

# Bert Callewaert

## List of Publications by Year in descending order

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Version: 2024-02-01

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	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. <i>Human Genetics</i> , 2022, 141, 65-80.	3.8	14
2	Expanded cardiovascular phenotype of Myhre syndrome includes tetralogy of Fallot suggesting a role for <i>SMAD4</i> in human neural crest defects. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1384-1395.	1.2	2
3	Identification of Codon 146 KRAS Variants in Isolated Epidermal Nevus and Multiple Lesions in Oculofacial Syndrome: Confirmation of the Phenotypic Continuum of Mosaic RASopathies. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4036.	4.1	4
4	Major response to adalimumab in patient with Sweet syndrome associated to an acquired cutis laxa. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2022, 36, .	2.4	3
5	Expanding the phenotypic spectrum of ALDH18A1-related autosomal recessive cutis laxa with a description of novel neuroradiological findings. <i>Clinical Dysmorphology</i> , 2022, 31, 66-70.	0.3	1
6	Expanding the Phenotype of B3GALNT2-Related Disorders. <i>Genes</i> , 2022, 13, 694.	2.4	3
7	LTBP1 promotes fibrillin incorporation into the extracellular matrix. <i>Matrix Biology</i> , 2022, 110, 60-75.	3.6	5
8	Biallelic variants in <i>ZNF142</i> lead to a syndromic neurodevelopmental disorder. <i>Clinical Genetics</i> , 2022, 102, 98-109.	2.0	6
9	Exploring the Mutational Landscape of Isolated Congenital Heart Defects: An Exome Sequencing Study Using Cardiac DNA. <i>Genes</i> , 2022, 13, 1214.	2.4	4
10	Cutis laxa: A comprehensive overview of clinical characteristics and pathophysiology. <i>Clinical Genetics</i> , 2021, 99, 53-66.	2.0	29
11	Expanding the molecular spectrum and the neurological phenotype related to <i>CAMTA1</i> variants. <i>Clinical Genetics</i> , 2021, 99, 259-268.	2.0	6
12	Arterial Tortuosity Syndrome: An Ascorbate Compartmentalization Disorder?. <i>Antioxidants and Redox Signaling</i> , 2021, 34, 875-889.	5.4	11
13	<i>IQSEC2</i> disorder: A new disease entity or a Rett spectrum continuum?. <i>Clinical Genetics</i> , 2021, 99, 462-474.	2.0	11
14	Lack of resemblance between Myhre syndrome and other "segmental progeroid" syndromes warrants restraint in applying this classification. <i>GeroScience</i> , 2021, 43, 459-461.	4.6	0
15	Loss-of-Function Variants in EFEMP1 Cause a Recognizable Connective Tissue Disorder Characterized by Cutis Laxa and Multiple Herniations. <i>Genes</i> , 2021, 12, 510.	2.4	6
16	Severe congenital cutis laxa: Identification of novel homozygous <i>LOX</i> gene variants in two families. <i>Clinical Genetics</i> , 2021, 100, 168-175.	2.0	3
17	Loss of zebrafish <i>atp6v1e1b</i> , encoding a subunit of vacuolar ATPase, recapitulates human ARCL type 2C syndrome and identifies multiple pathobiological signatures. <i>PLoS Genetics</i> , 2021, 17, e1009603.	3.5	3
18	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 1095-1114.	6.2	19

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19	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. <i>Genes</i> , 2021, 12, 1048.	2.4	6
20	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. <i>Journal of Experimental Medicine</i> , 2021, 218, .	8.5	43
21	Clinical and Molecular Delineation of Cutis Laxa Syndromes: Paradigms for Elastic Fiber Homeostasis. <i>Advances in Experimental Medicine and Biology</i> , 2021, 1348, 273-309.	1.6	4
22	Disproportion and dysmorphism in an adult Belgian population with Turner syndrome: risk factors for chronic diseases?. <i>Acta Clinica Belgica</i> , 2020, 75, 258-266.	1.2	2
23	A clinical scoring system for congenital contractural arachnodactyly. <i>Genetics in Medicine</i> , 2020, 22, 124-131.	2.4	17
24	A novel ADAMTS17 variant that causes Weill-Marchesani syndrome 4 alters fibrillin-1 and collagen type I deposition in the extracellular matrix. <i>Matrix Biology</i> , 2020, 88, 1-18.	3.6	35
25	Refinement of the clinical and mutational spectrum of <sc>UBE2A</sc> deficiency syndrome. <i>Clinical Genetics</i> , 2020, 98, 172-178.	2.0	5
26	New insights on the clinical variability of FKBP10 mutations. <i>European Journal of Medical Genetics</i> , 2020, 63, 103980.	1.3	2
27	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. <i>Science Advances</i> , 2020, 6, eaax0021.	10.3	56
28	Slc2a10 knock-out mice deficient in ascorbic acid synthesis recapitulate aspects of arterial tortuosity syndrome and display mitochondrial respiration defects. <i>Human Molecular Genetics</i> , 2020, 29, 1476-1488.	2.9	5
29	<i><sc>ATP</sc>6VOA2</i>-related cutis laxa in 10 novel patients: Focus on clinical variability and expansion of the phenotype. <i>Experimental Dermatology</i> , 2019, 28, 1142-1145.	2.9	14
30	Defining the Clinical, Molecular and Ultrastructural Characteristics in Occipital Horn Syndrome: Two New Cases and Review of the Literature. <i>Genes</i> , 2019, 10, 528.	2.4	23
31	Myhre syndrome: A first familial recurrence and broadening of the phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2494-2499.	1.2	16
32	Arterial tortuosity syndrome: 40 new families and literature review. <i>Genetics in Medicine</i> , 2018, 20, 1236-1245.	2.4	66
33	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. <i>Genetics in Medicine</i> , 2018, 20, 965-975.	2.4	67
34	Ophthalmic findings in patients with arterial tortuosity syndrome and carriers: A case series. <i>Ophthalmic Genetics</i> , 2018, 39, 29-34.	1.2	9
35	Genetic analysis of osteogenesis imperfecta in the <sc>P</sc>alestinian population: molecular screening of 49 affected families. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 15-26.	1.2	26
36	CRISPR/Cas9-mediated homology-directed repair by ssODNs in zebrafish induces complex mutational patterns resulting from genomic integration of repair-template fragments. <i>DMM Disease Models and Mechanisms</i> , 2018, 11, .	2.4	77

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37	Tailoring the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines for the Interpretation of Sequenced Variants in the <i>FBN1</i> Gene for Marfan Syndrome. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002039.	3.6	20
38	Mutations in <i>ATP6V1E1</i> or <i>ATP6V1A</i> Cause Autosomal-Recessive Cutis Laxa. <i>American Journal of Human Genetics</i> , 2017, 100, 216-227.	6.2	82
39	A novel case of autosomal dominant cutis laxa in a consanguineous family: report and literature review. <i>Clinical Dysmorphology</i> , 2017, 26, 142-147.	0.3	7
40	Confirmation of an <i>ARID2</i> defect in SWI/SNF-related intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3104-3108.	1.2	15
41	Imaging in cutis laxa syndrome caused by a dominant negative <i>ALDH18A1</i> mutation, with hypotheses for intracranial vascular tortuosity and wide perivascular spaces. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 912-920.	1.6	6
42	Small patella syndrome: New clinical and molecular insights into a consistent phenotype. <i>Clinical Genetics</i> , 2017, 92, 676-678.	2.0	10
43	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. <i>Genetics in Medicine</i> , 2017, 19, 691-700.	2.4	45
44	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	21.4	251
45	<i>GLUT10</i> Lacking in Arterial Tortuosity Syndrome Is Localized to the Endoplasmic Reticulum of Human Fibroblasts. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1820.	4.1	15
46	RIN2 syndrome: Expanding the clinical phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2408-2415.	1.2	8
47	Profiling of conserved non-coding elements upstream of <i>SHOX</i> and functional characterisation of the <i>SHOX</i> cis-regulatory landscape. <i>Scientific Reports</i> , 2016, 5, 17667.	3.3	27
48	BATCH-GE: Batch analysis of Next-Generation Sequencing data for genome editing assessment. <i>Scientific Reports</i> , 2016, 6, 30330.	3.3	82
49	Recurrent duplications of 17q12 associated with variable phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3038-3045.	1.2	22
50	Severe congenital neutropenia with neurological impairment due to a homozygous <i>VPS45</i> p.E238K mutation: A case report suggesting a genotype-phenotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3214-3218.	1.2	17
51	The Genetics of Soft Connective Tissue Disorders. <i>Annual Review of Genomics and Human Genetics</i> , 2015, 16, 229-255.	6.2	50
52	Gene panel sequencing in heritable thoracic aortic disorders and related entities – results of comprehensive testing in a cohort of 264 patients. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 9.	2.7	62
53	Redefining the MED13L syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1308-1317.	2.8	53
54	The diagnostic value of next generation sequencing in familial nonsyndromic congenital heart defects. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1822-1829.	1.2	48

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55	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. <i>American Journal of Human Genetics</i> , 2015, 97, 483-492.	6.2	70
56	Marfan Syndrome and Related Heritable Thoracic Aortic Aneurysms and Dissections. <i>Current Pharmaceutical Design</i> , 2015, 21, 4061-4075.	1.9	13
57	Absence of Cardiovascular Manifestations in a Haploinsufficient Tgfbr1 Mouse Model. <i>PLoS ONE</i> , 2014, 9, e89749.	2.5	9
58	Extensive clinical, hormonal and genetic screening in a large consecutive series of 46,XY neonates and infants with atypical sexual development. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 209.	2.7	44
59	Neonatal progeroid variant of Marfan syndrome with congenital lipodystrophy results from mutations at the 3' end of FBN1 gene. <i>European Journal of Medical Genetics</i> , 2014, 57, 230-234.	1.3	41
60	Exome sequencing identifies ZFPM2 as a cause of familial isolated congenital diaphragmatic hernia and possibly cardiovascular malformations. <i>European Journal of Medical Genetics</i> , 2014, 57, 247-252.	1.3	17
61	Severe congenital cutis laxa with cardiovascular manifestations due to homozygous deletions in ALDH18A1. <i>Molecular Genetics and Metabolism</i> , 2014, 112, 310-316.	1.1	41
62	Oto-facial syndrome and esophageal atresia, intellectual disability and zygomatic anomalies - expanding the phenotypes associated with EFTUD2 mutations. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 110.	2.7	56
63	Twenty patients including 7 probands with autosomal dominant cutis laxa confirm clinical and molecular homogeneity. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 36.	2.7	33
64	Deficiency for the ER-stress transducer OASIS causes severe recessive osteogenesis imperfecta in humans. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 154.	2.7	98
65	Novel MYH11 and ACTA2 mutations reveal a role for enhanced TGF $\beta$ <sup>2</sup> signaling in FTAAD. <i>International Journal of Cardiology</i> , 2013, 165, 314-321.	1.7	134
66	Thoracic aortic-aneurysm and dissection in association with significant mitral valve disease caused by mutations in TGFB2. <i>International Journal of Cardiology</i> , 2013, 165, 584-587.	1.7	58
67	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. <i>Human Mutation</i> , 2013, 34, 111-121.	2.5	67
68	Genes in Thoracic Aortic Aneurysms and Dissections - Do they Matter?: Translation and Integration of Research and Modern Genetic Techniques into Daily Clinical Practice. <i>Aorta</i> , 2013, 1, 135-145.	0.5	3
69	New insights into the molecular diagnosis and management of heritable thoracic aortic aneurysms and dissections. <i>Polish Archives of Internal Medicine</i> , 2013, 123, 693-700.	0.4	5
70	GLUT10 is required for the development of the cardiovascular system and the notochord and connects mitochondrial function to TGF $\beta$ <sup>2</sup> signaling. <i>Human Molecular Genetics</i> , 2012, 21, 1248-1259.	2.9	52
71	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 950-957.	6.2	95
72	Comprehensive molecular analysis demonstrates type V collagen mutations in over 90% of patients with classic EDS and allows to refine diagnostic criteria. <i>Human Mutation</i> , 2012, 33, 1485-1493.	2.5	133

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73	The new Ghent criteria for Marfan syndrome: what do they change?. <i>Clinical Genetics</i> , 2012, 81, 433-442.	2.0	90
74	New insights into the pathogenesis of autosomal dominant cutis laxa with report of five <i>ELN</i> mutations. <i>Human Mutation</i> , 2011, 32, 445-455.	2.5	116
75	Altered TGF $\beta$ signaling and cardiovascular manifestations in patients with autosomal recessive cutis laxa type I caused by fibulin-4 deficiency. <i>European Journal of Human Genetics</i> , 2010, 18, 895-901.	2.8	132
76	Short stature, severe aortic root dilation, skin hyperextensibility, extreme joint laxity and craniofacial dysmorphic features: a probable new syndrome. <i>Clinical Dysmorphology</i> , 2010, 19, 119-122.	0.3	0
77	The revised Ghent nosology for the Marfan syndrome. <i>Journal of Medical Genetics</i> , 2010, 47, 476-485.	3.2	1,677
78	Clinical and Molecular Study of 320 Children With Marfan Syndrome and Related Type I Fibrillinopathies in a Series of 1009 Proband With Pathogenic <i>FBN1</i> Mutations. <i>Pediatrics</i> , 2009, 123, 391-398.	2.1	146
79	Pathogenic <i>FBN1</i> 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. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 854-860.	1.2	40
80	Comprehensive clinical and molecular assessment of 32 probands with congenital contractural arachnodactyly: Report of 14 novel mutations and review of the literature. <i>Human Mutation</i> , 2009, 30, 334-341.	2.5	81
81	Clinical and mutation-type analysis from an international series of 198 probands with a pathogenic <i>FBN1</i> exons 24-32 mutation. <i>European Journal of Human Genetics</i> , 2009, 17, 491-501.	2.8	66
82	Unusual 8p inverted duplication deletion with telomere capture from 8q. <i>European Journal of Medical Genetics</i> , 2009, 52, 31-36.	1.3	26
83	Fortschritt in der Pathogenese des Marfan-Syndroms und verwandter Krankheiten. <i>Medizinische Genetik</i> , 2008, 20, 6-17.	0.2	2
84	Absence of arterial phenotype in mice with homozygous <i>slc2A10</i> missense substitutions. <i>Genesis</i> , 2008, 46, 385-389.	1.6	30
85	Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. <i>Human Mutation</i> , 2008, 29, 150-158.	2.5	295
86	Ehlers-Danlos syndromes and Marfan syndrome. <i>Best Practice and Research in Clinical Rheumatology</i> , 2008, 22, 165-189.	3.3	205
87	Contribution of molecular analyses in diagnosing Marfan syndrome and type I fibrillinopathies: an international study of 1009 probands. <i>Journal of Medical Genetics</i> , 2008, 45, 384-390.	3.2	83
88	Effect of Mutation Type and Location on Clinical Outcome in 1,013 Proband with Marfan Syndrome or Related Phenotypes and <i>FBN1</i> Mutations: An International Study. <i>American Journal of Human Genetics</i> , 2007, 81, 454-466.	6.2	485
89	Mutations in the facilitative glucose transporter <i>GLUT10</i> alter angiogenesis and cause arterial tortuosity syndrome. <i>Nature Genetics</i> , 2006, 38, 452-457.	21.4	354
90	Aneurysm Syndromes Caused by Mutations in the TGF $\beta$ Receptor. <i>New England Journal of Medicine</i> , 2006, 355, 788-798.	27.0	1,490

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91	Apoptotic enteropathy, gluten intolerance, and IBD-like inflammation associated with lipotoxicity in DGAT1 deficiency-related diarrhea: a case report of a 17-year-old patient and literature review. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 0, , .	2.8	3