

Meral Topcu

List of Publications by Year in descending order

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Version: 2024-02-01

19
papers

391
citations

1040056

9
h-index

839539

18
g-index

19
all docs

19
docs citations

19
times ranked

1289
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|--|------|-----------|
| 1 | Mutations in CLN7/MFSD8 are a common cause of variant late-infantile neuronal ceroid lipofuscinosis. <i>Brain</i> , 2009, 132, 810-819. | 7.6 | 116 |
| 2 | Management Strategies for CLN2 Disease. <i>Pediatric Neurology</i> , 2017, 69, 102-112. | 2.1 | 80 |
| 3 | Clinical outcomes of two patients with a novel pathogenic variant in ASNS: response to asparagine supplementation and review of the literature. <i>Human Genome Variation</i> , 2019, 6, 24. | 0.7 | 31 |
| 4 | MINPP1 prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. <i>Nature Communications</i> , 2020, 11, 6087. | 12.8 | 28 |
| 5 | Etiological yield of SNP microarrays in idiopathic intellectual disability. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 327-337. | 1.6 | 26 |
| 6 | Griscelli syndrome: Report of Three Cases. <i>Pediatric Pathology & Laboratory Medicine: Journal of the Society for Pediatric Pathology, Affiliated With the International Paediatric Pathology Association</i> , 1995, 15, 309-319. | 0.3 | 24 |
| 7 | Electrical status epilepticus during sleep: A study of 22 patients. <i>Brain and Development</i> , 2015, 37, 250-264. | 1.1 | 13 |
| 8 | Long-term effects of vagus nerve stimulation in refractory pediatric epilepsy: A single-center experience. <i>Epilepsy and Behavior</i> , 2020, 110, 107147. | 1.7 | 13 |
| 9 | Neurologic Involvement in Primary Immunodeficiency Disorders. <i>Journal of Child Neurology</i> , 2018, 33, 320-328. | 1.4 | 12 |
| 10 | Genetic and phenotypic features of patients with childhood ataxias diagnosed by next-generation sequencing gene panel. <i>Brain and Development</i> , 2020, 42, 6-18. | 1.1 | 12 |
| 11 | Management of CLN1 Disease: International Clinical Consensus. <i>Pediatric Neurology</i> , 2021, 120, 38-51. | 2.1 | 10 |
| 12 | The Complex Genetic Landscape of Hereditary Ataxias in Turkey and Implications in Clinical Practice. <i>Movement Disorders</i> , 2021, 36, 1676-1688. | 3.9 | 9 |
| 13 | Developmental abnormalities and mental retardation. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , 2013, 111, 211-217. | 1.8 | 5 |
| 14 | Unraveling neuronal ceroid lipofuscinosis type 2 (CLN2) disease: A tertiary center experience for determinants of diagnostic delay. <i>European Journal of Paediatric Neurology</i> , 2021, 33, 94-98. | 1.6 | 4 |
| 15 | Comprehensive clinical, biochemical, radiological and genetic analysis of 28 Turkish cases with suspected metachromatic leukodystrophy and their relatives. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100688. | 1.1 | 4 |
| 16 | Mystery Case: Pontine tegmental cap dysplasia in a neonate. <i>Neurology</i> , 2018, 91, e2100-e2101. | 1.1 | 2 |
| 17 | Letters to the editor. <i>Muscle and Nerve</i> , 1996, 19, 675-681. | 2.2 | 1 |
| 18 | Prenatal enzymatic diagnosis of lysosomal storage diseases using cultured amniotic cells, uncultured chorionic villus samples, and fetal blood cells: Hacettepe experience. <i>Prenatal Diagnosis</i> , 2019, 39, 1080-1085. | 2.3 | 1 |

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|----|---|-----|-----------|
| 19 | Effects of Johnstone pressure splints combined with neurodevelopmental therapy on spasticity and cutaneous sensory inputs in spastic cerebral palsy. <i>Developmental Medicine and Child Neurology</i> , 2001, 43, 307-313. | 2.1 | 0 |