Ilyas Okur

List of Publications by Year
in descending order

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49	565	686830 13	676716 22
papers	citations	h-index	g-index

56	56	56	1036
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Monocarboxylate Transporter 1 Deficiency and Ketone Utilization. New England Journal of Medicine, 2014, 371, 1900-1907.	13.9	86
2	IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel Î±-L-iduronidase (IDUA) alleles. Human Mutation, 2011, 32, E2189-E2210.	1.1	66
3	N-carbamylglutamate treatment for acute neonatal hyperammonemia in isovaleric acidemia. European Journal of Pediatrics, 2011, 170, 799-801.	1.3	37
4	Screening for Fabry disease in patients undergoing dialysis for chronic renal failure in Turkey: Identification of new case with novel mutation. Gene, 2013, 527, 42-47.	1.0	35
5	Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 236-245.	2.6	28
6	Vitamin D intoxication and hypercalcaemia in an infant treated with pamidronate infusions. European Journal of Pediatrics, 2004, 163, 163-165.	1.3	26
7	Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R. Journal of Inherited Metabolic Disease, 2011, 34, 225-231.	1.7	22
8	Structure-function analyses of microsomal triglyceride transfer protein missense mutations in abetalipoproteinemia and hypobetalipoproteinemia subjects. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2016, 1861, 1623-1633.	1.2	21
9	Quality of life in children treated with restrictive diet for inherited metabolic disease. Pediatrics International, 2013, 55, 428-433.	0.2	20
10	Very long-chain acyl CoA dehydrogenase deficiency which was accepted as infanticide. Forensic Science International, 2011, 210, e1-e3.	1.3	19
11	Audiologic evaluations of children with mucopolysaccharidosis. Brazilian Journal of Otorhinolaryngology, 2016, 82, 281-284.	0.4	19
12	Crisponi syndrome: A new case with additional features and new mutation in CRLF1. American Journal of Medical Genetics, Part A, 2008, 146A, 3237-3239.	0.7	16
13	Two novel deletions in hypotoniaâ€“cystinuria syndrome. Molecular Genetics and Metabolism, 2012, 107, 614-616.	0.5	16
14	3-Methylcrotonyl-CoA Carboxylase Deficiency: Phenotypic Variability in a Family. Journal of Child Neurology, 2009, 24, 478-481.	0.7	13
15	Oxidized low-density lipoprotein levels and carotid intima-media thickness as markers of early atherosclerosis in prepubertal obese children. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 657-62.	0.4	13
16	Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa. Annals of Nutrition and Metabolism, 2020, 76, 233-241.	1.0	12
17	Autism: Screening of inborn errors of metabolism and unexpected results. Autism Research, 2021, 14, 887-896.	2.1	12
18	Serum dipeptidyl peptidase-IV: A better screening test for early detection of mucopolysaccharidosis?. Clinica Chimica Acta, 2014, 431, 250-254.	0.5	10

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19	Nutritional Status of Syrian Refugees in Early Adolescence Living in Turkey. Journal of Immigrant and Minority Health, 2020, 22, 1149-1154.	0.8	8
20	ICV-administered tralesinidase alfa (BMN 250 NAGLU-IGF2) is well-tolerated and reduces heparan sulfate accumulation in the CNS of subjects with Sanfilippo syndrome type B (MPS IIIB). Molecular Genetics and Metabolism, 2019, 126, S40.	0.5	8
21	The levels of asymmetric dimethylarginine, homocysteine and carotid intima-media thickness in hypercholesterolemic children. Turkish Journal of Pediatrics, 2011, 53, 522-7.	0.3	8
22	The role of viral agents in aetiopathogenesis of acute rheumatic fever. Clinical Rheumatology, 2011, 30, 15-20.	1.0	7
23	Rapid screening of 10 common mutations in Turkish Gaucher patients using electronic DNA microarray. Blood Cells, Molecules, and Diseases, 2008, 40, 246-247.	0.6	6
24	High incidence of co-existing factors significantly modifying the phenotype in patients with Fabry disease. Gene, 2019, 687, 280-288.	1.0	6
25	CDH13 and LPHN3 Gene Polymorphisms in Attention-Deficit/Hyperactivity Disorder: Their Relation to Clinical Characteristics. Journal of Molecular Neuroscience, 2021, 71, 394-408.	1.1	5
26	Two patients from Turkey with a novel variant in the GM2A gene and review of the literature. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 805-812.	0.4	5
27	The Janus-faced manifestations of homozygous familial hypobetalipoproteinemia due to apolipoprotein B truncations. Journal of Clinical Lipidology, 2015, 9, 400-405.	0.6	4
28	Hypophosphatasia: is it an underdiagnosed disease even by expert physicians?. Journal of Bone and Mineral Metabolism, 2021, 39, 598-605.	1.3	4
29	Giant Bronchogenic Cyst Mimicking Tension Pneumothorax. Asian Cardiovascular and Thoracic Annals, 2006, 14, 244-246.	0.2	3
30	Lipid apheresis applications in childhood: Experience in the University Hospital of Gazi. Transfusion and Apheresis Science, 2008, 39, 235-240.	0.5	3
31	Epilepsy in Biotinidase Deficiency Is Distinct from Early Myoclonic Encephalopathy. Neuropediatrics, 2018, 49, 417-418.	0.3	3
32	Familial hyperphosphatemic tumoral calcinosis in an unusual and usual sites and dramatic improvement with the treatment of acetazolamide, sevelamer and topical sodium thiosulfate. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 813-816.	0.4	3
33	First successful concomitant therapy of immune tolerance induction therapy and desensitization in a CRIM-negative infantile Pompe patient. Journal of Pediatric Endocrinology and Metabolism, 2022, 35, 273-277.	0.4	3
34	Glikojen depo tip 1a ve tip 1b olgularÄ±nda klinik Ã¶zellikler ve hastalÄ±ÄŸÄ±n seyri. Turk Pediatri Arsivi, 2013, 48, 117-122.	0.9	2
35	In vitro translational readthrough by gentamicin and geneticin improves GLA activity in Fabry disease. Molecular Genetics and Metabolism, 2017, 120, S43.	0.5	2
36	Citrullinemia with an atypical presentation: Paroxysmal hypoventilation attacks. Journal of Pediatric Neurosciences, 2018, 13, 276.	0.2	2

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37	The first case with FBXL4 mutation successfully treated with a parenteral ketogenic diet for lactic acidosis. Journal of Parenteral and Enteral Nutrition, 2021, 45, 1788-1792.	1.3	1
38	Infantile Onset Glycogen Storage Disease Type 2: Case Report. Guncel Pediatri, 2014, 12, 131-134.	0.1	1
39	Clinical and event-based outcomes of patients with mucopolysaccharidosis VI receiving enzyme replacement therapy in Turkey: a case series. Orphanet Journal of Rare Diseases, 2021, 16, 438.	1.2	1
40	Vitamin D Levels and Bone Mineral Density in Inborn Errors of Metabolism Requiring Specialised Diets. Journal of the College of Physicians and Surgeons--Pakistan: JCPSP, 2019, 29, 1207-1211.	0.2	1
41	Hypercalcemia in glycogen storage disease type I patients of Turkish origin. Turkish Journal of Pediatrics, 2012, 54, 35-7.	0.3	1
42	Importance of family screening in Fabry disease: Reaching the bottom of the iceberg. Molecular Genetics and Metabolism, 2015, 114, S43.	0.5	0
43	Patient With Niemann-Pick Type C Presenting With a Jaw Mass Characterized With Lymph Node Involvement by Niemann-Pick Cells. Journal of Pediatric Hematology/Oncology, 2018, 40, 243-245.	0.3	0
44	Hematologic Findings of Inherited Metabolic Disease: They are More Than Expected. Journal of Pediatric Hematology/Oncology, 2018, 40, 355-359.	0.3	0
45	Ultra-Rare Disorder in a Young Girl with Lipodystrophy: Analbuminemia. Indian Journal of Pediatrics, 2021, 88, 723-723.	0.3	0
46	Congenital defects of glycosylation: Novel presentations with mainly neurological involvement and variable dysmorphic features. American Journal of Medical Genetics, Part A, 2021, 185, 2739-2747.	0.7	0
47	The Evaluation of Skeletal Manifestations in Patients with Gaucher Disease. Journal of Pediatric Research, 2021, 8, 257-261.	0.1	0
48	Association Between Soluble CD40 Ligand and Hypercholesterolemia in Children and Adolescents. Journal of Pediatric Research, 2017, 4, 1-5.	0.1	0
49	Expected or unexpected clinical findings in liver glycogen storage disease type IX: distinct clinical and molecular variability. Journal of Pediatric Endocrinology and Metabolism, 2022, .	0.4	0