# 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 | Congenital Hypotonia: Cracking a SAGA of consanguineous kindred harboring four genetic variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1849. | 1.2 | 6 |
| 2 | 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 |
| 3 | Concomitant congenital CMV infection and inherited liver diseases. European Journal of Medical Genetics, 2021, 64, 104249. | 1.3 | 2 |
| 4 | <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 |
| 5 | 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 |
| 6 | Mammalian Homologue NME3 of DYNAMO1 Regulates Peroxisome Division. International Journal of Molecular Sciences, 2020, 21, 8040. | 4.1 | 11 |
| 7 | 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 |
| 8 | Safety and Efficacy of Erythrocyte Encapsulated Thymidine Phosphorylase in Mitochondrial Neurogastrointestinal Encephalomyopathy. Journal of Clinical Medicine, 2019, 8, 457. | 2.4 | 36 |
| 9 | 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 |
| 10 | De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212. | 6.2 | 44 |
| 11 | 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 |
| 12 | Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578. | 2.5 | 22 |
| 13 | Newborn screening for cerebrotendinous xanthomatosis is the solution for early identification and treatment. Journal of Lipid Research, 2018, 59, 2214-2222. | 4.2 | 28 |
| 14 | De novo GRIN1 mutations: An emerging cause of severe early infantile encephalopathy. European Journal of Medical Genetics, 2017, 60, 317-320. | 1.3 | 32 |