# Ranad Shaheen

List of Publications by Year
in descending order

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Version: 2024-02-01

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|---|---|---|---|
| 151 papers | 8,462 citations | 29994 54 h-index | 56606 83 g-index |
| 159 all docs | 159 docs citations | 159 times ranked | 13248 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Reports, 2015, 10, 148-161. | 2.9 | 375 |
| 2 | Loss-of-function variant in DNASE1L3 causes a familial form of systemic lupus erythematosus. Nature Genetics, 2011, 43, 1186-1188. | 9.4 | 366 |
| 3 | The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45. | 9.4 | 251 |
| 4 | Identification and Partial Characterization of the Nonribosomal Peptide Synthetase Gene Responsible for Cereulide Production in Emetic Bacillus cereus. Applied and Environmental Microbiology, 2005, 71, 105-113. | 1.4 | 249 |
| 5 | Autozygome-guided exome sequencing in retinal dystrophy patients reveals pathogenetic mutations and novel candidate disease genes. Genome Research, 2013, 23, 236-247. | 2.4 | 226 |
| 6 | Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. American Journal of Human Genetics, 2011, 88, 536-547. | 2.6 | 196 |
| 7 | Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. Molecular Psychiatry, 2017, 22, 615-624. | 4.1 | 187 |
| 8 | Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes. Journal of Medical Genetics, 2012, 49, 234-241. | 1.5 | 164 |
| 9 | Novel CENPJ mutation causes Seckel syndrome. Journal of Medical Genetics, 2010, 47, 411-414. | 1.5 | 149 |
| 10 | Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117. | 2.6 | 147 |
| 11 | Genomic analysis of primordial dwarfism reveals novel disease genes. Genome Research, 2014, 24, 291-299. | 2.4 | 142 |
| 12 | Emetic toxin-producing strains of Bacillus cereus show distinct characteristics within the Bacillus cereus group. International Journal of Food Microbiology, 2006, 109, 132-138. | 2.1 | 141 |
| 13 | Mutation in WDR4 impairs tRNA m7G46 methylation and causes a distinct form of microcephalic primordial dwarfism. Genome Biology, 2015, 16, 210. | 3.8 | 132 |
| 14 | Study of autosomal recessive osteogenesis imperfecta in Arabia reveals a novel locus defined byTMEM38Bmutation. Journal of Medical Genetics, 2012, 49, 630-635. | 1.5 | 124 |
| 15 | Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429. | 1.8 | 122 |
| 16 | Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242. | 3.8 | 118 |
| 17 | Recessive Mutations in DOCK6, Encoding the Guanidine Nucleotide Exchange Factor DOCK6, Lead to Abnormal Actin Cytoskeleton Organization and Adams-Oliver Syndrome. American Journal of Human Genetics, 2011, 89, 328-333. | 2.6 | 115 |
| 18 | Mutations in EOGT Confirm the Genetic Heterogeneity of Autosomal-Recessive Adams-Oliver Syndrome. American Journal of Human Genetics, 2013, 92, 598-604. | 2.6 | 114 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Persistence strategies of Bacillus cereus spores isolated from dairy silo tanks. Food Microbiology, 2010, 27, 347-355. | 2.1 | 113 |
| 20 | A homozygous truncating mutation in PUS3 expands the role of tRNA modification in normal cognition. Human Genetics, 2016, 135, 707-713. | 1.8 | 112 |
| 21 | In search of triallelism in Bardetâ€“Biedl syndrome. European Journal of Human Genetics, 2012, 20, 420-427. | 1.4 | 111 |
| 22 | The genetic landscape of familial congenital hydrocephalus. Annals of Neurology, 2017, 81, 890-897. | 2.8 | 108 |
| 23 | Neu-Laxova Syndrome, an Inborn Error of Serine Metabolism, Is Caused by Mutations in PHGDH. American Journal of Human Genetics, 2014, 94, 898-904. | 2.6 | 93 |
| 24 | Mutation inADAT3, encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus. Journal of Medical Genetics, 2013, 50, 425-430. | 1.5 | 91 |
| 25 | Identification of embryonic lethal genes in humans by autozygosity mapping and exome sequencing in consanguineous families. Genome Biology, 2015, 16, 116. | 3.8 | 91 |
| 26 | Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540. | 1.8 | 89 |
| 27 | Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. Genetics in Medicine, 2016, 18, 554-562. | 1.1 | 89 |
| 28 | Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328. | 3.9 | 89 |
| 29 | Sperm bioassay for rapid detection of cereulide-producing Bacillus cereus in food and related environments. International Journal of Food Microbiology, 2004, 94, 175-183. | 2.1 | 88 |
| 30 | FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. American Journal of Human Genetics, 2009, 85, 414-418. | 2.6 | 86 |
| 31 | Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552. | 1.1 | 85 |
| 32 | Molecular autopsy in maternalâ€“fetal medicine. Genetics in Medicine, 2018, 20, 420-427. | 1.1 | 84 |
| 33 | Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. Genetics in Medicine, 2016, 18, 1244-1249. | 1.1 | 82 |
| 34 | Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549. | 9.4 | 81 |
| 35 | Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742. | 1.1 | 81 |
| 36 | Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79. | 2.6 | 77 |

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|---|---|---|---|
| 37 | Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. Genetics in Medicine, 2017, 19, 1144-1150. | 1.1 | 77 |
| 38 | Clinical and molecular characterisation of Bardet-Biedl syndrome in consanguineous populations: the power of homozygosity mapping. Journal of Medical Genetics, 2010, 47, 236-241. | 1.5 | 76 |
| 39 | Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36. | 2.6 | 74 |
| 40 | Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. Human Genetics, 2017, 136, 205-225. | 1.8 | 73 |
| 41 | A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. Human Mutation, 2011, 32, 573-578. | 1.1 | 72 |
| 42 | POC1A Truncation Mutation Causes a Ciliopathy in Humans Characterized by Primordial Dwarfism. American Journal of Human Genetics, 2012, 91, 330-336. | 2.6 | 70 |
| 43 | A founder CEP120 mutation in Jeune asphyxiating thoracic dystrophy expands the role of centriolar proteins in skeletal ciliopathies. Human Molecular Genetics, 2015, 24, 1410-1419. | 1.4 | 70 |
| 44 | ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. American Journal of Human Genetics, 2018, 103, 612-620. | 2.6 | 70 |
| 45 | Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945. | 3.7 | 70 |
| 46 | Occurrence of emetic toxin producing Bacillus cereus in the dairy production chain. International Dairy Journal, 2006, 16, 740-749. | 1.5 | 69 |
| 47 | The morbid genome of ciliopathies: an update. Genetics in Medicine, 2020, 22, 1051-1060. | 1.1 | 68 |
| 48 | Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. Human Genetics, 2016, 135, 1295-1298. | 1.8 | 65 |
| 49 | Mutations in FKBP10 cause both Bruck syndrome and isolated osteogenesis imperfecta in humans. American Journal of Medical Genetics, Part A, 2011, 155, 1448-1452. | 0.7 | 63 |
| 50 | Mutations of KIF14 cause primary microcephaly by impairing cytokinesis. Annals of Neurology, 2017, 82, 562-577. | 2.8 | 62 |
| 51 | Molecular characterization of retinitis pigmentosa in Saudi Arabia. Molecular Vision, 2009, 15, 2464-9. | 1.1 | 61 |
| 52 | Potential of selected infant food formulas for production of Bacillus cereus emetic toxin, cereulide. International Journal of Food Microbiology, 2006, 107, 287-294. | 2.1 | 60 |
| 53 | CC2D1A Regulates Human Intellectual and Social Function as well as NF-ÎºB Signaling Homeostasis. Cell Reports, 2014, 8, 647-655. | 2.9 | 60 |
| 54 | Increasing the sensitivity of clinical exome sequencing through improved filtration strategy. Genetics in Medicine, 2017, 19, 593-598. | 1.1 | 59 |

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|---|---|---|---|
| 55 | Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145. | 3.8 | 59 |
| 56 | A genomics approach to females with infertility and recurrent pregnancy loss. Human Genetics, 2020, 139, 605-613. | 1.8 | 59 |
| 57 | Genomic analysis of Meckelâ€“Gruber syndrome in Arabs reveals marked genetic heterogeneity and novel candidate genes. European Journal of Human Genetics, 2013, 21, 762-768. | 1.4 | 56 |
| 58 | Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. Genetics in Medicine, 2016, 18, 686-695. | 1.1 | 55 |
| 59 | PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. Human Genetics, 2019, 138, 231-239. | 1.8 | 53 |
| 60 | Mutations in SMG9, Encoding an Essential Component of Nonsense-Mediated Decay Machinery, Cause a Multiple Congenital Anomaly Syndrome in Humans and Mice. American Journal of Human Genetics, 2016, 98, 643-652. | 2.6 | 51 |
| 61 | Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217. | 2.6 | 51 |
| 62 | Allelic heterogeneity in inbred populations: The Saudi experience with AlstrÃ¶m syndrome as an illustrative example. American Journal of Medical Genetics, Part A, 2009, 149A, 662-665. | 0.7 | 48 |
| 63 | FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. American Journal of Human Genetics, 2010, 87, 306-307. | 2.6 | 47 |
| 64 | Cereulide-producing strains of Bacillus cereus show diversity. Archives of Microbiology, 2005, 184, 141-151. | 1.0 | 46 |
| 65 | Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616. | 1.1 | 46 |
| 66 | KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132. | 2.6 | 46 |
| 67 | Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. American Journal of Human Genetics, 2013, 93, 555-560. | 2.6 | 45 |
| 68 | WNT1 mutation with recessive osteogenesis imperfecta and profound neurological phenotype. Journal of Medical Genetics, 2013, 50, 491-492. | 1.5 | 45 |
| 69 | On the phenotypic spectrum of serine biosynthesis defects. Journal of Inherited Metabolic Disease, 2016, 39, 373-381. | 1.7 | 45 |
| 70 | Exome-based caseâ€“control association study using extreme phenotype design reveals novel candidates with protective effect in diabetic retinopathy. Human Genetics, 2016, 135, 193-200. | 1.8 | 45 |
| 71 | Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439. | 3.9 | 43 |
| 72 | Identification of a novel MKS locus defined byTMEM107mutation. Human Molecular Genetics, 2015, 24, 5211-5218. | 1.4 | 42 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659. | 1.1 | 41 |
| 74 | Molecular Characterization of Newborn Glaucoma Including a Distinct Aniridic Phenotype. Ophthalmic Genetics, 2011, 32, 138-142. | 0.5 | 40 |
| 75 | A novel syndrome of hypohidrosis and intellectual disability is linked to COG6 deficiency. Journal of Medical Genetics, 2013, 50, 431-436. | 1.5 | 40 |
| 76 | Cell-Intrinsic Adaptation Arising from Chronic Ablation of a Key Rho GTPase Regulator. Developmental Cell, 2016, 39, 28-43. | 3.1 | 40 |
| 77 | A null mutation in MICU2 causes abnormal mitochondrial calcium homeostasis and a severe neurodevelopmental disorder. Brain, 2017, 140, 2806-2813. | 3.7 | 38 |
| 78 | Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. Clinical Genetics, 2018, 93, 1210-1222. | 1.0 | 38 |
| 79 | PLAA Mutations Cause a Lethal Infantile Epileptic Encephalopathy by Disrupting Ubiquitin-Mediated Endolysosomal Degradation of Synaptic Proteins. American Journal of Human Genetics, 2017, 100, 706-724. | 2.6 | 37 |
| 80 | SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133. | 2.6 | 37 |
| 81 | Mutation inPLK4, encoding a master regulator of centriole formation, defines a novel locus for primordial dwarfism. Journal of Medical Genetics, 2014, 51, 814-816. | 1.5 | 35 |
| 82 | FBXO32, encoding a member of the SCF complex, is mutated in dilated cardiomyopathy. Genome Biology, 2016, 17, 2. | 3.8 | 35 |
| 83 | Mutations inTMEM231cause Meckelâ€“Gruber syndrome. Journal of Medical Genetics, 2013, 50, 160-162. | 1.5 | 34 |
| 84 | Matching Two Independent Cohorts ValidatesDPH1as a Gene Responsible for Autosomal Recessive Intellectual Disability with Short Stature, Craniofacial, and Ectodermal Anomalies. Human Mutation, 2015, 36, 1015-1019. | 1.1 | 32 |
| 85 | ADAT3â€related intellectual disability: Further delineation of the phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 1142-1147. | 0.7 | 32 |
| 86 | Hyperekplexia, microcephaly and simplified gyral pattern caused by novel ASNS mutations, case report. BMC Neurology, 2016, 16, 105. | 0.8 | 32 |
| 87 | Phenome-based approach identifies RIC1-linked Mendelian syndrome through zebrafish models, biobank associations and clinical studies. Nature Medicine, 2020, 26, 98-109. | 15.2 | 32 |
| 88 | A truncating mutation in B3GNT1 causes severe Walkerâ€“Warburg syndrome. Neurogenetics, 2013, 14, 243-245. | 0.7 | 31 |
| 89 | MDH1 deficiency is a metabolic disorder of the malateâ€“aspartate shuttle associated with early onset severe encephalopathy. Human Genetics, 2019, 138, 1247-1257. | 1.8 | 31 |
| 90 | Positional mapping ofPRKD1,NRP1andPRDM1as novel candidate disease genes in truncus arteriosus. Journal of Medical Genetics, 2015, 52, 322-329. | 1.5 | 30 |

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|---|---|---|---|
| 91 | Mutations in CIT, encoding citron rho-interacting serine/threonine kinase, cause severe primary microcephaly in humans. Human Genetics, 2016, 135, 1191-1197. | 1.8 | 30 |
| 92 | Identification of three novel <scp>ECEL1</scp> mutations in three families with distal arthrogryposis type <scp>5D</scp>. Clinical Genetics, 2014, 85, 568-572. | 1.0 | 29 |
| 93 | Congenital glaucoma and CYP1B1: an old story revisited. Human Genetics, 2019, 138, 1043-1049. | 1.8 | 29 |
| 94 | Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. Hepatology, 2020, 71, 2067-2079. | 3.6 | 28 |
| 95 | Environment driven cereulide production by emetic strains of Bacillus cereus. International Journal of Food Microbiology, 2008, 127, 60-67. | 2.1 | 27 |
| 96 | 3M Syndrome: An Easily Recognizable yet Underdiagnosed Cause of Proportionate Short Stature. Journal of Pediatrics, 2012, 161, 139-145.e1. | 0.9 | 27 |
| 97 | Biallelic variants in CTU2 cause DREAMâ€PL syndrome and impair thiolation of tRNA wobble U34. Human Mutation, 2019, 40, 2108-2120. | 1.1 | 25 |
| 98 | Bi-allelic Mutations in FAM149B1 Cause Abnormal Primary Cilium and a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2019, 104, 731-737. | 2.6 | 23 |
| 99 | The ECEL1-related strabismus phenotype is consistent with congenital cranial dysinnervation disorder. Journal of AAPOS, 2014, 18, 362-367. | 0.2 | 22 |
| 100 | Recessive, Deleterious Variants in SMG8 Expand the Role of Nonsense-Mediated Decay in Developmental Disorders in Humans. American Journal of Human Genetics, 2020, 107, 1178-1185. | 2.6 | 20 |
| 101 | The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725. | 1.1 | 19 |
| 102 | A novel mechanism for variable phenotypic expressivity in Mendelian diseases uncovered by an AU-rich element (ARE)-creating mutation. Genome Biology, 2017, 18, 144. | 3.8 | 19 |
| 103 | Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114. | 2.6 | 19 |
| 104 | THUMPD1 bi-allelic variants cause loss of tRNA acetylation and a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 587-600. | 2.6 | 19 |
| 105 | Clinical, biochemical and molecular characterization of peroxisomal diseases in Arabs. Clinical Genetics, 2011, 79, 60-70. | 1.0 | 18 |
| 106 | Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776. | 1.1 | 18 |
| 107 | Revisiting disease genes based on whole-exome sequencing in consanguineous populations. Human Genetics, 2015, 134, 1029-1034. | 1.8 | 17 |
| 108 | The syndrome dysmorphic facies, renal agenesis, ambiguous genitalia, microcephaly, polydactyly and lissencephaly (DREAMâ€PL): Report of two additional patients. American Journal of Medical Genetics, Part A, 2016, 170, 3222-3226. | 0.7 | 17 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Epileptic encephalopathy with continuous spike-and-wave during sleep maps to a homozygous truncating mutation in AMPA receptor component FRRS1L. Clinical Genetics, 2016, 90, 282-283. | 1.0 | 16 |
| 110 | Warsaw breakage syndrome: Further clinical and genetic delineation. American Journal of Medical Genetics, Part A, 2018, 176, 2404-2418. | 0.7 | 16 |
| 111 | An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. Human Genetics, 2020, 139, 1273-1283. | 1.8 | 16 |
| 112 | Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellarâ€vermis hypoplasia. American Journal of Medical Genetics, Part A, 2019, 179, 1053-1057. | 0.7 | 15 |
| 113 | GZF1 Mutations Expand the Genetic Heterogeneity of Larsen Syndrome. American Journal of Human Genetics, 2017, 100, 831-836. | 2.6 | 14 |
| 114 | Zellweger syndrome caused by PEX13 deficiency: Report of two novel mutations. American Journal of Medical Genetics, Part A, 2009, 149A, 1219-1223. | 0.7 | 13 |
| 115 | T (brachyury) is linked to a Mendelian form of neural tube defects in humans. Human Genetics, 2015, 134, 1139-1141. | 1.8 | 13 |
| 116 | Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. Orphanet Journal of Rare Diseases, 2016, 11, 52. | 1.2 | 13 |
| 117 | YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. Brain, 2020, 143, 2911-2928. | 3.7 | 13 |
| 118 | Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Frontiers in Genetics, 2020, 11, 580484. | 1.1 | 13 |
| 119 | Lethal variants in humans: lessons learned from a large molecular autopsy cohort. Genome Medicine, 2021, 13, 161. | 3.6 | 13 |
| 120 | Congenital disorder of glycosylation IIa: The trouble with diagnosing a dysmorphic inborn error of metabolism. American Journal of Medical Genetics, Part A, 2012, 158A, 245-246. | 0.7 | 12 |
| 121 | Mutations in known disease genes account for the majority of autosomal recessive retinal dystrophies. Clinical Genetics, 2018, 94, 554-563. | 1.0 | 12 |
| 122 | The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319. | 1.0 | 12 |
| 123 | The natural history of infantile neuroaxonal dystrophy. Orphanet Journal of Rare Diseases, 2020, 15, 109. | 1.2 | 11 |
| 124 | Missense NAA20 variantsimpairing the NatB protein N-terminal acetyltransferase cause autosomal recessivedevelopmental delay, intellectual disability, and microcephaly. Genetics in Medicine, 2021, 23, 2213-2218. | 1.1 | 11 |
| 125 | Autozygome maps dispensable DNA and reveals potential selective bias against nullizygosity. Genetics in Medicine, 2012, 14, 515-519. | 1.1 | 10 |
| 126 | Intrafamilial clinical heterogeneity of CSPP1â€related ciliopathy. American Journal of Medical Genetics, Part A, 2015, 167, 2478-2480. | 0.7 | 9 |

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|---|---|---|---|
| 127 | Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419. | 1.1 | 9 |
| 128 | Gonadal mosaicism for ACTA1 gene masquerading as autosomal recessive nemaline myopathy. American Journal of Medical Genetics, Part A, 2016, 170, 2219-2221. | 0.7 | 8 |
| 129 | Novel copy number variants and major limb reduction malformation: Report of three cases. American Journal of Medical Genetics, Part A, 2016, 170, 1245-1250. | 0.7 | 8 |
| 130 | Expanding the clinical and phenotypic heterogeneity associated with biallelic variants in ACO2. Annals of Clinical and Translational Neurology, 2020, 7, 1013-1028. | 1.7 | 8 |
| 131 | Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. Genetics in Medicine, 2020, 22, 2071-2080. | 1.1 | 7 |
| 132 | Survey of disorders of sex development in a large cohort of patients with diverse Mendelian phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 2789-2800. | 0.7 | 7 |
| 133 | GWAS signals revisited using human knockouts. Genetics in Medicine, 2018, 20, 64-68. | 1.1 | 6 |
| 134 | <scp>PLXNA2</scp> as a candidate gene in patients with intellectual disability. American Journal of Medical Genetics, Part A, 2021, 185, 3859-3865. | 0.7 | 6 |
| 135 | Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. JCI Insight, 2022, 7, . | 2.3 | 6 |
| 136 | Vanishing white matter disease caused by EIF2B2 mutation with the presentation of an adrenoleukodystrophy phenotype. Gene, 2012, 496, 141-143. | 1.0 | 5 |
| 137 | Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546. | 0.7 | 5 |
| 138 | Expanding the allelic disorders linked to TCTN1 to include Varadi syndrome (Orofaciodigital) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 | 0.7 | 4 |
| 139 | Further delineation of <scp>MYO18B</scp>â€related autosomal recessive <scp>Klippelâ€Feil</scp> syndrome with myopathy and facial dysmorphism. American Journal of Medical Genetics, Part A, 2021, 185, 370-376. | 0.7 | 4 |
| 140 | FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. American Journal of Human Genetics, 2010, 87, 571. | 2.6 | 3 |
| 141 | Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. American Journal of Human Genetics, 2011, 88, 677. | 2.6 | 3 |
| 142 | Further delineation of <scp>SMG9</scp>â€related heart and brain malformation syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1624-1630. | 0.7 | 3 |
| 143 | Transcriptome of CD8+ tumor-infiltrating T cells: a link between diabetes and colorectal cancer. Cancer Immunology, Immunotherapy, 2021, 70, 2625-2638. | 2.0 | 3 |
| 144 | Mutations in phospholipase C eta-1 (PLCH1) are associated with holoprosencephaly. Journal of Medical Genetics, 2022, 59, 358-365. | 1.5 | 3 |

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|---|---|---|---|
| 145 | Genetic testing results of children suspected to have Stickler syndrome type collagenopathy after ocular examination. Molecular Genetics & Genomic Medicine, 2021, 9, e1628. | 0.6 | 3 |
| 146 | Variable phenotypic expression of COG6 mutations. Journal of Medical Genetics, 2014, 51, 425.2-426. | 1.5 | 2 |
| 147 | A lethal phenotype associated with tissue plasminogen deficiency in humans. Human Genetics, 2016, 135, 1209-1211. | 1.8 | 2 |
| 148 | Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668. | 1.1 | 2 |
| 149 | FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. American Journal of Human Genetics, 2009, 85, 756. | 2.6 | 1 |
| 150 | Reply to â€œan extremely severe phenotype due to WDR81 nonsense mutationsâ€• Annals of Neurology, 2017, 82, 651-651. | 2.8 | 1 |
| 151 | Mitochondrial â€œdysmorphologyâ€•in variant classification. Human Genetics, 2022, 141, 55-64. | 1.8 | 0 |