

Marianne Rohrbach

List of Publications by Year in descending order

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63
papers

4,549
citations

147801

31
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123424

61
g-index

64
all docs

64
docs citations

64
times ranked

6094
citing authors

#	ARTICLE	IF	CITATIONS
1	The 2017 international classification of the Ehlers-Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	1.6	1,163
2	Exome Sequencing Identifies Truncating Mutations in Human SERPINF1 in Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2011, 88, 362-371.	6.2	316
3	Epidemiology of mucopolysaccharidoses. Molecular Genetics and Metabolism, 2017, 121, 227-240.	1.1	290
4	Mutations in the TGF β 2 Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromioclavicular and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14.	6.2	199
5	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. Human Mutation, 2012, 33, 343-350.	2.5	178
6	The Ehlers-Danlos syndromes, rare types. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 70-115.	1.6	168
7	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
8	Mutations in FKBP14 Cause a Variant of Ehlers-Danlos Syndrome with Progressive Kyphoscoliosis, Myopathy, and Hearing Loss. American Journal of Human Genetics, 2012, 90, 201-216.	6.2	136
9	Impaired antibacterial autophagy links granulomatous intestinal inflammation in Niemann-Pick disease type C1 and XIAP deficiency with NOD2 variants in Crohn's disease. Gut, 2017, 66, 1060-1073.	12.1	126
10	MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. Nature Communications, 2016, 7, 11920.	12.8	112
11	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	2.8	107
12	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. American Journal of Human Genetics, 2011, 88, 767-777.	6.2	106
13	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S, which Encode Subcomponents C1r and C1s of Complement. American Journal of Human Genetics, 2016, 99, 1005-1014.	6.2	100
14	Promotion of vesicular zinc efflux by ZIP13 and its implications for spondylocheiro dysplastic Ehlers-Danlos syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, E3530-8.	7.1	98
15	Revised recommendations for the management of Gaucher disease in children. European Journal of Pediatrics, 2013, 172, 447-458.	2.7	86
16	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	2.3	81
17	CRIM-negative infantile Pompe disease: 42-month treatment outcome. Journal of Inherited Metabolic Disease, 2010, 33, 751-757.	3.6	79
18	Phenotypic variability of the kyphoscoliotic type of Ehlers-Danlos syndrome (EDS VIA): clinical, molecular and biochemical delineation. Orphanet Journal of Rare Diseases, 2011, 6, 46.	2.7	75

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19	A study of the clinical and radiological features in a cohort of 93 patients with a <i>COL2A1</i> mutation causing spondyloepiphyseal dysplasia congenita or a related phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 461-475.	1.2	73
20	ZNF469 frequently mutated in the brittle cornea syndrome (BCS) is a single exon gene possibly regulating the expression of several extracellular matrix components. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 289-295.	1.1	61
21	Recessive osteogenesis imperfecta: Clinical, radiological, and molecular findings. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2012, 160C, 175-189.	1.6	53
22	The phenotype of the musculocontractural type of Ehlers-Danlos syndrome due to <i>CHST14</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 103-115.	1.2	53
23	LC-MS/MS based assay and reference intervals in children and adolescents for oxysterols elevated in Niemann-Pick diseases. <i>Clinical Biochemistry</i> , 2015, 48, 596-602.	1.9	50
24	Brittle cornea syndrome: recognition, molecular diagnosis and management. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 68.	2.7	48
25	Outcome of Patients with Classical Infantile Pompe Disease Receiving Enzyme Replacement Therapy in Germany. <i>JIMD Reports</i> , 2014, 20, 65-75.	1.5	47
26	Molecular Consequences of the SERPINH1/HSP47 Mutation in the Dachshund Natural Model of Osteogenesis Imperfecta. <i>Journal of Biological Chemistry</i> , 2015, 290, 17679-17689.	3.4	42
27	Characterization of new ACADSB gene sequence mutations and clinical implications in patients with 2-methylbutyrylglycinuria identified by newborn screening. <i>Molecular Genetics and Metabolism</i> , 2010, 100, 333-338.	1.1	41
28	Expanding the clinical and mutational spectrum of the Ehlers-Danlos syndrome, dermatosparaxis type. <i>Genetics in Medicine</i> , 2016, 18, 882-891.	2.4	37
29	Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta. <i>American Journal of Human Genetics</i> , 2019, 105, 836-843.	6.2	36
30	Obstructive sleep apnoea and quality of life in Ehlers-Danlos syndrome: a parallel cohort study. <i>Thorax</i> , 2017, 72, 729-735.	5.6	35
31	Hemizygous deletion of COL3A1, COL5A2, and MSTN causes a complex phenotype with aortic dissection: a lesson for and from true haploinsufficiency. <i>European Journal of Human Genetics</i> , 2010, 18, 1315-1321.	2.8	34
32	Measuring patient experiences in Fabry disease: validation of the Fabry-specific Pediatric Health and Pain Questionnaire (FPHQP). <i>Health and Quality of Life Outcomes</i> , 2012, 10, 116.	2.4	33
33	<i>COL1</i> -related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlers-Danlos syndrome overlap. <i>Clinical Genetics</i> , 2020, 97, 396-406.	2.0	27
34	A cohort of 17 patients with kyphoscoliotic Ehlers-Danlos syndrome caused by biallelic mutations in FKBP14: expansion of the clinical and mutational spectrum and description of the natural history. <i>Genetics in Medicine</i> , 2018, 20, 42-54.	2.4	26
35	Defect in proline synthesis: pyrroline-5-carboxylate reductase 1 deficiency leads to a complex clinical phenotype with collagen and elastin abnormalities. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 731-739.	3.6	25
36	Kyphoscoliotic type of Ehlers-Danlos Syndrome (EDS VIA) in six Egyptian patients presenting with a homogeneous clinical phenotype. <i>European Journal of Pediatrics</i> , 2015, 174, 105-112.	2.7	20

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37	A variant in the gene for GM-CSF, I117T, is associated with atopic asthma in a Swiss population of asthmatic children. <i>Journal of Allergy and Clinical Immunology</i> , 1999, 104, 247-248.	2.9	19
38	Swiss national guideline for reimbursement of enzyme replacement therapy in late-onset Pompe disease. <i>Journal of Neurology</i> , 2013, 260, 2279-2285.	3.6	19
39	Expanding the clinical and genetic spectrum of CAD deficiency: an epileptic encephalopathy treatable with uridine supplementation. <i>Genetics in Medicine</i> , 2020, 22, 1589-1597.	2.4	19
40	Paediatric Fabry disease: prognostic significance of ocular changes for disease severity. <i>BMC Ophthalmology</i> , 2016, 16, 202.	1.4	18
41	Quantification of muscle pathology in infantile Pompe disease. <i>Neuromuscular Disorders</i> , 2017, 27, 141-152.	0.6	18
42	Variant filtering, digenic variants, and other challenges in clinical sequencing: a lesson from fibrillinopathies. <i>Clinical Genetics</i> , 2020, 97, 235-245.	2.0	17
43	Clinical and Molecular Characterization of Classical-Like Ehlers-Danlos Syndrome Due to a Novel TNXB Variant. <i>Genes</i> , 2019, 10, 843.	2.4	16
44	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. <i>Genes</i> , 2021, 12, 65.	2.4	16
45	Transcriptome Profiling of Primary Skin Fibroblasts Reveal Distinct Molecular Features Between PLOD1- and FKBP14-Kyphoscoliotic Ehlers-Danlos Syndrome. <i>Genes</i> , 2019, 10, 517.	2.4	15
46	Early co-occurrence of a neurologic-psychiatric disease pattern in Niemann-Pick type C disease: a retrospective Swiss cohort study. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 176.	2.7	14
47	Extent, impact, and predictors of diagnostic delay in Pompe disease: A combined survey approach to unveil the diagnostic odyssey. <i>JIMD Reports</i> , 2019, 49, 89-95.	1.5	13
48	A new COL3A1 mutation in Ehlers-Danlos syndrome type IV. <i>Experimental Dermatology</i> , 2013, 22, 231-234.	2.9	12
49	Obstructive Sleep Apnoea in Children and Adolescents with Ehlers-Danlos Syndrome. <i>Respiration</i> , 2019, 97, 284-291.	2.6	12
50	The novel missense mutation Met48Lys in FKBP22 changes its structure and functions. <i>Scientific Reports</i> , 2020, 10, 497.	3.3	9
51	Urinary pyridinoline cross-links as biomarkers of osteogenesis imperfecta. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 104.	2.7	6
52	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. <i>American Journal of Human Genetics</i> , 2011, 89, 346.	6.2	5
53	Etiology of Carpal Tunnel Syndrome in a Large Cohort of Children. <i>Children</i> , 2021, 8, 624.	1.5	5
54	Increased augmentation index in patients with Ehlers-Danlos syndrome. <i>BMC Cardiovascular Disorders</i> , 2020, 20, 417.	1.7	4

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55	Marfan syndrome and related connective tissue disorders in the current era in Switzerland in 103 patients: medical and surgical management and impact of genetic testing. Swiss Medical Weekly, 2020, 150, w20189.	1.6	4
56	Stroke in Ehlers-Danlos Syndrome Kyphoscoliotic Type: Dissection or Vasculitis?. Pediatric Neurology, 2017, 74, 92-96.	2.1	2
57	Omics Profiling of S2P Mutant Fibroblasts as a Mean to Unravel the Pathomechanism and Molecular Signatures of X-Linked MBTPS2 Osteogenesis Imperfecta. Frontiers in Genetics, 2021, 12, 662751.	2.3	2
58	Is serum biotinidase enzyme activity a potential marker of perturbed glucose and lipid metabolism?. JIMD Reports, 2021, 57, 58-66.	1.5	1
59	Genetische Bindegewebskrankheiten. , 2014, , 1912-1925.		1
60	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. Springer Reference Medizin, 2019, , 1-25.	0.0	1
61	Hereditäre Bindegewebskrankheiten. Springer Reference Medizin, 2020, , 2835-2859.	0.0	1
62	Loss-of-function variants in exon 4 of TAB2 cause a recognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletal involvement. Genetics in Medicine, 2021, , .	2.4	1
63	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. , 2015, , 1-24.		0