656.	0.7	9
123	Mutations in PVRL4, Encoding Cell Adhesion Molecule Nectin-4, Cause Ectodermal Dysplasia-Syndactyly Syndrome. American Journal of Human Genetics, 2010, 87, 265-273.	6.2	98
124	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
125	Three novel mutations in the ANK membrane protein cause craniometaphyseal dysplasia with variable conductive hearing loss. American Journal of Medical Genetics, Part A, 2010, 152A, 870-874.	1.2	20
126	Clinical and laboratory phenotype associated with the aspirinâ€like defect. British Journal of Haematology, 2010, 148, 661-663.	2.5	3

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127	High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. European Journal of Human Genetics, 2010, 18, 1133-1140.	2.8	31
128	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180.	21.4	171
129	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
130	Two Italian patients with novel AAAS gene mutation expand allelic and phenotypic spectrum of triple A (Allgrove) syndrome. Clinical Genetics, 2010, 77, 298-301.	2.0	14
131	Joubert Syndrome and related disorders. Orphanet Journal of Rare Diseases, 2010, 5, 20.	2.7	325
132	Normal Cognitive Functions in Joubert Syndrome. Neuropediatrics, 2009, 40, 287-290.	0.6	35
133	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
134	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
135	The TOR1A polymorphism rs1182 and the risk of spread in primary blepharospasm. Movement Disorders, 2009, 24, 613-616.	3.9	35
136	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	3.9	43
137	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
138	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	21.4	383
139	Genome rearrangements in patients with blepharophimosis, mental retardation and hypothyroidism, soâ€called Youngâ€Simpson syndrome. Clinical Genetics, 2009, 76, 210-213.	2.0	3
140	Clinical, neuropsychological, neurophysiologic, and genetic features of a new Italian pedigree with familial cortical myoclonic tremor with epilepsy. Epilepsia, 2009, 50, 1284-1288.	5.1	40
141	A 6-year-old child with Fryns syndrome: Further delineation of the natural history of the condition in survivors. European Journal of Medical Genetics, 2009, 52, 421-425.	1.3	7
142	Subclinical sensory abnormalities in unaffected PINK1 heterozygotes. Journal of Neurology, 2008, 255, 1372-1377.	3.6	31
143	Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused byRASA1 mutations. Human Mutation, 2008, 29, 959-965.	2.5	382
144	Abnormal neuronal migration defect in the severe variant subtype of Adamsâ€“Oliver syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 1622-1623.	1.2	5

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145	Geroderma osteodysplastica maps to a 4 Mb locus on chromosome 1q24. American Journal of Medical Genetics, Part A, 2008, 146A, 3034-3037.	1.2	9
146	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	6.2	352
147	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	21.4	138
148	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	2.0	64
149	FAK-independent Î±vÎ²3 integrin-EGFR complexes rescue from anoikis matrix-defective fibroblasts. Biochimica Et Biophysica Acta - Molecular Cell Research, 2008, 1783, 1177-1188.	4.1	27
150	Genotypes and phenotypes of Joubert syndrome and related disorders. European Journal of Medical Genetics, 2008, 51, 1-23.	1.3	127
151	Diffusion Tensor Imaging in Joubert Syndrome. American Journal of Neuroradiology, 2007, 28, 1929-1933.	2.4	134
152	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
153	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	6.2	137
154	A novel patient with Cooks syndrome supports splitting from â€œclassicâ€•brachydactyly type B. American Journal of Medical Genetics, Part A, 2007, 143A, 195-199.	1.2	5
155	Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98.	2.5	66
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