

# ErtuÄrül Kiykim

## List of Publications by Year in descending order

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Version: 2024-02-01

51  
papers

518  
citations

933264

10  
h-index

752573

20  
g-index

51  
all docs

51  
docs citations

51  
times ranked

957  
citing authors

#	ARTICLE	IF	CITATIONS
1	Differences in the gut microbiota of healthy children and those with type 1 diabetes. <i>Pediatrics International</i> , 2014, 56, 336-343.	0.2	118
2	Spondyloocular Syndrome: Novel Mutations in <i>XYLT2</i> Gene and Expansion of the Phenotypic Spectrum. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 1577-1585.	3.1	43
3	Liver and/or kidney transplantation in amino and organic acid-related inborn errors of metabolism: An overview on European data. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 593-605.	1.7	34
4	Inherited metabolic disorders in Turkish patients with autism spectrum disorders. <i>Autism Research</i> , 2016, 9, 217-223.	2.1	30
5	Hereditary tyrosinemia type 1 in Turkey: Twenty year single-center experience. <i>Pediatrics International</i> , 2015, 57, 281-289.	0.2	27
6	Novel CLPB mutation in a patient with 3-methylglutaconic aciduria causing severe neurological involvement and congenital neutropenia. <i>Clinical Immunology</i> , 2016, 165, 1-3.	1.4	24
7	Early diagnosed cerebrotendinous xanthomatosis patients: clinical, neuroradiological characteristics and therapy results of a single center from Turkey. <i>Acta Neurologica Belgica</i> , 2019, 119, 343-350.	0.5	18
8	Telemedicine Applications in a Tertiary Pediatric Hospital in Turkey During COVID-19 Pandemic. <i>Telemedicine Journal and E-Health</i> , 2021, 27, 1180-1187.	1.6	16
9	Vitamin D levels in children and adolescents with autism. <i>Journal of International Medical Research</i> , 2020, 48, 030006052093463.	0.4	15
10	Screening Mucopolysaccharidosis Type IX in Patients with Juvenile Idiopathic Arthritis. <i>JIMD Reports</i> , 2015, 25, 21-24.	0.7	14
11	Biotinidase deficiency mimicking primary immune deficiencies. <i>BMJ Case Reports</i> , 2015, 2015, bcr2014209275-bcr2014209275.	0.2	11
12	Cobalamin C defect-related hemolytic uremic syndrome caused by new mutation in <i>MMACHC</i> . <i>Pediatrics International</i> , 2016, 58, 763-765.	0.2	10
13	Evaluation of dynamic thiol/disulphide homeostasis as a novel indicator of oxidative stress in maple syrup urine disease patients under treatment. <i>Metabolic Brain Disease</i> , 2017, 32, 179-184.	1.4	10
14	Oxidative stress among L-2-hydroxyglutaric aciduria disease patients: evaluation of dynamic thiol/disulfide homeostasis. <i>Metabolic Brain Disease</i> , 2019, 34, 283-288.	1.4	9
15	Challenges of following patients with inherited metabolic diseases during the COVID-19 outbreak. A cross-sectional online survey study. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2021, 34, 103-107.	0.4	9
16	Recurrence of carbamoyl phosphate synthetase 1 (CPS1) deficiency in Turkish patients: Characterization of a founder mutation by use of recombinant CPS1 from insect cells expression. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 267-273.	0.5	8
17	Hereditary Tyrosinemia Type 1 in Turkey. <i>Advances in Experimental Medicine and Biology</i> , 2017, 959, 157-172.	0.8	8
18	Capillary electrophoresis with capacitively coupled contactless conductivity detection for the determination of urinary ethylmalonic acid for the diagnosis of ethylmalonic aciduria. <i>Journal of Separation Science</i> , 2020, 43, 1365-1371.	1.3	8

#	ARTICLE	IF	CITATIONS
19	Severe lactic acidosis in an extremely low birth weight infant due to thiamine deficiency. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 693-695.	0.4	7
20	Evaluation of the reasons for the microvascular changes in patients with Fabry disease using optic coherence tomography angiography. <i>European Journal of Ophthalmology</i> , 2020, 31, 112067212097428.	0.7	7
21	Inborn errors of metabolism and coronavirus disease 2019: Evaluation of the metabolic outcome. <i>Pediatrics International</i> , 2022, 64, .	0.2	6
22	Clinical and neuroradiological approach to fucosidosis in a child with atypical presentation. <i>Annals of Indian Academy of Neurology</i> , 2015, 18, 471-4.	0.2	6
23	Screening of Free Carnitine and Acylcarnitine Status in Children With Familial Mediterranean Fever. <i>Archives of Rheumatology</i> , 2016, 31, 133-138.	0.3	5
24	Neonatal nonketotic hyperglycinemia: diffusion-weighted magnetic resonance imaging and diagnostic clues. <i>Acta Neurologica Belgica</i> , 2016, 116, 671-673.	0.5	5
25	Coagulation Disturbances in Patients with Argininemia. <i>Acta Haematologica</i> , 2018, 140, 221-225.	0.7	5
26	Continuous Renal Replacement Therapy with High Flow Rate Can Effectively, Safely, and Quickly Reduce Plasma Ammonia and Leucine Levels in Children. <i>Children</i> , 2019, 6, 53.	0.6	5
27	Multimodal imaging including optical coherence tomography angiography in patients with type B Niemann-Pick disease. <i>International Ophthalmology</i> , 2019, 39, 2545-2552.	0.6	5
28	Mercury intoxication resembling pediatric rheumatic diseases: case series and literature review. <i>Rheumatology International</i> , 2020, 40, 1333-1342.	1.5	5
29	Impact of sodium phenylbutyrate treatment in acute management of maple syrup urine disease attacks: a single-center experience. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2021, 34, 121-126.	0.4	5
30	Treatment of maple syrup urine disease with high flow hemodialysis in a neonate. <i>Turkish Journal of Pediatrics</i> , 2019, 61, 107.	0.3	5
31	Altered immune response in organic acidemia. <i>Pediatrics International</i> , 2022, 64, .	0.2	5
32	Idiopathic intracranial hypertension due to intralesional triamcenolone acetate. <i>Indian Pediatrics</i> , 2014, 51, 754-757.	0.2	4
33	Screening for Fabry Disease in Patients With Juvenile Systemic Lupus Erythematosus. <i>Archives of Rheumatology</i> , 2020, 35, 7-12.	0.3	4
34	Long-term N-carbamylglutamate treatment of hyperammonemia in patients with classic organic acidemias. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100715.	0.4	4
35	Two patients with novel missense mutation in the purine nucleoside phosphorylase gene without serious or recurrent infections. <i>Clinical and Experimental Neuroimmunology</i> , 2016, 7, 79-82.	0.5	3
36	Identifying and elucidating the roles of Y198N and Y204F mutations in the PAH enzyme through molecular dynamic simulations. <i>Journal of Biomolecular Structure and Dynamics</i> , 2021, , 1-12.	2.0	3

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37	COVID-19 triggered encephalopathic crisis in a patient with glutaric aciduria type 1. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2021, 34, 1611-1614.	0.4	3
38	Blood Pressure Variability in Fabry Disease Patients. <i>Nephron</i> , 2022, 146, 343-350.	0.9	3
39	Ornithine Aminotransferase Deficiency in Differential Diagnosis of Neonatal Hyperammonemia: A Case with a Novel OAT Gene Mutation. <i>Indian Journal of Pediatrics</i> , 2016, 83, 754-755.	0.3	2
40	Movement disorders in the early-diagnosed cerebrotendinous xanthomatosis: An electrophysiological study. <i>Parkinsonism and Related Disorders</i> , 2020, 80, 12-14.	1.1	2
41	Evaluation of the effect of chenodeoxycholic acid treatment on skeletal system findings in cerebrotendinous xanthomatosis patients. <i>Turk Pediatri Arsivi</i> , 2019, 54, 113-118.	0.9	2
42	L-2-hidroksiglutarik asidÄ¼ri hastalarÄ±nda klinik, nÄ¼roradyolojik ve genetik bulgularÄ±n deÄ¼erlendirilmesi. <i>Turk Pediatri Arsivi</i> , 2020, 55, 290-298.	0.9	2
43	Oculocutaneous tyrosinemia: A case report with delayed diagnosis and excellent response to dietary modification. <i>Indian Journal of Dermatology, Venereology and Leprology</i> , 2015, 81, 303.	0.2	1
44	Reflex Decay Test Can Reveal Ear Involvement in Fabry Disease. <i>Ear and Hearing</i> , 2021, Publish Ahead of Print, 1351-1357.	1.0	1
45	Postural tremor in L-2-hydroxyglutaric aciduria is associated with cerebellar atrophy. <i>Neurological Sciences</i> , 2021, , 1.	0.9	1
46	Lessons from two cases: is Fabry disease the correct diagnosis?. <i>BMJ Case Reports</i> , 2015, 2015, bcr2014208150-bcr2014208150.	0.2	0
47	MO055THE IMPACT OF THE COVID-19 PANDEMIC ON MOOD STATUS AND TREATMENT ADHERENCE IN PATIENTS WITH FABRY DISEASE. <i>Nephrology Dialysis Transplantation</i> , 2021, 36, .	0.4	0
48	Evaluation of dynamic thiol/disulfide homeostasis in hereditary tyrosinemia type 1 patients. <i>Pediatric Research</i> , 2021, , .	1.1	0
49	Screening Inherited Metabolic Disorders in Patients with Familial Mediterranean Fever. <i>Journal of Pediatric Research</i> , 2015, 2, 201-205.	0.1	0
50	A novel ABCD1 gene mutation in a patient with X-linked adrenoleukodystrophy with atypically normal plasma levels of very long chain fatty acids. <i>Marmara Medical Journal</i> , 2016, 29, 45.	0.2	0
51	Successful Hyperbaric Oxygen Treatment of Gangrenous Lesions due to Systemic Lupus Erythematosus. <i>Turkiye Klinikleri Pediatri</i> , 2017, 26, 103-106.	0.1	0