Pranoot Tanpaiboon

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8937738/publications.pdf

Version: 2024-02-01

26	478	759233 12	713466 21
papers	citations	h-index	g-index

26	26	26	896
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Long-term efficacy and safety of vestronidase alfa enzyme replacement therapy in pediatric subjects < 5 years with mucopolysaccharidosis VII. Molecular Genetics and Metabolism, 2022, 136, 28-37.	1.1	4
2	A patient with atypical presentation of chronic hepatosteatosis harboring a novel variant in the CPT1A gene. European Journal of Medical Genetics, 2021, 64, 104034.	1.3	2
3	Transforming the clinical outcome in CRIM-negative infantile Pompe disease identified via newborn screening: the benefits of early treatment with enzyme replacement therapy and immune tolerance induction. Genetics in Medicine, 2021, 23, 845-855.	2.4	26
4	Prenatal diagnosis of diencephalicâ€mesencephalic junction dysplasia: Fetal magnetic resonance imaging phenotypes, genetic diagnoses, and outcomes. Prenatal Diagnosis, 2021, 41, 778-790.	2.3	4
5	Infantile onset Sandhoff disease: clinical manifestation and a novel common mutation in Thai patients. BMC Pediatrics, 2021, 21, 22.	1.7	8
6	Noonan Syndrome in Thai Children. Indian Pediatrics, 2020, 57, 967-968.	0.4	0
7	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	3.8	17
8	Practical management of lysosomal storage disorders (LSDs). Translational Science of Rare Diseases, 2020, 4, 133-157.	1.5	5
9	GARSâ€related disease in infantile spinal muscular atrophy: Implications for diagnosis and treatment. American Journal of Medical Genetics, Part A, 2020, 182, 1167-1176.	1.2	15
10	Noonan Syndrome in Thai Children. Indian Pediatrics, 2020, 57, 967-968.	0.4	0
11	An immune tolerance approach using transient low-dose methotrexate in the ERT-naÃ¯ve setting of patients treated with a therapeutic protein: experience in infantile-onset Pompe disease. Genetics in Medicine, 2019, 21, 887-895.	2.4	28
12	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
13	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
14	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
15	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
16	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
17	Sustained immune tolerance induction in enzyme replacement therapyâ€“treated CRIM-negative patients with infantile Pompe disease. JCI Insight, 2017, 2, .	5.0	47
18	Role of elosulfase alfa in mucopolysaccharidosis IVA. The Application of Clinical Genetics, 2016, 9, 67.	3.0	14

#	Article	IF	Citations
19	Isolated methylmalonic acidemia with unusual presentation mimicking diabetic ketoacidosis. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 373-8.	0.9	16
20	Immune response to enzyme replacement therapies in lysosomal storage diseases and the role of immune tolerance induction. Molecular Genetics and Metabolism, 2016, 117, 66-83.	1.1	64
21	Survival and Developmental Milestones Among Pompe Registry Patients with Classic Infantile-Onset Pompe Disease with Different Timing of Initiation of Treatment with Enzyme Replacement Therapy. Journal of Neuromuscular Diseases, 2015, 2, S61-S62.	2.6	5
22	Elosulfase alfa for the treatment of mucopolysacchoridosis IVA. Expert Review of Endocrinology and Metabolism, 2015, 10, 569-579.	2.4	1
23	Survival and Developmental Milestones Among Pompe Registry Patients with Classic Infantile-Onset Pompe Disease with Different Timing of Initiation of Treatment with Enzyme Replacement Therapy. Journal of Neuromuscular Diseases, 2015, 2, S61-S62.	2.6	2
24	Noncompaction of the ventricular myocardium and hydrops fetalis in cobalamin C disease. Journal of Inherited Metabolic Disease, 2013, 36, 1085-1085.	3.6	0
25	Noncompaction of the Ventricular Myocardium and Hydrops Fetalis in Cobalamin C Disease. JIMD Reports, 2012, 10, 33-38.	1.5	22
26	IEM Digest. Molecular Genetics and Metabolism, 2005, 85, 2-6.	1.1	35