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List of Publications by Year in descending order

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932766 1199166 13 496 10 12 citations h-index g-index papers 14 14 14 616 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	A crossâ€sectional, prospective ocular motor study in 72 patients with Niemannâ€Pick disease type C. European Journal of Neurology, 2021, 28, 3040-3050.	1.7	7
2	Sebelipase alfa for lysosomal acid lipase deficiency: 5â€year treatment experience from a phase 2 openâ€label extension study. Liver International, 2020, 40, 2203-2214.	1.9	15
3	Otorhinolaryngological manifestations in 61 patients with mucopolysaccharidosis. International Journal of Pediatric Otorhinolaryngology, 2020, 135, 110137.	0.4	13
4	Evaluation of different suspicion indices in identifying patients with Niemann-Pick disease Type C in clinical practice: a post hoc analysis of a retrospective chart review. Orphanet Journal of Rare Diseases, 2019, 14, 161.	1.2	5
5	Disease characteristics, prognosis and miglustat treatment effects on disease progression in patients with Niemann-Pick disease Type C: an international, multicenter, retrospective chart review. Orphanet Journal of Rare Diseases, 2019, 14, 32.	1.2	37
6	Oculomotor and Vestibular Findings in Gaucher Disease Type 3 and Their Correlation with Neurological Findings. Frontiers in Neurology, 2017, 8, 711.	1.1	20
7	Clinical Features of Lysosomal Acid Lipase Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2015, 61, 619-625.	0.9	92
8	Acetyl- <scp>dl</scp> -leucine in Niemann-Pick type C. Neurology, 2015, 85, 1368-1375.	1.5	88
9	Sebelipase alfa over 52weeks reduces serum transaminases, liver volume and improves serum lipids in patients with lysosomal acid lipase deficiency. Journal of Hepatology, 2014, 61, 1135-1142.	1.8	81
10	Effect of rapid cessation of enzyme replacement therapy: A report of 5 more cases. Molecular Genetics and Metabolism, 2014, 111, 212-213.	0.5	14
11	Clinical effect and safety profile of recombinant human lysosomal acid lipase in patients With cholesteryl ester storage disease. Hepatology, 2013, 58, 950-957.	3.6	99
12	The Use of Elevated Doses of Genistein-Rich Soy Extract in the Gene Expression-Targeted Isoflavone Therapy for Sanfilippo Disease Patients. JIMD Reports, 2011, 5, 21-25.	0.7	25
13	Enzyme replacement therapy for Gaucher disease in twin pregnancy. International Journal of Gynecology and Obstetrics, 2009, 106, 64-66.	1.0	О