# Marianne Rohrbach

List of Publications by Year
in descending order



| 63 | 4,549 | 147801 | 123424 |
|---|---|---|---|
| papers | citations | 31 h-index | 61 g-index |

| 64 | 64 | 64 | 6094 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Is serum biotinidase enzyme activity a potential marker of perturbed glucose and lipid metabolism?. JIMD Reports, 2021, 57, 58-66. | 1.5 | 1 |
| 2 | Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. Genes, 2021, 12, 65. | 2.4 | 16 |
| 3 | 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 |
| 4 | Etiology of Carpal Tunnel Syndrome in a Large Cohort of Children. Children, 2021, 8, 624. | 1.5 | 5 |
| 5 | Loss-of-function variants in exon 4 of TAB2 causeÂaÂrecognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletalÂinvolvement. Genetics in Medicine, 2021, , . | 2.4 | 1 |
| 6 | Variant filtering, digenic variants, and other challenges in clinical sequencing: a lesson from fibrillinopathies. Clinical Genetics, 2020, 97, 235-245. | 2.0 | 17 |
| 7 | COL1â€related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlersâ€Danlos syndrome overlap. Clinical Genetics, 2020, 97, 396-406. | 2.0 | 27 |
| 8 | The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250. | 6.2 | 138 |
| 9 | Increased augmentation index in patients with Ehlers-Danlos syndrome. BMC Cardiovascular Disorders, 2020, 20, 417. | 1.7 | 4 |
| 10 | Expanding the clinical and genetic spectrum of CAD deficiency: an epileptic encephalopathy treatable with uridine supplementation. Genetics in Medicine, 2020, 22, 1589-1597. | 2.4 | 19 |
| 11 | The novel missense mutation Met48Lys in FKBP22 changes its structure and functions. Scientific Reports, 2020, 10, 497. | 3.3 | 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. Swiss Medical Weekly, 2020, 150, w20189. | 1.6 | 4 |
| 13 | HereditÄre Bindegewebskrankheiten. Springer Reference Medizin, 2020, , 2835-2859. | 0.0 | 1 |
| 14 | Transcriptome Profiling of Primary Skin Fibroblasts Reveal Distinct Molecular Features Between PLOD1- and FKBP14-Kyphoscoliotic Ehlersâ€“Danlos Syndrome. Genes, 2019, 10, 517. | 2.4 | 15 |
| 15 | Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta. American Journal of Human Genetics, 2019, 105, 836-843. | 6.2 | 36 |
| 16 | Extent, impact, and predictors of diagnostic delay in Pompe disease: A combined survey approach to unveil the diagnostic odyssey. JIMD Reports, 2019, 49, 89-95. | 1.5 | 13 |
| 17 | Clinical and Molecular Characterization of Classical-Like Ehlers-Danlos Syndrome Due to a Novel TNXB Variant. Genes, 2019, 10, 843. | 2.4 | 16 |
| 18 | Obstructive Sleep Apnoea in Children and Adolescents with Ehlers-Danlos Syndrome. Respiration, 2019, 97, 284-291. | 2.6 | 12 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | HereditÃre Bindegewebskrankheiten bei Kindern und Jugendlichen. Springer Reference Medizin, 2019, , 1-25. | 0.0 | 1 |
| 20 | A cohort of 17 patients with kyphoscoliotic Ehlersâ€“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. | 2.4 | 26 |
| 21 | Impaired antibacterial autophagy links granulomatous intestinal inflammation in Niemannâ€“Pick disease type C1 and XIAP deficiency with NOD2 variants in Crohn's disease. Gut, 2017, 66, 1060-1073. | 12.1 | 126 |
| 22 | Obstructive sleep apnoea and quality of life in Ehlers-Danlos syndrome: a parallel cohort study. Thorax, 2017, 72, 729-735. | 5.6 | 35 |
| 23 | Stroke in Ehlers-Danlos Syndrome Kyphoscoliotic Type: Dissection or Vasculitis?. Pediatric Neurology, 2017, 74, 92-96. | 2.1 | 2 |
| 24 | Epidemiology of mucopolysaccharidoses. Molecular Genetics and Metabolism, 2017, 121, 227-240. | 1.1 | 290 |
| 25 | The Ehlersâ€“Danlos syndromes, rare types. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 70-115. | 1.6 | 168 |
| 26 | The 2017 international classification of the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26. | 1.6 | 1,163 |
| 27 | Quantification of muscle pathology in infantile Pompe disease. Neuromuscular Disorders, 2017, 27, 141-152. | 0.6 | 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. | 6.2 | 100 |
| 29 | Paediatric Fabry disease: prognostic significance of ocular changes for disease severity. BMC Ophthalmology, 2016, 16, 202. | 1.4 | 18 |
| 30 | 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 |
| 31 | MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. Nature Communications, 2016, 7, 11920. | 12.8 | 112 |
| 32 | Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891. | 2.4 | 37 |
| 33 | Urinary pyridinoline cross-links as biomarkers of osteogenesis imperfecta. Orphanet Journal of Rare Diseases, 2015, 10, 104. | 2.7 | 6 |
| 34 | Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123. | 2.3 | 81 |
| 35 | A study of the clinical and radiological features in a cohort of 93 patients with a COL2A1 mutation causing spondyloepiphyseal dysplasia congenita or a related phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 461-475. | 1.2 | 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. | 2.7 | 20 |

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|---|---|---|---|
| 37 | LC-MS/MS based assay and reference intervals in children and adolescents for oxysterols elevated in Niemannâ€“Pick diseases. Clinical Biochemistry, 2015, 48, 596-602. | 1.9 | 50 |
| 38 | Molecular Consequences of the SERPINH1/HSP47 Mutation in the Dachshund Natural Model of Osteogenesis Imperfecta. Journal of Biological Chemistry, 2015, 290, 17679-17689. | 3.4 | 42 |
| 39 | HereditÃre 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. Orphanet Journal of Rare Diseases, 2014, 9, 176. | 2.7 | 14 |
| 41 | Outcome of Patients with Classical Infantile Pompe Disease Receiving Enzyme Replacement Therapy in Germany. JIMD Reports, 2014, 20, 65-75. | 1.5 | 47 |
| 42 | Genetische Bindegewebskrankheiten. , 2014, , 1912-1925. | | 1 |
| 43 | Brittle cornea syndrome: recognition, molecular diagnosis and management. Orphanet Journal of Rare Diseases, 2013, 8, 68. | 2.7 | 48 |
| 44 | Swiss national guideline for reimbursement of enzyme replacement therapy in late-onset Pompe disease. Journal of Neurology, 2013, 260, 2279-2285. | 3.6 | 19 |
| 45 | Revised recommendations for the management of Gaucher disease in children. European Journal of Pediatrics, 2013, 172, 447-458. | 2.7 | 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. Molecular Genetics and Metabolism, 2013, 109, 289-295. | 1.1 | 61 |
| 47 | A new <scp>COL3A1</scp> mutation in <scp>E</scp>hlersâ€“<scp>D</scp>anlos syndrome type <scp>IV</scp>. Experimental Dermatology, 2013, 22, 231-234. | 2.9 | 12 |
| 48 | EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19. | 2.8 | 107 |
| 49 | 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 |
| 50 | Measuring patient experiences in Fabry disease: validation of the Fabry-specific Pediatric Health and Pain Questionnaire (FPHPQ). Health and Quality of Life Outcomes, 2012, 10, 116. | 2.4 | 33 |
| 51 | Recessive osteogenesis imperfecta: Clinical, radiological, and molecular findings. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 175-189. | 1.6 | 53 |
| 52 | 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 |
| 53 | Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. Human Mutation, 2012, 33, 343-350. | 2.5 | 178 |
| 54 | 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 |

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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. | 6.2 | 106 |
| 56 | Mutations in the TGFÎ² Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14. | 6.2 | 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. | 6.2 | 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. | 3.6 | 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. | 2.7 | 75 |
| 60 | CRIMâ€negative infantile Pompe disease: 42â€month treatment outcome. Journal of Inherited Metabolic Disease, 2010, 33, 751-757. | 3.6 | 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. | 2.8 | 34 |
| 62 | Characterization of new ACADSB gene sequence mutations and clinical implications in patients with 2-methylbutyrylglycinuria identified by newborn screening. Molecular Genetics and Metabolism, 2010, 100, 333-338. | 1.1 | 41 |
| 63 | A variant in the gene for GM-CSF, I117T, is associated with atopic asthma in a Swiss population of asthmatic childrenâ˜†â˜†â˜†â˜…â˜…â˜…. Journal of Allergy and Clinical Immunology, 1999, 104, 247-248. | 2.9 | 19 |