# Aysegul Tokatli

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8527972/publications.pdf

Version: 2024-02-01

| 21 | 244 | 933447 10 | 996975 15 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 22 | 22 | 22 | 494 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Oral health status of children and young adults with maple syrup urine disease in Turkey. BMC Oral Health, 2021, 21, 8. | 2.3 | 1 |
| 2 | Sensory, voluntary, and motor postural control in children and adolescents with mucopolysaccharidosis. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 583-589. | 0.9 | 0 |
| 3 | Evaluation of Cardiac Findings in Mucopolysaccharidosis Type III Patients. Journal of Pediatric Research, 2021, 8, 195-201. | 0.2 | 0 |
| 4 | DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant. Metabolic Brain Disease, 2021, 36, 1405-1410. | 2.9 | 8 |
| 5 | Oral health status of children with phenylketonuria. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 361-365. | 0.9 | 8 |
| 6 | Comment on: â€œMultiple acylâ€'CoA dehydrogenase deficiency in elderly carriersâ€•. Journal of Neurology, 2020, 267, 1209-1210. | 3.6 | 1 |
| 7 | Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department. European Journal of Pediatrics, 2020, 179, 1107-1114. | 2.7 | 4 |
| 8 | Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe. American Journal of Medical Genetics, Part A, 2020, 182, 705-712. | 1.2 | 10 |
| 9 | Retrospective evaluation of 85 patients with urea cycle disorders: one center experience, three new mutations. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 721-728. | 0.9 | 3 |
| 10 | The effectiveness of enzyme replacement therapy on cardiac findings in patients with mucopolysaccharidosis. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 1049-1053. | 0.9 | 9 |
| 11 | Post-mortem detection of FLAD1 mutations in 2 Turkish siblings with hypotonia in early infancy. Neuromuscular Disorders, 2018, 28, 787-790. | 0.6 | 14 |
| 12 | Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride. Gynecological Endocrinology, 2017, 33, 19-20. | 1.7 | 3 |
| 13 | Genotypicâ€phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey. American Journal of Medical Genetics, Part A, 2017, 173, 2954-2967. | 1.2 | 17 |
| 14 | Hereditary Dopamine Transporter Deficiency Syndrome: Challenges in Diagnosis and Treatment. Neuropediatrics, 2017, 48, 049-052. | 0.6 | 28 |
| 15 | Evaluation and identification of IDUA gene mutations in Turkishpatients with mucopolysaccharidosis type I. Turkish Journal of Medical Sciences, 2016, 46, 404-408. | 0.9 | 11 |
| 16 | Key features and clinical variability of COG6-CDG. Molecular Genetics and Metabolism, 2015, 116, 163-170. | 1.1 | 49 |
| 17 | Lack of prolidase causes a bone phenotype both in human and in mouse. Bone, 2015, 72, 53-64. | 2.9 | 23 |
| 18 | Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia. European Journal of Medical Genetics, 2014, 57, 596-601. | 1.3 | 15 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | High prevalence of cerebral venous sinus thrombosis (CVST) as presentation of cystathionine beta-synthase deficiency in childhood: Molecular and clinical findings of Turkish probands. Gene, 2014, 534, 197-203. | 2.2 | 13 |
| 20 | Acetaminophen-induced hepatotoxicity in a glutathione synthetase-deficient patient. Turkish Journal of Pediatrics, 2007, 49, 75-6. | 0.6 | 10 |
| 21 | Molecular analysis of 16 Turkish families with DHPR deficiency using denaturing gradient gel electrophoresis (DGGE). Human Genetics, 2000, 107, 546-553. | 3.8 | 16 |