Nathalie Guffon

List of Publications by Year
in descending order

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		759233	996975
19	1,559	12	15
papers	citations	h-index	g-index

20	20	20	1214
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Oral treatment for mucopolysaccharidosis <scp>VI</scp>: Outcomes of the first phase <scp>IIa</scp> study with odiparcil. Journal of Inherited Metabolic Disease, 2022, 45, 340-352.	3.6	7
2	Long term disease burden post-transplantation: three decades of observations in 25 Hurler patients successfully treated with hematopoietic stem cell transplantation (HSCT). Orphanet Journal of Rare Diseases, 2021, 16, 60.	2.7	27
3	Clinical outcomes in a series of 18 patients with long chain fatty acids oxidation disorders treated with triheptanoin for a median duration of 22Âmonths. Molecular Genetics and Metabolism, 2021, 132, 227-233.	1.1	9
4	Maladies de Gaucher, de Niemann-Pick par dÃ©ficit en sphingomyÃ©linase acide et de Niemann-Pick type C. Revue Francophone Des Laboratoires, 2021, 2021, 30-34.	0.0	0
5	Aspects cliniques des mucopolysaccharidoses et oligosaccharidoses. Revue Francophone Des Laboratoires, 2021, 2021, 20-29.	0.0	0
6	ThÃ©rapeutiques actuelles et perspectives. Revue Francophone Des Laboratoires, 2021, 2021, 67-70.	0.0	0
7	A rare late progression form of Sly syndrome mucopolysaccharidosis. JIMD Reports, 2019, 49, 1-6.	1.5	8
8	Growth impairment and limited range of joint motion in children should raise suspicion of an attenuated form of mucopolysaccharidosis: expert opinion. European Journal of Pediatrics, 2019, 178, 593-603.	2.7	22
9	Diagnosis, quality of life, and treatment of patients with Hunter syndrome in the French healthcare system: a retrospective observational study. Orphanet Journal of Rare Diseases, 2015, 10, 43.	2.7	47
10	Natural disease history and characterisation of SUMF1 molecular defects in ten unrelated patients with multiple sulfatase deficiency. Orphanet Journal of Rare Diseases, 2015, 10, 31.	2.7	33
11	Natural history and galsulfase treatment in mucopolysaccharidosis VI (MPS VI, Maroteauxâ€“Lamy) Tj ETQq1 1 0.784314 rgBT /Overlock American Journal of Medical Genetics, Part A, 2014, 164, 1953-1964.	1.2	74
12	The Morquio A Clinical Assessment Program: Baseline results illustrating progressive, multisystemic clinical impairments in Morquio A subjects. Molecular Genetics and Metabolism, 2013, 109, 54-61.	1.1	117
13	Evaluation of Miglustat Treatment in Patients with Type III Mucopolysaccharidosis: A Randomized, Double-Blind, Placebo-Controlled Study. Journal of Pediatrics, 2011, 159, 838-844.e1.	1.8	28
14	Enzyme replacement therapy for mucopolysaccharidosis VI: evaluation of longâ€term pulmonary function in patients treated with recombinant human Nâ€acetylgalactosamine 4â€sulfatase. Journal of Inherited Metabolic Disease, 2010, 33, 51-60.	3.6	80
15	Home treatment with intravenous enzyme replacement therapy with idursulfase for mucopolysaccharidosis type II â€" data from the Hunter Outcome Survey. Molecular Genetics and Metabolism, 2010, 101, 123-129.	1.1	26
16	Mucopolysaccharidosis type II (Hunter syndrome): a clinical review and recommendations for treatment in the era of enzyme replacement therapy. European Journal of Pediatrics, 2008, 167, 267-277.	2.7	418
17	Long-term follow-up of endurance and safety outcomes during enzyme replacement therapy for mucopolysaccharidosis VI: Final results of three clinical studies of recombinant human N-acetylgalactosamine 4-sulfatase. Molecular Genetics and Metabolism, 2008, 94, 469-475.	1.1	198
18	Enzyme replacement therapy for mucopolysaccharidosis VI: A phase 3, randomized, double-blind, placebo-controlled, multinational study of recombinant human N-acetylgalactosamine 4-sulfatase (recombinant human arylsulfatase B or rhASB) and follow-on, open-label extension study. Journal of Pediatrics, 2006, 148, 533-539.e6.	1.8	335

#	Article	IF	Citations
19	Threshold effect of urinary glycosaminoglycans and the walk test as indicators of disease progression in a survey of subjects with Mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). , 2005, 134A, 144-150.		130