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	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
2	Ehlers-Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	1,624
3	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
4	Update of the UMD-FBN1mutation database and creation of anFBN1polymorphism database. Human Mutation, 2003, 22, 199-208.	2.5	299
5	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
6	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
7	Achondrogenesis type IB is caused by mutations in the diastrophic dysplasia sulphate transporter gene. Nature Genetics, 1996, 12, 100-102.	21.4	219
8	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
9	Homozygosity mapping of the gene for alkaptonuria to chromosome 3q2. Nature Genetics, 1993, 5, 201-204.	21.4	128
10	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
11	New insights into the performance of human whole-exome capture platforms. Nucleic Acids Research, 2015, 43, e76-e76.	14.5	103
12	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
13	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
14	Molecular defects of type III procollagen in Ehlers-Danlos syndrome type IV. Human Genetics, 1989, 82, 104-108.	3.8	97
15	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
16	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
17	Invited editorial comment: Osteogenesis imperfecta: Update and perspective. American Journal of Medical Genetics Part A, 1984, 17, 429-435.	2.4	59
18	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

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19	Intestinal glucose transport: Evidence for a membrane trafficâ€“based pathway in humans. Gastroenterology, 2003, 124, 34-39.	1.3	59
20	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
21	Decreased extracellular deposition of fibrillin and decorin in neonatal Marfan syndrome fibroblasts. Human Genetics, 1993, 90, 511-5.	3.8	56
22	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
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	Phenotypic and genotypic overlap between atelosteogenesis type 2 and diastrophic dysplasia. Human Genetics, 1996, 98, 657-661.	3.8	54
25	The phenotype of the musculocontractural type of EhlersâĐanlos syndrome due to CHST14 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 103-115.	1.2	53
26	Casting new light on the clinical spectrum of neonatal severe hyperparathyroidism. Clinical Endocrinology, 1999, 50, 691-693.	2.4	47
27	Schwartz-Jampel syndrome type 2 and Stï¿½ve-Wiedemann syndrome: A case for ?Lumping?. , 1998, 78, 150-154.		43
28	Intracellular degradation of newly synthesized collagen is conformation-dependent. FEBS Letters, 1981, 133, 142-144.	2.8	40
29	EhlersâĐanlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	40
30	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
31	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
32	Short Communication. Glycogenosis Ib: Neutrophil Microbicidal Defects Due to Impaired Hexose Monophosphate Shunt. Pediatric Research, 1984, 18, 297-299.	2.3	35
33	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
34	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
35	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
36	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

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37	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
38	Ehlersâ€Danlos syndrome type IV D: an autosomal recessive disorder. Clinical Genetics, 1984, 25, 278-287.	2.0	28
39	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
40	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
41	Marfan Syndrome and Other Microfibrillar Disorders. , 0, , 585-626.		20
42	The Renal Handling of Carnitine in Patients with Selective Tuhulopathy and with Fanconi Syndrome. Pediatric Research, 1987, 21, 201-204.	2.3	19
43	Sibs affected with both Ehlers-Danlos syndrome type VI and cystic fibrosis. , 1998, 78, 455-460.		18
44	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
45	Variant filtering, digenic variants, and other challenges in clinical sequencing: a lesson from fibrillinopathies. Clinical Genetics, 2020, 97, 235-245.	2.0	17
46	Persistent and Transient Distal Renal Tubular Acidosis with Bicarbonate Wasting. Pediatric Research, 1975, 9, 767-773.	2.3	16
47	Skeletal Dysplasias Related to Defects in Sulfate Metabolism. , 0, , 939-960.		15
48	Ehlers-Danlos syndrome type VIII and leukodystrophy. American Journal of Medical Genetics Part A, 1996, 66, 239-240.	2.4	14
49	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
50	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
51	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
52	IN VITRO PROTEOGLYCAN SULFATION DERIVED FROM SULFHYDRYL COMPOUNDS IN SULFATE TRANSPORTER CHONDRODYSPLASIAS. Fetal and Pediatric Pathology, 2003, 22, 311-321.	0.3	8
53	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
54	IN VITRO PROTEOGLYCAN SULFATION DERIVED FROM SULFHYDRYL COMPOUNDS IN SULFATE TRANSPORTER CHONDRODYSPLASIAS. Fetal and Pediatric Pathology, 2003, 22, 311-321.	0.3	4

#	Article	IF	Citations
55	Genetische Bindegewebskrankheiten. , 2014, , 1912-1925.		1
56	HereditÃre Bindegewebskrankheiten bei Kindern und Jugendlichen. Springer Reference Medizin, 2019, , 1-25.	0.0	1
57	HereditÃre Bindegewebskrankheiten. Springer Reference Medizin, 2020, , 2835-2859.	0.0	1
58	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
59	Osteoporosis-Pseudoglioma Syndrome. , 0, , 1119-1121.		0
60	The boy with massive glucosuria. Nephrology Dialysis Transplantation, 2004, 19, 1319-1320.	0.7	0
61	Richard Gitzelmann (23rd February 1930â€“31st October 2013). European Journal of Pediatrics, 2014, 173, 695-697.	2.7	0
62	1. Das Ehlers-Danlos-Syndrom: Klinik â€' Einteilung â€' Pathogenese â€' Genetik. , 2017, , 1-30.		0
63	HereditÃre Bindegewebskrankheiten bei Kindern und Jugendlichen. , 2015, , 1-24.		0
64	Obituary for Claude Bachmann, <scp>MD</scp> (1941â€“2022). Journal of Inherited Metabolic Disease, 2022, 45, 657-658.	3.6	0