

Markus Ries,, Mhsc, Fcp

List of Publications by Year in descending order

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93
papers

4,545
citations

136950

32
h-index

102487

66
g-index

105
all docs

105
docs citations

105
times ranked

2986
citing authors

#	ARTICLE	IF	CITATIONS
1	Fabry disease defined: baseline clinical manifestations of 366 patients in the Fabry Outcome Survey. <i>European Journal of Clinical Investigation</i> , 2004, 34, 236-242.	3.4	701
2	Long-term therapy with agalsidase alfa for Fabry disease: safety and effects on renal function in a home infusion setting. <i>Nephrology Dialysis Transplantation</i> , 2006, 21, 345-354.	0.7	246
3	Enzyme replacement therapy with agalsidase alfa in patients with Fabry's disease: an analysis of registry data. <i>Lancet, The</i> , 2009, 374, 1986-1996.	13.7	246
4	The Mainz Severity Score Index: a new instrument for quantifying the Anderson's Fabry disease phenotype, and the response of patients to enzyme replacement therapy. <i>Clinical Genetics</i> , 2004, 65, 299-307.	2.0	228
5	Randomized, controlled trial of miglustat in Gaucher's disease type 3. <i>Annals of Neurology</i> , 2008, 64, 514-522.	5.3	223
6	Cardiac manifestations of Anderson's Fabry disease in heterozygous females. <i>Journal of the American College of Cardiology</i> , 2002, 40, 1668-1674.	2.8	177
7	The early clinical phenotype of Fabry disease: a study on 35 European children and adolescents. <i>European Journal of Pediatrics</i> , 2003, 162, 767-772.	2.7	176
8	Pediatric Fabry Disease. <i>Pediatrics</i> , 2005, 115, e344-e355.	2.1	171
9	Enzyme-Replacement Therapy With Agalsidase Alfa in Children With Fabry Disease. <i>Pediatrics</i> , 2006, 118, 924-932.	2.1	156
10	Agalsidase Alfa and Kidney Dysfunction in Fabry Disease. <i>Journal of the American Society of Nephrology: JASN</i> , 2009, 20, 1132-1139.	6.1	148
11	Weekly Enzyme Replacement Therapy May Slow Decline of Renal Function in Patients with Fabry Disease Who Are on Long-Term Biweekly Dosing. <i>Journal of the American Society of Nephrology: JASN</i> , 2007, 18, 1576-1583.	6.1	116
12	The Relationship of Vascular Glycolipid Storage to Clinical Manifestations of Fabry Disease. <i>Medicine (United States)</i> , 2005, 84, 261-268.	1.0	111
13	Disease manifestations and X inactivation in heterozygous females with Fabry disease. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2006, 95, 30-38.	1.5	93
14	Critical assessment of chitotriosidase analysis in the rational laboratory diagnosis of children with Gaucher disease and Niemann-Pick disease type A/B and C. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 647-652.	3.6	86
15	Enzyme replacement therapy and intraepidermal innervation density in Fabry disease. <i>Muscle and Nerve</i> , 2006, 34, 53-56.	2.2	83
16	Pathological findings in a patient with Fabry disease who died after 2.5 years of enzyme replacement. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2006, 448, 337-343.	2.8	80
17	Thirty-four novel mutations of the GLA gene in 121 patients with Fabry disease. <i>Human Mutation</i> , 2005, 25, 412-412.	2.5	74
18	Neuropathic and cerebrovascular correlates of hearing loss in Fabry disease. <i>Brain</i> , 2006, 130, 143-150.	7.6	63

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19	The Saccadic and Neurological Deficits in Type 3 Gaucher Disease. PLoS ONE, 2011, 6, e22410.	2.5	62
20	Four-Year Prospective Clinical Trial of Agalsidase Alfa in Children with Fabry Disease. Journal of Pediatrics, 2010, 156, 832-837.e1.	1.8	54
21	Use of gabapentin to reduce chronic neuropathic pain in Fabry disease. Journal of Inherited Metabolic Disease, 2003, 26, 413-414.	3.6	53
22	Proteomics of specific treatment-related alterations in Fabry disease: A strategy to identify biological abnormalities. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 2873-2878.	7.1	53
23	Ultra-orphan diseases: a quantitative analysis of the natural history of molybdenum cofactor deficiency. Genetics in Medicine, 2015, 17, 965-970.	2.4	45
24	Quantitative dysmorphology assessment in Fabry disease. Genetics in Medicine, 2006, 8, 96-101.	2.4	43
25	Enzyme replacement therapy and beyondâ€”in memoriam Roscoe O. Brady, M.D. (1923â€”2016). Journal of Inherited Metabolic Disease, 2017, 40, 343-356.	3.6	42
26	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. Genetics in Medicine, 2017, 19, 983-988.	2.4	42
27	Parapelvic kidney cysts: A distinguishing feature with high prevalence in Fabry disease. Kidney International, 2004, 66, 978-982.	5.2	41
28	Pressure for drug development in lysosomal storage disorders â€” a quantitative analysis thirty years beyond the US orphan drug act. Orphanet Journal of Rare Diseases, 2015, 10, 46.	2.7	40
29	Cardiac Involvement in Anderson-Fabry Disease. Journal of the American Society of Nephrology: JASN, 2002, 13, S147-S149.	6.1	39
30	Fabry Disease: A Disorder of Childhood Onset. Pediatric Neurology, 2016, 64, 10-20.	2.1	38
31	Enzyme Replacement in Fabry Disease: Pharmacokinetics and Pharmacodynamics of Agalsidase Alfa in Children and Adolescents. Journal of Clinical Pharmacology, 2007, 47, 1222-1230.	2.0	37
32	Clinical Manifestation in Female Fabry Disease Patients. , 2001, 136, 245-250.		35
33	Fabry's diseaseâ€”an important risk factor for stroke. Lancet, The, 2005, 366, 1754-1756.	13.7	34
34	A prospective 10-year study of individualized, intensified enzyme replacement therapy in advanced Fabry disease. Journal of Inherited Metabolic Disease, 2015, 38, 1129-1136.	3.6	33
35	Clinical characteristics of 248 patients with Krabbe disease: quantitative natural history modeling based on published cases. Genetics in Medicine, 2019, 21, 2208-2215.	2.4	33
36	Ultra-orphan lysosomal storage diseases: A cross-sectional quantitative analysis of the natural history of alpha-mannosidosis. Journal of Inherited Metabolic Disease, 2019, 42, 975-983.	3.6	26

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37	Neurological Manifestation of Fabry Disease in Females. , 2001, 136, 241-244.		24
38	A cross-sectional quantitative analysis of the natural history of Farber disease: an ultra-orphan condition with rheumatologic and neurological cardinal disease features. Genetics in Medicine, 2018, 20, 524-530.	2.4	24
39	Cognitive and Behavioral Consequences of Pediatric Delirium: A Pilot Study*. Pediatric Critical Care Medicine, 2018, 19, e531-e537.	0.5	22
40	Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. Genetics in Medicine, 2013, 15, 983-989.	2.4	21
41	Quantitative natural history characterization in a cohort of 142 published cases of patients with galactosialidosisâ€”A crossâ€”sectional study. Journal of Inherited Metabolic Disease, 2019, 42, 295-302.	3.6	21
42	Novel Treatments for Rare Cancers: The U.S. Orphan Drug Act Is Deliveringâ€”A Cross-Sectional Analysis. Oncologist, 2016, 21, 487-493.	3.7	20
43	Enzyme Replacement Therapy in Orphan and Ultra-Orphan Diseases. Pharmacoeconomics, 2007, 25, 201-208.	3.3	19
44	Ten Years after the International Committee of Medical Journal Editorsâ€™ Clinical Trial Registration Initiative, One Quarter of Phase 3 Pediatric Epilepsy Clinical Trials Still Remain Unpublished: A Cross Sectional Analysis. PLoS ONE, 2016, 11, e0144973.	2.5	18
45	FDA orphan drug designations for lysosomal storage disorders â€” a cross-sectional analysis. PLoS ONE, 2020, 15, e0230898.	2.5	17
46	Thirty Years of Orphan Drug Legislation and the Development of Drugs to Treat Rare Seizure Conditions: A Cross Sectional Analysis. PLoS ONE, 2016, 11, e0161660.	2.5	17
47	Challenging behavior in mucopolysaccharidoses types Iâ€”III and day-to-day coping strategies: a cross sectional explorative study. Orphanet Journal of Rare Diseases, 2020, 15, 275.	2.7	16
48	De novo variants in <i>CEL2F2</i> that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. Human Mutation, 2021, 42, 66-76.	2.5	16
49	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	3.6	16
50	Public Health Leadership in a VUCA World Environment: Lessons Learned during the Regional Emergency Rollout of SARS-CoV-2 Vaccinations in Heidelberg, Germany, during the COVID-19 Pandemic. Vaccines, 2021, 9, 887.	4.4	16
51	The COVID-19 Infodemic: Mechanism, Impact, and Counter-Measuresâ€”A Review of Reviews. Sustainability, 2022, 14, 2605.	3.2	16
52	A cross-sectional quantitative analysis of the natural history of free sialic acid storage diseaseâ€”an ultra-orphan multisystemic lysosomal storage disorder. Genetics in Medicine, 2019, 21, 347-352.	2.4	14
53	Critical appraisal of genotype assessment in molybdenum cofactor deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 801-811.	3.6	13
54	Defining the hidden evidence in autism research. Forty per cent of rigorously designed clinical trials remain unpublished â€”a crossâ€”sectional analysis. International Journal of Methods in Psychiatric Research, 2017, 26, .	2.1	13

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55	Skin-impedance in Fabry Disease: A prospective, controlled, non-randomized clinical study. BMC Neurology, 2008, 8, 41.	1.8	12
56	Fabry Disease: Angiokeratoma, Biomarker, and the Effect of Enzyme Replacement Therapy on Kidney Function. Archives of Dermatology, 2005, 141, 904-5; author reply 905-6.	1.4	11
57	Disasters in Germany and France: An Analysis of the Emergency Events Database From a Pediatric Perspective. Disaster Medicine and Public Health Preparedness, 2019, 13, 958-965.	1.3	11
58	Apoptotic abnormalities in differential gene expression in peripheral blood mononuclear cells from children with Fabry disease. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 48-52.	1.5	10
59	Screening for Respiratory Syncytial Virus and Isolation Strategies in Children Hospitalized With acute Respiratory Tract Infection. Medicine (United States), 2014, 93, e144.	1.0	10
60	Angiokeratoma and Pain, But Not Fabry Disease: Considerations for Differential Diagnosis. , 2001, 136, 256-259.		9
61	Anderson-Fabry Disease in Children and Adolescents. , 2001, 136, 251-255.		9
62	Clinical Research in Vulnerable Populations: Variability and Focus of Institutional Review Boards™ Responses. PLoS ONE, 2015, 10, e0135997.	2.5	9
63	Publication status of completed registered studies in paediatric appendicitis: a cross-sectional analysis. BMJ Open, 2018, 8, e021684.	1.9	9
64	Patterns of extreme temperature-related catastrophic events in Europe including the Russian Federation: a cross-sectional analysis of the Emergency Events Database. BMJ Open, 2021, 11, e046359.	1.9	9
65	An Assessment of Publication Status of Pediatric Liver Transplantation Studies. PLoS ONE, 2016, 11, e0168251.	2.5	8
66	Cross-sectional quantitative analysis of the natural history of TUBA1A and TUBB2B tubulinopathies. Genetics in Medicine, 2021, 23, 516-523.	2.4	8
67	Novel treatments for rare rheumatologic disorders: analysis of the impact of 30 years of the US orphan drug act. Orphanet Journal of Rare Diseases, 2016, 11, 60.	2.7	6
68	The drug development pipeline for glioblastoma – A cross sectional assessment of the FDA Orphan Drug Product designation database. PLoS ONE, 2021, 16, e0252924.	2.5	6
69	Schiffmann R, Rapkiewicz A, Abu-Asab M, Ries M, Askari H, Tsokos M, Quezado M. Pathological findings in a patient with Fabry disease, who died after 2.5 years of enzyme replacement. Virchows Arch. 2005 Nov 29; 147: 7. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2006. 448. 873-873.	2.8	5
70	Disease manifestations and X inactivation in heterozygous females with Fabry disease. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 30-38.	1.5	5
71	Quantitative retrospective natural history modeling of WDR45-related developmental and epileptic encephalopathy – a systematic cross-sectional analysis of 160 published cases. Autophagy, 2022, 18, 1715-1727.	9.1	5
72	Cross-sectional analysis on publication status and age representation of clinical studies addressing mechanical ventilation and ventilator-induced lung injury in infants and children. BMJ Open, 2018, 8, e023524.	1.9	4

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73	Publication bias in pediatric emergence delirium: a cross-sectional analysis of ClinicalTrials.gov and ClinicalTrialsRegister.eu. <i>BMJ Open</i> , 2020, 10, e037346.	1.9	4
74	Clinical Trials on Diabetic Nephropathy: A Cross-Sectional Analysis. <i>Diabetes Therapy</i> , 2019, 10, 229-243.	2.5	3
75	Disease awareness or subtle product placement? Orphan diseases featured in the television series "House, M.D." - a cross-sectional analysis. <i>BMC Medical Ethics</i> , 2020, 21, 20.	2.4	3
76	Intuitive Global Insight Into COVID-19 Clinical Research Activities" The "COVID-19 Map of Hope" <i>Journal of Clinical Pharmacology</i> , 2020, 60, 826-827.	2.0	3
77	Missing trial results: analysis of the current publication rate of studies in pediatric dialysis from 2003 to 2020. <i>Pediatric Nephrology</i> , 2023, 38, 227-236.	1.7	3
78	Novel orphan medicines and abandoned pathways - the US Orphan Drug Act of 1983 and the impact on rare rheumatologic diseases and lysosomal storage disorders. <i>Molecular and Cellular Pediatrics</i> , 2015, 2, A1.	1.8	1
79	Decision-making in acute viral bronchiolitis: A universal guideline and a publication gap. <i>PLoS ONE</i> , 2020, 15, e0237801.	2.5	1
80	A Cross-sectional Quantitative Analysis of the Natural History of Alpha-mannosidosis. <i>Neuropediatrics</i> , 2019, 50, .	0.6	1
81	Publication rate and research topics of studies in pediatric kidney transplantation. <i>Pediatric Transplantation</i> , 2022, 26, e14262.	1.0	1
82	Differential diagnosis of Gaucher and Niemann-Pick disease in early childhood. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2007, 91, 159-159.	1.5	0
83	Punctate calcifications in lysosomal storage disorders. <i>Clinical Dysmorphology</i> , 2009, 18, 172-177.	0.3	0
84	A prospective 10year study of individualized, intensified enzyme replacement therapy in advanced Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2015, 114, S103.	1.1	0
85	Spectrum of Clinical Research in Juvenile Idiopathic Arthritis: A Cross-Sectional Analysis of Registered Studies in Clinicaltrials.gov and Clinicaltrialsregister.eu. <i>Biomedicines</i> , 2021, 9, 1860.	3.2	0
86	FDA orphan drug designations for lysosomal storage disorders " a cross-sectional analysis. , 2020, 15, e0230898.		0
87	FDA orphan drug designations for lysosomal storage disorders " a cross-sectional analysis. , 2020, 15, e0230898.		0
88	FDA orphan drug designations for lysosomal storage disorders " a cross-sectional analysis. , 2020, 15, e0230898.		0
89	FDA orphan drug designations for lysosomal storage disorders " a cross-sectional analysis. , 2020, 15, e0230898.		0
90	Decision-making in acute viral bronchiolitis: A universal guideline and a publication gap. , 2020, 15, e0237801.		0

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91	Decision-making in acute viral bronchiolitis: A universal guideline and a publication gap. , 2020, 15, e0237801.		0
92	Decision-making in acute viral bronchiolitis: A universal guideline and a publication gap. , 2020, 15, e0237801.		0
93	Decision-making in acute viral bronchiolitis: A universal guideline and a publication gap. , 2020, 15, e0237801.		0