Beyhan TÃ¼ysÃ¼z

List of Publications by Year
in descending order

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114 papers	5,373 citations	94433 37 h-index	88630 70 g-index
116 all docs	116 docs citations	116 times ranked	9630 citing authors

#	Article	IF	Citations
1	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. Nature, 2010, 467, 207-210.	27.8	457
2	The gene encoding gigaxonin, a new member of the cytoskeletal BTB/kelch repeat family, is mutated in giant axonal neuropathy. Nature Genetics, 2000, 26, 370-374.	21.4	372
3	IFT80, which encodes a conserved intraflagellar transport protein, is mutated in Jeune asphyxiating thoracic dystrophy. Nature Genetics, 2007, 39, 727-729.	21.4	310
4	Recessive Robinow syndrome, allelic to dominant brachydactyly type B, is caused by mutation of ROR2. Nature Genetics, 2000, 25, 419-422.	21.4	277
5	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	28.9	228
6	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	21.4	201
7	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
8	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
9	The Essential Role of Centrosomal NDE1 in Human Cerebral Cortex Neurogenesis. American Journal of Human Genetics, 2011, 88, 523-535.	6.2	146
10	Recessive loss of function of the neuronal ubiquitin hydrolase UCHL1 leads to early-onset progressive neurodegeneration. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 3489-3494.	7.1	144
11	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651.	8.2	136
12	Hennekam syndrome can be caused by FAT4 mutations and be allelic to Van Maldergem syndrome. Human Genetics, 2014, 133, 1161-1167.	3.8	122
13	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	2.9	115
14	Idiopathic Hyperphosphatasia andTNFRSF11BMutations: Relationships Between Phenotype and Genotype. Journal of Bone and Mineral Research, 2003, 18, 2095-2104.	2.8	113
15	Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2013, 93, 932-944.	6.2	108
16	Recessive LAMC3 mutations cause malformations of occipital cortical development. Nature Genetics, 2011, 43, 590-594.	21.4	102
17	XYLT1 Mutations in Desbuquois Dysplasia Type 2. American Journal of Human Genetics, 2014, 94, 405-414.	6.2	92
18	Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778.	8.2	82

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19	A novel mutation in EED associated with overgrowth. Journal of Human Genetics, 2015, 60, 339-342.	2.3	75
20	The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): A review of clinical features, radiographic features, and WISP3 mutations in 63 affected individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 217-229.	1.6	74
21	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
22	Mutations in ANKLE2, a ZIKA Virus Target, Disrupt an Asymmetric Cell Division Pathway in Drosophila Neuroblasts to Cause Microcephaly. Developmental Cell, 2019, 51, 713-729.e6.	7.0	71
23	Mutations in the prostaglandin transporter encoding geneSLCO2A1Cause primary hypertrophic osteoarthropathy and isolated digital clubbing. Human Mutation, 2012, 33, 660-664.	2.5	69
24	Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2759-2767.	3.6	67
25	Functional deficiencies of sulfite oxidase: Differential diagnoses in neonates presenting with intractable seizures and cystic encephalomalacia. Brain and Development, 2010, 32, 544-549.	1.1	66
26	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.	2.5	66
27	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	1.1	57
28	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
29	Autosomal recessive spastic tetraplegia caused by AP4M1 and AP4B1 gene mutation: Expansion of the facial and neuroimaging features. American Journal of Medical Genetics, Part A, 2014, 164, 1677-1685.	1.2	55
30	Clinical variability of asphyxiating thoracic dystrophy (Jeune) syndrome: Evaluation and classification of 13 patients. American Journal of Medical Genetics, Part A, 2009, 149A, 1727-1733.	1.2	54
31	Omaniâ€type spondyloepiphyseal dysplasia with cardiac involvement caused by a missense mutation in CHST3. Clinical Genetics, 2009, 75, 375-383.	2.0	54
32	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	2.5	47
33	A novel matrix metalloproteinase 2 (MMP2) terminal hemopexin domain mutation in a family with multicentric osteolysis with nodulosis and arthritis with cardiac defects. European Journal of Human Genetics, 2009, 17, 565-572.	2.8	46
34	Brain Malformations Associated With Knobloch Syndromeâ€”Review of Literature, Expanding Clinical Spectrum, and Identification of Novel Mutations. Pediatric Neurology, 2014, 51, 806-813.e8.	2.1	43
35	Spondyloocular Syndrome: Novel Mutations in XYLT2 Gene and Expansion of the Phenotypic Spectrum. Journal of Bone and Mineral Research, 2016, 31, 1577-1585.	2.8	43
36	Molecular investigations to improve diagnostic accuracy in patients with ARC syndrome. Human Mutation, 2009, 30, E330-E337.	2.5	40

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37	SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects. Nature Communications, 2018, 9, 3087.	12.8	39
38	Is the novel SCKL3 at 14q23 the predominant Seckel locus?. European Journal of Human Genetics, 2003, 11, 851-857.	2.8	38
39	Disruptions in asymmetric centrosome inheritance and WDR62-Aurora kinase B interactions in primary microcephaly. Scientific Reports, 2017, 7, 43708.	3.3	37
40	The lysosomal storage disorders mucolipidosis type II, type III alpha/beta, and type III gamma: Update onGNPTABandGNPTGmutations. Human Mutation, 2019, 40, 842-864.	2.5	36
41	Unilateral Cerebellar Hypoplasia with Different Clinical Features. Cerebellum, 2011, 10, 49-60.	2.5	32
42	DOCK6Mutations Are Responsible for a Distinct Autosomal-Recessive Variant of Adams-Oliver Syndrome Associated with Brain and Eye Anomalies. Human Mutation, 2015, 36, 593-598.	2.5	32
43	The phenotype caused by PYCR1 mutations corresponds to geroderma osteodysplasticum rather than autosomal recessive cutis laxa type 2. American Journal of Medical Genetics, Part A, 2011, 155, 134-140.	1.2	31
44	Ocular Manifestations of Juvenile Paget Disease. JAMA Ophthalmology, 2010, 128, 698.	2.4	29
45	GORAB Missense Mutations Disrupt RAB6 and ARF5 Binding and Golgi Targeting. Journal of Investigative Dermatology, 2015, 135, 2368-2376.	0.7	28
46	IMPAD1 mutations in two Catelâ€Manzke like patients. American Journal of Medical Genetics, Part A, 2012, 158A, 2183-2187.	1.2	27
47	Growth charts of Turkish children with Down syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2656-2664.	1.2	24
48	Calcitonin treatment in osteoectasia with hyperphosphatasia (juvenile Paget's disease): radiographic changes after treatment. Pediatric Radiology, 1999, 29, 838-841.	2.0	23
49	Prevalence of Praderâ€“Willi Syndrome among Infants with Hypotonia. Journal of Pediatrics, 2014, 164, 1064-1067.	1.8	23
50	Two novel mutations in XYLT2 cause spondyloocular syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3195-3200.	1.2	22
51	Detection of Y Chromosomal Material in Patients with a 45,X Karyotype by PCR Method. Tohoku Journal of Experimental Medicine, 2007, 211, 243-249.	1.2	21
52	Testotoxicosis: Report of Two Cases, One with a Novel Mutation in LHCGR Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2015, 7, 242-248.	0.9	21
53	Novel VLDLR microdeletion identified in two Turkish siblings with pachygyria and pontocerebellar atrophy. Neurogenetics, 2010, 11, 319-325.	1.4	19
54	Clinical variability of Waardenburgâ€“Shah syndrome in patients with proximal 13q deletion syndrome including the endothelinâ€B receptor locus. American Journal of Medical Genetics, Part A, 2009, 149A, 2290-2295.	1.2	18

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55	Primary hypertrophic osteoarthropathy caused by homozygous deletion in HPGD gene in a family: changing clinical and radiological findings with long-term follow-up. Rheumatology International, 2014, 34, 1539-1544.	3.0	18
56	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	7.6	18
57	Prenatal Diagnosis of Arthrogryposis multiplex congenita with Increased Nuchal Translucency but without Any Underlying Fetal Neurogenic or Myogenic Pathology. Fetal Diagnosis and Therapy, 2002, 17, 29-33.	1.4	16
58	A Novel de novo FZD2 Mutation in a Patient with Autosomal Dominant Omodysplasia. Molecular Syndromology, 2017, 8, 318-324.	0.8	16
59	Spondyloenchondrodysplasia: Clinical variability in three cases. , 2004, 128A, 185-189.		15
60	Spondyloepimetaphyseal dysplasia Pakistani type: Expansion of the phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 1300-1308.	1.2	15
61	Genotypeâ€“phenotype investigation of 35 patients from 11 unrelated families with camptodactylyâ€“arthropathyâ€“coxa varaâ€“pericarditis (<scp>CACP</scp>) syndrome. Molecular Genetics & Genomic Medicine, 2018, 6, 230-248.	1.2	15
62	Novel NTRK1 mutations cause hereditary sensory and autonomic neuropathy type IV: demonstration of a founder mutation in the Turkish population. Neurogenetics, 2008, 9, 119-125.	1.4	14
63	A New SPINK5 Donor Splice Site Mutation in Siblings with Netherton Syndrome. Acta Dermato-Venereologica, 2010, 90, 95-96.	1.3	14
64	A patient with a novel homozygous missense mutation in FTO and concomitant nonsense mutation in CETP. Journal of Human Genetics, 2016, 61, 395-403.	2.3	14
65	Mucolipidosis type III gamma: Three novel mutation and genotype-phenotype study in eleven patients. Gene, 2018, 642, 398-407.	2.2	14
66	SOPH syndrome in three affected individuals showing similarities with progeroid cutis laxa conditions in early infancy. Journal of Human Genetics, 2019, 64, 609-616.	2.3	14
67	Mucolipidosis type IV in a Turkish boy associated with a novel MCOLN1 mutation. Brain and Development, 2009, 31, 702-705.	1.1	13
68	Genome sequencing in families with congenital limb malformations. Human Genetics, 2021, 140, 1229-1239.	3.8	13
69	Phenotypic Expansion of Congenital Disorder of Glycosylation Due to SRD5A3 Null Mutation. JIMD Reports, 2015, 26, 7-12.	1.5	11
70	Recessive MYF5 Mutations Cause External Ophthalmoplegia, Rib, and Vertebral Anomalies. American Journal of Human Genetics, 2018, 103, 115-124.	6.2	11
71	Patient with the mesomelic dysplasia, Nievergelt syndrome, and cerebellovermian agenesis and cataracts. American Journal of Medical Genetics Part A, 2002, 109, 206-210.	2.4	10
72	Osteogenesis imperfecta in 140 Turkish families: Molecular spectrum and, comparison of long-term clinical outcome of those with COL1A1/A2 and biallelic variants. Bone, 2022, 155, 116293.	2.9	10

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73	Oral and craniofacial manifestations of Ellisâ€“van Creveld syndrome: Case series. Journal of Cranio-Maxillo-Facial Surgery, 2016, 44, 919-924.	1.7	9
74	Clinical Characteristics and Growth Hormone Treatment in Patients with Prader-Willi Syndrome. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 308-319.	0.9	9
75	The time of onset of abnormal calcification in spondylometaepiphyseal dysplasia, short limb-abnormal calcification type. Pediatric Radiology, 2009, 39, 84-89.	2.0	8
76	A patient with Duchenne muscular dystrophy and autism demonstrates a hemizygous deletion affecting Dystrophin. American Journal of Medical Genetics, Part A, 2010, 152A, 1039-1042.	1.2	8
77	Four Novel SCN1A Mutations in Turkish Patients With Severe Myoclonic Epilepsy of Infancy (SMEI). Journal of Child Neurology, 2010, 25, 1265-1268.	1.4	8
78	Multiple small hyperintense lesions in the subcortical white matter on cranial MR images in two Turkish brothers with cold-induced sweating syndrome caused by a novel missense mutation in the CRLF1 gene. Brain and Development, 2013, 35, 596-601.	1.1	8
79	Skeletal and molecular findings in 51 Cleidocranial dysplasia patients from Turkey. American Journal of Medical Genetics, Part A, 2021, 185, 2488-2495.	1.2	8
80	A microduplication of the Rubinsteinâ€“Taybi region on 16p13.3 in a girl with a bilateral complete cleft lip and palate and severe mental retardation. Clinical Dysmorphology, 2012, 21, 204-207.	0.3	7
81	Mutation spectrum and pivotal features for differential diagnosis of Mucopolysaccharidosis IVA patients with severe and attenuated phenotype. Gene, 2019, 704, 59-67.	2.2	7
82	Congenital generalized lipodystrophy: The evaluation of clinical follow-up findings in a series of five patients with type 1 and two patients with type 4. European Journal of Medical Genetics, 2020, 63, 103819.	1.3	7
83	Restrictive Dermopathy in a Turkish Newborn. Pediatric Dermatology, 2011, 28, 408-411.	0.9	6
84	Macrocephaly-Capillary Malformation Syndrome in a Newborn With Tetralogy of Fallot and Sagittal Sinus Thrombosis. Journal of Child Neurology, 2013, 28, 115-119.	1.4	6
85	Clinical and Molecular Characterization of Fanconi Anemia Patients in Turkey. Molecular Syndromology, 2020, 11, 183-196.	0.8	6
86	Resolution of sclerotic lesions of dysosteosclerosis due to biallelic SLC29A3 variant in a Turkish girl. American Journal of Medical Genetics, Part A, 2021, 185, 2271-2277.	1.2	6
87	Picture of the Month. JAMA Pediatrics, 1999, 153, 765.	3.0	5
88	A Novel GJC2 Mutation Associated with Hypomyelination and MÃ¼llerian Agenesis Syndrome: Coincidence or a New Entity?. Neuropediatrics, 2012, 43, 159-161.	0.6	5
89	Chromosome 14q11.2-q21.1 duplication: a rare cause of West syndrome. Epileptic Disorders, 2018, 20, 219-224.	1.3	5
90	Combined in vitro and in silico analyses of missense mutations in GNPTAB provide new insights into the molecular bases of mucolipidosis II and III alpha/beta. Human Mutation, 2020, 41, 133-139.	2.5	5

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91	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
92	Renal involvement in patients with mucolipidosis IIIalpha/beta: Causal relation or coâ€occurrence?. American Journal of Medical Genetics, Part A, 2016, 170, 1187-1195.	1.2	4
93	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
94	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
95	Long-Term Follow-Up Outcomes of 19 Patients with Osteogenesis Imperfecta Type XI and Bruck Syndrome Type I Caused by FKBP10 Variants. Calcified Tissue International, 2021, 109, 633-644.	3.1	4
96	Natural history of facial and skeletal features from neonatal period to adulthood in a 3M syndrome cohort with biallelic CUL7 or OBSL1 variants. European Journal of Medical Genetics, 2021, 64, 104346.	1.3	4
97	A splice site mutation in the <scp>TSEN2</scp> causes a new syndrome with craniofacial and central nervous system malformations, and atypical hemolytic uremic syndrome. Clinical Genetics, 2022, 101, 346-358.	2.0	4
98	Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type: longitudinal observation of radiographic findings in a child heterozygous for a KIF22 mutation. Pediatric Radiology, 2015, 45, 771-776.	2.0	3
99	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
100	A Novel Mutation of KIF11 in a Child with 22q11.2 Deletion Syndrome Associated with MCLMR. Molecular Syndromology, 2018, 9, 266-270.	0.8	3
101	Biallelic BICD2 variant is a novel candidate for Cohen-like syndrome. Journal of Human Genetics, 2022, 67, 553-556.	2.3	3
102	Seven patients with Smith-McCort dysplasia 2: Four novel nonsense variants in RAB33B and follow-up findings. European Journal of Medical Genetics, 2021, 64, 104248.	1.3	2
103	A patient with mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, keratodermia syndrome caused by AP1B1 gene variant. Clinical Dysmorphology, 2021, 30, 54-57.	0.3	2
104	Longâ€term followâ€up findings in a Turkish girl with osteogenesis imperfecta type <scp>XX</scp> caused by a homozygous <scp>MESD</scp> variant. American Journal of Medical Genetics, Part A, 2022, 188, 1639-1646.	1.2	2
105	Specific early signs and long-term follow-up findings of progressive pseudorheumatoid dysplasia (PPRD) in the Turkish cohort. Rheumatology, 2022, 61, 3693-3703.	1.9	2
106	Short trunk stature, brachydactyly, and platyspondyly in three sibs: A new form of brachyolmia or a new skeletal dysplasia?. , 2003, 119A, 375-380.		1
107	Three Offspring with Cri-du-Chat Syndrome from Phenotypically Normal Parents. Molecular Syndromology, 2020, 11, 97-103.	0.8	1
108	Spondylometaepiphyseal Dysplasia Short Limb-Abnormal Calcification Type in Turkish Patients Reveals a Novel Mutation and New Features. Molecular Syndromology, 2022, 13, 23-37.	0.8	1

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109	The Methylation Status in the Chromosome 11p15.5 Region and Metabolic Disorders in Children with Syndromic and Nonsyndromic Intrauterine Growth Restriction. Molecular Syndromology, 2022, 13, 108-116.	0.8	1
110	Response to Kouwenberg et al. â€œRecognizable Phenotype With Common Occurrence of Microcephaly, Psychomotor Retardation, But No Spontaneous Bone Fractures in ARCL2B Due to PYCR1 Mutationsâ€• American Journal of Medical Genetics, Part A, 2011, 155, 2333-2334.	1.2	0
111	Neurofibromatosis Type 1 in Children: A Single-Center Experience. , 2021, 56, 339-343.		0
112	Two novel variants and follow-up findings in four children with Bloom syndrome from two families. Clinical Dysmorphology, 2021, Publish Ahead of Print, 31-35.	0.3	0
113	Follow-up Findings in a Turkish Girl with Pseudohypoparathyroidism Type Ia Caused by a Novel Heterozygous Mutation in the GNAS Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 74-79.	0.9	0
114	Effects of Long-Term Pamidronate Treatment on Bone Density and Fracture Rate in 65 Osteogenesis Imperfecta Patients. , 2023, 56, 474-478.		0