Melis KÃ¶se

List of Publications by Year
in descending order

27 papers	117 citations	1477746 6 h-index	1473754 9 g-index
27 all docs	27 docs citations	27 times ranked	148 citing authors

#	Article	IF	Citations
1	Expanding the spectrum of VAC14 related pediatric-onset neurological disease; striatonigral degeneration with brainstem involvement. European Journal of Medical Genetics, 2021, 64, 104117.	0.7	3
2	Neuronal ceroid lipofuscinosis: genetic and phenotypic spectrum of 14 patients from Turkey. Neurological Sciences, 2021, 42, 1103-1111.	0.9	7
3	The utility of next-generation sequencing technologies in diagnosis of Mendelian mitochondrial diseases and reflections on clinical spectrum. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 417-430.	0.4	8
4	The utility of reverse phenotyping: a case of lysinuric protein intolerance presented with childhood osteoporosis. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 957-960.	0.4	2
5	Outcomes of Dyslipidemia Screening Program in School-aged Children. Journal of Pediatric Research, 2021, 8, 155-160.	0.1	0
6	Clinical findings in five Turkish patients with citrin deficiency and identification of a novel mutation on SLC25A13. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 157-163.	0.4	8
7	SURF1 related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. Molecular Genetics and Metabolism Reports, 2020, 25, 100657.	0.4	10
8	A new mutation associated with Pierson syndrome. Archivos Argentinos De Pediatria, 2020, 118, e288-e291.	0.3	1
9	Challenges in the management of an ignored cause of hyperammonemic encephalopathy: pyruvate carboxylase deficiency. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 569-574.	0.4	4
10	Cascade screening and treatment of children with familial hypercholesterolemia in Turkey. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1251-1256.	0.4	5
11	Different clinical presentation in a patient with two novel pathogenic variants of the fbxl4 gene. Turkish Journal of Pediatrics, 2020, 62, 652.	0.3	4
12	Clinical and Molecular Features of Our Pompe Patients: Single-Center Experience. Medical Journal of Bakirkoy, 2020, , .	0.0	1
13	Congenital Disorder of Glycosylation: Clinical and Molecular Characteristics of 9 Patients from Turkey. Journal of Dr Behcet Uz Children S Hospital, 2020, , .	0.1	2
14	Coexistence of Gaucher Disease and severe congenital neutropenia. Blood Cells, Molecules, and Diseases, 2019, 76, 1-6.	0.6	5
15	Clinical and molecular characteristics and time of diagnosis of patients with classical galactosemia in an unscreened population in Turkey. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 675-681.	0.4	6
16	Inborn errors of metabolism: a three-year experience. Journal of Dr Behcet Uz Children S Hospital, 2019, , .	0.1	1
17	Klasik Nonketotik Hiperglisinemi TanÄ±lÄ± Bebekte Ä°laca DirenÃ§li Epilepsi Tedavisinde Ketojenik Diyet UygulamasÄ±: Bir Olgu Sunumu. Journal of Nutrition and Dietetics, 2019, 47, 100-104.	0.1	0
18	Evaluation of Cardiovascular Involvement and Cytokine Levels in Patients with Mucopolysaccharidosis. Journal of Pediatric Research, 2019, 6, 121-127.	0.1	1

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19	Patients with Gaucher type 1: Switching from imiglucerase to miglustat therapy. Blood Cells, Molecules, and Diseases, 2018, 68, 180-184.	0.6	5
20	Single center experience of biotinidase deficiency: 259 patients and six novel mutations. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 917-926.	0.4	23
21	The Second Case of Saposin A Deficiency and Altered Autophagy. JIMD Reports, 2018, 44, 43-54.	0.7	6
22	Experience of the ketogenic diet therapy of the ketogenic diet team in a third-line childrenâ€™s hospital. Journal of Dr Behcet Uz Children S Hospital, 2018, , .	0.1	0
23	A Patient with MSUD: Acute Management with Sodium Phenylacetate/Sodium Benzoate and Sodium Phenylbutyrate. Case Reports in Pediatrics, 2017, 2017, 1-4.	0.2	5
24	Presentation and management of classical urea cycle disorders: lessons from our experience. Journal of Dr Behcet Uz Children S Hospital, 2016, , .	0.1	0
25	Two novel mutations in acid Î±-glucosidase gene in two patients with Pompe disease. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 1265-7.	0.4	2
26	One Year Experience of PheburaneÂ® (Sodium Phenylbutyrate) Treatment in a Patient with Argininosuccinate Lyase Deficiency. JIMD Reports, 2014, 19, 31-33.	0.7	6
27	Clinical, Neuroimaging, and Genetic Features of the Patients with L-2-Hydroxyglutaric Aciduria. Journal of Pediatric Research, 0, , 39-43.	0.1	2