

Hİlya Kayserili

List of Publications by Year in descending order

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Version: 2024-02-01

200
papers

15,690
citations

19636

61
h-index

19726

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. <i>Cell</i> , 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). <i>Cell</i> , 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. <i>American Journal of Human Genetics</i> , 2002, 71, 1033-1043. | 2.6 | 636 |
| 4 | Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. <i>Science</i> , 2014, 343, 506-511. | 6.0 | 466 |
| 5 | 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 |
| 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. <i>Nature Genetics</i> , 2005, 37, 468-470. | 9.4 | 334 |
| 7 | 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 |
| 8 | TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. <i>Nature Genetics</i> , 2011, 43, 189-196. | 9.4 | 326 |
| 9 | 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 |
| 10 | 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 |
| 11 | 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 |
| 12 | Mutations in WNT1 Cause Different Forms of Bone Fragility. <i>American Journal of Human Genetics</i> , 2013, 92, 565-574. | 2.6 | 240 |
| 13 | Responsible implementation of expanded carrier screening. <i>European Journal of Human Genetics</i> , 2016, 24, e1-e12. | 1.4 | 240 |
| 14 | CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. <i>Cell</i> , 2014, 157, 651-663. | 13.5 | 228 |
| 15 | Mutations in PYCR1 cause cutis laxa with progeroid features. <i>Nature Genetics</i> , 2009, 41, 1016-1021. | 9.4 | 211 |
| 16 | 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 |
| 17 | CEP152 is a genome maintenance protein disrupted in Seckel syndrome. <i>Nature Genetics</i> , 2011, 43, 23-26. | 9.4 | 201 |
| 18 | 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 |

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|----|---|------|-----------|
| 19 | Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. <i>Nature Genetics</i> , 2012, 44, 581-585. | 9.4 | 191 |
| 20 | Mutations in different components of FGF signaling in LADD syndrome. <i>Nature Genetics</i> , 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. <i>Human Molecular Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2003, 73, 957-966. | 2.6 | 174 |
| 23 | Harlequin Ichthyosis. <i>Archives of Dermatology</i> , 2011, 147, 681. | 1.7 | 145 |
| 24 | CantÃ³ Syndrome Is Caused by Mutations in ABCC9. <i>American Journal of Human Genetics</i> , 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. <i>Journal of Medical Genetics</i> , 2014, 51, 659-668. | 1.5 | 141 |
| 26 | RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. <i>Nature</i> , 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. <i>Journal of Medical Genetics</i> , 2008, 46, 192-197. | 1.5 | 138 |
| 28 | CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. <i>American Journal of Human Genetics</i> , 2007, 81, 104-113. | 2.6 | 137 |
| 29 | 17Î±-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 |
| 30 | 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 |
| 31 | PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. <i>JCI Insight</i> , 2016, 1, . | 2.3 | 134 |
| 32 | 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 |
| 33 | Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. <i>Human Molecular Genetics</i> , 2009, 18, 2149-2165. | 1.4 | 115 |
| 34 | Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. <i>Human Mutation</i> , 2010, 31, 1142-1154. | 1.1 | 111 |
| 35 | Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. <i>Nature Genetics</i> , 2015, 47, 528-534. | 9.4 | 111 |
| 36 | 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 |

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|----|--|-----|-----------|
| 37 | 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 |
| 38 | DVL1 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 |
| 39 | The molecular mechanism underlying Roberts syndrome involves loss of ESCO2 acetyltransferase activity. <i>Human Molecular Genetics</i> , 2008, 17, 2172-2180. | 1.4 | 108 |
| 40 | 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 |
| 41 | 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 |
| 42 | Distinguishing the four genetic causes of jouberts syndrome-related disorders. <i>Annals of Neurology</i> , 2005, 57, 513-519. | 2.8 | 104 |
| 43 | ALX4 dysfunction disrupts craniofacial and epidermal development. <i>Human Molecular Genetics</i> , 2009, 18, 4357-4366. | 1.4 | 103 |
| 44 | Spectrum of HSPG2 (Perlecan) mutations in patients with Schwartz-Jampel syndrome. <i>Human Mutation</i> , 2006, 27, 1082-1091. | 1.1 | 98 |
| 45 | 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 |
| 46 | Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 905-914. | 2.6 | 90 |
| 47 | 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 |
| 48 | 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 |
| 49 | The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307. | 1.1 | 80 |
| 50 | 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 |
| 51 | 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 |
| 52 | 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 |
| 53 | De novo mutations in PLXND1 and REV3L cause Möbius syndrome. <i>Nature Communications</i> , 2015, 6, 7199. | 5.8 | 76 |
| 54 | Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. <i>Human Mutation</i> , 2014, 35, 76-85. | 1.1 | 74 |

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|----|--|------|-----------|
| 55 | Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. <i>Human Genetics</i> , 2012, 131, 1761-1773. | 1.8 | 73 |
| 56 | C5orf42 is the major gene responsible for OFD syndrome type VI. <i>Human Genetics</i> , 2014, 133, 367-377. | 1.8 | 71 |
| 57 | 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 |
| 58 | RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. <i>Journal of Clinical Investigation</i> , 2015, 125, 3585-3599. | 3.9 | 69 |
| 59 | Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. <i>Nature Genetics</i> , 2012, 44, 709-713. | 9.4 | 68 |
| 60 | Frequency of renal malformations in Turner syndrome: analysis of 82 Turkish children. <i>Pediatric Nephrology</i> , 2000, 14, 1111-1114. | 0.9 | 67 |
| 61 | 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 |
| 62 | 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 |
| 63 | 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 |
| 64 | 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 |
| 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. <i>Journal of Medical Genetics</i> , 2010, 47, 30-37. | 1.5 | 65 |
| 66 | 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 |
| 67 | 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 |
| 68 | 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 |
| 69 | 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 |
| 70 | Mutations in ACTR1 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 |
| 71 | 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 |
| 72 | <i>OTX2</i> mutations contribute to the otocephaly-dysgnathia complex. <i>Journal of Medical Genetics</i> , 2012, 49, 373-379. | 1.5 | 58 |

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|----|--|-----|-----------|
| 73 | 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 |
| 74 | Genotype-phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 352-361. | 0.5 | 57 |
| 75 | De novo <i>WNT5A</i> -associated autosomal dominant Robinow syndrome suggests specificity of genotype and phenotype. <i>Clinical Genetics</i> , 2015, 87, 34-41. | 1.0 | 56 |
| 76 | Mutations in <i>CDK5</i> and <i>RAP2</i> cause Seckel syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 467-480. | 0.6 | 55 |
| 77 | 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 |
| 78 | <i>TCTEX1D2</i> mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. <i>Nature Communications</i> , 2015, 6, 7074. | 5.8 | 51 |
| 79 | <i>GLI1</i> inactivation is associated with developmental phenotypes overlapping with Ellis-van Creveld syndrome. <i>Human Molecular Genetics</i> , 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 <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 |
| 82 | Evaluation of Clinical Manifestations in Patients with Severe Lymphedema with and without <i>CCBE1</i> Mutations. <i>Molecular Syndromology</i> , 2013, 4, 107-113. | 0.3 | 45 |
| 83 | Whole-Exome Sequencing Identifies Novel Variants for Tooth Agenesis. <i>Journal of Dental Research</i> , 2018, 97, 49-59. | 2.5 | 44 |
| 84 | Cohesin complex-associated holoprosencephaly. <i>Brain</i> , 2019, 142, 2631-2643. | 3.7 | 43 |
| 85 | Identification of loss-of-function mutations of <i>SLC35D1</i> 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 |
| 86 | A Novel Semiquantitative Polymerase Chain Reaction/Enzyme Digestion-Based Method for Detection of Large Scale Deletions/Conversions of the <i>CYP21</i> 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 |
| 87 | Down Syndrome Diagnosis Based on Gabor Wavelet Transform. <i>Journal of Medical Systems</i> , 2012, 36, 3205-3213. | 2.2 | 39 |
| 88 | A novel <i>ICK</i> mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. <i>Cilia</i> , 2016, 5, 8. | 1.8 | 37 |
| 89 | Unusual molecular findings in autosomal recessive spinal muscular atrophy.. <i>Journal of Medical Genetics</i> , 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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 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. <i>American Journal of Human Genetics</i> , 2017, 101, 391-403. | 2.6 | 35 |
| 92 | Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. <i>PLoS Genetics</i> , 2017, 13, e1006683. | 1.5 | 35 |
| 93 | Variants in members of the cadherin-catenin complex, CDH1 and CTNND1, cause blepharochelodonic syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 210-219. | 1.4 | 34 |
| 94 | 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 |
| 95 | Oral manifestations of 17 patients affected with mucopolysaccharidosis type VI. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 263-268. | 1.7 | 32 |
| 96 | DOCK6 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 |
| 97 | 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 |
| 98 | Teratogenicity of Antiepileptic Drugs. <i>Clinical Psychopharmacology and Neuroscience</i> , 2017, 15, 19-27. | 0.9 | 30 |
| 99 | Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. <i>Nature Communications</i> , 2020, 11, 4589. | 5.8 | 30 |
| 100 | Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. <i>Neuron</i> , 2020, 107, 82-94.e6. | 3.8 | 30 |
| 101 | Mild nasal malformations and parietal foramina caused by homozygous ALX4 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 236-244. | 0.7 | 29 |
| 102 | Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by PAPS2 Mutations. <i>Human Mutation</i> , 2013, 34, 1381-1386. | 1.1 | 29 |
| 103 | A mutation in the signal sequence of LRP5 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 |
| 104 | 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 |
| 105 | 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 |
| 106 | MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctive cerebellar, ocular, craniofacial and genital features (COFG). <i>Journal of Medical Genetics</i> , 2019, 56, 100-107. | 1.9 | 25 |
| 107 | A loss-of-function NUA2 mutation in humans causes anencephaly due to impaired Hippo-YAP signaling. <i>Journal of Experimental Medicine</i> , 2020, 217, . | 4.2 | 25 |
| 108 | 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 |

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|-----|--|-----|-----------|
| 109 | Candidate locus analysis for PHACE syndrome. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Human Genetics</i> , 2018, 137, 689-703. | 1.8 | 24 |
| 111 | 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 |
| 112 | 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 |
| 113 | Seckel syndrome: report of a case. <i>Journal of Clinical Pediatric Dentistry</i> , 2002, 26, 305-309. | 0.5 | 22 |
| 114 | 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 |
| 115 | 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 |
| 116 | Enamel-Renal-Gingival syndrome, hypodontia, and a novel <i>FAM20A</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2124-2128. | 0.7 | 21 |
| 117 | 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 |
| 118 | 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 |
| 119 | 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 |
| 120 | Clinical manifestations of 17 patients affected with mucopolysaccharidosis type VI and eight novel <i>ARSB</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Journal of Inherited Metabolic Disease</i> , 2003, 26, 17-23. | 1.7 | 19 |
| 122 | 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 |
| 123 | Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. <i>Human Mutation</i> , 2017, 38, 524-531. | 1.1 | 18 |
| 124 | Pallister-Killian syndrome: clinical, cytogenetic and molecular findings in 15 cases. <i>Molecular Cytogenetics</i> , 2018, 11, 45. | 0.4 | 18 |
| 125 | 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 |
| 126 | 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 |

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|-----|--|-----|-----------|
| 127 | Skull defects, alopecia, hypertelorism, and notched alae nasi caused by homozygous <i>ALX4</i> 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 <i>IRX5</i> 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 Human-specific Neuroepithelial Tissue for Modeling Gene and Drug-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 years' experience in Istanbul. Acta Cardiologica, 2012, 67, 649-655. | 0.3 | 12 |
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