Hanna Mandel

List of Publications by Year
in descending order

14	327	933447 10	1058476 14
papers	citations	h-index	g-index

14	14	14	583
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. Journal of Inherited Metabolic Disease, 2021, 44, 376-387.	3.6	47
2	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	6.2	44
3	Erythrocyte Encapsulated Thymidine Phosphorylase for the Treatment of Patients with Mitochondrial Neurogastrointestinal Encephalomyopathy: Study Protocol for a Multi-Centre, Multiple Dose, Open Label Trial. Journal of Clinical Medicine, 2019, 8, 1096.	2.4	39
4	Safety and Efficacy of Erythrocyte Encapsulated Thymidine Phosphorylase in Mitochondrial Neurogastrointestinal Encephalomyopathy. Journal of Clinical Medicine, 2019, 8, 457.	2.4	36
5	Two separate functions of NME3 critical for cell survival underlie a neurodegenerative disorder. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 566-574.	7.1	36
6	De novo GRIN1 mutations: An emerging cause of severe early infantile encephalopathy. European Journal of Medical Genetics, 2017, 60, 317-320.	1.3	32
7	Newborn screening for cerebrotendinous xanthomatosis is the solution for early identification and treatment. Journal of Lipid Research, 2018, 59, 2214-2222.	4.2	28
8	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
9	Sedaghatian-type spondylometaphyseal dysplasia: Whole exome sequencing in neonatal dry blood spots enabled identification of a novel variant in GPX4. European Journal of Medical Genetics, 2020, 63, 104020.	1.3	12
10	Mammalian Homologue NME3 of DYNAMO1 Regulates Peroxisome Division. International Journal of Molecular Sciences, 2020, 21, 8040.	4.1	11
11	<scp>COG6â€CDG</scp>: Expanding the phenotype with emphasis on glycosylation defects involved in the causation of male disorders of sex development. Clinical Genetics, 2020, 98, 402-407.	2.0	8
12	Congenital Hypotonia: Cracking a SAGA of consanguineous kindred harboring four genetic variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1849.	1.2	6
13	Severe infantile epileptic encephalopathy associated with D-glyceric aciduria: report of a novel case and review. Metabolic Brain Disease, 2019, 34, 557-563.	2.9	4
14	Concomitant congenital CMV infection and inherited liver diseases. European Journal of Medical Genetics, 2021, 64, 104249.	1.3	2