Robin H Lachmann

List of Publications by Year
in descending order

40	1,852	331670 21	315739 38
papers	citations	h-index	g-index

42	42	42	3251
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Lost in translationâ€"Challenges in drug development for inherited metabolic diseases. Journal of Inherited Metabolic Disease, 2022, 45, 381-382.	3.6	2
2	A randomized, placebo-controlled clinical trial evaluating olipudase alfa enzyme replacement therapy for chronic acid sphingomyelinase deficiency (ASMD) in adults: One-year results. Genetics in Medicine, 2022, 24, 1425-1436.	2.4	30
3	Independent Registries Are Cost-Effective Tools to Provide Mandatory Postauthorization Surveillance for Orphan Medicinal Products. Value in Health, 2021, 24, 268-273.	0.3	7
4	Longâ€term cognitive and psychosocial outcomes in adults with phenylketonuria. Journal of Inherited Metabolic Disease, 2021, 44, 1353-1368.	3.6	24
5	Burosumab treatment in adults with X-linked hypophosphataemia: 96-week patient-reported outcomes and ambulatory function from a randomised phase 3 trial and open-label extension. RMD Open, 2021, 7, e001714.	3.8	26
6	In-depth phenotyping for clinical stratification of Gaucher disease. Orphanet Journal of Rare Diseases, 2021, 16, 431.	2.7	11
7	Treating lysosomal storage disorders: What have we learnt?. Journal of Inherited Metabolic Disease, 2020, 43, 125-132.	3.6	20
8	SLC37A4-CDG: Mislocalization of the glucose-6-phosphate transporter to the Golgi causes a new congenital disorder of glycosylation. Molecular Genetics and Metabolism Reports, 2020, 25, 100636.	1.1	4
9	Registries for orphan drugs: generating evidence or marketing tools?. Orphanet Journal of Rare Diseases, 2020, 15, 235.	2.7	18
10	Ammonia: what adult neurologists need to know. Practical Neurology, 2020, , practneurol-2020-002654.	1.1	2
11	Long-term efficacy of olipudase alfa in adults with acid sphingomyelinase deficiency (ASMD): Further clearance of hepatic sphingomyelin is associated with additional improvements in pro- and anti-atherogenic lipid profiles after 42Âmonths of treatment. Molecular Genetics and Metabolism, 2020, 131, 245-252.	1.1	28
12	Proposed Stages of Myocardial Phenotype Development in FabryÂDisease. JACC: Cardiovascular Imaging, 2019, 12, 1673-1683.	5.3	91
13	The neurological and psychological phenotype of adult patients with earlyâ€treated phenylketonuria: A systematic review. Journal of Inherited Metabolic Disease, 2019, 42, 209-219.	3.6	42
14	Outcome of adult patients with Xâ€linked hypophosphatemia caused by PHEX gene mutations. Journal of Inherited Metabolic Disease, 2018, 41, 865-876.	3.6	103
15	Increased resting cerebral blood flow in adult Fabry disease. Neurology, 2018, 90, e1379-e1385.	1.1	19
16	Olipudase alfa for treatment of acid sphingomyelinase deficiency (ASMD): safety and efficacy in adults treated for 30 months. Journal of Inherited Metabolic Disease, 2018, 41, 829-838.	3.6	92
17	GYG1 causing progressive limb girdle myopathy with onset during teenage years (polyglucosan body) Tj ETQq1 1 0.784314 rgBT /Ove	0.6	8
18	Mucolipidosis type III, a series of adult patients. Journal of Inherited Metabolic Disease, 2018, 41, 839-848.	3.6	14

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19	Isolated aortic root dilation in homocystinuria. Journal of Inherited Metabolic Disease, 2018, 41, 109-115.	3.6	11
20	Cardiac Phenotype of Prehypertrophic Fabry Disease. Circulation: Cardiovascular Imaging, 2018, 11, e007168.	2.6	58
21	Valproate-induced hyperammonemia - uncovering an underlying inherited metabolic disorder: a case report. Journal of Medical Case Reports, 2018, 12, 134.	0.8	15
22	Annual severity increment score as a tool for stratifying patients with Niemann-Pick disease type C and for recruitment to clinical trials. Orphanet Journal of Rare Diseases, 2018, 13, 143.	2.7	41
23	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
24	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. Journal of Medical Genetics, 2017, 54, 288-296.	3.2	262
25	Expanding the phenotype in argininosuccinic aciduria: need for new therapies. Journal of Inherited Metabolic Disease, 2017, 40, 357-368.	3.6	55
26	Issues with European guidelines for phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 681-683.	11.4	26
27	Homocysteine and methylmalonate: when should I measure them and what do they mean?. Practical Neurology, 2016, 16, 328-333.	1.1	1
28	Position statement on the role of healthcare professionals, patient organizations and industry in European Reference Networks. Orphanet Journal of Rare Diseases, 2016, 11, 7.	2.7	14
29	Urinary excretion and metabolism of miglustat and valproate in patients with Niemannâ€“Pick type C1 disease: One- and two-dimensional solution-state 1 H NMR studies. Journal of Pharmaceutical and Biomedical Analysis, 2016, 117, 276-288.	2.8	4
30	Muddâ€™s disease (MAT I/III deficiency): a survey of data for MAT1A homozygotes and compound heterozygotes. Orphanet Journal of Rare Diseases, 2015, 10, 99.	2.7	39
31	A National Pilot Study to Investigate the Effects of Sub-Maximal Aerobic Exercise in Adults with Late-Onset Pompe Disease. Journal of Neuromuscular Diseases, 2015, 2, S67-S68.	2.6	0
32	Chronic kidney disease and an uncertain diagnosis of Fabry disease: Approach to a correct diagnosis. Molecular Genetics and Metabolism, 2015, 114, 242-247.	1.1	51
33	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057.	3.6	186
34	Ten-year outcome of enzyme replacement therapy with agalsidase beta in patients with Fabry disease. Journal of Medical Genetics, 2015, 52, 353-358.	3.2	266
35	A National Pilot Study to Investigate the Effects of Sub-Maximal Aerobic Exercise in Adults with Late-Onset Pompe Disease. Journal of Neuromuscular Diseases, 2015, 2, S67-S68.	2.6	0
36	Enzyme replacement therapy for lysosomal storage diseases. Current Opinion in Pediatrics, 2011, 23, 588-593.	2.0	94

#	Article	IF	Citations
37	Treatments for lysosomal storage disorders. Biochemical Society Transactions, 2010, 38, 1465-1468.	3.4	25
38	Substrate-reduction therapy with miglustat for glycosphingolipid storage disorders affecting the brain. Expert Review of Endocrinology and Metabolism, 2009, 4, 217-224.	2.4	2
39	An Adult with Type 2 Citrullinemia Presenting in Europe. New England Journal of Medicine, 2008, 358, 1408-1409.	27.0	26
40	Very longâ€term outcomes in 23 patients with <scp>cblA</scp> type methylmalonic acidemia. Journal of Inherited Metabolic Disease, 0, , .	3.6	2