GÃ¶zde YeÅil

List of Publications by Year
in descending order

87 papers	2,190 citations	361413 20 h-index	254184 43 g-index
89 all docs	89 docs citations	89 times ranked	5212 citing authors

#	Article	IF	Citations
1	A Novel Mutation in the TRIP11 Gene: Diagnostic Approach from Relatively Common Skeletal Dysplasias to an Extremely Rare Odontochondrodysplasia. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2022, 14, 475-480.	0.9	1
2	Evolution and longâ€term outcomes of combined immunodeficiency due to CARMIL2 deficiency. Allergy: European Journal of Allergy and Clinical Immunology, 2022, 77, 1004-1019.	5.7	19
3	Investigation of <u>MBL2</u> and <u>NOS3</u> functional gene variants in suspected COVID-19 PCR (â€“) patients. Pathogens and Global Health, 2022, 116, 178-184.	2.3	6
4	Biallelic loss of TRAPPC9 function links vesicle trafficking pathway to autosomal recessive intellectual disability. Journal of Human Genetics, 2022, 67, 279-284.	2.3	8
5	Functional loss of ubiquitinâ€specific protease 14 may lead to a novel distal arthrogryposis phenotype. Clinical Genetics, 2022, 101, 421-428.	2.0	1
6	Parents of ataxiaâ€telangiectasia patients display a distinct cellular immune phenotype mimicking ATMâ€mutated patients. Pediatric Allergy and Immunology, 2021, 32, 349-357.	2.6	3
7	Cause of recurrent rhabdomyolysis, carnitine palmitoyltransferase II deficiency and novel pathogenic mutation. Ideggyogyaszati Szemle, 2021, 74, 135-138.	0.7	1
8	Strong mesangial IgA stainingâ€”does it always refer to IgA nephropathy in a patient with proteinuria and hematuria? Answers. Pediatric Nephrology, 2021, 36, 2043-2045.	1.7	1
9	Strong mesangial IgA stainingâ€”does it always refer to IgA nephropathy in a patient with proteinuria and hematuria? Questions. Pediatric Nephrology, 2021, 36, 2039-2041.	1.7	0
10	Investigation of (epi)genotype causes and followâ€up manifestations in the patients with classical and atypical phenotype of Beckwithâ€Wiedemann spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 1721-1731.	1.2	5
11	Mannose binding lectin gene 2 (rs1800450) missense variant may contribute to development and severity of COVID-19 infection. Infection, Genetics and Evolution, 2021, 89, 104717.	2.3	34
12	Neurofibromatosis type 1: Expanded variant spectrum with multiplex ligationâ€dependent probe amplification and genotypeâ€“phenotype correlation in 138 Turkish patients. Annals of Human Genetics, 2021, 85, 155-165.	0.8	4
13	Expanding the clinical phenotype of <scp>RASopathies</scp> in 38 Turkish patients, including the rare <scp>LZTR1</scp>, <scp>RAF1</scp>, <scp>RIT1</scp> variants, and large deletion in <scp>NF1</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3623-3633.	1.2	4
14	Familial atypical hemolytic uremic syndrome with positive p.S1191L (c.3572C>T) mutation on the CFH gene: A single-center experience. Balkan Journal of Medical Genetics, 2021, 24, 81-88.	0.5	0
15	Two patients with chronic mucocutaneous candidiasis caused by TRAF3IP2 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 256-261.e2.	2.9	10
16	Is There a Link between Circadian Clock Protein PERIOD 3 (PER3) (rs57875989) Variant and the Severity of COVID-19 Infection?. Current Medical Science, 2021, 41, 1075-1080.	1.8	2
17	Evaluation of the parentsâ€™ anxiety levels before and after the diagnosis of their child with a rare genetic disease: the necessity of psychological support. Orphanet Journal of Rare Diseases, 2021, 16, 402.	2.7	14
18	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	6.2	38

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19	A Novel Mutation of HINT1 Gene in an Adolescent Female with Axonal Neuropathy and Neuromyotonia. Journal of Pediatric Neurology, 2021, 19, 180-182.	0.2	0
20	Broad-spectrum XX and XY gonadal dysgenesis in patients with a homozygous L193S variant in PPP2R3C. European Journal of Endocrinology, 2021, 186, 65-72.	3.7	1
21	Rare cause of severe hypertension in an adolescent boy presenting with short stature: Questions. Pediatric Nephrology, 2020, 35, 403-404.	1.7	1
22	Rare cause of severe hypertension in an adolescent boy presenting with short stature: Answers. Pediatric Nephrology, 2020, 35, 405-407.	1.7	4
23	A rare cause of hypertension in childhood: Questions. Pediatric Nephrology, 2020, 35, 77-78.	1.7	1
24	A rare cause of hypertension in childhood: Answers. Pediatric Nephrology, 2020, 35, 79-82.	1.7	5
25	Vanishing white matter disease with different faces. Child's Nervous System, 2020, 36, 353-361.	1.1	6
26	Diagnostic Modalities Based on Flow Cytometry for Chronic Granulomatous Disease: A Multicenter Study in a Well-Defined Cohort. Journal of Allergy and Clinical Immunology: in Practice, 2020, 8, 3525-3534.e1.	3.8	7
27	Expanding Clinical Phenotype of TRAPPC12-Related Childhood Encephalopathy: Two Cases and Review of Literature. Neuropediatrics, 2020, 51, 430-434.	0.6	3
28	Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. European Journal of Human Genetics, 2020, 28, 1243-1264.	2.8	27
29	Early-onset rapidly progressive myoclonic epilepsy associated with G392R likely pathogenic variant in SERPINI1. Seizure: the Journal of the British Epilepsy Association, 2020, 80, 181-182.	2.0	5
30	L-2-hidroksiglutarik asidÃ¼ri hastalarÄ±nda klinik, nÃ¶roradyolojik ve genetik bulgularÄ±n deÄŸerlendirilmesi. Turk Pediatri Arsivi, 2020, 55, 290-298.	0.9	2
31	A Case of Sotos Syndrome Caused by a Novel Variant in the NSD1 Gene: A Proposed Rationale to Treat Accompanying Precocious Puberty. Acta Endocrinologica, 2020, 16, 245-249.	0.3	3
32	Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycleâ€opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 2056-2066.	1.2	15
33	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	6.2	74
34	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067.	3.6	53
35	Evaluation of growth and puberty in a child with a novel TBX19 gene mutation and review of the literature. Hormones, 2019, 18, 229-236.	1.9	6
36	P74â€...Patient with intermittent posture abnormality: an alexander disease case report. , 2019, , .		0

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37	P94â€…Cytochrome P450 oxidoreductase enzyme deficiency: a case report. , 2019, , .		0
38	A Rare Cause of Adrenal Insufficiency â€“ Isolated ACTH Deficiency Due to TBX19 Mutation: Long-Term Follow-Up of Two Cases and Review of the Literature. Hormone Research in Paediatrics, 2019, 92, 395-403.	1.8	4
39	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctivecerebellar,ocular, craniofacial andgenital features (COFG) Tj ETQq1 1 0.784314 rgBT /Overlock 10	3.4	25
40	Early diagnosed cerebrotendinous xanthomatosis patients: clinical, neuroradiological characteristics and therapy results of a single center from Turkey. Acta Neurologica Belgica, 2019, 119, 343-350.	1.1	18
41	PPP2R3C gene variants cause syndromic 46,XY gonadal dysgenesis and impaired spermatogenesis in humans. European Journal of Endocrinology, 2019, 180, 291-309.	3.7	18
42	Correlation Between DTI Findings and Volume of Corpus Callosum in Children with AUTISM. Current Medical Imaging, 2019, 15, 895-899.	0.8	26
43	Novel PTCH1 Gene Mutation in a Patient with Gorlinâ€“Goltz Syndrome. Annals of Dermatology, 2019, 31, S10.	0.9	3
44	Pseudohypoparathyroidism Type Ia with Normocalcemia. BezmiÃ¢lem Science, 2019, 7, 170-173.	0.2	0
45	A Case with Laron Syndrome. BezmiÃ¢lem Science, 2019, 7, 251-254.	0.2	1
46	Early Diagnosis of Fanconi-Bickel Syndrome and a Novel Mutation inSLC2A2 Gene. Haseki Tip Bulteni, 2019, 57, 328-331.	0.3	0
47	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	2.4	104
48	Spinal muscular atrophy with progressive myoclonic epilepsy linked to mutations in ASAH1. Clinical Neurology and Neurosurgery, 2018, 164, 47-49.	1.4	8
49	Mucolipidosis type III gamma: Three novel mutation and genotype-phenotype study in eleven patients. Gene, 2018, 642, 398-407.	2.2	14
50	Expanding the Phenotype of Homozygous KCNMA1 Mutations; Dyskinesia, Epilepsy, Intellectual Disability, Cerebellar and Corticospinal Tract Atrophy. Balkan Medical Journal, 2018, 35, 336-339.	0.8	40
51	Longitudinal Follow-Up of Two Patients with Dysspondyloenchondromatosis due to Novel Heterozygous Mutations in <i>COL2A1</i>. Molecular Syndromology, 2018, 9, 134-140.	0.8	3
52	Vanishing white matter disease with a novel EIF2B5 mutation: A 10-year follow-up. Clinical Neurology and Neurosurgery, 2018, 171, 190-193.	1.4	3
53	Familial amyloid polyneuropathy due to p.ALA140 SER mutation. Neurology India, 2018, 66, 238.	0.4	0
54	The Contribution of DTI in Determining the Relationship of Epilepsy and Brain Lesions in Children with Tuberous Sclerosis. Current Medical Imaging, 2018, 14, 401-406.	0.8	0

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55	A novel EPM2A mutation in a patient with Lafora disease presenting with early parkinsonism symptoms in childhood. Seizure: the Journal of the British Epilepsy Association, 2017, 51, 77-79.	2.0	8
56	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
57	Hereditary spastic paraplegia type 35 caused by a novel fa2h mutation. Turkish Journal of Pediatrics, 2017, 59, 329.	0.6	10
58	A case with Rubinstein-Taybi syndrome: A novel frameshift mutation in the CREBBP gene. Turkish Journal of Pediatrics, 2017, 59, 601-603.	0.6	5
59	Endocrinological Evaluations of a Neurofibromatosis Type 1 Cohort: Is it Necessary to Evaluate Autoimmune Thyroiditis in Neurofibromatosis Type 1?. Balkan Medical Journal, 2017, 34, 522-526.	0.8	9
60	Novel CLPB mutation in a patient with 3-methylglutaconic aciduria causing severe neurological involvement and congenital neutropenia. Clinical Immunology, 2016, 165, 1-3.	3.2	24
61	MRI and MRS findings in fucosidosis; a rare lysosomal storage disease. Brain and Development, 2016, 38, 435-438.	1.1	15
62	Microcephaly, dysmorphic features, corneal dystrophy, hairy nipples, underdeveloped labioscrotal folds, and small cerebellum in four patients. American Journal of Medical Genetics, Part A, 2016, 170, 1391-1399.	1.2	5
63	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	6.2	66
64	Cytogenetic Analysis of 65 Women with Premature Ovarian Insufficiency. Journal of Clinical and Analytical Medicine, 2016, 7, .	0.1	0
65	Homozygous Loss-of-function Mutations inSOHLH1in Patients With Nonsyndromic Hypergonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E808-E814.	3.6	29
66	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
67	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. Nature Genetics, 2015, 47, 73-77.	21.4	130
68	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651.	8.2	136
69	Report of a patient with Templeâ€“Baraitser syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 848-851.	1.2	8
70	Stuveâ€“Wiedemann syndrome: Is it underrecognized?. American Journal of Medical Genetics, Part A, 2014, 164, 2200-2205.	1.2	11
71	The drug-transporter gene MDR1 C3435T and G2677T/A polymorphisms and the risk of multidrug-resistant epilepsy in Turkish children. Molecular Biology Reports, 2014, 41, 331-336.	2.3	37
72	The Effect of Genetic Polymorphisms of Cytochrome P450 CYP2C9, CYP2C19, and CYP2D6 on Drug-Resistant Epilepsy in Turkish Children. Molecular Diagnosis and Therapy, 2014, 18, 229-236.	3.8	21

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73	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	28.9	189
74	Prevalence of X-aneuploidies, X-structural abnormalities and 46,XY sex reversal in Turkish women with primary amenorrhea or premature ovarian insufficiency. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2014, 182, 211-215.	1.1	18
75	Comparison of metabolic profile and abdominal fat distribution between karyotypically normal women with premature ovarian insufficiency and age matched controls. Maturitas, 2014, 79, 306-310.	2.4	37
76	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
77	Congenital Agenesis of Scrotum and Labia Majora in Siblings. Urology, 2013, 81, 421-423.	1.0	12
78	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused byPAPSS2Mutations. Human Mutation, 2013, 34, 1381-1386.	2.5	29
79	A Giant Ovarian Cyst in a Neonate with Classical 21-Hydroxylase Deficiency with Very High Testosterone Levels Demonstrating a High-Dose Hook Effect. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 151-153.	0.9	10
80	A Novel GJC2 Mutation Associated with Hypomyelination and MÃ¼llerian Agenesis Syndrome: Coincidence or a New Entity?. Neuropediatrics, 2012, 43, 159-161.	0.6	5
81	An infant with spinal muscular atrophy and tetrology of Fallot. Clinical Neurology and Neurosurgery, 2012, 114, 1033-1034.	1.4	1
82	Restrictive Dermopathy in a Turkish Newborn. Pediatric Dermatology, 2011, 28, 408-411.	0.9	6
83	Warburg Micro syndrome in a Turkish boy. Clinical Dysmorphology, 2007, 16, 89-93.	0.3	18
84	Evaluation of mental retardation - Part 1: Etiologic classification of 4659 patients with mental retardation or multiple congenital abnormality and mental retardation. Journal of Pediatric Neurosciences, 2007, 2, 45.	0.3	2
85	Evaluation of mental retardation - Part 2: The factors that elucidate the etiologic diagnosis of the patients with mental retardation or multiple congenital abnormality and mental retardation. Journal of Pediatric Neurosciences, 2007, 2, 53.	0.3	0
86	Facial Dysmorphism in Leigh Syndrome With SURF-1 Mutation and COX Deficiency. Pediatric Neurology, 2006, 34, 486-489.	2.1	14
87	Proteus syndrome with agenesis of the rectus abdominis. British Journal of Dermatology, 2006, 155, 1094-1095.	1.5	0