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	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
2	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
3	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	28.9	189
4	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
5	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651.	8.2	136
6	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. Nature Genetics, 2015, 47, 73-77.	21.4	130
7	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	2.4	104
8	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
9	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
10	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
11	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
12	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
13	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
14	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
15	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
16	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused byPAPSS2Mutations. Human Mutation, 2013, 34, 1381-1386.	2.5	29
17	Homozygous Loss-of-function Mutations inSOHLH1in Patients With Nonsyndromic Hypergonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E808-E814.	3.6	29
18	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

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19	Correlation Between DTI Findings and Volume of Corpus Callosum in Children with AUTISM. Current Medical Imaging, 2019, 15, 895-899.	0.8	26
20	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctivecerebellar,ocular, craniofacial andgenital features (COFG) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 69	3.2	25
21	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
22	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
23	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
24	Warburg Micro syndrome in a Turkish boy. Clinical Dysmorphology, 2007, 16, 89-93.	0.3	18
25	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
26	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
27	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
28	MRI and MRS findings in fucosidosis; a rare lysosomal storage disease. Brain and Development, 2016, 38, 435-438.	1.1	15
29	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
30	Facial Dysmorphism in Leigh Syndrome With SURF-1 Mutation and COX Deficiency. Pediatric Neurology, 2006, 34, 486-489.	2.1	14
31	Mucolipidosis type III gamma: Three novel mutation and genotype-phenotype study in eleven patients. Gene, 2018, 642, 398-407.	2.2	14
32	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
33	Congenital Agenesis of Scrotum and Labia Majora in Siblings. Urology, 2013, 81, 421-423.	1.0	12
34	Stuveâ€“Wiedemann syndrome: Is it underrecognized?. American Journal of Medical Genetics, Part A, 2014, 164, 2200-2205.	1.2	11
35	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
36	Two patients with chronic mucocutaneous candidiasis caused by TRAF3IP2 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 256-261.e2.	2.9	10

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37	Hereditary spastic paraplegia type 35 caused by a novel fa2h mutation. Turkish Journal of Pediatrics, 2017, 59, 329.	0.6	10
38	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
39	Report of a patient with Templeâ€“Baraitser syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 848-851.	1.2	8
40	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
41	Spinal muscular atrophy with progressive myoclonic epilepsy linked to mutations in ASAH1. Clinical Neurology and Neurosurgery, 2018, 164, 47-49.	1.4	8
42	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
43	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
44	Restrictive Dermopathy in a Turkish Newborn. Pediatric Dermatology, 2011, 28, 408-411.	0.9	6
45	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
46	Vanishing white matter disease with different faces. Child's Nervous System, 2020, 36, 353-361.	1.1	6
47	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
48	A Novel GJC2 Mutation Associated with Hypomyelination and MÃ¼llerian Agenesis Syndrome: Coincidence or a New Entity?. Neuropediatrics, 2012, 43, 159-161.	0.6	5
49	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
50	A rare cause of hypertension in childhood: Answers. Pediatric Nephrology, 2020, 35, 79-82.	1.7	5
51	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
52	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
53	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
54	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

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55	Rare cause of severe hypertension in an adolescent boy presenting with short stature: Answers. Pediatric Nephrology, 2020, 35, 405-407.	1.7	4
56	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
57	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
58	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
59	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
60	Expanding Clinical Phenotype of TRAPPC12-Related Childhood Encephalopathy: Two Cases and Review of Literature. Neuropediatrics, 2020, 51, 430-434.	0.6	3
61	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
62	Novel PTCH1 Gene Mutation in a Patient with Gorlinâ€“Goltz Syndrome. Annals of Dermatology, 2019, 31, S10.	0.9	3
63	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
64	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
65	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
66	L-2-hidroksiglutarik asidÃ¼ri hastalarÄ±nda klinik, nÃ¶roradyolojik ve genetik bulgularÄ±n deÄŸerlendirilmesi. Turk Pediatri Arsivi, 2020, 55, 290-298.	0.9	2
67	An infant with spinal muscular atrophy and tetrology of Fallot. Clinical Neurology and Neurosurgery, 2012, 114, 1033-1034.	1.4	1
68	Rare cause of severe hypertension in an adolescent boy presenting with short stature: Questions. Pediatric Nephrology, 2020, 35, 403-404.	1.7	1
69	A rare cause of hypertension in childhood: Questions. Pediatric Nephrology, 2020, 35, 77-78.	1.7	1
70	Cause of recurrent rhabdomyolysis, carnitine palmitoyltransferase II deficiency and novel pathogenic mutation. Ideggyogyaszati Szemle, 2021, 74, 135-138.	0.7	1
71	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
72	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

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73	A Case with Laron Syndrome. BezmiÃ¢lem Science, 2019, 7, 251-254.	0.2	1
74	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
75	Functional loss of ubiquitinâ€specific protease 14 may lead to a novel distal arthrogryposis phenotype. Clinical Genetics, 2022, 101, 421-428.	2.0	1
76	Proteus syndrome with agenesis of the rectus abdominis. British Journal of Dermatology, 2006, 155, 1094-1095.	1.5	0
77	P74â€…Patient with intermittent posture abnormality: an alexander disease case report. , 2019, , .		0
78	P94â€…Cytochrome P450 oxidoreductase enzyme deficiency: a case report. , 2019, , .		0
79	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
80	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
81	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
82	Cytogenetic Analysis of 65 Women with Premature Ovarian Insufficiency. Journal of Clinical and Analytical Medicine, 2016, 7, .	0.1	0
83	Familial amyloid polyneuropathy due to p.ALA140 SER mutation. Neurology India, 2018, 66, 238.	0.4	0
84	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
85	Pseudohypoparathyroidism Type Ia with Normocalcemia. BezmiÃ¢lem Science, 2019, 7, 170-173.	0.2	0
86	Early Diagnosis of Fanconi-Bickel Syndrome and a Novel Mutation inSLC2A2 Gene. Haseki Tip Bulteni, 2019, 57, 328-331.	0.3	0
87	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