Beat Steinmann

List of Publications by Year
in descending order

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64	7,201	147801	168389
papers	citations	31 h-index	53 g-index

67	67	67	7012
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Obituary for Claude Bachmann, <scp>MD</scp> (1941â€“2022). Journal of Inherited Metabolic Disease, 2022, 45, 657-658.	3.6	0
2	Celiprolol but not losartan improves the biomechanical integrity of the aorta in a mouse model of vascular Ehlersâ€“Danlos syndrome. Cardiovascular Research, 2020, 116, 457-465.	3.8	21
3	Variant filtering, digenic variants, and other challenges in clinical sequencing: a lesson from fibrillinopathies. Clinical Genetics, 2020, 97, 235-245.	2.0	17
4	HereditÃre Bindegewebskrankheiten. Springer Reference Medizin, 2020, , 2835-2859.	0.0	1
5	HereditÃre Bindegewebskrankheiten bei Kindern und Jugendlichen. Springer Reference Medizin, 2019, , 1-25.	0.0	1
6	Inherited metabolic disorders presenting as hypoxic ischaemic encephalopathy: A case series of patients presenting at a tertiary care hospital in Pakistan. JPMA the Journal of the Pakistan Medical Association, 2019, 69, 432-436.	0.2	1
7	1. Das Ehlers-Danlos-Syndrom: Klinik â€' Einteilung â€' Pathogenese â€' Genetik. , 2017, , 1-30.		0
8	The phenotype of the musculocontractural type of Ehlersâ€Danlos syndrome due to CHST14 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 103-115.	1.2	53
9	A summary of molecular genetic findings in fructose-1,6-bisphosphatase deficiency with a focus on a common long-range deletion and the role of MLPA analysis. Orphanet Journal of Rare Diseases, 2016, 11, 44.	2.7	28
10	New insights into the performance of human whole-exome capture platforms. Nucleic Acids Research, 2015, 43, e76-e76.	14.5	103
11	HereditÃre Bindegewebskrankheiten bei Kindern und Jugendlichen. , 2015, , 1-24.		0
12	Richard Gitzelmann (23rd February 1930â€“31st October 2013). European Journal of Pediatrics, 2014, 173, 695-697.	2.7	0
13	Genetische Bindegewebskrankheiten. , 2014, , 1912-1925.		1
14	Clinical utility gene card for: Ehlersâ€“Danlos syndrome types Iâ€“VII and variants - update 2012. European Journal of Human Genetics, 2013, 21, 118-118.	2.8	37
15	Promotion of vesicular zinc efflux by ZIP13 and its implications for spondylocheiro dysplastic Ehlersâ€“Danlos syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, E3530-8.	7.1	98
16	Generalized Arterial Calcification of Infancy and Pseudoxanthoma Elasticum Can Be Caused by Mutations in Either ENPP1 or ABCC6. American Journal of Human Genetics, 2012, 90, 25-39.	6.2	274
17	Ehlers-Danlos Syndrome Type VI in a 17-Year-Old Iranian Boy with Severe Muscular Weakness - A Diagnostic Challenge?. Iranian Journal of Pediatrics, 2010, 20, 358-62.	0.3	5
18	Spondylocheiro Dysplastic Form of the Ehlers-Danlos Syndromeâ€"An Autosomal-Recessive Entity Caused by Mutations in the Zinc Transporter Gene SLC39A13. American Journal of Human Genetics, 2008, 82, 1290-1305.	6.2	191

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19	Differential diagnosis of muscular hypotonia in infants: The kyphoscoliotic type of Ehlersâ€“Danlos syndrome (EDS VI). Neuromuscular Disorders, 2008, 18, 210-214.	0.6	34
20	Automated HPLC Assay for Urinary Collagen Cross-links: Effect of Age, Menopause, and Metabolic Bone Diseases. Clinical Chemistry, 2008, 54, 1546-1553.	3.2	32
21	A bioinformatics framework for genotypeâ€“phenotype correlation in humans with Marfan syndrome caused by FBN1 gene mutations. Journal of Biomedical Informatics, 2006, 39, 171-183.	4.3	12
22	Nevo syndrome is allelic to the kyphoscoliotic type of the Ehlers-Danlos syndrome (EDS VIA). American Journal of Medical Genetics, Part A, 2005, 133A, 158-164.	1.2	58
23	Mutation analysis of the PLOD1 gene: An efficient multistep approach to the molecular diagnosis of the kyphoscoliotic type of Ehlers-Danlos syndrome (EDS VIA). Molecular Genetics and Metabolism, 2005, 86, 269-276.	1.1	55
24	The boy with massive glucosuria. Nephrology Dialysis Transplantation, 2004, 19, 1319-1320.	0.7	0
25	Update of the UMD-FBN1mutation database and creation of anFBN1polymorphism database. Human Mutation, 2003, 22, 199-208.	2.5	299
26	Intestinal glucose transport: Evidence for a membrane trafficâ€“based pathway in humans. Gastroenterology, 2003, 124, 34-39.	1.3	59
27	IN VITRO PROTEOGLYCAN SULFATION DERIVED FROM SULFHYDRYL COMPOUNDS IN SULFATE TRANSPORTER CHONDRODYSPLASIAS. Fetal and Pediatric Pathology, 2003, 22, 311-321.	0.3	8
28	IN VITRO PROTEOGLYCAN SULFATION DERIVED FROM SULFHYDRYL COMPOUNDS IN SULFATE TRANSPORTER CHONDRODYSPLASIAS. Fetal and Pediatric Pathology, 2003, 22, 311-321.	0.3	4
29	The kyphoscoliotic type of Ehlersâ€“Danlos syndrome (type VI): differential effects on the hydroxylation of lysine in collagens I and II revealed by analysis of cross-linked telopeptides from urine. Molecular Genetics and Metabolism, 2002, 76, 211-216.	1.1	55
30	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
31	Calvarial â€œdoughnut lesionsâ€• Clinical spectrum of the syndrome, report on a case, and review of the literature. American Journal of Medical Genetics Part A, 2001, 99, 238-243.	2.4	17
32	Compound heterozygosity for a disease-causing G1489D and disease-modifying G530S substitution inCOL5A1 of a patient with the classical type of Ehlers-Danlos syndrome: An explanation of intrafamilial variability?. , 2000, 90, 72-79.		36
33	Casting new light on the clinical spectrum of neonatal severe hyperparathyroidism. Clinical Endocrinology, 1999, 50, 691-693.	2.4	47
34	Ehlers-Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	1,624
35	Schwartz-Jampel syndrome type 2 and Stï¿½ve-Wiedemann syndrome: A case for ?Lumping?. , 1998, 78, 150-154.		43
36	Sibs affected with both Ehlers-Danlos syndrome type VI and cystic fibrosis. , 1998, 78, 455-460.		18

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37	Proteoglycan sulfation in cartilage and cell cultures from patients with sulfate transporter chondrodysplasias: Relationship to clinical severity and indications on the role of intracellular sulfate production. Matrix Biology, 1998, 17, 361-369.	3.6	59
38	Ehlersâ€Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	40
39	A Rare Branch-Point Mutation Is Associated with Missplicing of Fibrillin-2 in a Large Family with Congenital Contractural Arachnodactyly. American Journal of Human Genetics, 1997, 60, 1389-1398.	6.2	73
40	Mutations in GLUT2, the gene for the liver-type glucose transporter, in patients with Fanconi-Bickel syndrome. Nature Genetics, 1997, 17, 324-326.	21.4	304
41	Phenotypic and genotypic overlap between atelosteogenesis type 2 and diastrophic dysplasia. Human Genetics, 1996, 98, 657-661.	3.8	54
42	A chondrodysplasia family produced by mutations in thediastrophic dysplasia sulfate transporter gene: Genotype/phenotype correlations. American Journal of Medical Genetics Part A, 1996, 63, 144-147.	2.4	114
43	Ehlers-Danlos syndrome type VIII and leukodystrophy. American Journal of Medical Genetics Part A, 1996, 66, 239-240.	2.4	14
44	Achondrogenesis type IB is caused by mutations in the diastrophic dysplasia sulphate transporter gene. Nature Genetics, 1996, 12, 100-102.	21.4	219
45	Clinical, ultrastructural and biochemical studies in two sibs with Ehlersâ€Danlos syndrome type VIâ€Bâ€like features. Clinical Genetics, 1994, 46, 417-422.	2.0	9
46	Long-term follow-up of two sibs with Larsen syndrome possibly due to parental germ-line mosaicism. American Journal of Medical Genetics Part A, 1993, 47, 187-197.	2.4	28
47	Decreased extracellular deposition of fibrillin and decorin in neonatal Marfan syndrome fibroblasts. Human Genetics, 1993, 90, 511-5.	3.8	56
48	Homozygosity mapping of the gene for alkaptonuria to chromosome 3q2. Nature Genetics, 1993, 5, 201-204.	21.4	128
49	Normal Production, Nature, and Extent of Intracellular Degradation of Newly Synthesized Collagen in Fibroblasts from a Patient with Prolidase Deficiency. Connective Tissue Research, 1993, 29, 23-30.	2.3	9
50	Genetic Linkage of the Marfan Syndrome, Ectopia Lentis, and Congenital Contractural Arachnodactyly to the Fibrillin Genes on Chromosomes 15 and 5. New England Journal of Medicine, 1992, 326, 905-909.	27.0	257
51	A homozygous stop codon in the lysyl hydroxylase gene in two siblings with Ehlersâ€“Danlos syndrome type VI. Nature Genetics, 1992, 2, 228-231.	21.4	101
52	Ehlers-Danlos syndrome type IV: A subset of patients distinguished by low serum levels of the amino-terminal propeptide of type III procollagen. American Journal of Medical Genetics Part A, 1989, 34, 68-71.	2.4	28
53	Molecular defects of type III procollagen in Ehlers-Danlos syndrome type IV. Human Genetics, 1989, 82, 104-108.	3.8	97
54	The Renal Handling of Carnitine in Patients with Selective Tuhulopathy and with Fanconi Syndrome. Pediatric Research, 1987, 21, 201-204.	2.3	19

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55	Structural study of a mutant type I collagen from a patient with lethal osteogenesis imperfecta containing an intramolecular disulfide bond in the triple-helical domain. FEBS Letters, 1986, 198, 213-216.	2.8	30
56	Short Communication. Glycogenosis Ib: Neutrophil Microbicidal Defects Due to Impaired Hexose Monophosphate Shunt. Pediatric Research, 1984, 18, 297-299.	2.3	35
57	Invited editorial comment: Osteogenesis imperfecta: Update and perspective. American Journal of Medical Genetics Part A, 1984, 17, 429-435.	2.4	59
58	Ehlersâ€Danlos syndrome type IV D: an autosomal recessive disorder. Clinical Genetics, 1984, 25, 278-287.	2.0	28
59	Ascorbate deficiency results in decreased collagen production: Under-hydroxylation of proline leads to increased intracellular degradation. Archives of Biochemistry and Biophysics, 1983, 226, 681-686.	3.0	59
60	Intracellular degradation of newly synthesized collagen is conformation-dependent. FEBS Letters, 1981, 133, 142-144.	2.8	40
61	Persistent and Transient Distal Renal Tubular Acidosis with Bicarbonate Wasting. Pediatric Research, 1975, 9, 767-773.	2.3	16
62	Marfan Syndrome and Other Microfibrillar Disorders. , 0, , 585-626.		20
63	Skeletal Dysplasias Related to Defects in Sulfate Metabolism. , 0, , 939-960.		15
64	Osteoporosis-Pseudoglioma Syndrome. , 0, , 1119-1121.		0