Bart L Loeys

List of Publications by Year
in descending order

238 papers	23,884 citations	13332 70 h-index	9346 148 g-index
249 all docs	249 docs citations	249 times ranked	21060 citing authors

#	Article	IF	Citations
1	The revised Ghent nosology for the Marfan syndrome. Journal of Medical Genetics, 2010, 47, 476-485.	1.5	1,677
2	Losartan, an AT1 Antagonist, Prevents Aortic Aneurysm in a Mouse Model of Marfan Syndrome. Science, 2006, 312, 117-121.	6.0	1,591
3	A syndrome of altered cardiovascular, craniofacial, neurocognitive and skeletal development caused by mutations in TGFBR1 or TGFBR2. Nature Genetics, 2005, 37, 275-281.	9.4	1,543
4	Aneurysm Syndromes Caused by Mutations in the TGF-Î² Receptor. New England Journal of Medicine, 2006, 355, 788-798.	13.9	1,490
5	Angiotensin II Blockade and Aortic-Root Dilation in Marfan's Syndrome. New England Journal of Medicine, 2008, 358, 2787-2795.	13.9	767
6	Angiotensin II type 1 receptor blockade attenuates TGF-Î²â€“induced failure of muscle regeneration in multiple myopathic states. Nature Medicine, 2007, 13, 204-210.	15.2	603
7	Effect of Mutation Type and Location on Clinical Outcome in 1,013 Probands with Marfan Syndrome or Related Phenotypes and FBN1 Mutations: An International Study. American Journal of Human Genetics, 2007, 81, 454-466.	2.6	485
8	Atenolol versus Losartan in Children and Young Adults with Marfan's Syndrome. New England Journal of Medicine, 2014, 371, 2061-2071.	13.9	457
9	Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. American Journal of Human Genetics, 2007, 80, 485-494.	2.6	445
10	Loeysâ€“Dietz syndrome: a primer for diagnosis and management. Genetics in Medicine, 2014, 16, 576-587.	1.1	435
11	Noncanonical TGFÎ² Signaling Contributes to Aortic Aneurysm Progression in Marfan Syndrome Mice. Science, 2011, 332, 358-361.	6.0	422
12	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nature Genetics, 2012, 44, 922-927.	9.4	391
13	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. Nature Genetics, 2005, 37, 282-288.	9.4	367
14	Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. Nature Genetics, 2006, 38, 452-457.	9.4	354
15	The molecular genetics of Marfan syndrome and related disorders. Journal of Medical Genetics, 2006, 43, 769-787.	1.5	347
16	Update of the UMD-FBN1mutation database and creation of anFBN1polymorphism database. Human Mutation, 2003, 22, 199-208.	1.1	299
17	Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. Human Mutation, 2008, 29, 150-158.	1.1	295
18	Homozygosity for a missense mutation in fibulin-5 (FBLN5) results in a severe form of cutis laxa. Human Molecular Genetics, 2002, 11, 2113-2118.	1.4	283

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19	Early Surgical Experience With Loeys-Dietz: A New Syndrome of Aggressive Thoracic Aortic Aneurysm Disease. Annals of Thoracic Surgery, 2007, 83, S757-S763.	0.7	254
20	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. Journal of Medical Genetics, 2009, 46, 511-523.	1.5	250
21	Mutation of TBCE causes hypoparathyroidismâ€“Âretardationâ€“dysmorphism and autosomal recessive Kennyâ€“Caffey syndrome. Nature Genetics, 2002, 32, 448-452.	9.4	248
22	Circulating Transforming Growth Factor-Î² in Marfan Syndrome. Circulation, 2009, 120, 526-532.	1.6	246
23	Mutations in a TGF-Î² Ligand, TGFB3, CauseÂSyndromic Aortic Aneurysms andÂDissections. Journal of the American College of Cardiology, 2015, 65, 1324-1336.	1.2	238
24	Mutations in the TGF-Î² repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. Nature Genetics, 2012, 44, 1249-1254.	9.4	237
25	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
26	Extending the phenotype of recurrent rearrangements of 16p11.2: Deletions in mentally retarded patients without autism and in normal individuals. European Journal of Medical Genetics, 2009, 52, 77-87.	0.7	226
27	Phenotypic spectrum of the SMAD3-related aneurysmsâ€“osteoarthritis syndrome. Journal of Medical Genetics, 2012, 49, 47-57.	1.5	221
28	Comprehensive molecular screening of theFBN1gene favors locus homogeneity of classical Marfan syndrome. Human Mutation, 2004, 24, 140-146.	1.1	210
29	Mutations ofVMD2Splicing Regulators Cause Nanophthalmos and Autosomal Dominant Vitreoretinochoroidopathy (ADVIRC). , 2004, 45, 3683.		205
30	Ehlers-Danlos syndromes and Marfan syndrome. Best Practice and Research in Clinical Rheumatology, 2008, 22, 165-189.	1.4	205
31	Genotype and Phenotype Analysis of 171 Patients Referred for Molecular Study of the Fibrillin-1 Gene FBN1 Because of Suspected Marfan Syndrome. Archives of Internal Medicine, 2001, 161, 2447-2454.	4.3	204
32	Mutations in Fibrillin-1 Cause Congenital Scleroderma: Stiff Skin Syndrome. Science Translational Medicine, 2010, 2, 23ra20.	5.8	195
33	Differences in manifestations of Marfan syndrome, Ehlers-Danlos syndrome, and Loeys-Dietz syndrome. Annals of Cardiothoracic Surgery, 2017, 6, 582-594.	0.6	192
34	Clinical Validity of Genes for Heritable Thoracic Aortic Aneurysm and Dissection. Journal of the American College of Cardiology, 2018, 72, 605-615.	1.2	190
35	Recent progress towards a molecular understanding of Marfan syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 139C, 4-9.	0.7	176
36	Familial thoracic aortic dilation and bicommissural aortic valve: A prospective analysis of natural history and inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 1960-1967.	0.7	176

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37	Pseudoxanthoma Elasticum-Like Phenotype with Cutis Laxa and Multiple Coagulation Factor Deficiency Represents a Separate Genetic Entity. Journal of Investigative Dermatology, 2007, 127, 581-587.	0.3	168
38	A Dominant-NegativeGFI1BMutation in the Gray Platelet Syndrome. New England Journal of Medicine, 2014, 370, 245-253.	13.9	152
39	Genetics of Thoracic Aortic Aneurysm. Circulation Research, 2013, 113, 327-340.	2.0	151
40	Clinical and Molecular Study of 320 Children With Marfan Syndrome and Related Type I Fibrillinopathies in a Series of 1009 Probands With Pathogenic FBN1 Mutations. Pediatrics, 2009, 123, 391-398.	1.0	146
41	Mutations in KIF11 Cause Autosomal-Dominant Microcephaly Variably Associated with Congenital Lymphedema and Chorioretinopathy. American Journal of Human Genetics, 2012, 90, 356-362.	2.6	138
42	Aggressive Cardiovascular Phenotype of Aneurysms-Osteoarthritis Syndrome Caused by Pathogenic SMAD3 Variants. Journal of the American College of Cardiology, 2012, 60, 397-403.	1.2	135
43	Novel MYH11 and ACTA2 mutations reveal a role for enhanced TGFÎ² signaling in FTAAD. International Journal of Cardiology, 2013, 165, 314-321.	0.8	134
44	Cardiovascular manifestations in men and women carrying a FBN1 mutation. European Heart Journal, 2010, 31, 2223-2229.	1.0	133
45	Altered TGFÎ² signaling and cardiovascular manifestations in patients with autosomal recessive cutis laxa type I caused by fibulin-4 deficiency. European Journal of Human Genetics, 2010, 18, 895-901.	1.4	132
46	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
47	Marfan Syndrome and Related Disorders: 25 Years of Gene Discovery. Human Mutation, 2016, 37, 524-531.	1.1	125
48	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. American Journal of Human Genetics, 2016, 99, 174-187.	2.6	124
49	The Loeysâ€“Dietz syndrome: an update for the clinician. Current Opinion in Cardiology, 2010, 25, 546-551.	0.8	121
50	The molecular basis of classic Ehlers-Danlos syndrome: A comprehensive study of biochemical and molecular findings in 48 unrelated patients. Human Mutation, 2005, 25, 28-37.	1.1	117
51	New insights into the pathogenesis of autosomalâ€dominant cutis laxa with report of fiveELNmutations. Human Mutation, 2011, 32, 445-455.	1.1	116
52	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	1.1	116
53	Arterial Tortuosity. Hypertension, 2019, 73, 951-960.	1.3	110
54	Loeys-Dietz Syndrome. Advances in Experimental Medicine and Biology, 2014, 802, 95-105.	0.8	106

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55	Neuroradiologic Manifestations of Loeys-Dietz Syndrome Type 1. American Journal of Neuroradiology, 2009, 30, 1614-1619.	1.2	104
56	Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Human Molecular Genetics, 2013, 22, 2590-2602.	1.4	103
57	ROBO4 variants predispose individuals to bicuspid aortic valve and thoracic aortic aneurysm. Nature Genetics, 2019, 51, 42-50.	9.4	101
58	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	1.1	97
59	Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. Genetics in Medicine, 2017, 19, 386-395.	1.1	94
60	A mutation update for the FLNC gene in myopathies and cardiomyopathies. Human Mutation, 2020, 41, 1091-1111.	1.1	92
61	Challenges for CNV interpretation in clinical molecular karyotyping: Lessons learned from a 1001 sample experience. European Journal of Medical Genetics, 2009, 52, 398-403.	0.7	90
62	The new Ghent criteria for Marfan syndrome: what do they change?. Clinical Genetics, 2012, 81, 433-442.	1.0	90
63	Candidate Gene Resequencing in a Large Bicuspid Aortic Valve-Associated Thoracic Aortic Aneurysm Cohort: SMAD6 as an Important Contributor. Frontiers in Physiology, 2017, 8, 400.	1.3	85
64	Fibulin-5 mutations: mechanisms of impaired elastic fiber formation in recessive cutis laxa. Human Molecular Genetics, 2006, 15, 3379-3386.	1.4	84
65	Contribution of molecular analyses in diagnosing Marfan syndrome and type I fibrillinopathies: an international study of 1009 probands. Journal of Medical Genetics, 2008, 45, 384-390.	1.5	83
66	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. Circulation, 2020, 142, 324-338.	1.6	83
67	Novel clinico-molecular insights in pseudoxanthoma elasticum provide an efficient molecular screening method and a comprehensive diagnostic flowchart. Human Mutation, 2008, 29, 205-205.	1.1	82
68	Comprehensive clinical and molecular assessment of 32 probands with congenital contractural arachnodactyly: Report of 14 novel mutations and review of the literature. Human Mutation, 2009, 30, 334-341.	1.1	81
69	Aetiology and management of hereditary aortopathy. Nature Reviews Cardiology, 2017, 14, 197-208.	6.1	75
70	Loeys-Dietz Syndrome: MDCT Angiography Findings. American Journal of Roentgenology, 2007, 189, W29-W35.	1.0	74
71	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	2.6	73
72	What Is New in Dilatation of the Ascending Aorta?. Circulation, 2011, 123, 924-928.	1.6	72

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73	Applying massive parallel sequencing to molecular diagnosis of Marfan and Loeys-Dietz syndromes. Human Mutation, 2011, 32, 1053-1062.	1.1	71
74	Infantile Restrictive Cardiomyopathy Resulting From a Mutation in the Cardiac Troponin T Gene. Pediatrics, 2006, 117, 1830-1833.	1.0	68
75	Clinical and mutation-type analysis from an international series of 198 probands with a pathogenic FBN1 exons 24â€“32 mutation. European Journal of Human Genetics, 2009, 17, 491-501.	1.4	66
76	Musculoskeletal Findings of Loeys-Dietz Syndrome. Journal of Bone and Joint Surgery - Series A, 2010, 92, 1876-1883.	1.4	66
77	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	1.1	66
78	Biallelic Loss of Proprioception-Related PIEZO2 Causes Muscular Atrophy with Perinatal Respiratory Distress, Arthrogryposis, and Scoliosis. American Journal of Human Genetics, 2016, 99, 1206-1216.	2.6	65
79	PORCNmutations in focal dermal hypoplasia: coping with lethality. Human Mutation, 2009, 30, E618-E628.	1.1	64
80	Aneurysm of the Pulmonary Artery, a Systematic Review and Critical Analysis of Current Literature. Congenital Heart Disease, 2016, 11, 102-109.	0.0	64
81	An AugmentedABCA4Screen Targeting Noncoding Regions Reveals a Deep Intronic Founder Variant in Belgian Stargardt Patients. Human Mutation, 2015, 36, 39-42.	1.1	61
82	New RAB3GAP1 mutations in patients with Warburg Micro Syndrome from different ethnic backgrounds and a possible founder effect in the Danish. European Journal of Human Genetics, 2010, 18, 1100-1106.	1.4	60
83	VariantDB: a flexible annotation and filtering portal for next generation sequencing data. Genome Medicine, 2014, 6, 74.	3.6	60
84	The Ghent Marfan Trial â€" A randomized, double-blind placebo controlled trial with losartan in Marfan patients treated with Î²-blockers. International Journal of Cardiology, 2012, 157, 354-358.	0.8	59
85	Characteristics of children and young adults with Marfan syndrome and aortic root dilation in a randomized trial comparing atenolol and losartan therapy. American Heart Journal, 2013, 165, 828-835.e3.	1.2	59
86	Expert consensus recommendations on the cardiogenetic care for patients with thoracic aortic disease and their first-degree relatives. International Journal of Cardiology, 2018, 258, 243-248.	0.8	59
87	A critical analysis of minor cardiovascular criteria in the diagnostic evaluation of patients with Marfan syndrome. Genetics in Medicine, 2006, 8, 401-408.	1.1	58
88	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.	0.7	58
89	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	2.6	58
90	COL5A1 signal peptide mutations interfere with protein secretion and cause classic Ehlers-Danlos syndrome. Human Mutation, 2009, 30, E395-E403.	1.1	57

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91	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. Genetics in Medicine, 2021, 23, 47-58.	1.1	57
92	A Decade of Discovery in the Genetic Understanding of Thoracic Aortic Disease. Canadian Journal of Cardiology, 2016, 32, 13-25.	0.8	55
93	Homozygous Missense Mutation in Fibulin-5 in an Iranian Autosomal Recessive Cutis Laxa Pedigree and Associated Haplotype. Journal of Investigative Dermatology, 2006, 126, 1506-1509.	0.3	53
94	Marfan syndrome. Current Opinion in Pediatrics, 2012, 24, 498-504.	1.0	53
95	Does monosymptomatic enuresis exist? A molecular genetic exploration of 32 families with enuresis/incontinence. BJU International, 2002, 90, 76-83.	1.3	50
96	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	1.4	48
97	Utility of molecular analyses in the exploration of extreme intrafamilial variability in the Marfan syndrome. Clinical Genetics, 2007, 72, 188-198.	1.0	47
98	Polymorphisms in the lectin pathway genes as a possible cause of early chronic Pseudomonas aeruginosa colonization in cystic fibrosis patients. Human Immunology, 2012, 73, 1175-1183.	1.2	47
99	Recognizing the tenascinâ€X deficient type of Ehlersâ€“Danlos syndrome: a crossâ€sectional study in 17 patients. Clinical Genetics, 2017, 91, 411-425.	1.0	46
100	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants ofCOH1. Human Mutation, 2009, 30, E404-E420.	1.1	44
101	Design and rationale of a prospective, collaborative meta-analysis of all randomized controlled trials of angiotensin receptor antagonists in Marfan syndrome, based on individual patient data: A report from the Marfan Treatment Trialists' Collaboration. American Heart Journal, 2015, 169, 605-612.	1.2	44
102	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. European Journal of Human Genetics, 2017, 25, 43-51.	1.4	44
103	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50 Report of eight cases including a living child and further evidence for autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 1268-1281.	0.7	43
104	An Integrated Framework to Quantitatively Link Mouse-Specific Hemodynamics to Aneurysm Formation in Angiotensin II-infused ApoE â^'/â^' mice. Annals of Biomedical Engineering, 2011, 39, 2430-2444.	1.3	43
105	Extracellular Matrix in Vascular Disease, Part 2/4. Journal of the American College of Cardiology, 2020, 75, 2189-2203.	1.2	43
106	Pathogenic FBN1 mutations in 146 adults not meeting clinical diagnostic criteria for Marfan syndrome: Further delineation of type 1 fibrillinopathies and focus on patients with an isolated major criterion. American Journal of Medical Genetics, Part A, 2009, 149A, 854-860.	0.7	40
107	Replacing Vascular Corrosion Casting by In Vivo Micro-CT Imaging for Building 3D Cardiovascular Models in Mice. Molecular Imaging and Biology, 2011, 13, 78-86.	1.3	40
108	TGF-Î² signalopathies as a paradigm for translational medicine. European Journal of Medical Genetics, 2015, 58, 695-703.	0.7	39

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109	Genetic counselling and testing in adults with congenital heart disease: A consensus document of the ESC Working Group of Grown-Up Congenital Heart Disease, the ESC Working Group on Aorta and Peripheral Vascular Disease and the European Society of Human Genetics. European Journal of Preventive Cardiology, 2020, 27, 1423-1435.	0.8	38
110	Clinical and electrophysiological findings in autosomal dominant vitreoretinochoroidopathy: report of a new pedigree. , 2001, 239, 575-582.		36
111	A clinical appraisal of different Z-score equations for aortic root assessment in the diagnostic evaluation of Marfan syndrome. Genetics in Medicine, 2013, 15, 528-532.	1.1	35
112	AnFBN1Deep Intronic Mutation in a Familial Case of Marfan Syndrome: An Explanation for Genetically Unsolved Cases?. Human Mutation, 2014, 35, 571-574.	1.1	34
113	Confirmation of the role of pathogenic SMAD6 variants in bicuspid aortic valve-related aortopathy. European Journal of Human Genetics, 2019, 27, 1044-1053.	1.4	32
114	Interpretation and actionability of genetic variants in cardiomyopathies: a position statement from the European Society of Cardiology Council on cardiovascular genomics. European Heart Journal, 2022, 43, 1901-1916.	1.0	32
115	The Meier-Gorlin syndrome, or ear-patella-short stature syndrome, in sibs. American Journal of Medical Genetics Part A, 1999, 84, 61-67.	2.4	31
116	Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. Human Molecular Genetics, 2015, 24, 1106-1118.	1.4	31
117	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31
118	Absence of arterial phenotype in mice with homozygous slc2A10 missense substitutions. Genesis, 2008, 46, 385-389.	0.8	30
119	Enrichment of Rare Variants in Loeysâ€“Dietz Syndrome Genes in Spontaneous Coronary Artery Dissection but Not in Severe Fibromuscular Dysplasia. Circulation, 2020, 142, 1021-1024.	1.6	30
120	Biglycan in the Skeleton. Journal of Histochemistry and Cytochemistry, 2020, 68, 747-762.	1.3	30
121	Genotypeâ€“phenotype correlation in eight new patients with a deletion encompassing 2q31.1. American Journal of Medical Genetics, Part A, 2010, 152A, 1213-1224.	0.7	29
122	Renal insufficiency, a frequent complication with age in oralâ€facialâ€digital syndrome type I. Clinical Genetics, 2010, 77, 258-265.	1.0	29
123	17q24.2 microdeletions: a new syndromal entity with intellectual disability, truncal obesity, mood swings and hallucinations. European Journal of Human Genetics, 2012, 20, 534-539.	1.4	28
124	Clinical utility gene card for: Hereditary thoracic aortic aneurysm and dissection including next-generation sequencing-based approaches. European Journal of Human Genetics, 2016, 24, 146-150.	1.4	28
125	Predictors of Bicuspid Aortic Valveâ€“Associated Aortopathy in Childhood. Circulation: Cardiovascular Imaging, 2020, 13, e009717.	1.3	28
126	The Impact of Simplified Boundary Conditions and Aortic Arch Inclusion on CFD Simulations in the Mouse Aorta: A Comparison With Mouse-specific Reference Data. Journal of Biomechanical Engineering, 2011, 133, 121006.	0.6	27

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127	Unusual 8p inverted duplication deletion with telomere capture from 8q. European Journal of Medical Genetics, 2009, 52, 31-36.	0.7	26
128	Educational paper. European Journal of Pediatrics, 2013, 172, 997-1005.	1.3	26
129	Health-Related Quality of Life in Children and Young Adults with Marfan Syndrome. Journal of Pediatrics, 2019, 204, 250-255.e1.	0.9	26
130	Clinical utility gene card for: Marfan syndrome type 1 and related phenotypes [FBN1]. European Journal of Human Genetics, 2010, 18, 1071-1071.	1.4	25
131	Angiotensin receptor blockers: a panacea for Marfan syndrome and related disorders?. Drug Discovery Today, 2015, 20, 262-266.	3.2	25
132	Bi-allelic Loss-of-Function Mutations in the NPR-C Receptor Result in Enhanced Growth and Connective Tissue Abnormalities. American Journal of Human Genetics, 2018, 103, 288-295.	2.6	25
133	Novel pathogenicSMAD2variants in five families with arterial aneurysm and dissection: further delineation of the phenotype. Journal of Medical Genetics, 2019, 56, 220-227.	1.5	25
134	Whole exome sequencing is an efficient, sensitive and specific method of mutation detection in osteogenesis imperfecta and Marfan syndrome. BoneKEy Reports, 2013, 2, 456.	2.7	24
135	Copy number variation analysis in bicuspid aortic valve-related aortopathy identifies TBX20 as a contributing gene. European Journal of Human Genetics, 2019, 27, 1033-1043.	1.4	24
136	Fungal intracranial aneurysm in a child with familial chronic mucocutaneous candidiasis. European Journal of Pediatrics, 1999, 158, 650-652.	1.3	23
137	GENETIC FIBRILLINOPATHIES : NEW INSIGHTS IN MOLECULAR DIAGNOSIS AND CLINICAL MANAGEMENT. Acta Clinica Belgica, 2003, 58, 3-11.	0.5	23
138	The influence of aortic dimensions on calculated wall shear stress in the mouse aortic arch. Computer Methods in Biomechanics and Biomedical Engineering, 2009, 12, 491-499.	0.9	23
139	Clinical utility gene card for: Loeys-Dietz syndrome (TGFBR1/2) and related phenotypes. European Journal of Human Genetics, 2011, 19, 1108-1108.	1.4	23
140	The genetic architecture of non-syndromic thoracic aortic aneurysm. Heart, 2015, 101, 1678-1684.	1.2	23
141	Defining the Clinical, Molecular and Ultrastructural Characteristics in Occipital Horn Syndrome: Two New Cases and Review of the Literature. Genes, 2019, 10, 528.	1.0	23
142	European reference network for rare vascular diseases (VASCERN) consensus statement for the screening and management of patients with pathogenic ACTA2 variants. Orphanet Journal of Rare Diseases, 2019, 14, 264.	1.2	23
143	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	1.1	23
144	A dynamic mucin mRNA signature associates with COVID-19 disease presentation and severity. JCI Insight, 2021, 6, .	2.3	23

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145	Bruck syndrome: neonatal presentation and natural course in three patients. Pediatric Radiology, 1998, 28, 781-789.	1.1	22
146	Developmental delay and connective tissue disorder in four patients sharing a common microdeletion at 6q13-14. Journal of Medical Genetics, 2010, 47, 717-720.	1.5	22
147	The neuromuscular differential diagnosis of joint hypermobility. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 23-42.	0.7	22
148	The 8th international research symposium on the Marfan Syndrome and related conditions. American Journal of Medical Genetics, Part A, 2012, 158A, 42-49.	0.7	21
149	Telomeric refinement of the MCKD1 locuson chromosome 1q21**See Editorial by Bichet and Fujiwara, p. 864.. Kidney International, 2004, 66, 580-585.	2.6	20
150	Identification of Mutations in the PRDM5 Gene in Brittle Cornea Syndrome. Cornea, 2016, 35, 853-859.	0.9	18
151	FLNA mutations in surviving males presenting with connective tissue findings: two new case reports and review of the literature. BMC Medical Genetics, 2018, 19, 140.	2.1	18
152	Aortic aneurysm/dissection and osteogenesis imperfecta: Four new families and review of the literature. Bone, 2019, 121, 191-195.	1.4	18
153	Bone lessons from Marfan syndrome and related disorders: fibrillin, TGF-B and BMP at the balance of too long and too short. Pediatric Endocrinology Reviews, 2013, 10 Suppl 2, 417-23.	1.2	18
154	Severe aortopathy due to fibulin-4 deficiency: molecular insights, surgical strategy, and a review of the literature. European Journal of Pediatrics, 2014, 173, 671-5.	1.3	17
155	NovelLMNAmutations cause an aggressive atypical neonatal progeria without progerin accumulation. Journal of Medical Genetics, 2016, 53, 776-785.	1.5	17
156	Partial anomalous pulmonary venous return in Turner syndrome. European Journal of Radiology, 2017, 95, 141-146.	1.2	17
157	Cutis laxa of the autosomal recessive type in a consanguineous family. European Journal of Dermatology, 2003, 13, 529-33.	0.3	17
158	Genetic variations in toll-like receptor pathway and lung function decline in Cystic Fibrosis patients. Human Immunology, 2013, 74, 1649-1655.	1.2	16
159	The search for genotype/phenotype correlation in Marfan syndrome: to be or not to be?. European Heart Journal, 2016, 37, 3291-3293.	1.0	16
160	Loeys-Dietz Syndrome. Advances in Experimental Medicine and Biology, 2021, 1348, 251-264.	0.8	16
161	Identification of copy number variants associated with BPES-like phenotypes. Human Genetics, 2008, 124, 489-498.	1.8	15
162	A new sporadic case of early-onset Loeys-Dietz syndrome due to the recurrent mutation p.R528C in theTGFBR2 gene substantiates interindividual clinical variability. Journal of Applied Genetics, 2009, 50, 405-410.	1.0	15

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163	The diagnostic value of the facial features of Marfan syndrome. Journal of Children's Orthopaedics, 2010, 4, 545-551.	0.4	15
164	Congenital glucoseâ€“galactose malabsorption: a novel deletion within the SLC5A1 gene. European Journal of Pediatrics, 2013, 172, 409-411.	1.3	15
165	Targeted Next-Generation Sequencing of 51 Genes Involved in Primary Electrical Disease. Journal of Molecular Diagnostics, 2017, 19, 445-459.	1.2	15
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