

Marianne Rohrbach

List of Publications by Year in descending order

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63
papers

4,549
citations

147566

31
h-index

123241

61
g-index

64
all docs

64
docs citations

64
times ranked

6094
citing authors

#	ARTICLE	IF	CITATIONS
1	Is serum biotinidase enzyme activity a potential marker of perturbed glucose and lipid metabolism?. <i>JIMD Reports</i> , 2021, 57, 58-66.	0.7	1
2	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. <i>Genes</i> , 2021, 12, 65.	1.0	16
3	Omics Profiling of S2P Mutant Fibroblasts as a Mean to Unravel the Pathomechanism and Molecular Signatures of X-Linked MBTPS2 Osteogenesis Imperfecta. <i>Frontiers in Genetics</i> , 2021, 12, 662751.	1.1	2
4	Etiology of Carpal Tunnel Syndrome in a Large Cohort of Children. <i>Children</i> , 2021, 8, 624.	0.6	5
5	Loss-of-function variants in exon 4 of TAB2 cause a recognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletal involvement. <i>Genetics in Medicine</i> , 2021, , .	1.1	1
6	Variant filtering, digenic variants, and other challenges in clinical sequencing: a lesson from fibrillinopathies. <i>Clinical Genetics</i> , 2020, 97, 235-245.	1.0	17
7	<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.	1.0	27
8	The Genetic Landscape and Epidemiology of Phenylketonuria. <i>American Journal of Human Genetics</i> , 2020, 107, 234-250.	2.6	138
9	Increased augmentation index in patients with Ehlers-Danlos syndrome. <i>BMC Cardiovascular Disorders</i> , 2020, 20, 417.	0.7	4
10	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.	1.1	19
11	The novel missense mutation Met48Lys in FKBP22 changes its structure and functions. <i>Scientific Reports</i> , 2020, 10, 497.	1.6	9
12	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. <i>Swiss Medical Weekly</i> , 2020, 150, w20189.	0.8	4
13	Hereditäre Bindegewebskrankheiten. <i>Springer Reference Medizin</i> , 2020, , 2835-2859.	0.0	1
14	Transcriptome Profiling of Primary Skin Fibroblasts Reveal Distinct Molecular Features Between PLOD1- and FKBP14-Kyphoscoliotic Ehlers-Danlos Syndrome. <i>Genes</i> , 2019, 10, 517.	1.0	15
15	Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta. <i>American Journal of Human Genetics</i> , 2019, 105, 836-843.	2.6	36
16	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.	0.7	13
17	Clinical and Molecular Characterization of Classical-Like Ehlers-Danlos Syndrome Due to a Novel TNXB Variant. <i>Genes</i> , 2019, 10, 843.	1.0	16
18	Obstructive Sleep Apnoea in Children and Adolescents with Ehlers-Danlos Syndrome. <i>Respiration</i> , 2019, 97, 284-291.	1.2	12

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19	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. Springer Reference Medizin, 2019, , 1-25.	0.0	1
20	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. Genetics in Medicine, 2018, 20, 42-54.	1.1	26
21	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.	6.1	126
22	Obstructive sleep apnoea and quality of life in Ehlers-Danlos syndrome: a parallel cohort study. Thorax, 2017, 72, 729-735.	2.7	35
23	Stroke in Ehlers-Danlos Syndrome Kyphoscoliotic Type: Dissection or Vasculitis?. Pediatric Neurology, 2017, 74, 92-96.	1.0	2
24	Epidemiology of mucopolysaccharidoses. Molecular Genetics and Metabolism, 2017, 121, 227-240.	0.5	290
25	The Ehlers-Danlos syndromes, rare types. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 70-115.	0.7	168
26	The 2017 international classification of the Ehlers-Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	0.7	1,163
27	Quantification of muscle pathology in infantile Pompe disease. Neuromuscular Disorders, 2017, 27, 141-152.	0.3	18
28	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.	2.6	100
29	Paediatric Fabry disease: prognostic significance of ocular changes for disease severity. BMC Ophthalmology, 2016, 16, 202.	0.6	18
30	The phenotype of the musculocontractural type of Ehlers-Danlos syndrome due to <i>CHST14</i> mutations. American Journal of Medical Genetics, Part A, 2016, 170, 103-115.	0.7	53
31	MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. Nature Communications, 2016, 7, 11920.	5.8	112
32	Expanding the clinical and mutational spectrum of the Ehlers-Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891.	1.1	37
33	Urinary pyridinoline cross-links as biomarkers of osteogenesis imperfecta. Orphanet Journal of Rare Diseases, 2015, 10, 104.	1.2	6
34	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81
35	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. American Journal of Medical Genetics, Part A, 2015, 167, 461-475.	0.7	73
36	Kyphoscoliotic type of Ehlers-Danlos Syndrome (EDS VIA) in six Egyptian patients presenting with a homogeneous clinical phenotype. European Journal of Pediatrics, 2015, 174, 105-112.	1.3	20

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37	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.	0.8	50
38	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.	1.6	42
39	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. , 2015, , 1-24.		0
40	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.	1.2	14
41	Outcome of Patients with Classical Infantile Pompe Disease Receiving Enzyme Replacement Therapy in Germany. <i>JIMD Reports</i> , 2014, 20, 65-75.	0.7	47
42	Genetische Bindegewebskrankheiten. , 2014, , 1912-1925.		1
43	Brittle cornea syndrome: recognition, molecular diagnosis and management. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 68.	1.2	48
44	Swiss national guideline for reimbursement of enzyme replacement therapy in late-onset Pompe disease. <i>Journal of Neurology</i> , 2013, 260, 2279-2285.	1.8	19
45	Revised recommendations for the management of Gaucher disease in children. <i>European Journal of Pediatrics</i> , 2013, 172, 447-458.	1.3	86
46	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.	0.5	61
47	A new <i>COL3A1</i> mutation in Ehlers-Danlos syndrome type IV. <i>Experimental Dermatology</i> , 2013, 22, 231-234.	1.4	12
48	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. <i>European Journal of Human Genetics</i> , 2012, 20, 11-19.	1.4	107
49	Promotion of vesicular zinc efflux by ZIP13 and its implications for spondylocheiro dysplastic Ehlers-Danlos syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, E3530-8.	3.3	98
50	Measuring patient experiences in Fabry disease: validation of the Fabry-specific Pediatric Health and Pain Questionnaire (FPHPQ). <i>Health and Quality of Life Outcomes</i> , 2012, 10, 116.	1.0	33
51	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.	0.7	53
52	Mutations in FKBP14 Cause a Variant of Ehlers-Danlos Syndrome with Progressive Kyphoscoliosis, Myopathy, and Hearing Loss. <i>American Journal of Human Genetics</i> , 2012, 90, 201-216.	2.6	136
53	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. <i>Human Mutation</i> , 2012, 33, 343-350.	1.1	178
54	Exome Sequencing Identifies Truncating Mutations in Human SERPINF1 in Autosomal-Recessive Osteogenesis Imperfecta. <i>American Journal of Human Genetics</i> , 2011, 88, 362-371.	2.6	316

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55	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.	2.6	106
56	Mutations in the TGF β 2 Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14.	2.6	199
57	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. American Journal of Human Genetics, 2011, 89, 346.	2.6	5
58	Defect in proline synthesis: pyrroline-5-carboxylate reductase 1 deficiency leads to a complex clinical phenotype with collagen and elastin abnormalities. Journal of Inherited Metabolic Disease, 2011, 34, 731-739.	1.7	25
59	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.	1.2	75
60	CRIM α -negative infantile Pompe disease: 42 α -month treatment outcome. Journal of Inherited Metabolic Disease, 2010, 33, 751-757.	1.7	79
61	Hemizygous deletion of COL3A1, COL5A2, and MSTN causes a complex phenotype with aortic dissection: a lesson for and from true haploinsufficiency. European Journal of Human Genetics, 2010, 18, 1315-1321.	1.4	34
62	Characterization of new ACADSB gene sequence mutations and clinical implications in patients with 2-methylbutyryl-glycinuria identified by newborn screening. Molecular Genetics and Metabolism, 2010, 100, 333-338.	0.5	41
63	A variant in the gene for GM-CSF, I117T, is associated with atopic asthma in a Swiss population of asthmatic children. Journal of Allergy and Clinical Immunology, 1999, 104, 247-248.	1.5	19