# HÏ‹lya Kayserili

List of Publications by Year
in descending order

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|---|---|---|---|
| 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 | Expanding the spectrum of syndromic PPP2R3Câ€related <scp>XY</scp> gonadal dysgenesis to <scp>XX</scp> gonadal dysgenesis. Clinical Genetics, 2022, 101, 221-232. | 1.0 | 3 |
| 2 | Functional loss of ubiquitinâ€specific protease 14 may lead to a novel distal arthrogryposis phenotype. Clinical Genetics, 2022, 101, 421-428. | 1.0 | 1 |
| 3 | A New Family with a Novel <b><i>OTUD6B</i></b> Mutation: Practicing Whole Exome Sequencing for Antenatal Diagnosis of Tetralogy of Fallot. Molecular Syndromology, 2022, 13, 206-211. | 0.3 | 2 |
| 4 | Biallelic TERT variant leads to Hoyeraalâ€“Hreidarsson syndrome with additional dyskeratosis congenita findings. American Journal of Medical Genetics, Part A, 2022, 188, 1226-1232. | 0.7 | 2 |
| 5 | Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918. | 1.1 | 8 |
| 6 | Evaluation of growth, puberty, osteoporosis, and the response to longâ€term bisphosphonate therapy in four patients with osteoporosisâ€pseudoglioma syndrome. American Journal of Medical Genetics, Part A, 2022, , . | 0.7 | 2 |
| 7 | Human model of IRX5 mutations reveals key role for this transcription factor in ventricular conduction. Cardiovascular Research, 2021, 117, 2092-2107. | 1.8 | 17 |
| 8 | A Micropatterned Humanâ€Specific Neuroepithelial Tissue for Modeling Gene and Drugâ€Induced Neurodevelopmental Defects. Advanced Science, 2021, 8, 2001100. | 5.6 | 13 |
| 9 | 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 |
| 10 | 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 |
| 11 | 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 |
| 12 | ESHG PPPC Comments on postmortem use of genetic data for research purposes. European Journal of Human Genetics, 2020, 28, 144-146. | 1.4 | 3 |
| 13 | A rare cause of chronic hyponatremia in an infant: Questions. Pediatric Nephrology, 2020, 35, 241-242. | 0.9 | 0 |
| 14 | Heterozygous pathogenic variants inGLI1are a common finding in isolated postaxial polydactyly A/B. Human Mutation, 2020, 41, 265-276. | 1.1 | 6 |
| 15 | 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 |
| 16 | Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589. | 5.8 | 30 |
| 17 | Characteristic dental pattern with hypodontia and short roots in Fraser syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1681-1689. | 0.7 | 7 |
| 18 | Clinical exome sequencing in neuromuscular diseases: an experience from Turkey. Neurological Sciences, 2020, 41, 2157-2164. | 0.9 | 10 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 19 | Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. Neuron, 2020, 107, 82-94.e6. | 3.8 | 30 |
| 20 | A novel shoulder disability staging system for scapulothoracic arthrodesis in patients with facioscapulohumeral dystrophy. Orthopaedics and Traumatology: Surgery and Research, 2020, 106, 701-707. | 0.9 | 6 |
| 21 | Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643. | 3.7 | 43 |
| 22 | European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. European Journal of Human Genetics, 2019, 27, 1763-1773. | 1.4 | 78 |
| 23 | 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 |
| 24 | Turkish Ectodermal Dysplasia Cohort: From Phenotype to Genotype in 17 Families. Cytogenetic and Genome Research, 2019, 157, 189-196. | 0.6 | 7 |
| 25 | Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling. Neuromuscular Disorders, 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. American Journal of Human Genetics, 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. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 1403-1406. | 0.4 | 3 |
| 28 | Terminal osseous dysplasia with pigmentary defects (TODPD) in a Turkish girl with new skin findings. American Journal of Medical Genetics, Part A, 2019, 179, 123-129. | 0.7 | 4 |
| 29 | Sclerosing bone dysplasias with hallmarks of dysosteosclerosis in four patients carrying mutations in SLC29A3 and TCIRG1. Bone, 2019, 120, 495-503. | 1.4 | 23 |
| 30 | The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307. | 1.1 | 80 |
| 31 | 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 |
| 32 | 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 21 | 1.3 | 25 |
| 33 | 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 |
| 34 | CLINICAL CLASSIFICATION OF RADIAL RAY DEFECTS AND RESEARCH INTO ETIOPATHOGENESIS. European Oral Research, 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. Ä°stanbul TÄ±p FakÃ¼ltesi Dergisi, 2019, 82, 5-11. | 0.1 | 0 |
| 36 | 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 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | Whole-Exome Sequencing Identifies Novel Variants for Tooth Agenesis. Journal of Dental Research, 2018, 97, 49-59. | 2.5 | 44 |
| 40 | The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. American Journal of Medical Genetics, Part A, 2018, 176, 2740-2750. | 0.7 | 6 |
| 41 | Pallister-Killian syndrome: clinical, cytogenetic and molecular findings in 15 cases. Molecular Cytogenetics, 2018, 11, 45. | 0.4 | 18 |
| 42 | RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. Nature, 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. Human Genetics, 2018, 137, 689-703. | 1.8 | 24 |
| 44 | 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 |
| 45 | 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 |
| 46 | A facioscapulohumeralis muscularis dystrophia kezelÃ©sÃ©nek multidiszciplinÃ¡ris megkÃ¶zelÃtÃ©se. Ideggyogyaszati Szemle, 2018, 71, 337-342. | 0.4 | 0 |
| 47 | Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. Human Mutation, 2017, 38, 524-531. | 1.1 | 18 |
| 48 | De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255. | 9.4 | 88 |
| 49 | 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 |
| 50 | 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 |
| 51 | 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 |
| 52 | GLI1 inactivation is associated with developmental phenotypes overlapping with Ellisâ€“van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571. | 1.4 | 50 |
| 53 | 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 |
| 54 | Loss-of-function mutations in Carboxypeptidase D cause a new syndrome with lymphedema and sensorineural hearing loss. Mechanisms of Development, 2017, 145, S32. | 1.7 | 0 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 55 | PYCR2 Protects from Neurodegeneration by Controlling Oligodendrocyte Maturation and Glycinemia through SHMT2. Mechanisms of Development, 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. American Journal of Medical Genetics, Part A, 2017, 173, 3136-3142. | 0.7 | 9 |
| 57 | Teratogenicity of Antiepileptic Drugs. Clinical Psychopharmacology and Neuroscience, 2017, 15, 19-27. | 0.9 | 30 |
| 58 | Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683. | 1.5 | 35 |
| 59 | â€œHomozygous, and compound heterozygous mutation in 3 Turkish family with Jervell and Lange-Nielsen syndrome: case reportsâ€• BMC Medical Genetics, 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. Nature Medicine, 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. World Journal of Radiology, 2017, 9, 365-370. | 0.5 | 3 |
| 62 | Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864. | 1.1 | 134 |
| 63 | Ungual squamous cell carcinoma in a patient with Mal de Meleda. JDDG - Journal of the German Society of Dermatology, 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. Journal of Investigative Dermatology, 2016, 136, S243. | 0.3 | 0 |
| 65 | Characteristic calcaneal ossification: an additional early radiographic finding in infants with fibrodysplasia ossificans progressiva. Pediatric Radiology, 2016, 46, 1568-1572. | 1.1 | 7 |
| 66 | 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 |
| 67 | Holtâ€“Oram syndrome because of the novel TBX5 mutation c.481A>C. Clinical Dysmorphology, 2016, 25, 192-194. | 0.1 | 1 |
| 68 | Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615. | 1.5 | 55 |
| 69 | ALX4 related parietal foramina mimicking encephalocele in prenatal period. Prenatal Diagnosis, 2016, 36, 591-593. | 1.1 | 4 |
| 70 | Microcephaly, dysmorphic features, corneal dystrophy, hairy nipples, underdeveloped labioscrotal folds, and small cerebellum in four patients. American Journal of Medical Genetics, Part A, 2016, 170, 1391-1399. | 0.7 | 5 |
| 71 | Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2759-2767. | 1.8 | 67 |
| 72 | Loss of Iroquois homeobox transcription factors 3 and 5 in osteoblasts disrupts cranial mineralization. Bone Reports, 2016, 5, 86-95. | 0.2 | 21 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 73 | A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8. | 1.8 | 37 |
| 74 | 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 |
| 75 | Responsible implementation of expanded carrier screening. European Journal of Human Genetics, 2016, 24, e1-e12. | 1.4 | 240 |
| 76 | PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, . | 2.3 | 134 |
| 77 | Prenatal ultrasonographic diagnosis of generalized arterial calcification of infancy. Journal of Clinical Ultrasound, 2015, 43, 50-54. | 0.4 | 10 |
| 78 | Mutations in  <scp>CDK</scp> 5 <scp>RAP</scp> 2  cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480. | 0.6 | 55 |
| 79 | Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 128. | 1.2 | 46 |
| 80 | Sclerosteosis (craniotubular hyperostosis-syndactyly) with complex hyperphalangy of the index finger. Pediatric Radiology, 2015, 45, 1239-1243. | 1.1 | 7 |
| 81 | De novo mutations in PLXND1 and REV3L cause MÃ¶bius syndrome. Nature Communications, 2015, 6, 7199. | 5.8 | 76 |
| 82 | A severe collodion phenotype in the newborn period associated with a homozygous missense mutation inALOX12B. British Journal of Dermatology, 2015, 173, 285-287. | 1.4 | 2 |
| 83 | 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 |
| 84 | 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 |
| 85 | Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025. | 13.5 | 1,725 |
| 86 | 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 |
| 87 | Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534. | 9.4 | 111 |
| 88 | TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 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. BMC Medical Genetics, 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. European Journal of Human Genetics, 2015, 23, 729-735. | 1.4 | 26 |

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|---|---|---|---|
| 91 | 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 |
| 92 | RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 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. Journal of Medical Genetics, 2014, 51, 659-668. | 1.5 | 141 |
| 94 | 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 |
| 95 | 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 |
| 96 | Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. Human Mutation, 2014, 35, 76-85. | 1.1 | 74 |
| 97 | Enamelâ€"Renalâ€"Gingival syndrome, hypodontia, and a novel FAM20A mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2124-2128. | 0.7 | 21 |
| 98 | Mild nasal clefting may be predictive for ALX4 heterozygotes. American Journal of Medical Genetics, Part A, 2014, 164, 2054-2058. | 0.7 | 9 |
| 99 | A patient with a de-novo deletion 3p25.3 and features overlapping with Rubinsteinâ€"Taybi syndrome. Clinical Dysmorphology, 2014, 23, 67-70. | 0.1 | 3 |
| 100 | Clinicogenetic Study of Turkish Patients With Syndromic Craniosynostosis and Literature Review. Pediatric Neurology, 2014, 50, 482-490. | 1.0 | 14 |
| 101 | C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377. | 1.8 | 71 |
| 102 | Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511. | 6.0 | 466 |
| 103 | CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663. | 13.5 | 228 |
| 104 | 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 |
| 105 | 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 |
| 106 | Oral manifestations of 17 patients affected with mucopolysaccharidosis type VI. Journal of Inherited Metabolic Disease, 2014, 37, 263-268. | 1.7 | 32 |
| 107 | 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 |
| 108 | Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914. | 2.6 | 90 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 109 | Evaluation of Clinical Manifestations in Patients with Severe Lymphedema with and without CCBE1 Mutations. Molecular Syndromology, 2013, 4, 107-113. | 0.3 | 45 |
| 110 | Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome. Human Genetics, 2013, 132, 885-898. | 1.8 | 77 |
| 111 | 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 |
| 112 | Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361. | 0.5 | 57 |
| 113 | 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 |
| 114 | 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 |
| 115 | Multiple synostoses syndrome inÂthree members of a family displaying a novel mutation inÂNOGGIN gene. Journal of Plastic, Reconstructive and Aesthetic Surgery, 2013, 66, e287-e289. | 0.5 | 3 |
| 116 | Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574. | 2.6 | 240 |
| 117 | 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 |
| 118 | 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 |
| 119 | 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 |
| 120 | Prenatal diagnosis of frontonasal dysplasia with anterior encephalocele. Journal of the Turkish German Gynecology Association, 2013, 14, 50-52. | 0.2 | 7 |
| 121 | Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused byPAPSS2Mutations. Human Mutation, 2013, 34, 1381-1386. | 1.1 | 29 |
| 122 | Genotypic and phenotypic analysis of 396 individuals with mutations inSonic Hedgehog. Journal of Medical Genetics, 2012, 49, 473-479. | 1.5 | 67 |
| 123 | A large duplication involving the IHH locus mimics acrocallosal syndrome. European Journal of Human Genetics, 2012, 20, 639-644. | 1.4 | 14 |
| 124 | Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585. | 9.4 | 191 |
| 125 | OTX2mutations contribute to the otocephaly-dysgnathia complex. Journal of Medical Genetics, 2012, 49, 373-379. | 1.5 | 58 |
| 126 | Cardiovascular abnormalities in Williams syndrome: 20 yearsâ€™ experience in Istanbul. Acta Cardiologica, 2012, 67, 649-655. | 0.3 | 12 |

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|---|---|---|---|
| 127 | Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773. | 1.8 | 73 |
| 128 | Down Syndrome Diagnosis Based on Gabor Wavelet Transform. Journal of Medical Systems, 2012, 36, 3205-3213. | 2.2 | 39 |
| 129 | Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713. | 9.4 | 68 |
| 130 | Mutations in BCKD-kinase Lead to a Potentially Treatable Form of Autism with Epilepsy. Science, 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. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2012, 114, e42-e46. | 0.2 | 8 |
| 132 | Candidate locus analysis for PHACE syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1363-1367. | 0.7 | 24 |
| 133 | Multiple supernumerary molars, anterior openbite, and large ear lobules in mucopolysaccharidosis type VI patient. American Journal of Medical Genetics, Part A, 2012, 158A, 1798-1800. | 0.7 | 5 |
| 134 | Mutations in RIPK4 Cause the Autosomal-Recessive Form of Popliteal Pterygium Syndrome. American Journal of Human Genetics, 2012, 90, 76-85. | 2.6 | 80 |
| 135 | CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101. | 2.6 | 141 |
| 136 | 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 |
| 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 | CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26. | 9.4 | 201 |
| 139 | TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196. | 9.4 | 326 |
| 140 | A novel homozygous COL11A2 deletion causes a Câ€terminal protein truncation with incomplete mRNA decay in a Turkish patient. American Journal of Medical Genetics, Part A, 2011, 155, 180-185. | 0.7 | 3 |
| 141 | Harlequin Ichthyosis. Archives of Dermatology, 2011, 147, 681. | 1.7 | 145 |
| 142 | Tumor spectrum in children with Noonan syndrome and SOS1 or RAF1 mutations. Genes Chromosomes and Cancer, 2010, 49, 242-252. | 1.5 | 57 |
| 143 | Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 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. Journal of Medical Genetics, 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. Journal of Medical Genetics, 2010, 47, 262-267. | 1.5 | 67 |
| 146 | LI-FRAUMENI SYNDROME IN A TURKISH FAMILY. Pediatric Hematology and Oncology, 2010, 27, 297-305. | 0.3 | 4 |
| 147 | P106. Congenital hypertelorism and osteopenia: A novel autosomal recessive disease. Differentiation, 2010, 80, S52. | 1.0 | 0 |
| 148 | ALX4 dysfunction disrupts craniofacial and epidermal development. Human Molecular Genetics, 2009, 18, 4357-4366. | 1.4 | 103 |
| 149 | Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 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. Journal of Medical Genetics, 2009, 46, 562-568. | 1.5 | 41 |
| 151 | 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 |
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