HÏ‹lya Kayserili

List of Publications by Year
in descending order

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200 papers	15,690 citations	19608 61 h-index	19690 117 g-index
215 all docs	215 docs citations	215 times ranked	22700 citing authors

#	Article	IF	Citations
1	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025.	13.5	1,725
2	Activation-Induced Cytidine Deaminase (AID) Deficiency Causes the Autosomal Recessive Form of the Hyper-IgM Syndrome (HIGM2). Cell, 2000, 102, 565-575.	13.5	1,489
3	Mutations in the O-Mannosyltransferase Gene POMT1 Give Rise to the Severe Neuronal Migration Disorder Walker-Warburg Syndrome. American Journal of Human Genetics, 2002, 71, 1033-1043.	2.6	636
4	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	6.0	466
5	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	9.4	383
6	Roberts syndrome is caused by mutations in ESCO2, a human homolog of yeast ECO1 that is essential for the establishment of sister chromatid cohesion. Nature Genetics, 2005, 37, 468-470.	9.4	334
7	p63 Gene Mutations in EEC Syndrome, Limb-Mammary Syndrome, and Isolated Split Handâ€“Split Foot Malformation Suggest a Genotype-Phenotype Correlation. American Journal of Human Genetics, 2001, 69, 481-492.	2.6	331
8	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	9.4	326
9	Mutations in the AHI1 Gene, Encoding Jouberin, Cause Joubert Syndrome with Cortical Polymicrogyria. American Journal of Human Genetics, 2004, 75, 979-987.	2.6	275
10	Mutations in BCKD-kinase Lead to a Potentially Treatable Form of Autism with Epilepsy. Science, 2012, 338, 394-397.	6.0	272
11	Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndrome. Nature Genetics, 2000, 25, 423-426.	9.4	242
12	Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574.	2.6	240
13	Responsible implementation of expanded carrier screening. European Journal of Human Genetics, 2016, 24, e1-e12.	1.4	240
14	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	13.5	228
15	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	9.4	211
16	Genotypic and Phenotypic Spectrum in Tricho-Rhino-Phalangeal Syndrome Types I and III. American Journal of Human Genetics, 2001, 68, 81-91.	2.6	205
17	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	9.4	201
18	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	2.6	196

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19	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585.	9.4	191
20	Mutations in different components of FGF signaling in LADD syndrome. Nature Genetics, 2006, 38, 414-417.	9.4	190
21	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	1.4	190
22	Mutations in Capillary Morphogenesis Gene-2 Result in the Allelic Disorders Juvenile Hyaline Fibromatosis and Infantile Systemic Hyalinosis. American Journal of Human Genetics, 2003, 73, 957-966.	2.6	174
23	Harlequin Ichthyosis. Archives of Dermatology, 2011, 147, 681.	1.7	145
24	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	2.6	141
25	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	1.5	141
26	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. Nature, 2018, 557, 564-569.	13.7	141
27	Epigenetic mutations of the imprinted IGF2-H19 domain in Silver-Russell syndrome (SRS): results from a large cohort of patients with SRS and SRS-like phenotypes. Journal of Medical Genetics, 2008, 46, 192-197.	1.5	138
28	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	2.6	137
29	17Â-Hydroxysteroid Dehydrogenase-3 Deficiency: Diagnosis, Phenotypic Variability, Population Genetics, and Worldwide Distribution of Ancient and de Novo Mutations. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 4713-4721.	1.8	136
30	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	1.1	134
31	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	2.3	134
32	Exome sequencing identifiesDYNC2H1mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. Journal of Medical Genetics, 2013, 50, 309-323.	1.5	127
33	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	1.4	115
34	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 2010, 31, 1142-1154.	1.1	111
35	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	9.4	111
36	BBS4 Is a Minor Contributor to Bardet-Biedl Syndrome and May Also Participate in Triallelic Inheritance. American Journal of Human Genetics, 2002, 71, 22-29.	2.6	110

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37	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	2.6	110
38	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	2.6	110
39	The molecular mechanism underlying Roberts syndrome involves loss of ESCO2 acetyltransferase activity. Human Molecular Genetics, 2008, 17, 2172-2180.	1.4	108
40	Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2013, 93, 932-944.	2.6	108
41	Loss of desmoplakin isoform I causes early onset cardiomyopathy and heart failure in a Naxos-like syndrome. Journal of Medical Genetics, 2005, 43, e05-e05.	1.5	106
42	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	2.8	104
43	ALX4 dysfunction disrupts craniofacial and epidermal development. Human Molecular Genetics, 2009, 18, 4357-4366.	1.4	103
44	Spectrum ofHSPG2(Perlecan) mutations in patients with Schwartz-Jampel syndrome. Human Mutation, 2006, 27, 1082-1091.	1.1	98
45	Deficiency for the ER-stress transducer OASIS causes severe recessive osteogenesis imperfecta in humans. Orphanet Journal of Rare Diseases, 2013, 8, 154.	1.2	98
46	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914.	2.6	90
47	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
48	Mutations in RIPK4 Cause the Autosomal-Recessive Form of Popliteal Pterygium Syndrome. American Journal of Human Genetics, 2012, 90, 76-85.	2.6	80
49	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	1.1	80
50	European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. European Journal of Human Genetics, 2019, 27, 1763-1773.	1.4	78
51	Recessive osteogenesis imperfecta caused by LEPRE1 mutations: clinical documentation and identification of the splice form responsible for prolyl 3-hydroxylation. Journal of Medical Genetics, 2009, 46, 233-241.	1.5	77
52	Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome. Human Genetics, 2013, 132, 885-898.	1.8	77
53	De novo mutations in PLXND1 and REV3L cause MÃ¶bius syndrome. Nature Communications, 2015, 6, 7199.	5.8	76
54	Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. Human Mutation, 2014, 35, 76-85.	1.1	74

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55	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	1.8	73
56	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	1.8	71
57	Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	9.4	70
58	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	3.9	69
59	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	9.4	68
60	Frequency of renal malformations in Turner syndrome: analysis of 82 Turkish children. Pediatric Nephrology, 2000, 14, 1111-1114.	0.9	67
61	Identification of 11 novel mutations in eight BBS genes by high-resolution homozygosity mapping. Journal of Medical Genetics, 2010, 47, 262-267.	1.5	67
62	Genotypic and phenotypic analysis of 396 individuals with mutations inSonic Hedgehog. Journal of Medical Genetics, 2012, 49, 473-479.	1.5	67
63	Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2759-2767.	1.8	67
64	Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	9.4	66
65	Phenotypic variability in 49 cases of ESCO2 mutations, including novel missense and codon deletion in the acetyltransferase domain, correlates with ESCO2 expression and establishes the clinical criteria for Roberts syndrome. Journal of Medical Genetics, 2010, 47, 30-37.	1.5	65
66	Recontacting patients in clinical genetics services: recommendations of the European Society of Human Genetics. European Journal of Human Genetics, 2019, 27, 169-182.	1.4	65
67	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	1.4	64
68	Homozygous mutation inNUP107leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. Journal of Medical Genetics, 2017, 54, 399-403.	1.5	62
69	Spectrum of mutations in the Fanconi anaemia group G gene, FANCG/XRCC9. European Journal of Human Genetics, 2000, 8, 861-868.	1.4	61
70	Mutations in ACTRT1 and its enhancer RNA elements lead to aberrant activation of Hedgehog signaling in inherited and sporadic basal cell carcinomas. Nature Medicine, 2017, 23, 1226-1233.	15.2	59
71	Mutations in thelipoma HMGIC fusion partner-like 5 (LHFPL5)gene cause autosomal recessive nonsyndromic hearing loss. Human Mutation, 2006, 27, 633-639.	1.1	58
72	OTX2mutations contribute to the otocephaly-dysgnathia complex. Journal of Medical Genetics, 2012, 49, 373-379.	1.5	58

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73	Tumor spectrum in children with Noonan syndrome and SOS1 or RAF1 mutations. Genes Chromosomes and Cancer, 2010, 49, 242-252.	1.5	57
74	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	0.5	57
75	De novo <scp>WNT5A</scp>â€associated autosomal dominant Robinow syndrome suggests specificity of genotype and phenotype. Clinical Genetics, 2015, 87, 34-41.	1.0	56
76	Mutations in <scp>CDK</scp> 5 <scp>RAP</scp> 2 cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480.	0.6	55
77	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	1.5	55
78	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	5.8	51
79	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellisâ€“van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571.	1.4	50
80	Seven cases of Wiedemann-Beckwith syndrome, including the first reported case of mosaic paternal isodisomy along the whole chromosome 11. , 1998, 79, 347-353.		49
81	Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 128.	1.2	46
82	Evaluation of Clinical Manifestations in Patients with Severe Lymphedema with and without CCBE1 Mutations. Molecular Syndromology, 2013, 4, 107-113.	0.3	45
83	Whole-Exome Sequencing Identifies Novel Variants for Tooth Agenesis. Journal of Dental Research, 2018, 97, 49-59.	2.5	44
84	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	3.7	43
85	Identification of loss-of-function mutations of SLC35D1 in patients with Schneckenbecken dysplasia, but not with other severe spondylodysplastic dysplasias group diseases. Journal of Medical Genetics, 2009, 46, 562-568.	1.5	41
86	A Novel Semiquantitative Polymerase Chain Reaction/Enzyme Digestion-Based Method for Detection of Large Scale Deletions/Conversions of the CYP21 Gene and Mutation Screening in Turkish Families with 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 5893-5897.	1.8	39
87	Down Syndrome Diagnosis Based on Gabor Wavelet Transform. Journal of Medical Systems, 2012, 36, 3205-3213.	2.2	39
88	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8.	1.8	37
89	Unusual molecular findings in autosomal recessive spinal muscular atrophy.. Journal of Medical Genetics, 1996, 33, 469-474.	1.5	36
90	Identification and Functional Assessment of Novel and Known Insulin Receptor Mutations in Five Patients with Syndromes of Severe Insulin Resistance. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 4251-4257.	1.8	35

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91	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	2.6	35
92	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	1.5	35
93	Variants in members of the cadherinâ€“catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. European Journal of Human Genetics, 2018, 26, 210-219.	1.4	34
94	Recontacting or not recontacting? A survey of current practices in clinical genetics centres in Europe. European Journal of Human Genetics, 2018, 26, 946-954.	1.4	33
95	Oral manifestations of 17 patients affected with mucopolysaccharidosis type VI. Journal of Inherited Metabolic Disease, 2014, 37, 263-268.	1.7	32
96	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.	1.1	32
97	Cleidocranial dysplasia: Clinical, endocrinologic and molecular findings in 15 patients from 11 families. European Journal of Medical Genetics, 2017, 60, 163-168.	0.7	31
98	Teratogenicity of Antiepileptic Drugs. Clinical Psychopharmacology and Neuroscience, 2017, 15, 19-27.	0.9	30
99	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	5.8	30
100	Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. Neuron, 2020, 107, 82-94.e6.	3.8	30
101	Mild nasal malformations and parietal foramina caused by homozygous ALX4 mutations. American Journal of Medical Genetics, Part A, 2012, 158A, 236-244.	0.7	29
102	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused byPAPSS2Mutations. Human Mutation, 2013, 34, 1381-1386.	1.1	29
103	A mutation in the signal sequence ofLRP5in a family with an osteoporosis-pseudoglioma syndrome (OPPG)-like phenotype indicates a novel disease mechanism for trinucleotide repeats. Human Mutation, 2009, 30, 641-648.	1.1	27
104	Points to consider for prioritizing clinical genetic testing services: a European consensus process oriented at accountability for reasonableness. European Journal of Human Genetics, 2015, 23, 729-735.	1.4	26
105	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. American Journal of Human Genetics, 2019, 105, 1126-1147.	2.6	25
106	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 13	1.3	25
107	A loss-of-function NUAK2 mutation in humans causes anencephaly due to impaired Hippo-YAP signaling. Journal of Experimental Medicine, 2020, 217, .	4.2	25
108	A new syndrome, congenital extraocular muscle fibrosis with ulnar hand anomalies, maps to chromosome 21qter. Journal of Medical Genetics, 2005, 42, 408-415.	1.5	24

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109	Candidate locus analysis for PHACE syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1363-1367.	0.7	24
110	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. Human Genetics, 2018, 137, 689-703.	1.8	24
111	Congenital heart disease in children with DownÂ's syndrome: Turkish experience of 13 years. Acta Cardiologica, 2008, 63, 585-589.	0.3	23
112	Sclerosing bone dysplasias with hallmarks of dysosteosclerosis in four patients carrying mutations in SLC29A3 and TCIRG1. Bone, 2019, 120, 495-503.	1.4	23
113	Seckel syndrome: report of a case. Journal of Clinical Pediatric Dentistry, 2002, 26, 305-309.	0.5	22
114	An Insulin Receptor Mutant (Asp707â†' Ala), Involved in Leprechaunism, Is Processed and Transported to the Cell Surface but Unable to Bind Insulin. Journal of Biological Chemistry, 1996, 271, 18719-18724.	1.6	21
115	Detection of Y Chromosomal Material in Patients with a 45,X Karyotype by PCR Method. Tohoku Journal of Experimental Medicine, 2007, 211, 243-249.	0.5	21
116	Enamelâ€"Renalâ€"Gingival syndrome, hypodontia, and a novel FAM20A mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2124-2128.	0.7	21
117	Loss of Iroquois homeobox transcription factors 3 and 5 in osteoblasts disrupts cranial mineralization. Bone Reports, 2016, 5, 86-95.	0.2	21
118	A homozygous pathogenic missense variant broadens the phenotypic and mutational spectrum of CREB3L1-related osteogenesis imperfecta. Human Molecular Genetics, 2019, 28, 1801-1809.	1.4	21
119	Orodental findings of a family with lacrimo-auriculo-dento digital (LADD) syndrome. Oral Surgery Oral Medicine Oral Pathology Oral Radiology and Endodontics, 2008, 106, e33-e44.	1.6	20
120	Clinical manifestations of 17 patients affected with mucopolysaccharidosis type VI and eight novel ARSB mutations. American Journal of Medical Genetics, Part A, 2014, 164, 1443-1453.	0.7	20
121	Molecular analyses of the HGO gene mutations in Turkish alkaptonuria patients suggest that the R58fs mutation originated from Central Asia and was spread throughout Europe and Anatolia by human migrations. Journal of Inherited Metabolic Disease, 2003, 26, 17-23.	1.7	19
122	Etiologic Evaluation in 247 Children with Global Developmental Delay at Istanbul, Turkey. Journal of Tropical Pediatrics, 2005, 51, 310-313.	0.7	18
123	Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. Human Mutation, 2017, 38, 524-531.	1.1	18
124	Pallister-Killian syndrome: clinical, cytogenetic and molecular findings in 15 cases. Molecular Cytogenetics, 2018, 11, 45.	0.4	18
125	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	1.4	18
126	Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling. Neuromuscular Disorders, 2019, 29, 601-613.	0.3	18

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127	Skull defects, alopecia, hypertelorism, and notched alae nasi caused by homozygous ALX4 gene mutation. American Journal of Medical Genetics, Part A, 2014, 164, 1322-1327.	0.7	17
128	Variants affecting diverse domains of MEPE are associated with two distinct bone disorders, a craniofacial bone defect and otosclerosis. Genetics in Medicine, 2019, 21, 1199-1208.	1.1	17
129	Human model of IRX5 mutations reveals key role for this transcription factor in ventricular conduction. Cardiovascular Research, 2021, 117, 2092-2107.	1.8	17
130	Molecular genetic screening of MBS1 locus on chromosome 13 for microdeletions and exclusion of FGF9, GSH1 and CDX2 as causative genes in patients with Moebius syndrome. European Journal of Medical Genetics, 2009, 52, 315-320.	0.7	16
131	An unusual presentation of Kabuki syndrome with orbital cysts, microphthalmia, and cholestasis with bile duct paucity. American Journal of Medical Genetics, Part A, 2016, 170, 3282-3288.	0.7	16
132	Overarching control of autophagy and DNA damage response by CHD6 revealed by modeling a rare human pathology. Nature Communications, 2021, 12, 3014.	5.8	16
133	Molecular characterisation of a new case of microphthalmia with linear skin defects (MLS). Journal of Medical Genetics, 2001, 38, 411-417.	1.5	15
134	A large duplication involving the IHH locus mimics acrocallosal syndrome. European Journal of Human Genetics, 2012, 20, 639-644.	1.4	14
135	Clinicogenetic Study of Turkish Patients With Syndromic Craniosynostosis and Literature Review. Pediatric Neurology, 2014, 50, 482-490.	1.0	14
136	A Micropatterned Humanâ€Specific Neuroepithelial Tissue for Modeling Gene and Drugâ€Induced Neurodevelopmental Defects. Advanced Science, 2021, 8, 2001100.	5.6	13
137	Evaluation of coronary artery abnormalities in Williams syndrome patients using myocardial perfusion scintigraphy and CT angiography. Cardiology Journal, 2012, 19, 301-308.	0.5	13
138	Cardiovascular abnormalities in Williams syndrome: 20 yearsâ€™ experience in Istanbul. Acta Cardiologica, 2012, 67, 649-655.	0.3	12
139	Novel DDR2 mutation identified by whole exome sequencing in a Moroccan patient with spondyloâ€metaâ€epiphyseal dysplasia, short limbâ€abnormal calcification type. American Journal of Medical Genetics, Part A, 2016, 170, 460-465.	0.7	12
140	Glycine to tryptophan substitution in type I collagen in a patient with OI type III: a unique collagen mutation. Journal of Medical Genetics, 2000, 37, 371-375.	1.5	11
141	The identification of small supernumerary marker chromosomes; the experiences of 15,792 fetal karyotyping from Turkey. European Journal of Medical Genetics, 2006, 49, 207-214.	0.7	11
142	Craniodentofacial Manifestations In Hallermann-Streiff Syndrome. Cranio - Journal of Craniomandibular Practice, 2009, 27, 33-38.	0.6	11
143	Newly described clinical features in two siblings with MACS syndrome and a novel mutation in RIN2. American Journal of Medical Genetics, Part A, 2014, 164, 484-489.	0.7	11
144	A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1015-1022.	0.7	11

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145	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	2.6	11
146	Prenatal ultrasonographic diagnosis of generalized arterial calcification of infancy. Journal of Clinical Ultrasound, 2015, 43, 50-54.	0.4	10
147	Clinical exome sequencing in neuromuscular diseases: an experience from Turkey. Neurological Sciences, 2020, 41, 2157-2164.	0.9	10
148	Screening of Deletions in SMN, NAIP and BTF2p44 Genes in Turkish Spinal Muscular Atrophy Patients. Human Heredity, 2000, 50, 162-165.	0.4	9
149	Mild nasal clefting may be predictive for ALX4 heterozygotes. American Journal of Medical Genetics, Part A, 2014, 164, 2054-2058.	0.7	9
150	Twins with hereditary sensory and autonomic neuropathy type IV with preserved periodontal sensation. European Journal of Medical Genetics, 2014, 57, 240-246.	0.7	9
151	A new hereditary congenital facial palsy case supports arg5 in HOX-DNA binding domain as possible hot spot for mutations. European Journal of Medical Genetics, 2015, 58, 358-363.	0.7	9
152	Clinical delineation of a subtype of frontonasal dysplasia with creased nasal ridge and upper limb anomalies: Report of six unrelated patients. American Journal of Medical Genetics, Part A, 2017, 173, 3136-3142.	0.7	9
153	Short Rib Polydactyly Syndrome Type 3 with Absence of Fibulae (Verma-Naumoff Syndrome). Fetal Diagnosis and Therapy, 2005, 20, 410-414.	0.6	8
154	Short Femurs Detected at 25 and 31 Weeks of Gestation Diagnosed as Leroy I-Cell Disease in the Postnatal Period: A Report of Two Cases. Fetal Diagnosis and Therapy, 2007, 22, 198-202.	0.6	8
155	A novel c.1255G>T (p.D419Y) mutation in SH3BP2 gene causes cherubism in a Turkish family. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2012, 114, e42-e46.	0.2	8
156	Skeletal and molecular findings in 51 Cleidocranial dysplasia patients from Turkey. American Journal of Medical Genetics, Part A, 2021, 185, 2488-2495.	0.7	8
157	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	1.1	8
158	Identification of point mutations in Turkish DMD/BMD families using multiplex-single stranded conformation analysis (SSCA). European Journal of Human Genetics, 1999, 7, 765-770.	1.4	7
159	Studies on the pathogenesis of Costello syndrome. Journal of Medical Genetics, 2003, 40, 37e-37.	1.5	7
160	Prenatal diagnosis of frontonasal dysplasia with anterior encephalocele. Journal of the Turkish German Gynecology Association, 2013, 14, 50-52.	0.2	7
161	Sclerosteosis (craniotubular hyperostosis-syndactyly) with complex hyperphalangy of the index finger. Pediatric Radiology, 2015, 45, 1239-1243.	1.1	7
162	Characteristic calcaneal ossification: an additional early radiographic finding in infants with fibrodysplasia ossificans progressiva. Pediatric Radiology, 2016, 46, 1568-1572.	1.1	7

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