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	Current progress in clinical, molecular, and genetic aspects of adult fibromuscular dysplasia. Cardiovascular Research, 2022, 118, 65-83.	1.8	14
2	Molecular characterization and investigation of the role of genetic variation in phenotypic variability and response to treatment in a large pediatric Marfan syndrome cohort. Genetics in Medicine, 2022, 24, 1045-1053.	1.1	13
3	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
4	Update on the molecular landscape of thoracic aortic aneurysmal disease. Current Opinion in Cardiology, 2022, Publish Ahead of Print, .	0.8	3
5	Morpho-functional comparison of differentiation protocols to create iPSC-derived cardiomyocytes. Biology Open, 2022, 11, .	0.6	3
6	Generation of two induced pluripotent stem cell (iPSC) lines (BBANTWi006-A, BBANTWi007-A) from Brugada syndrome patients carrying an SCN5A mutation. Stem Cell Research, 2022, 60, 102719.	0.3	5
7	Diagnostic yield of genetic testing in heart transplant recipients with prior cardiomyopathy. Journal of Heart and Lung Transplantation, 2022, 41, 1218-1227.	0.3	7
8	The fibrillinopathies: New insights with focus on the paradigm of opposing phenotypes for both FBN1 and FBN2. Human Mutation, 2022, 43, 815-831.	1.1	7
9	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	1.1	1
10	Genome-Wide Epistasis for Cardiovascular Severity in Marfan Study Design: Patient Organization Driven Research. Aorta, 2022, , .	0.1	0
11	Isolated aneurysmal disease as an underestimated finding in individuals with JAG1 pathogenic variants. Human Mutation, 2022, 43, 1824-1828.	1.1	3
12	Clinical characterization of the first Belgian SCN5A founder mutation cohort. Europace, 2021, 23, 918-927.	0.7	3
13	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
14	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
15	PTGIR, a susceptibility gene for fibromuscular dysplasia?. Cardiovascular Research, 2021, 117, 990-992.	1.8	1
16	iPSC-Cardiomyocyte Models of Brugada Syndromeâ€"Achievements, Challenges and Future Perspectives. International Journal of Molecular Sciences, 2021, 22, 2825.	1.8	13
17	Comparability of different Z-score equations for aortic root dimensions in children with Marfan syndrome. Cardiology in the Young, 2021, 31, 1962-1968.	0.4	5
18	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	2.6	10

#	Article	IF	Citations
19	Novel LOX Variants in Five Families with Aortic/Arterial Aneurysm and Dissection with Variable Connective Tissue Findings. International Journal of Molecular Sciences, 2021, 22, 7111.	1.8	7
20	A dynamic mucin mRNA signature associates with COVID-19 disease presentation and severity. JCI Insight, 2021, 6, .	2.3	23
21	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	0.6	6
22	Two novel presentations of KCNMA1â€related pathologyâ€“â€“Expanding the clinical phenotype of a rare channelopathy. Molecular Genetics & Genomic Medicine, 2021, 9, e1797.	0.6	6
23	Molecular autopsy and subsequent functional analysis reveal de novo DSG2 mutation as cause of sudden death. European Journal of Medical Genetics, 2021, 64, 104322.	0.7	0
24	Clinically relevant variants in a large cohort of Indian patients with Marfan syndrome and related disorders identified by next-generation sequencing. Scientific Reports, 2021, 11, 764.	1.6	7
25	Loeys-Dietz Syndrome. Advances in Experimental Medicine and Biology, 2021, 1348, 251-264.	0.8	16
26	Meester-Loeys Syndrome. Advances in Experimental Medicine and Biology, 2021, 1348, 265-272.	0.8	4
27	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
28	The Role of Genetics in Risk Stratification of Thoracic Aortic Aneurysm Dissection. Hearts, 2020, 1, 50-61.	0.4	0
29	Compound Heterozygous SCN5A Mutations in Severe Sodium Channelopathy With Brugada Syndrome: A Case Report. Frontiers in Cardiovascular Medicine, 2020, 7, 117.	1.1	3
30	Chondrodysplasias and Aneurysmal Thoracic Aortopathy: An Emerging Tale of Molecular Intersection. Trends in Molecular Medicine, 2020, 26, 783-795.	3.5	2
31	Hide and seek: Somatic SMAD3 mutations in melorheostosis. Journal of Experimental Medicine, 2020, 217, .	4.2	1
32	Delineation of a new fibrillino-2-pathy with evidence for a role of FBN2 in the pathogenesis of carpal tunnel syndrome. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107085.	1.5	4
33	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
34	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
35	Variants in ADRB1 and CYP2C9: Association with Response to Atenolol and Losartan in Marfan Syndrome. Journal of Pediatrics, 2020, 222, 213-220.e5.	0.9	8
36	Predictors of Bicuspid Aortic Valveâ€“Associated Aortopathy in Childhood. Circulation: Cardiovascular Imaging, 2020, 13, e009717.	1.3	28

#	Article	IF	Citations
37	Biglycan in the Skeleton. Journal of Histochemistry and Cytochemistry, 2020, 68, 747-762.	1.3	30
38	A mutation update for the FLNC gene in myopathies and cardiomyopathies. Human Mutation, 2020, 41, 1091-1111.	1.1	92
39	Blood biomarkers in patients with bicuspid aortic valve disease. Journal of Cardiology, 2020, 76, 287-294.	0.8	3
40	Extracellular Matrix in Vascular Disease, Part 2/4. Journal of the American College of Cardiology, 2020, 75, 2189-2203.	1.2	43
41	Bicuspid Aortic Valve. , 2020, , 345-360.		0
42	Optical Mapping in hiPSC-CM and Zebrafish to Resolve Cardiac Arrhythmias. Hearts, 2020, 1, 181-199.	0.4	2
43	Pathophysiology and Principles of Management of Hereditary Aneurysmal Aortopathies. , 2020, , 293-316.		0
44	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
45	Identification and characterization of a novel FBN1 gene variant in an extended family with variable clinical phenotype of Marfan syndrome. Connective Tissue Research, 2019, 60, 146-154.	1.1	2
46	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
47	Progressive Pulmonary Artery Dilatation is Associated with Type B Aortic Dissection in Patients with Marfan Syndrome. Journal of Clinical Medicine, 2019, 8, 1848.	1.0	4
48	Molecular Signature of CAID Syndrome: Noncanonical Roles of SGO1 in Regulation of TGF-Î² Signaling and Epigenomics. Cellular and Molecular Gastroenterology and Hepatology, 2019, 7, 411-431.	2.3	11
49	Aortic aneurysm/dissection and osteogenesis imperfecta: Four new families and review of the literature. Bone, 2019, 121, 191-195.	1.4	18
50	Aortic Valve Surgery in Nonelderly Patients: Insights Gained From AVIATOR. Seminars in Thoracic and Cardiovascular Surgery, 2019, 31, 643-649.	0.4	10
51	Clinical Aspects of Heritable Connective Tissue Disorders. , 2019, , 523-530.		0
52	Arterial Tortuosity. Hypertension, 2019, 73, 951-960.	1.3	110
53	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
54	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

#	Article	IF	Citations
55	Cardiogeneticsbank@UZA: A Collection of DNA, Tissues, and Cell Lines as a Translational Tool. Frontiers in Medicine, 2019, 6, 198.	1.2	1
56	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
57	Comparison of biomechanical properties in ascending aortic aneurysms of patients with congenital bicuspid aortic valve and Marfan syndrome. International Journal of Cardiology, 2019, 278, 65-69.	0.8	8
58	ROBO4 variants predispose individuals to bicuspid aortic valve and thoracic aortic aneurysm. Nature Genetics, 2019, 51, 42-50.	9.4	101
59	Health-Related Quality of Life in Children and Young Adults with Marfan Syndrome. Journal of Pediatrics, 2019, 204, 250-255.e1.	0.9	26
60	Genetics of Marfan Syndrome and Loeys-Dietz Syndrome. , 2019, , 561-566.		0
61	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
62	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	1.1	116
63	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	1.1	66
64	Left ventricular non-compaction with Ebstein anomaly attributed to a TPM1 mutation. European Journal of Medical Genetics, 2018, 61, 8-10.	0.7	12
65	First evidence of maternally inherited mosaicism in TGFBR1 and subtle primary myocardial changes in Loeys-Dietz syndrome: a case report. BMC Medical Genetics, 2018, 19, 170.	2.1	4
66	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
67	Predictors of Rapid Aortic Root Dilation and Referral for Aortic Surgery in Marfan Syndrome. Pediatric Cardiology, 2018, 39, 1453-1461.	0.6	14
68	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
69	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
70	Severe Phenotype of Cutis Laxa Type 1B with Antenatal Signs due to a Novel Homozygous Nonsense Mutation in EFEMP2. Molecular Syndromology, 2018, 9, 190-196.	0.3	5
71	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31
72	Marfan Syndrome and Related Disorders. , 2018, , 589-615.		2

#	Article	IF	Citations
73	Aetiology and management of hereditary aortopathy. Nature Reviews Cardiology, 2017, 14, 197-208.	6.1	75
74	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
75	Targeted Next-Generation Sequencing of 51 Genes Involved in Primary Electrical Disease. Journal of Molecular Diagnostics, 2017, 19, 445-459.	1.2	15
76	Metatarsal bony syndactyly in 2 fetuses with Smithâ€Lemliâ€Opitz syndrome: An underâ€recognized part of the clinical spectrum. Clinical Genetics, 2017, 92, 342-343.	1.0	1
77	Partial anomalous pulmonary venous return in Turner syndrome. European Journal of Radiology, 2017, 95, 141-146.	1.2	17
78	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
79	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
80	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
81	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
82	Differences in manifestations of Marfan syndrome, Ehlers-Danlos syndrome, and Loeys-Dietz syndrome. Annals of Cardiothoracic Surgery, 2017, 6, 582-594.	0.6	192
83	Molecular Insights into Bicuspid Aortic Valve Development and the associated aortopathy. AIMS Molecular Science, 2017, 4, 478-508.	0.3	1
84	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
85	Aneurysm of the Pulmonary Artery, a Systematic Review and Critical Analysis of Current Literature. Congenital Heart Disease, 2016, 11, 102-109.	0.0	64
86	Marfan Syndrome and Related Disorders: 25 Years of Gene Discovery. Human Mutation, 2016, 37, 524-531.	1.1	125
87	Identification of Mutations in the PRDM5 Gene in Brittle Cornea Syndrome. Cornea, 2016, 35, 853-859.	0.9	18
88	NovelLMNAmutations cause an aggressive atypical neonatal progeria without progerin accumulation. Journal of Medical Genetics, 2016, 53, 776-785.	1.5	17
89	Identification of FBN1 gene mutations in Ukrainian Marfan syndrome patients. Genetical Research, 2016, 98, e13.	0.3	2
90	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

#	Article	IF	Citations
91	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
92	Genpanels, een recente innovatie in de moleculair-genetische laboratoria en een wereld van verschil. Bijblijven (Amsterdam, Netherlands), 2016, 32, 16-24.	0.0	0
93	Genetic Testing in Thoracic Aortic Diseaseâ€"When, Why, and How?. Canadian Journal of Cardiology, 2016, 32, 131-134.	0.8	12
94	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
95	A Decade of Discovery in the Genetic Understanding of Thoracic Aortic Disease. Canadian Journal of Cardiology, 2016, 32, 13-25.	0.8	55
96	Bicuspid Aortic Valve. , 2016, , 295-308.		1
97	An AugmentedABCA4Screen Targeting Noncoding Regions Reveals a Deep Intronic Founder Variant in Belgian Stargardt Patients. Human Mutation, 2015, 36, 39-42.	1.1	61
98	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	1.1	97
99	TGF-Î² signalopathies as a paradigm for translational medicine. European Journal of Medical Genetics, 2015, 58, 695-703.	0.7	39
100	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
101	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
102	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
103	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
104	Cervical artery dissections and type A aortic dissection in a family with a novel missense COL3A1 mutation of vascular type Ehlersâ€"Danlos syndrome. European Journal of Medical Genetics, 2015, 58, 634-636.	0.7	6
105	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	2.6	73
106	The genetic architecture of non-syndromic thoracic aortic aneurysm. Heart, 2015, 101, 1678-1684.	1.2	23
107	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	2.6	58
108	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

#	Article	IF	Citations
109	Angiotensin receptor blockers: a panacea for Marfan syndrome and related disorders?. Drug Discovery Today, 2015, 20, 262-266.	3.2	25
110	Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. Human Molecular Genetics, 2015, 24, 1106-1118.	1.4	31
111	Genetics of sudden cardiac death in the young. Clinical Genetics, 2015, 88, 101-113.	1.0	12
112	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
113	Absence of Cardiovascular Manifestations in a Haploinsufficient Tgfbr1 Mouse Model. PLoS ONE, 2014, 9, e89749.	1.1	9
114	VariantDB: a flexible annotation and filtering portal for next generation sequencing data. Genome Medicine, 2014, 6, 74.	3.6	60
115	Response to Pyeritz et al.. Genetics in Medicine, 2014, 16, 642-644.	1.1	3
116	Transforming Growth Factor Beta and Bone. , 2014, , 211-216.		0
117	Atenolol versus Losartan in Children and Young Adults with Marfan's Syndrome. New England Journal of Medicine, 2014, 371, 2061-2071.	13.9	457
118	Loeys-Dietz Syndrome. Advances in Experimental Medicine and Biology, 2014, 802, 95-105.	0.8	106
119	3q27.3 microdeletional syndrome: a recognisable clinical entity associating dysmorphic features, marfanoid habitus, intellectual disability and psychosis with mood disorder. Journal of Medical Genetics, 2014, 51, 21-27.	1.5	12
120	A Dominant-NegativeGFI1BMutation in the Gray Platelet Syndrome. New England Journal of Medicine, 2014, 370, 245-253.	13.9	152
121	Loeysâ€“Dietz syndrome: a primer for diagnosis and management. Genetics in Medicine, 2014, 16, 576-587.	1.1	435
122	Intermittent Brugada syndrome in an anorexic adolescent girl. Journal of Cardiology Cases, 2014, 10, 81-84.	0.2	1
123	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
124	Bladder exstrophyâ€“epispadias complex and tripleâ€X syndrome: Incidental finding or causality?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 797-800.	1.6	1
125	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
126	Congenital glucoseâ€“galactose malabsorption: a novel deletion within the SLC5A1 gene. European Journal of Pediatrics, 2013, 172, 409-411.	1.3	15

#	Article	IF	Citations
127	Genetic variations in toll-like receptor pathway and lung function decline in Cystic Fibrosis patients. Human Immunology, 2013, 74, 1649-1655.	1.2	16
128	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
129	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
130	Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Human Molecular Genetics, 2013, 22, 2590-2602.	1.4	103
131	Educational paper. European Journal of Pediatrics, 2013, 172, 997-1005.	1.3	26
132	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
133	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
134	Genetics of Thoracic Aortic Aneurysm. Circulation Research, 2013, 113, 327-340.	2.0	151
135	A Dominant-Negative GFI1B Mutation in Gray Platelet Syndrome. Blood, 2013, 122, LBA-3-LBA-3.	0.6	1
136	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
137	Nasal speech in patients with 12q15 microdeletions. European Journal of Human Genetics, 2012, 20, 367-367.	1.4	0
138	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
139	Marfan syndrome. Current Opinion in Pediatrics, 2012, 24, 498-504.	1.0	53
140	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nature Genetics, 2012, 44, 922-927.	9.4	391
141	Phenotypic spectrum of the SMAD3-related aneurysmsâ€“osteoarthritis syndrome. Journal of Medical Genetics, 2012, 49, 47-57.	1.5	221
142	Mutations in the TGF-Î² repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. Nature Genetics, 2012, 44, 1249-1254.	9.4	237
143	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
144	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

#	Article	IF	Citations
145	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
146	Loeys-Dietz syndrome: A possible solution for Akhenatenâ€™s and his familyâ€™s mystery syndrome. South African Medical Journal, 2012, 102, 661.	0.2	3
147	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
148	The new Ghent criteria for Marfan syndrome: what do they change?. Clinical Genetics, 2012, 81, 433-442.	1.0	90
149	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
150	Preventing the aortic complications of Marfan syndrome: a caseâ€example of translational genomic medicine. British Journal of Clinical Pharmacology, 2011, 72, 6-17.	1.1	7
151	Nasal speech and hypothyroidism are common hallmarks of 12q15 microdeletions. European Journal of Human Genetics, 2011, 19, 1032-1037.	1.4	11
152	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
153	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
154	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
155	New insights into the pathogenesis of autosomalâ€dominant cutis laxa with report of fiveELNmutations. Human Mutation, 2011, 32, 445-455.	1.1	116
156	Applying massive parallel sequencing to molecular diagnosis of Marfan and Loeys-Dietz syndromes. Human Mutation, 2011, 32, 1053-1062.	1.1	71
157	Noncanonical TGFÎ² Signaling Contributes to Aortic Aneurysm Progression in Marfan Syndrome Mice. Science, 2011, 332, 358-361.	6.0	422
158	What Is New in Dilatation of the Ascending Aorta?. Circulation, 2011, 123, 924-928.	1.6	72
159	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
160	The Loeysâ€“Dietz syndrome: an update for the clinician. Current Opinion in Cardiology, 2010, 25, 546-551.	0.8	121
161	Musculoskeletal Findings of Loeys-Dietz Syndrome. Journal of Bone and Joint Surgery - Series A, 2010, 92, 1876-1883.	1.4	66
162	The diagnostic value of the facial features of Marfan syndrome. Journal of Children's Orthopaedics, 2010, 4, 545-551.	0.4	15

#	Article	IF	Citations
163	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
164	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
165	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
166	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
167	Renal insufficiency, a frequent complication with age in oralâ€facialâ€digital syndrome type I. Clinical Genetics, 2010, 77, 258-265.	1.0	29
168	Mutations in Fibrillin-1 Cause Congenital Scleroderma: Stiff Skin Syndrome. Science Translational Medicine, 2010, 2, 23ra20.	5.8	195
169	Short stature, severe aortic root dilation, skin hyperextensibility, extreme joint laxity and craniofacial dysmorphic features: a probable new syndrome. Clinical Dysmorphology, 2010, 19, 119-122.	0.1	0
170	Resolving in-vivo flow fields in the systemic circulation of the mouse through combined ultrasound imaging and computational fluid dynamics. , 2010, , .		0
171	Cardiovascular manifestations in men and women carrying a FBN1 mutation. European Heart Journal, 2010, 31, 2223-2229.	1.0	133
172	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
173	Expanding the phenotype of sudden cardiac deathâ€”An unusual presentation of a family with a Lamin A/C mutation. International Journal of Cardiology, 2010, 138, 97-99.	0.8	8
174	The revised Ghent nosology for the Marfan syndrome. Journal of Medical Genetics, 2010, 47, 476-485.	1.5	1,677
175	A Turkish patient of typical Loeys-Dietz syndrome with a TGFBR2 mutation. Genetic Counseling, 2010, 21, 225-32.	0.1	2
176	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
177	Circulating Transforming Growth Factor-Î² in Marfan Syndrome. Circulation, 2009, 120, 526-532.	1.6	246
178	Neuroradiologic Manifestations of Loeys-Dietz Syndrome Type 1. American Journal of Neuroradiology, 2009, 30, 1614-1619.	1.2	104
179	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
180	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

#	Article	IF	Citations
181	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
182	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants ofCOH1. Human Mutation, 2009, 30, E404-E420.	1.1	44
183	COL5A1 signal peptide mutations interfere with protein secretion and cause classic Ehlers-Danlos syndrome. Human Mutation, 2009, 30, E395-E403.	1.1	57
184	PORCNmutations in focal dermal hypoplasia: coping with lethality. Human Mutation, 2009, 30, E618-E628.	1.1	64
185	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
186	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
187	Unusual 8p inverted duplication deletion with telomere capture from 8q. European Journal of Medical Genetics, 2009, 52, 31-36.	0.7	26
188	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
189	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
190	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
191	Identification of copy number variants associated with BPES-like phenotypes. Human Genetics, 2008, 124, 489-498.	1.8	15
192	Absence of arterial phenotype in mice with homozygous slc2A10 missense substitutions. Genesis, 2008, 46, 385-389.	0.8	30
193	Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. Human Mutation, 2008, 29, 150-158.	1.1	295
194	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
195	Ehlers-Danlos syndromes and Marfan syndrome. Best Practice and Research in Clinical Rheumatology, 2008, 22, 165-189.	1.4	205
196	Aortic dissection in a young man with Loeysâ€“Dietz syndrome. Journal of Thoracic and Cardiovascular Surgery, 2008, 135, 1174-1175.e1.	0.4	11
197	Angiotensin II Blockade and Aortic-Root Dilation in Marfan's Syndrome. New England Journal of Medicine, 2008, 358, 2787-2795.	13.9	767
198	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

#	Article	IF	Citations
199	Loeys-Dietz Syndrome: MDCT Angiography Findings. American Journal of Roentgenology, 2007, 189, W29-W35.	1.0	74
200	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
201	Overall intelligibility, language, articulation, voice and resonance characteristics in a child with Shprintzenâ€“Goldberg syndrome. International Journal of Pediatric Otorhinolaryngology, 2007, 71, 721-728.	0.4	9
202	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
203	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
204	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 552 Td (a 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
205	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
206	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
207	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
208	Utility of molecular analyses in the exploration of extreme intrafamilial variability in the Marfan syndrome. Clinical Genetics, 2007, 72, 188-198.	1.0	47
209	New insights into the pathogenesis and treatment of arterial aneurysms and dissections. Current Cardiovascular Risk Reports, 2007, 1, 401-409.	0.8	3
210	Infantile Restrictive Cardiomyopathy Resulting From a Mutation in the Cardiac Troponin T Gene. Pediatrics, 2006, 117, 1830-1833.	1.0	68
211	Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. Nature Genetics, 2006, 38, 452-457.	9.4	354
212	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
213	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
214	The molecular genetics of Marfan syndrome and related disorders. Journal of Medical Genetics, 2006, 43, 769-787.	1.5	347
215	Losartan, an AT1 Antagonist, Prevents Aortic Aneurysm in a Mouse Model of Marfan Syndrome. Science, 2006, 312, 117-121.	6.0	1,591
216	Fibulin-5 mutations: mechanisms of impaired elastic fiber formation in recessive cutis laxa. Human Molecular Genetics, 2006, 15, 3379-3386.	1.4	84

#	Article	IF	Citations
217	Aneurysm Syndromes Caused by Mutations in the TGF-Î² Receptor. New England Journal of Medicine, 2006, 355, 788-798.	13.9	1,490
218	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
219	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
220	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
221	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
222	Mutations ofVMD2Splicing Regulators Cause Nanophthalmos and Autosomal Dominant Vitreoretinochoroidopathy (ADVIRC). , 2004, 45, 3683.		205
223	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
224	DUP25 remains unconfirmed. American Journal of Medical Genetics Part A, 2004, 131A, 320-321.	2.4	4
225	Comprehensive molecular screening of theFBN1gene favors locus homogeneity of classical Marfan syndrome. Human Mutation, 2004, 24, 140-146.	1.1	210
226	Update of the UMD-FBN1mutation database and creation of anFBN1polymorphism database. Human Mutation, 2003, 22, 199-208.	1.1	299
227	GENETIC FIBRILLINOPATHIES : NEW INSIGHTS IN MOLECULAR DIAGNOSIS AND CLINICAL MANAGEMENT. Acta Clinica Belgica, 2003, 58, 3-11.	0.5	23
228	Cutis laxa of the autosomal recessive type in a consanguineous family. European Journal of Dermatology, 2003, 13, 529-33.	0.3	17
229	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
230	Linear and whorled nevoid hypermelanosis with bilateral giant cerebral aneurysms. American Journal of Medical Genetics Part A, 2002, 112, 95-98.	2.4	9
231	Does monosymptomatic enuresis exist? A molecular genetic exploration of 32 families with enuresis/incontinence. BJU International, 2002, 90, 76-83.	1.3	50
232	Mutation of TBCE causes hypoparathyroidismâ€“Âretardationâ€“dysmorphism and autosomal recessive Kennyâ€“Caffey syndrome. Nature Genetics, 2002, 32, 448-452.	9.4	248
233	Clinical and electrophysiological findings in autosomal dominant vitreoretinochoroidopathy: report of a new pedigree. , 2001, 239, 575-582.		36
234	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

#	Article	IF	Citations
235	Fungal intracranial aneurysm in a child with familial chronic mucocutaneous candidiasis. European Journal of Pediatrics, 1999, 158, 650-652.	1.3	23
236	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
237	Bruck syndrome: neonatal presentation and natural course in three patients. Pediatric Radiology, 1998, 28, 781-789.	1.1	22
238	Connective Tissue Disorders. , 0, , 537-546.		0