

Hİlya Kayserili

List of Publications by Year in descending order

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Version: 2024-02-01

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	Expanding the spectrum of syndromic <i>PPP2R3C</i> -related <i>XY</i> gonadal dysgenesis to <i>XX</i> gonadal dysgenesis. <i>Clinical Genetics</i> , 2022, 101, 221-232.	1.0	3
2	Functional loss of ubiquitin-specific protease 14 may lead to a novel distal arthrogryposis phenotype. <i>Clinical Genetics</i> , 2022, 101, 421-428.	1.0	1
3	A New Family with a Novel <i>OTUD6B</i> Mutation: Practicing Whole Exome Sequencing for Antenatal Diagnosis of Tetralogy of Fallot. <i>Molecular Syndromology</i> , 2022, 13, 206-211.	0.3	2
4	Biallelic <i>TERT</i> variant leads to Hoyeraal-Hreidarsson syndrome with additional dyskeratosis congenita findings. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1226-1232.	0.7	2
5	Phenotypic and mutational spectrum of <i>ROR2</i> -related Robinow syndrome. <i>Human Mutation</i> , 2022, 43, 900-918.	1.1	8
6	Evaluation of growth, puberty, osteoporosis, and the response to long-term bisphosphonate therapy in four patients with osteoporosis-pseudoglioma syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	0.7	2
7	Human model of <i>IRX5</i> mutations reveals key role for this transcription factor in ventricular conduction. <i>Cardiovascular Research</i> , 2021, 117, 2092-2107.	1.8	17
8	A Micropatterned Human-specific Neuroepithelial Tissue for Modeling Gene and Drug-induced Neurodevelopmental Defects. <i>Advanced Science</i> , 2021, 8, 2001100.	5.6	13
9	Skeletal and molecular findings in 51 Cleidocranial dysplasia patients from Turkey. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2488-2495.	0.7	8
10	Overarching control of autophagy and DNA damage response by CHD6 revealed by modeling a rare human pathology. <i>Nature Communications</i> , 2021, 12, 3014.	5.8	16
11	Loss of <i>C2orf69</i> defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 1301-1317.	2.6	11
12	ESHG PPC Comments on postmortem use of genetic data for research purposes. <i>European Journal of Human Genetics</i> , 2020, 28, 144-146.	1.4	3
13	A rare cause of chronic hyponatremia in an infant: Questions. <i>Pediatric Nephrology</i> , 2020, 35, 241-242.	0.9	0
14	Heterozygous pathogenic variants in <i>GLI1</i> are a common finding in isolated postaxial polydactyly A/B. <i>Human Mutation</i> , 2020, 41, 265-276.	1.1	6
15	A loss-of-function <i>NUAK2</i> mutation in humans causes anencephaly due to impaired Hippo-YAP signaling. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	25
16	Loss of <i>MTX2</i> causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. <i>Nature Communications</i> , 2020, 11, 4589.	5.8	30
17	Characteristic dental pattern with hypodontia and short roots in Fraser syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1681-1689.	0.7	7
18	Clinical exome sequencing in neuromuscular diseases: an experience from Turkey. <i>Neurological Sciences</i> , 2020, 41, 2157-2164.	0.9	10

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19	Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. <i>Neuron</i> , 2020, 107, 82-94.e6.	3.8	30
20	A novel shoulder disability staging system for scapulothoracic arthrodesis in patients with facioscapulohumeral dystrophy. <i>Orthopaedics and Traumatology: Surgery and Research</i> , 2020, 106, 701-707.	0.9	6
21	Cohesin complex-associated holoprosencephaly. <i>Brain</i> , 2019, 142, 2631-2643.	3.7	43
22	European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. <i>European Journal of Human Genetics</i> , 2019, 27, 1763-1773.	1.4	78
23	A homozygous pathogenic missense variant broadens the phenotypic and mutational spectrum of CREB3L1-related osteogenesis imperfecta. <i>Human Molecular Genetics</i> , 2019, 28, 1801-1809.	1.4	21
24	Turkish Ectodermal Dysplasia Cohort: From Phenotype to Genotype in 17 Families. <i>Cytogenetic and Genome Research</i> , 2019, 157, 189-196.	0.6	7
25	Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling. <i>Neuromuscular Disorders</i> , 2019, 29, 601-613.	0.3	18
26	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. <i>American Journal of Human Genetics</i> , 2019, 105, 1126-1147.	2.6	25
27	Zoledronate-responsive calcitriol-mediated hypercalcemia in a 5-year-old case with squamous cell carcinoma on the background of xeroderma pigmentosum. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2019, 32, 1403-1406.	0.4	3
28	Terminal osseous dysplasia with pigmentary defects (TODPD) in a Turkish girl with new skin findings. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 123-129.	0.7	4
29	Sclerosing bone dysplasias with hallmarks of dysosteosclerosis in four patients carrying mutations in SLC29A3 and TCIRG1. <i>Bone</i> , 2019, 120, 495-503.	1.4	23
30	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	1.1	80
31	Recontacting patients in clinical genetics services: recommendations of the European Society of Human Genetics. <i>European Journal of Human Genetics</i> , 2019, 27, 169-182.	1.4	65
32	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctive cerebellar, ocular, craniofacial and genital features (COFG) <i>Tj ETQq0 0 0 rgBT 10verlock 20 Tf 50 2</i>	1.0	10
33	Variants affecting diverse domains of MEPE are associated with two distinct bone disorders, a craniofacial bone defect and otosclerosis. <i>Genetics in Medicine</i> , 2019, 21, 1199-1208.	1.1	17
34	CLINICAL CLASSIFICATION OF RADIAL RAY DEFECTS AND RESEARCH INTO ETIOPATHOGENESIS. <i>European Oral Research</i> , 2019, 81, 127-138.	0.5	0
35	APPLICATION OF MLPA (MULTIPLEX LIGATION-DEPENDENT PROBE AMPLIFICATION) IN FETUSES WITH AN ABNORMAL SONOGRAM AND NORMAL KARYOTYPE. <i>Ä°stanbul TÄ±p FakÄ°ltesi Dergisi</i> , 2019, 82, 5-11.	0.1	0
36	A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1015-1022.	0.7	11

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37	Recontacting or not recontacting? A survey of current practices in clinical genetics centres in Europe. <i>European Journal of Human Genetics</i> , 2018, 26, 946-954.	1.4	33
38	Variants in members of the cadherin-catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 210-219.	1.4	34
39	Whole-Exome Sequencing Identifies Novel Variants for Tooth Agenesis. <i>Journal of Dental Research</i> , 2018, 97, 49-59.	2.5	44
40	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2740-2750.	0.7	6
41	Pallister-Killian syndrome: clinical, cytogenetic and molecular findings in 15 cases. <i>Molecular Cytogenetics</i> , 2018, 11, 45.	0.4	18
42	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. <i>Nature</i> , 2018, 557, 564-569.	13.7	141
43	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. <i>Human Genetics</i> , 2018, 137, 689-703.	1.8	24
44	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscaphodysplasia. <i>European Journal of Human Genetics</i> , 2018, 26, 1611-1622.	1.4	18
45	Biallelic loss of human CTNNA2, encoding β -catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. <i>Nature Genetics</i> , 2018, 50, 1093-1101.	9.4	70
46	A facioscapulohumeralis muscularis dystrophia kezeli nek multidisziplinaris megkezelése. <i>Ideggyogyaszati Szemle</i> , 2018, 71, 337-342.	0.4	0
47	Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. <i>Human Mutation</i> , 2017, 38, 524-531.	1.1	18
48	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. <i>Nature Genetics</i> , 2017, 49, 249-255.	9.4	88
49	Biallelic mutations in the 3' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. <i>Nature Genetics</i> , 2017, 49, 457-464.	9.4	66
50	Homozygous mutation in <i>NUP107</i> leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. <i>Journal of Medical Genetics</i> , 2017, 54, 399-403.	1.5	62
51	Cleidocranial dysplasia: Clinical, endocrinologic and molecular findings in 15 patients from 11 families. <i>European Journal of Medical Genetics</i> , 2017, 60, 163-168.	0.7	31
52	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellis-van Creveld syndrome. <i>Human Molecular Genetics</i> , 2017, 26, 4556-4571.	1.4	50
53	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. <i>American Journal of Human Genetics</i> , 2017, 101, 391-403.	2.6	35
54	Loss-of-function mutations in Carboxypeptidase D cause a new syndrome with lymphedema and sensorineural hearing loss. <i>Mechanisms of Development</i> , 2017, 145, S32.	1.7	0

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55	PYCR2 Protects from Neurodegeneration by Controlling Oligodendrocyte Maturation and Glycinemia through SHMT2. <i>Mechanisms of Development</i> , 2017, 145, S116-S117.	1.7	0
56	Clinical delineation of a subtype of frontonasal dysplasia with creased nasal ridge and upper limb anomalies: Report of six unrelated patients. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3136-3142.	0.7	9
57	Teratogenicity of Antiepileptic Drugs. <i>Clinical Psychopharmacology and Neuroscience</i> , 2017, 15, 19-27.	0.9	30
58	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. <i>PLoS Genetics</i> , 2017, 13, e1006683.	1.5	35
59	Homozygous, and compound heterozygous mutation in 3 Turkish family with Jervell and Lange-Nielsen syndrome: case reports. <i>BMC Medical Genetics</i> , 2017, 18, 114.	2.1	0
60	Mutations in ACTRT1 and its enhancer RNA elements lead to aberrant activation of Hedgehog signaling in inherited and sporadic basal cell carcinomas. <i>Nature Medicine</i> , 2017, 23, 1226-1233.	15.2	59
61	Imatinib response of gastrointestinal stromal tumor patients with germline mutation on KIT exon 13: A family report. <i>World Journal of Radiology</i> , 2017, 9, 365-370.	0.5	3
62	Mutation Update for Kabuki Syndrome Genes <i>KMT2D</i> and <i>KDM6A</i> and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. <i>Human Mutation</i> , 2016, 37, 847-864.	1.1	134
63	Ungual squamous cell carcinoma in a patient with Mal de Meleda. <i>JDDG - Journal of the German Society of Dermatology</i> , 2016, 14, 514-516.	0.4	1
64	485 Mutations in ACTRT1 and its transcribed non-coding elements lead to aberrant activation of the Hedgehog signaling pathway in inherited and sporadic basal cell carcinomas. <i>Journal of Investigative Dermatology</i> , 2016, 136, S243.	0.3	0
65	Characteristic calcaneal ossification: an additional early radiographic finding in infants with fibrodysplasia ossificans progressiva. <i>Pediatric Radiology</i> , 2016, 46, 1568-1572.	1.1	7
66	An unusual presentation of Kabuki syndrome with orbital cysts, microphthalmia, and cholestasis with bile duct paucity. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3282-3288.	0.7	16
67	Holt-Oram syndrome because of the novel TBX5 mutation c.481A>C. <i>Clinical Dysmorphology</i> , 2016, 25, 192-194.	0.1	1
68	Mutations in <i>CEP120</i> cause Joubert syndrome as well as complex ciliopathy phenotypes. <i>Journal of Medical Genetics</i> , 2016, 53, 608-615.	1.5	55
69	<i>ALX4</i> related parietal foramina mimicking encephalocele in prenatal period. <i>Prenatal Diagnosis</i> , 2016, 36, 591-593.	1.1	4
70	Microcephaly, dysmorphic features, corneal dystrophy, hairy nipples, underdeveloped labioscrotal folds, and small cerebellum in four patients. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1391-1399.	0.7	5
71	Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 2759-2767.	1.8	67
72	Loss of Iroquois homeobox transcription factors 3 and 5 in osteoblasts disrupts cranial mineralization. <i>Bone Reports</i> , 2016, 5, 86-95.	0.2	21

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73	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. <i>Cilia</i> , 2016, 5, 8.	1.8	37
74	Novel <i>DDR2</i> mutation identified by whole exome sequencing in a Moroccan patient with spondyloepiphyseal dysplasia, short limb abnormal calcification type. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 460-465.	0.7	12
75	Responsible implementation of expanded carrier screening. <i>European Journal of Human Genetics</i> , 2016, 24, e1-e12.	1.4	240
76	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. <i>JCI Insight</i> , 2016, 1, .	2.3	134
77	Prenatal ultrasonographic diagnosis of generalized arterial calcification of infancy. <i>Journal of Clinical Ultrasound</i> , 2015, 43, 50-54.	0.4	10
78	Mutations in <i>CDK5</i> & <i>RAP2</i> cause Seckel syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 467-480.	0.6	55
79	Novel <i>MASP1</i> mutations are associated with an expanded phenotype in 3MC1 syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 128.	1.2	46
80	Sclerosteosis (craniotubular hyperostosis-syndactyly) with complex hyperphalangy of the index finger. <i>Pediatric Radiology</i> , 2015, 45, 1239-1243.	1.1	7
81	De novo mutations in <i>PLXND1</i> and <i>REV3L</i> cause M ^u bius syndrome. <i>Nature Communications</i> , 2015, 6, 7199.	5.8	76
82	A severe collodion phenotype in the newborn period associated with a homozygous missense mutation in <i>ALOX12B</i> . <i>British Journal of Dermatology</i> , 2015, 173, 285-287.	1.4	2
83	A new hereditary congenital facial palsy case supports <i>arg5</i> in HOX-DNA binding domain as possible hot spot for mutations. <i>European Journal of Medical Genetics</i> , 2015, 58, 358-363.	0.7	9
84	<i>DOCK6</i> Mutations Are Responsible for a Distinct Autosomal-Recessive Variant of Adams-Oliver Syndrome Associated with Brain and Eye Anomalies. <i>Human Mutation</i> , 2015, 36, 593-598.	1.1	32
85	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. <i>Cell</i> , 2015, 161, 1012-1025.	13.5	1,725
86	<i>DVL1</i> Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 612-622.	2.6	110
87	Biallelic mutations in <i>SNX14</i> cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. <i>Nature Genetics</i> , 2015, 47, 528-534.	9.4	111
88	<i>TCTEX1D2</i> mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. <i>Nature Communications</i> , 2015, 6, 7074.	5.8	51
89	Determining the genome-wide kinship coefficient seems unhelpful in distinguishing consanguineous couples with a high versus low risk for adverse reproductive outcome. <i>BMC Medical Genetics</i> , 2015, 16, 50.	2.1	1
90	Points to consider for prioritizing clinical genetic testing services: a European consensus process oriented at accountability for reasonableness. <i>European Journal of Human Genetics</i> , 2015, 23, 729-735.	1.4	26

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91	<i>De novo</i> WNT5A-associated autosomal dominant Robinow syndrome suggests specificity of genotype and phenotype. <i>Clinical Genetics</i> , 2015, 87, 34-41.	1.0	56
92	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. <i>Journal of Clinical Investigation</i> , 2015, 125, 3585-3599.	3.9	69
93	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. <i>Journal of Medical Genetics</i> , 2014, 51, 659-668.	1.5	141
94	Newly described clinical features in two siblings with MACS syndrome and a novel mutation in RIN2. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 484-489.	0.7	11
95	Skull defects, alopecia, hypertelorism, and notched alae nasi caused by homozygous ALX4 gene mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1322-1327.	0.7	17
96	Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. <i>Human Mutation</i> , 2014, 35, 76-85.	1.1	74
97	Enamel "Renal" Gingival syndrome, hypodontia, and a novel FAM20A mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2124-2128.	0.7	21
98	Mild nasal clefting may be predictive for ALX4 heterozygotes. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2054-2058.	0.7	9
99	A patient with a de-novo deletion 3p25.3 and features overlapping with Rubinstein-Taybi syndrome. <i>Clinical Dysmorphology</i> , 2014, 23, 67-70.	0.1	3
100	Clinicogenetic Study of Turkish Patients With Syndromic Craniosynostosis and Literature Review. <i>Pediatric Neurology</i> , 2014, 50, 482-490.	1.0	14
101	C5orf42 is the major gene responsible for OFD syndrome type VI. <i>Human Genetics</i> , 2014, 133, 367-377.	1.8	71
102	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. <i>Science</i> , 2014, 343, 506-511.	6.0	466
103	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. <i>Cell</i> , 2014, 157, 651-663.	13.5	228
104	Clinical manifestations of 17 patients affected with mucopolysaccharidosis type VI and eight novel ARSB mutations. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1443-1453.	0.7	20
105	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. <i>American Journal of Human Genetics</i> , 2014, 95, 285-293.	2.6	110
106	Oral manifestations of 17 patients affected with mucopolysaccharidosis type VI. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 263-268.	1.7	32
107	Twins with hereditary sensory and autonomic neuropathy type IV with preserved periodontal sensation. <i>European Journal of Medical Genetics</i> , 2014, 57, 240-246.	0.7	9
108	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 905-914.	2.6	90

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109	Evaluation of Clinical Manifestations in Patients with Severe Lymphedema with and without CCBE1 Mutations. <i>Molecular Syndromology</i> , 2013, 4, 107-113.	0.3	45
110	Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome. <i>Human Genetics</i> , 2013, 132, 885-898.	1.8	77
111	Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy. <i>American Journal of Human Genetics</i> , 2013, 93, 932-944.	2.6	108
112	Genotype-phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 352-361.	0.5	57
113	Deficiency for the ER-stress transducer OASIS causes severe recessive osteogenesis imperfecta in humans. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 154.	1.2	98
114	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. <i>American Journal of Human Genetics</i> , 2013, 93, 915-925.	2.6	196
115	Multiple synostoses syndrome in three members of a family displaying a novel mutation in NOGGIN gene. <i>Journal of Plastic, Reconstructive and Aesthetic Surgery</i> , 2013, 66, e287-e289.	0.5	3
116	Mutations in WNT1 Cause Different Forms of Bone Fragility. <i>American Journal of Human Genetics</i> , 2013, 92, 565-574.	2.6	240
117	A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. <i>Human Molecular Genetics</i> , 2013, 22, 5121-5135.	1.4	190
118	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 1074-1078.	1.4	64
119	Exome sequencing identifies <i>DYNC2H1</i> mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. <i>Journal of Medical Genetics</i> , 2013, 50, 309-323.	1.5	127
120	Prenatal diagnosis of frontonasal dysplasia with anterior encephalocele. <i>Journal of the Turkish German Gynecology Association</i> , 2013, 14, 50-52.	0.2	7
121	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by <i>PAPSS2</i> Mutations. <i>Human Mutation</i> , 2013, 34, 1381-1386.	1.1	29
122	Genotypic and phenotypic analysis of 396 individuals with mutations in <i>Sonic Hedgehog</i> . <i>Journal of Medical Genetics</i> , 2012, 49, 473-479.	1.5	67
123	A large duplication involving the IHH locus mimics acrocallosal syndrome. <i>European Journal of Human Genetics</i> , 2012, 20, 639-644.	1.4	14
124	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. <i>Nature Genetics</i> , 2012, 44, 581-585.	9.4	191
125	<i>OTX2</i> mutations contribute to the otocephaly-dysgnathia complex. <i>Journal of Medical Genetics</i> , 2012, 49, 373-379.	1.5	58
126	Cardiovascular abnormalities in Williams syndrome: 20 years'™ experience in Istanbul. <i>Acta Cardiologica</i> , 2012, 67, 649-655.	0.3	12

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127	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. <i>Human Genetics</i> , 2012, 131, 1761-1773.	1.8	73
128	Down Syndrome Diagnosis Based on Gabor Wavelet Transform. <i>Journal of Medical Systems</i> , 2012, 36, 3205-3213.	2.2	39
129	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. <i>Nature Genetics</i> , 2012, 44, 709-713.	9.4	68
130	Mutations in <i>BCKD-kinase</i> Lead to a Potentially Treatable Form of Autism with Epilepsy. <i>Science</i> , 2012, 338, 394-397.	6.0	272
131	A novel c.1255G>T (p.D419Y) mutation in SH3BP2 gene causes cherubism in a Turkish family. <i>Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology</i> , 2012, 114, e42-e46.	0.2	8
132	Candidate locus analysis for PHACE syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1363-1367.	0.7	24
133	Multiple supernumerary molars, anterior openbite, and large ear lobules in mucopolysaccharidosis type VI patient. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1798-1800.	0.7	5
134	Mutations in RIPK4 Cause the Autosomal-Recessive Form of Popliteal Pterygium Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 76-85.	2.6	80
135	CantÃ© Syndrome Is Caused by Mutations in ABCC9. <i>American Journal of Human Genetics</i> , 2012, 90, 1094-1101.	2.6	141
136	Mild nasal malformations and parietal foramina caused by homozygous <i>ALX4</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 236-244.	0.7	29
137	Evaluation of coronary artery abnormalities in Williams syndrome patients using myocardial perfusion scintigraphy and CT angiography. <i>Cardiology Journal</i> , 2012, 19, 301-308.	0.5	13
138	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. <i>Nature Genetics</i> , 2011, 43, 23-26.	9.4	201
139	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. <i>Nature Genetics</i> , 2011, 43, 189-196.	9.4	326
140	A novel homozygous <i>COL11A2</i> deletion causes a C-terminal protein truncation with incomplete mRNA decay in a Turkish patient. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 180-185.	0.7	3
141	Harlequin Ichthyosis. <i>Archives of Dermatology</i> , 2011, 147, 681.	1.7	145
142	Tumor spectrum in children with Noonan syndrome and <i>SOS1</i> or <i>RAF1</i> mutations. <i>Genes Chromosomes and Cancer</i> , 2010, 49, 242-252.	1.5	57
143	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. <i>Human Mutation</i> , 2010, 31, 1142-1154.	1.1	111
144	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. <i>Journal of Medical Genetics</i> , 2010, 47, 30-37.	1.5	65

#	ARTICLE	IF	CITATIONS
145	Identification of 11 novel mutations in eight BBS genes by high-resolution homozygosity mapping. <i>Journal of Medical Genetics</i> , 2010, 47, 262-267.	1.5	67
146	LI-FRAUMENI SYNDROME IN A TURKISH FAMILY. <i>Pediatric Hematology and Oncology</i> , 2010, 27, 297-305.	0.3	4
147	P106. Congenital hypertelorism and osteopenia: A novel autosomal recessive disease. <i>Differentiation</i> , 2010, 80, S52.	1.0	0
148	ALX4 dysfunction disrupts craniofacial and epidermal development. <i>Human Molecular Genetics</i> , 2009, 18, 4357-4366.	1.4	103
149	Loss-of-function mutations in ATP6VOA2 impair vesicular trafficking, tropoelastin secretion and cell survival. <i>Human Molecular Genetics</i> , 2009, 18, 2149-2165.	1.4	115
150	Identification of loss-of-function mutations of SLC35D1 in patients with Schneckenbecken dysplasia, but not with other severe spondylodysplastic dysplasias group diseases. <i>Journal of Medical Genetics</i> , 2009, 46, 562-568.	1.5	41
151	A mutation in the signal sequence of <i>LRP5</i> in a family with an osteoporosis-pseudoglioma syndrome (OPPG)-like phenotype indicates a novel disease mechanism for trinucleotide repeats. <i>Human Mutation</i> , 2009, 30, 641-648.	1.1	27
152	Mutations in PYCR1 cause cutis laxa with progeroid features. <i>Nature Genetics</i> , 2009, 41, 1016-1021.	9.4	211
153	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidylinositol signaling to the ciliopathies. <i>Nature Genetics</i> , 2009, 41, 1032-1036.	9.4	383
154	Recessive osteogenesis imperfecta caused by LEPRE1 mutations: clinical documentation and identification of the splice form responsible for prolyl 3-hydroxylation. <i>Journal of Medical Genetics</i> , 2009, 46, 233-241.	1.5	77
155	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. <i>European Journal of Medical Genetics</i> , 2009, 52, 315-320.	0.7	16
156	Mutational screening of BASP1 and transcribed processed pseudogene TP ^g -BASP1 in patients with Moebius syndrome. <i>Journal of Genetics and Genomics</i> , 2009, 36, 251-256.	1.7	3
157	Gorlin's Syndrome: Case Report and Management Protocol. <i>Balkan Journal of Medical Genetics</i> , 2009, 12, 61-64.	0.5	2
158	Craniofacial Manifestations In Hallermann-Streiff Syndrome. <i>Cranio - Journal of Craniomandibular Practice</i> , 2009, 27, 33-38.	0.6	11
159	Orofacial findings of a family with lacrimo-auriculo-dento digital (LADD) syndrome. <i>Oral Surgery Oral Medicine Oral Pathology Oral Radiology and Endodontics</i> , 2008, 106, e33-e44.	1.6	20
160	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. <i>Journal of Medical Genetics</i> , 2008, 46, 192-197.	1.5	138
161	The molecular mechanism underlying Roberts syndrome involves loss of ESCO2 acetyltransferase activity. <i>Human Molecular Genetics</i> , 2008, 17, 2172-2180.	1.4	108
162	Congenital heart disease in children with Down's syndrome: Turkish experience of 13 years. <i>Acta Cardiologica</i> , 2008, 63, 585-589.	0.3	23

#	ARTICLE	IF	CITATIONS
163	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. <i>Fetal Diagnosis and Therapy</i> , 2007, 22, 198-202.	0.6	8
164	Detection of Y Chromosomal Material in Patients with a 45,X Karyotype by PCR Method. <i>Tohoku Journal of Experimental Medicine</i> , 2007, 211, 243-249.	0.5	21
165	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndrome-Related Disorders. <i>American Journal of Human Genetics</i> , 2007, 81, 104-113.	2.6	137
166	Evaluation of mental retardation - Part 1: Etiologic classification of 4659 patients with mental retardation or multiple congenital abnormality and mental retardation. <i>Journal of Pediatric Neurosciences</i> , 2007, 2, 45.	0.2	2
167	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. <i>Journal of Pediatric Neurosciences</i> , 2007, 2, 53.	0.2	0
168	The identification of small supernumerary marker chromosomes; the experiences of 15,792 fetal karyotyping from Turkey. <i>European Journal of Medical Genetics</i> , 2006, 49, 207-214.	0.7	11
169	Mutations in different components of FGF signaling in LADD syndrome. <i>Nature Genetics</i> , 2006, 38, 414-417.	9.4	190
170	Mutations in the lipoma HMGIC fusion partner-like 5 (LHFPL5) gene cause autosomal recessive nonsyndromic hearing loss. <i>Human Mutation</i> , 2006, 27, 633-639.	1.1	58
171	Spectrum of HSPG2 (Perlecan) mutations in patients with Schwartz-Jampel syndrome. <i>Human Mutation</i> , 2006, 27, 1082-1091.	1.1	98
172	Roberts syndrome is caused by mutations in ESCO2, a human homolog of yeast ECO1 that is essential for the establishment of sister chromatid cohesion. <i>Nature Genetics</i> , 2005, 37, 468-470.	9.4	334
173	Distinguishing the four genetic causes of Joubert syndrome-related disorders. <i>Annals of Neurology</i> , 2005, 57, 513-519.	2.8	104
174	Loss of desmoplakin isoform I causes early onset cardiomyopathy and heart failure in a Naxos-like syndrome. <i>Journal of Medical Genetics</i> , 2005, 43, e05-e05.	1.5	106
175	A new syndrome, congenital extraocular muscle fibrosis with ulnar hand anomalies, maps to chromosome 21qter. <i>Journal of Medical Genetics</i> , 2005, 42, 408-415.	1.5	24
176	Etiologic Evaluation in 247 Children with Global Developmental Delay at Istanbul, Turkey. <i>Journal of Tropical Pediatrics</i> , 2005, 51, 310-313.	0.7	18
177	Short Rib Polydactyly Syndrome Type 3 with Absence of Fibulae (Verma-Naumoff Syndrome). <i>Fetal Diagnosis and Therapy</i> , 2005, 20, 410-414.	0.6	8
178	Mutations in the AHI1 Gene, Encoding Joubertin, Cause Joubert Syndrome with Cortical Polymicrogyria. <i>American Journal of Human Genetics</i> , 2004, 75, 979-987.	2.6	275
179	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. <i>Journal of Inherited Metabolic Disease</i> , 2003, 26, 17-23.	1.7	19
180	Mutations in Capillary Morphogenesis Gene-2 Result in the Allelic Disorders Juvenile Hyaline Fibromatosis and Infantile Systemic Hyalinosis. <i>American Journal of Human Genetics</i> , 2003, 73, 957-966.	2.6	174

#	ARTICLE	IF	CITATIONS
181	Studies on the pathogenesis of Costello syndrome. <i>Journal of Medical Genetics</i> , 2003, 40, 37e-37.	1.5	7
182	Identification and Functional Assessment of Novel and Known Insulin Receptor Mutations in Five Patients with Syndromes of Severe Insulin Resistance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 4251-4257.	1.8	35
183	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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 5893-5897.	1.8	39
184	Seckel syndrome: report of a case. <i>Journal of Clinical Pediatric Dentistry</i> , 2002, 26, 305-309.	0.5	22
185	BBS4 Is a Minor Contributor to Bardet-Biedl Syndrome and May Also Participate in Triallelic Inheritance. <i>American Journal of Human Genetics</i> , 2002, 71, 22-29.	2.6	110
186	Mutations in the O-Mannosyltransferase Gene POMT1 Give Rise to the Severe Neuronal Migration Disorder Walker-Warburg Syndrome. <i>American Journal of Human Genetics</i> , 2002, 71, 1033-1043.	2.6	636
187	Genotypic and Phenotypic Spectrum in Tricho-Rhino-Phalangeal Syndrome Types I and III. <i>American Journal of Human Genetics</i> , 2001, 68, 81-91.	2.6	205
188	p63 Gene Mutations in EEC Syndrome, Limb-Mammary Syndrome, and Isolated Split Hand Split Foot Malformation Suggest a Genotype-Phenotype Correlation. <i>American Journal of Human Genetics</i> , 2001, 69, 481-492.	2.6	331
189	Molecular characterisation of a new case of microphthalmia with linear skin defects (MLS). <i>Journal of Medical Genetics</i> , 2001, 38, 411-417.	1.5	15
190	Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndrome. <i>Nature Genetics</i> , 2000, 25, 423-426.	9.4	242
191	Spectrum of mutations in the Fanconi anaemia group G gene, FANCG/XRCC9. <i>European Journal of Human Genetics</i> , 2000, 8, 861-868.	1.4	61
192	Frequency of renal malformations in Turner syndrome: analysis of 82 Turkish children. <i>Pediatric Nephrology</i> , 2000, 14, 1111-1114.	0.9	67
193	Screening of Deletions in SMN, NAIP and BTF2p44 Genes in Turkish Spinal Muscular Atrophy Patients. <i>Human Heredity</i> , 2000, 50, 162-165.	0.4	9
194	Glycine to tryptophan substitution in type I collagen in a patient with OI type III: a unique collagen mutation. <i>Journal of Medical Genetics</i> , 2000, 37, 371-375.	1.5	11
195	Activation-Induced Cytidine Deaminase (AID) Deficiency Causes the Autosomal Recessive Form of the Hyper-IgM Syndrome (HIGM2). <i>Cell</i> , 2000, 102, 565-575.	13.5	1,489
196	Identification of point mutations in Turkish DMD/BMD families using multiplex-single stranded conformation analysis (SSCA). <i>European Journal of Human Genetics</i> , 1999, 7, 765-770.	1.4	7
197	17 β -Hydroxysteroid Dehydrogenase-3 Deficiency: Diagnosis, Phenotypic Variability, Population Genetics, and Worldwide Distribution of Ancient and de Novo Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 4713-4721.	1.8	136
198	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

#	ARTICLE	IF	CITATIONS
199	An Insulin Receptor Mutant (Asp707→ Ala), Involved in Leprechaunism, Is Processed and Transported to the Cell Surface but Unable to Bind Insulin. <i>Journal of Biological Chemistry</i> , 1996, 271, 18719-18724.	1.6	21
200	Unusual molecular findings in autosomal recessive spinal muscular atrophy.. <i>Journal of Medical Genetics</i> , 1996, 33, 469-474.	1.5	36