Halise Neslihan Ã–nenli Mungan

List of Publications by Year
in descending order

42 papers	1,264 citations	840119 11 h-index	360668 35 g-index
46 all docs	46 docs citations	46 times ranked	1563 citing authors

#	Article	IF	Citations
1	Herediter Tirozinemi Tip-1 ve Tip-1 Diabetes Mellitus BirlikteliÄŸinde Diyet YÃ¶netimi: Olgu Sunumu. Journal of Nutrition and Dietetics, 2022, 49, 115-120.	0.1	0
2	A rare cause of hydrops fetalis in two Gaucher disease type 2 patients with a novel mutation. Metabolic Brain Disease, 2022, , .	1.4	2
3	The outcome of 41 Late-Diagnosed Turkish GA-1 Patients: A Candidate for the Turkish NBS. Neuropediatrics, 2021, 52, 358-369.	0.3	4
4	Morquio A syndrome and effect of enzyme replacement therapy in different age groups of Turkish patients: a case series. Orphanet Journal of Rare Diseases, 2021, 16, 144.	1.2	5
5	Mucopolysaccharidosis Type-II with Pathognomonic Skin Appearance: A Case with Pebbling Sign. Journal of Pediatric Research, 2021, 8, 206-208.	0.1	0
6	Clinical and molecular findings in 37 Turkish patients with isolated methylmalonic acidemia. Turkish Journal of Medical Sciences, 2021, 51, 1220-1228.	0.4	1
7	Coexistence of molybdenum cofactor deficiency type A and hypertrophic pyloric stenosis, a new case. Turk Pediatri Arsivi, 2020, 56, 78-80.	0.9	0
8	Congenital erythropoietic porphyria with erythrodontia: A case report. International Journal of Paediatric Dentistry, 2019, 29, 542-548.	1.0	4
9	Mucopolysaccharidosis type VI, 9 sibling pairs and 1 set of three siblings: single center experience from Turkey. Molecular Genetics and Metabolism, 2018, 123, S101.	0.5	0
10	A case with Pallister-Killian syndrome misdiagnosed as mucopolysaccharidosis. Molecular Genetics and Metabolism, 2018, 123, S100.	0.5	0
11	Early onset alpha-mannosidosis: A Turkish case. Molecular Genetics and Metabolism, 2018, 123, S100-S101.	0.5	0
12	An ignored cause of red urine in children: rhabdomyolysis due to carnitine palmitoyltransferase II (CPT-II) deficiency. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 237-239.	0.4	3
13	Improved metabolic control in tetrahydrobiopterin (BH4), responsive phenylketonuria with sapropterin administered in two divided doses vs. a single daily dose. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 713-718.	0.4	6
14	Prospective Turkish Cohort Study to Investigate the Frequency of Niemann-Pick Disease Type C Mutations in Consanguineous Families with at Least One Homozygous Family Member. Molecular Diagnosis and Therapy, 2017, 21, 643-651.	1.6	5
15	Combination of two different homozygote mutations in Pompe disease. Pediatrics International, 2016, 58, 241-243.	0.2	1
16	Tyrosinemia type 1 and irreversible neurologic crisis after one month discontinuation of nitisone. Metabolic Brain Disease, 2016, 31, 1181-1183.	1.4	12
17	Propionic acidemia: a Turkish case report of a successful pregnancy, labor and lactation. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 863-6.	0.4	3
18	Brown-Vialetto-Van Laere syndrome: two siblings with a new mutation and dramatic therapeutic effect of high-dose riboflavin. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 227-31.	0.4	7

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19	Homozygous familial hypobetalipoproteinemia: A Turkish case carrying a missense mutation in apolipoprotein B. Clinica Chimica Acta, 2016, 452, 185-190.	0.5	11
20	Clinical findings and effect of sodium hydrogen carbonate in patients with glutathione synthetase deficiency. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 481-5.	0.4	5
21	A Desensitization Method to Maintain Enzyme Replacement Therapy in Mucopolysaccharidosis Type VI. Journal of Investigational Allergology and Clinical Immunology, 2016, 26, 130-132.	0.6	3
22	A Case Report of a Very Rare Association of Tyrosinemia type I and Pancreatitis Mimicking Neurologic Crisis of Tyrosinemia Type I. Balkan Medical Journal, 2016, 33, 370-372.	0.3	6
23	A 17-Year-Old Girl with Chronic Intermittent Abdominal Pain. Pediatric Annals, 2015, 44, 139-141.	0.3	3
24	Two Novel Missense Mutations in Nonketotic Hyperglycinemia. Journal of Child Neurology, 2015, 30, 789-792.	0.7	5
25	An asymptomatic mother diagnosed with 3-methylcrotonyl-CoA carboxylase deficiency after newborn screening. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 669-71.	0.4	5
26	Primary systemic carnitine deficiency: a Turkish case with a novel homozygous SLC22A5 mutation and 14 years follow-up. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1179-81.	0.4	5
27	Hashimoto's encephalopathy: four cases and review of literature. International Journal of Neuroscience, 2014, 124, 302-306.	0.8	30
28	Loss-of-Function Mutations inPNPLA6Encoding Neuropathy Target Esterase Underlie Pubertal Failure and Neurological Deficits in Gordon Holmes Syndrome. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2067-E2075.	1.8	92
29	Multiple sulfatase deficiency: A case series of four children. Annals of Indian Academy of Neurology, 2013, 16, 720.	0.2	10
30	Distribution of Gene Mutations Associated with Familial Normosmic Idiopathic Hypogonadotropic Hypogonadism. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 121-126.	0.4	28
31	Chromium levels in healthy and newly diagnosed type 1 diabetic children. Pediatrics International, 2012, 54, 780-785.	0.2	12
32	A homozygous recurring mutation in WISP3 causing progressive pseudorheumatoid arthropathy. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 105-8.	0.4	14
33	Wolcott-Rallison syndrome due to the same mutation (W522X) in EIF2AK3 in two unrelated families and review of the literature*. Pediatric Diabetes, 2010, 11, 279-285.	1.2	43
34	TAC3 and TACR3 mutations in familial hypogonadotropic hypogonadism reveal a key role for Neurokinin B in the central control of reproduction. Nature Genetics, 2009, 41, 354-358.	9.4	817
35	Ambulatory Blood Pressure Monitoring and Serum Nitric Oxide Concentration in Type 1 Diabetic Children. Endocrine Journal, 2009, 56, 477-485.	0.7	12
36	Molecular Genetic Analysis of Normosmic Hypogonadotropic Hypogonadism in a Turkish Population: Identification and Detailed Functional Characterization of a Novel Mutation in the Gonadotropin-Releasing Hormone Receptor Gene. Neuroendocrinology, 2006, 84, 301-308.	1.2	41

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37	The importance of arginine mutation for the evolutionary structure and function of phenylalanine hydroxylase gene. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2006, 601, 39-45.	0.4	5
38	Bone calcium changes during diabetic ketoacidosis: A comparison with lactic acidosis due to volume depletion. Bone, 2005, 37, 122-127.	1.4	21
39	Three Children with Triple A Syndrome due to a Mutation (R478X) in the AAAS Gene. Hormone Research in Paediatrics, 2004, 61, 3-6.	0.8	13
40	Bone Mineral Changes in Acute Metabolic Acidosis due to Acute Gastroenteritis. Calcified Tissue International, 2004, 75, 380-383.	1.5	3
41	Seroprevalence of rubella in school girls and pregnant women. European Journal of Epidemiology, 2002, 18, 81-84.	2.5	21
42	Primary Hyperparathyroidism in an Infant with Three Parathyroid Glands and Pulmonary Calcinosis. Journal of Pediatric Endocrinology and Metabolism, 2001, 14, 1173-1176.	0.4	6