

# Aysegul Tokatli

## List of Publications by Year in descending order

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Version: 2024-02-01

21  
papers

244  
citations

933447

10  
h-index

996975

15  
g-index

22  
all docs

22  
docs citations

22  
times ranked

494  
citing authors

#	ARTICLE	IF	CITATIONS
1	Key features and clinical variability of COG6-CDG. <i>Molecular Genetics and Metabolism</i> , 2015, 116, 163-170.	1.1	49
2	Hereditary Dopamine Transporter Deficiency Syndrome: Challenges in Diagnosis and Treatment. <i>Neuropediatrics</i> , 2017, 48, 049-052.	0.6	28
3	Lack of prolidase causes a bone phenotype both in human and in mouse. <i>Bone</i> , 2015, 72, 53-64.	2.9	23
4	Genotypic and phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2954-2967.	1.2	17
5	Molecular analysis of 16 Turkish families with DHPR deficiency using denaturing gradient gel electrophoresis (DGGE). <i>Human Genetics</i> , 2000, 107, 546-553.	3.8	16
6	Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia. <i>European Journal of Medical Genetics</i> , 2014, 57, 596-601.	1.3	15
7	Post-mortem detection of FLAD1 mutations in 2 Turkish siblings with hypotonia in early infancy. <i>Neuromuscular Disorders</i> , 2018, 28, 787-790.	0.6	14
8	High prevalence of cerebral venous sinus thrombosis (CVST) as presentation of cystathionine beta-synthase deficiency in childhood: Molecular and clinical findings of Turkish probands. <i>Gene</i> , 2014, 534, 197-203.	2.2	13
9	Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I. <i>Turkish Journal of Medical Sciences</i> , 2016, 46, 404-408.	0.9	11
10	Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 705-712.	1.2	10
11	Acetaminophen-induced hepatotoxicity in a glutathione synthetase-deficient patient. <i>Turkish Journal of Pediatrics</i> , 2007, 49, 75-6.	0.6	10
12	The effectiveness of enzyme replacement therapy on cardiac findings in patients with mucopolysaccharidosis. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2019, 32, 1049-1053.	0.9	9
13	Oral health status of children with phenylketonuria. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2020, 33, 361-365.	0.9	8
14	DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant. <i>Metabolic Brain Disease</i> , 2021, 36, 1405-1410.	2.9	8
15	Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department. <i>European Journal of Pediatrics</i> , 2020, 179, 1107-1114.	2.7	4
16	Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride. <i>Gynecological Endocrinology</i> , 2017, 33, 19-20.	1.7	3
17	Retrospective evaluation of 85 patients with urea cycle disorders: one center experience, three new mutations. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2020, 33, 721-728.	0.9	3
18	Comment on: "Multiple acyl-CoA dehydrogenase deficiency in elderly carriers". <i>Journal of Neurology</i> , 2020, 267, 1209-1210.	3.6	1

#	ARTICLE	IF	CITATIONS
19	Oral health status of children and young adults with maple syrup urine disease in Turkey. BMC Oral Health, 2021, 21, 8.	2.3	1
20	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
21	Evaluation of Cardiac Findings in Mucopolysaccharidosis Type III Patients. Journal of Pediatric Research, 2021, 8, 195-201.	0.2	0