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	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025.	28.9	1,725
2	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
3	Formation of new chromatin domains determines pathogenicity of genomic duplications. Nature, 2016, 538, 265-269.	27.8	582
4	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
5	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
6	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	21.4	368
7	Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. Nature Genetics, 2006, 38, 452-457.	21.4	354
8	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
9	Joubert Syndrome and related disorders. Orphanet Journal of Rare Diseases, 2010, 5, 20.	2.7	325
10	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
11	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
12	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
13	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
14	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	21.4	157
15	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	21.4	138
16	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
17	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
18	Diffusion Tensor Imaging in Joubert Syndrome. American Journal of Neuroradiology, 2007, 28, 1929-1933.	2.4	134

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19	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
20	Genotypes and phenotypes of Joubert syndrome and related disorders. European Journal of Medical Genetics, 2008, 51, 1-23.	1.3	127
21	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	5.3	125
22	Management of pain and fatigue in the joint hypermobility syndrome (a.k.a. Ehlersâ€“Danlos syndrome,) Tj ETQq0 0 0 rgBT /Overlock 10 Medical Genetics, Part A, 2012, 158A, 2055-2070.	1.2	124
23	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
24	A locus for autosomal dominant keratoconus maps to human chromosome 3p14-q13. Journal of Medical Genetics, 2004, 41, 188-192.	3.2	118
25	Ehlersâ€“Danlos syndrome, classical type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 27-39.	1.6	116
26	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
27	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
28	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
29	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
30	Park6â€linked parkinsonism occurs in several european families. Annals of Neurology, 2002, 51, 14-18.	5.3	98
31	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
32	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
33	Description, Nomenclature, and Mapping of a Novel Cerebello-Renal Syndrome with the Molar Tooth Malformation. American Journal of Human Genetics, 2003, 73, 663-670.	6.2	91
34	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
35	Human Fibroblasts with Mutations in COL5A1 and COL3A1 Genes Do Not Organize Collagens and Fibronectin in the Extracellular Matrix, Down-regulate Î±2Î²1 Integrin, and Recruit Î±vÎ²3 Instead of Î±5Î²1 Integrin. Journal of Biological Chemistry, 2004, 279, 18157-18168.	3.4	90
36	Small fiber neuropathy is a common feature of Ehlers-Danlos syndromes. Neurology, 2016, 87, 155-159.	1.1	90

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37	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
38	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
39	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
40	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
41	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	2.5	74
42	Mutations in the NHEJ Component XRCC4 Cause Primordial Dwarfism. American Journal of Human Genetics, 2015, 96, 412-424.	6.2	71
43	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
44	Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98.	2.5	66
45	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	2.0	64
46	Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. Orphanet Journal of Rare Diseases, 2012, 7, 4.	2.7	64
47	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
48	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
49	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
50	Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. Movement Disorders, 2003, 18, 1047-1051.	3.9	58
51	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
52	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
53	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	1.1	57
54	Assessing the role ofDRD5 andDYT1 in two different caseâ€“control series with primary blepharospasm. Movement Disorders, 2007, 22, 162-166.	3.9	54

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55	Majewski osteodysplastic primordial dwarfism type II (MOPD II) complicated by stroke: Clinical report and review of cerebral vascular anomalies. American Journal of Medical Genetics, Part A, 2005, 139A, 212-215.	1.2	52
56	Characterization of ANKRD11 mutations in humans and mice related to KBG syndrome. Human Genetics, 2015, 134, 181-190.	3.8	52
57	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
58	Cellular and Molecular Mechanisms in the Pathogenesis of Classical, Vascular, and Hypermobile Ehlersâ€'Danlos Syndromes. Genes, 2019, 10, 609.	2.4	46
59	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
60	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
61	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
62	Novel locus for autosomal dominant pure hereditary spastic paraplegia (SPG19) maps to chromosome 9q33-q34. Annals of Neurology, 2002, 51, 681-685.	5.3	44
63	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
64	Familial blepharospasm is inherited as an autosomal dominant trait and relates to a novel unassigned gene. Movement Disorders, 2003, 18, 207-212.	3.9	43
65	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	3.9	43
66	Exclusion of candidate genes in a family with arterial tortuosity syndrome. American Journal of Medical Genetics Part A, 2004, 126A, 221-228.	2.4	42
67	GLUT10 deficiency leads to oxidative stress and non-canonical Îµvβ3 integrin-mediated TGFβ signalling associated with extracellular matrix disarray in arterial tortuosity syndrome skin fibroblasts. Human Molecular Genetics, 2015, 24, 6769-6787.	2.9	42
68	Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3β,5Îµ,6β-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
69	KBG syndrome. Orphanet Journal of Rare Diseases, 2006, 1, 50.	2.7	41
70	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
71	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
72	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

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73	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
74	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
75	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
76	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
77	KBG syndrome in a cohort of Italian patients. American Journal of Medical Genetics Part A, 2004, 131A, 144-149.	2.4	36
78	Normal Cognitive Functions in Joubert Syndrome. Neuropediatrics, 2009, 40, 287-290.	0.6	35
79	The TOR1A polymorphism rs1182 and the risk of spread in primary blepharospasm. Movement Disorders, 2009, 24, 613-616.	3.9	35
80	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
81	Antenatal presentation of the oculo-auriculo-vertebral spectrum (OAVS). American Journal of Medical Genetics, Part A, 2006, 140A, 1573-1579.	1.2	34
82	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
83	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
84	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
85	Recurrent triploidy of maternal origin. European Journal of Human Genetics, 2003, 11, 972-974.	2.8	31
86	Subclinical sensory abnormalities in unaffected PINK1 heterozygotes. Journal of Neurology, 2008, 255, 1372-1377.	3.6	31
87	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
88	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
89	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
90	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

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91	Molecular Genetics and Pathogenesis of Ehlersâ€“Danlos Syndrome and Related Connective Tissue Disorders. Genes, 2020, 11, 547.	2.4	29
92	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
93	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
94	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. Journal of Clinical Investigation, 2019, 129, 5374-5380.	8.2	27
95	Autosomal dominant hereditary benign telangiectasia maps to the CMC1 locus for capillary malformation on chromosome 5q14. Journal of Medical Genetics, 2003, 40, 849-853.	3.2	26
96	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. Journal of Human Genetics, 2007, 52, 1011-1017.	2.3	25
97	The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803.	3.9	25
98	p63â€dependent and independent mechanisms of nectinâ€1 and nectinâ€4 regulation in the epidermis. Experimental Dermatology, 2015, 24, 114-119.	2.9	25
99	Glucose transporter type 10â€”lacking in arterial tortuosity syndromeâ€”facilitates dehydroascorbic acid transport. FEBS Letters, 2016, 590, 1630-1640.	2.8	25
100	A family of fibronectin mRNAs in human normal and transformed cells. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1986, 868, 207-214.	2.4	24
101	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
102	Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. Movement Disorders, 2002, 17, 392-397.	3.9	23
103	Expanding the Clinical and Mutational Spectrum of Recessive AEBP1-Related Classical-Like Ehlers-Danlos Syndrome. Genes, 2019, 10, 135.	2.4	23
104	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
105	Multiplex ligationâ€dependent probe amplification assay for simultaneous detection of Parkinson's disease gene rearrangements. Movement Disorders, 2007, 22, 2274-2278.	3.9	20
106	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
107	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
108	Clinical utility gene card for: Joubert Syndrome - update 2013. European Journal of Human Genetics, 2013, 21, 1187-1187.	2.8	20

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109	Genotypic Categorization of Loeys-Dietz Syndrome Based on 24 Novel Families and Literature Data. Genes, 2019, 10, 764.	2.4	20
110	H2AFY promoter deletion causes PITX1 endoactivation and Liebenberg syndrome. Journal of Medical Genetics, 2019, 56, 246-251.	3.2	20
111	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
112	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
113	Bruchâ€™s membrane abnormalities in PRDM5-related brittle cornea syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 145.	2.7	19
114	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
115	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
116	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
117	Kinematic and Diffusion Tensor Imaging Definition of Familial Marcus Gunn Jaw-Winking Synkinesis. PLoS ONE, 2012, 7, e51749.	2.5	18
118	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
119	Ablepharon-macrostomia syndrome in a 46-year-old woman. American Journal of Medical Genetics Part A, 2004, 127A, 96-98.	2.4	17
120	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
121	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
122	Clinical and Molecular Characterization of Classical-Like Ehlers-Danlos Syndrome Due to a Novel TNXB Variant. Genes, 2019, 10, 843.	2.4	16
123	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
124	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
125	A novel family with an unusual early-onset generalized dystonia. Movement Disorders, 2005, 20, 81-86.	3.9	14
126	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

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127	Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. Italian Journal of Pediatrics, 2020, 46, 130.	2.6	14
128	Partial lipodystrophy associated with muscular dystrophy of unknown genetic origin. Muscle and Nerve, 2014, 49, 928-930.	2.2	13
129	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
130	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
131	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
132	Primary Focal Hyperhidrosis in a New Family Not Linked to Known Loci. Dermatology, 2011, 223, 335-342.	2.1	12
133	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. European Journal of Human Genetics, 2018, 26, 1266-1271.	2.8	12
134	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
135	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
136	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
137	A Single Strand Conformation Polymorphism-Based Carrier Test for Spinal Muscular Atrophy. Genetic Testing and Molecular Biomarkers, 2001, 5, 33-37.	1.7	11
138	Cerebral cavernous malformations associated to meningioma: High penetrance in a novel family mutated in thePDCD10gene. Neuroradiology Journal, 2015, 28, 289-293.	1.2	11
139	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
140	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.1	11
141	Absence of Correlation Between BMP-4 Polymorphism and Postmenopausal Osteoporosis in Italian Women. Calcified Tissue International, 2000, 67, 93-94.	3.1	10
142	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
143	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	3.3	10
144	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

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145	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
146	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
147	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
148	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
149	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
150	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
151	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
152	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
153	Quantitative ultrasound at the phalanges in a cohort of monozygotic twins of different ages. Radiologia Medica, 2015, 120, 277-282.	7.7	6
154	Barber-Say Syndrome and Ablepharon-Macrostomia Syndrome: A Patient's View. Molecular Syndromology, 2017, 8, 172-178.	0.8	6
155	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
156	Delineation of MidXq28â€duplication syndrome distal to MECP2 and proximal to RAB39B genes. Clinical Genetics, 2019, 96, 246-253.	2.0	6
157	Primary hypothyroidism and osteopenia associated with Neuhauser syndrome. American Journal of Medical Genetics Part A, 2002, 111, 412-414.	2.4	5
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