

Mustafa Dogan

List of Publications by Year in descending order

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Version: 2024-02-01

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2258059

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| # | ARTICLE | IF | CITATIONS |
|---|---|-----|-----------|
| 1 | Chorioretinal dystrophy, hypogonadotropic hypogonadism, and cerebellar ataxia: Boucher-Neuhauser syndrome due to a homozygous (c.3524C>G (p.Ser1175Cys)) variant in <i>PNPLA6</i> gene. <i>Ophthalmic Genetics</i> , 2021, 42, 276-282. | 1.2 | 7 |
| 2 | Clinical and molecular findings in a Turkish family with an ultra-rare condition, ELP2-related neurodevelopmental disorder. <i>Molecular Biology Reports</i> , 2021, 48, 701-708. | 2.3 | 4 |
| 3 | Biallelic Novel USP53 Splicing Variant Disrupting the Gene Function that Causes Cholestasis Phenotype and Review of the Literature. <i>Molecular Syndromology</i> , 2022, 13, 471-484. | 0.8 | 4 |
| 4 | Study of ten causal genes in Turkish patients with clinically suspected maturity-onset diabetes of the young (MODY) using a targeted next-generation sequencing panel. <i>Molecular Biology Reports</i> , 0, , . | 2.3 | 4 |
| 5 | Deficiency of alkaline ceramidase 3 with infancy-onset progressive leukoencephalopathy: a second case report. <i>Acta Neurologica Belgica</i> , 2021, 121, 1867-1870. | 1.1 | 2 |
| 6 | Clinical, radiological and computational studies on two novel GNPTG variants causing mucopolipidosis III gamma phenotypes with varying severity. <i>Molecular Biology Reports</i> , 2021, 48, 1465-1474. | 2.3 | 2 |
| 7 | A case with Turner syndrome and alopecia areata. <i>OrtadoÄŸu TÄ±p Dergisi</i> , 2019, 11, 631-633. | 0.1 | 0 |