Bert Callewaert

List of Publications by Year in descending order

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91 papers	7,714 citations	94433 37 h-index	53230 85 g-index
96 all docs	96 docs citations	96 times ranked	8670 citing authors

#	Article	IF	Citations
1	The revised Ghent nosology for the Marfan syndrome. Journal of Medical Genetics, 2010, 47, 476-485.	3.2	1,677
2	Aneurysm Syndromes Caused by Mutations in the TGF-Î² Receptor. New England Journal of Medicine, 2006, 355, 788-798.	27.0	1,490
3	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.	6.2	485
4	Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. Nature Genetics, 2006, 38, 452-457.	21.4	354
5	Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. Human Mutation, 2008, 29, 150-158.	2.5	295
6	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
7	Ehlers-Danlos syndromes and Marfan syndrome. Best Practice and Research in Clinical Rheumatology, 2008, 22, 165-189.	3.3	205
8	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.	2.1	146
9	Novel MYH11 and ACTA2 mutations reveal a role for enhanced TGFÎ² signaling in FTAAD. International Journal of Cardiology, 2013, 165, 314-321.	1.7	134
10	Comprehensive molecular analysis demonstrates type V collagen mutations in over 90% of patients with classic EDS and allows to refine diagnostic criteria. Human Mutation, 2012, 33, 1485-1493.	2.5	133
11	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.	2.8	132
12	New insights into the pathogenesis of autosomalâ€dominant cutis laxa with report of fiveELNmutations. Human Mutation, 2011, 32, 445-455.	2.5	116
13	Deficiency for the ER-stress transducer OASIS causes severe recessive osteogenesis imperfecta in humans. Orphanet Journal of Rare Diseases, 2013, 8, 154.	2.7	98
14	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	6.2	95
15	The new Ghent criteria for Marfan syndrome: what do they change?. Clinical Genetics, 2012, 81, 433-442.	2.0	90
16	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.	3.2	83
17	BATCH-GE: Batch analysis of Next-Generation Sequencing data for genome editing assessment. Scientific Reports, 2016, 6, 30330.	3.3	82
18	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82

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19	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.	2.5	81
20	CRISPR/Cas9-mediated homology-directed repair by ssODNs in zebrafish induces complex mutational patterns resulting from genomic integration of repair-template fragments. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	77
21	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. American Journal of Human Genetics, 2015, 97, 483-492.	6.2	70
22	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	2.5	67
23	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
24	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.	2.8	66
25	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	2.4	66
26	Gene panel sequencing in heritable thoracic aortic disorders and related entities â€“ results of comprehensive testing in a cohort of 264 patients. Orphanet Journal of Rare Diseases, 2015, 10, 9.	2.7	62
27	Thoracic aortic-aneurysm and dissection in association with significant mitral valve disease caused by mutations in TGFB2. International Journal of Cardiology, 2013, 165, 584-587.	1.7	58
28	Oto-facial syndrome and esophageal atresia, intellectual disability and zygomatic anomalies - expanding the phenotypes associated with EFTUD2 mutations. Orphanet Journal of Rare Diseases, 2013, 8, 110.	2.7	56
29	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. Science Advances, 2020, 6, eaax0021.	10.3	56
30	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
31	GLUT10 is required for the development of the cardiovascular system and the notochord and connects mitochondrial function to TGFÎ² signaling. Human Molecular Genetics, 2012, 21, 1248-1259.	2.9	52
32	The Genetics of Soft Connective Tissue Disorders. Annual Review of Genomics and Human Genetics, 2015, 16, 229-255.	6.2	50
33	The diagnostic value of next generation sequencing in familial nonsyndromic congenital heart defects. American Journal of Medical Genetics, Part A, 2015, 167, 1822-1829.	1.2	48
34	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
35	Extensive clinical, hormonal and genetic screening in a large consecutive series of 46,XY neonates and infants with atypical sexual development. Orphanet Journal of Rare Diseases, 2014, 9, 209.	2.7	44
36	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43

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37	Neonatal progeroid variant of Marfan syndrome with congenital lipodystrophy results from mutations at the 3â€² end of FBN1 gene. European Journal of Medical Genetics, 2014, 57, 230-234.	1.3	41
38	Severe congenital cutis laxa with cardiovascular manifestations due to homozygous deletions in ALDH18A1. Molecular Genetics and Metabolism, 2014, 112, 310-316.	1.1	41
39	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.	1.2	40
40	A novel ADAMTS17 variant that causes Weill-Marchesani syndrome 4 alters fibrillin-1 and collagen type I deposition in the extracellular matrix. Matrix Biology, 2020, 88, 1-18.	3.6	35
41	Twenty patients including 7 probands with autosomal dominant cutis laxa confirm clinical and molecular homogeneity. Orphanet Journal of Rare Diseases, 2013, 8, 36.	2.7	33
42	Absence of arterial phenotype in mice with homozygous slc2A10 missense substitutions. Genesis, 2008, 46, 385-389.	1.6	30
43	Cutis laxa: A comprehensive overview of clinical characteristics and pathophysiology. Clinical Genetics, 2021, 99, 53-66.	2.0	29
44	Profiling of conserved non-coding elements upstream of SHOX and functional characterisation of the SHOX cis-regulatory landscape. Scientific Reports, 2016, 5, 17667.	3.3	27
45	Unusual 8p inverted duplication deletion with telomere capture from 8q. European Journal of Medical Genetics, 2009, 52, 31-36.	1.3	26
46	Genetic analysis of osteogenesis imperfecta in the <scp>P</scp>alestinian population: molecular screening of 49 affected families. Molecular Genetics & Genomic Medicine, 2018, 6, 15-26.	1.2	26
47	Defining the Clinical, Molecular and Ultrastructural Characteristics in Occipital Horn Syndrome: Two New Cases and Review of the Literature. Genes, 2019, 10, 528.	2.4	23
48	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	1.2	22
49	Tailoring the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines for the Interpretation of Sequenced Variants in the FBN1 Gene for Marfan Syndrome. Circulation Genomic and Precision Medicine, 2018, 11, e002039.	3.6	20
50	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
51	Exome sequencing identifies ZFPM2 as a cause of familial isolated congenital diaphragmatic hernia and possibly cardiovascular malformations. European Journal of Medical Genetics, 2014, 57, 247-252.	1.3	17
52	Severe congenital neutropenia with neurological impairment due to a homozygous VPS45 p.E238K mutation: A case report suggesting a genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2015, 167, 3214-3218.	1.2	17
53	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	2.4	17
54	Myhre syndrome: A first familial recurrence and broadening of the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 2494-2499.	1.2	16

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55	Confirmation of an ARID2 defect in SWI/SNFâ€related intellectual disability. American Journal of Medical Genetics, Part A, 2017, 173, 3104-3108.	1.2	15
56	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
57	<scp>ATP</scp>6V0A2â€related cutis laxa in 10 novel patients: Focus on clinical variability and expansion of the phenotype. Experimental Dermatology, 2019, 28, 1142-1145.	2.9	14
58	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	3.8	14
59	Marfan Syndrome and Related Heritable Thoracic Aortic Aneurysms and Dissections. Current Pharmaceutical Design, 2015, 21, 4061-4075.	1.9	13
60	Arterial Tortuosity Syndrome: An Ascorbate Compartmentalization Disorder?. Antioxidants and Redox Signaling, 2021, 34, 875-889.	5.4	11
61	<scp>IQSEC2</scp> disorder: A new disease entity or a Rett spectrum continuum?. Clinical Genetics, 2021, 99, 462-474.	2.0	11
62	Small patella syndrome: New clinical and molecular insights into a consistent phenotype. Clinical Genetics, 2017, 92, 676-678.	2.0	10
63	Absence of Cardiovascular Manifestations in a Haploinsufficient Tgfbr1 Mouse Model. PLoS ONE, 2014, 9, e89749.	2.5	9
64	Ophthalmic findings in patients with arterial tortuosity syndrome and carriers: A case series. Ophthalmic Genetics, 2018, 39, 29-34.	1.2	9
65	RIN2 syndrome: Expanding the clinical phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2408-2415.	1.2	8
66	A novel case of autosomal dominant cutis laxa in a consanguineous family: report and literature review. Clinical Dysmorphology, 2017, 26, 142-147.	0.3	7
67	Imaging in cutis laxa syndrome caused by a dominant negative ALDH18A1 mutation, with hypotheses for intracranial vascular tortuosity andÂwide perivascular spaces. European Journal of Paediatric Neurology, 2017, 21, 912-920.	1.6	6
68	Expanding the molecular spectrum and the neurological phenotype related to <scp>CAMTA1</scp> variants. Clinical Genetics, 2021, 99, 259-268.	2.0	6
69	Loss-of-Function Variants in EFEMP1 Cause a Recognizable Connective Tissue Disorder Characterized by Cutis Laxa and Multiple Herniations. Genes, 2021, 12, 510.	2.4	6
70	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. Genes, 2021, 12, 1048.	2.4	6
71	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
72	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	2.0	5

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73	Slc2a10 knock-out mice deficient in ascorbic acid synthesis recapitulate aspects of arterial tortuosity syndrome and display mitochondrial respiration defects. Human Molecular Genetics, 2020, 29, 1476-1488.	2.9	5
74	New insights into the molecular diagnosis and management of heritable thoracic aortic aneurysms and dissections. Polish Archives of Internal Medicine, 2013, 123, 693-700.	0.4	5
75	LTBP1 promotes fibrillin incorporation into the extracellular matrix. Matrix Biology, 2022, 110, 60-75.	3.6	5
76	Clinical and Molecular Delineation of Cutis Laxa Syndromes: Paradigms for Elastic Fiber Homeostasis. Advances in Experimental Medicine and Biology, 2021, 1348, 273-309.	1.6	4
77	Identification of Codon 146 KRAS Variants in Isolated Epidermal Nevus and Multiple Lesions in Oculoectodermal Syndrome: Confirmation of the Phenotypic Continuum of Mosaic RASopathies. International Journal of Molecular Sciences, 2022, 23, 4036.	4.1	4
78	Exploring the Mutational Landscape of Isolated Congenital Heart Defects: An Exome Sequencing Study Using Cardiac DNA. Genes, 2022, 13, 1214.	2.4	4
79	Genes in Thoracic Aortic Aneurysms and Dissections - Do they Matter?: Translation and Integration of Research and Modern Genetic Techniques into Daily Clinical Practice. Aorta, 2013, 1, 135-145.	0.5	3
80	Severe congenital cutis laxa: Identification of novel homozygous <scp>LOX</scp> gene variants in two families. Clinical Genetics, 2021, 100, 168-175.	2.0	3
81	Loss of zebrafish atp6v1e1b, encoding a subunit of vacuolar ATPase, recapitulates human ARCL type 2C syndrome and identifies multiple pathobiological signatures. PLoS Genetics, 2021, 17, e1009603.	3.5	3
82	Major response to adalimumab in patient with Sweet syndrome associated to an acquired cutis laxa. Journal of the European Academy of Dermatology and Venereology, 2022, 36, .	2.4	3
83	Expanding the Phenotype of B3GALNT2-Related Disorders. Genes, 2022, 13, 694.	2.4	3
84	Apoptotic enteropathy, gluten intolerance, and IBD-like inflammation associated with lipotoxicity in DGAT1 deficiencyâ€“related diarrhea: a case report of a 17-year-old patient and literature review. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 0, , .	2.8	3
85	Fortschritt in der Pathogenese des Marfan-Syndroms und verwandter Krankheiten. Medizinische Genetik, 2008, 20, 6-17.	0.2	2
86	Disproportion and dysmorphism in an adult Belgian population with Turner syndrome: risk factors for chronic diseases?. Acta Clinica Belgica, 2020, 75, 258-266.	1.2	2
87	New insights on the clinical variability of FKBP10 mutations. European Journal of Medical Genetics, 2020, 63, 103980.	1.3	2
88	Expanded cardiovascular phenotype of Myhre syndrome includes tetralogy of Fallot suggesting a role for <scp>SMAD4</scp> in human neural crest defects. American Journal of Medical Genetics, Part A, 2022, 188, 1384-1395.	1.2	2
89	Expanding the phenotypic spectrum of ALDH18A1-related autosomal recessive cutis laxa with a description of novel neuroradiological findings. Clinical Dysmorphology, 2022, 31, 66-70.	0.3	1
90	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.	0.3	0

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91	Lack of resemblance between Myhre syndrome and other â€œsegmental progeroidâ€•syndromes warrants restraint in applying this classification. GeroScience, 2021, 43, 459-461.	4.6	0