Fowzan S Alkuraya

List of Publications by Year
in descending order

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473	19,746	13332 70	32181 105
papers	citations	h-index	g-index

516	516	516	28526
all docs	docs citations	times ranked	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	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. Nature Genetics, 2016, 48, 1071-1076.	9.4	314
4	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
5	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	9.4	251
6	LPS-responsive beige-like anchor (LRBA) gene mutation in a family with inflammatory bowel disease and combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2012, 130, 481-488.e2.	1.5	232
7	Mutations in CCNO result in congenital mucociliary clearance disorder with reduced generation of multiple motile cilia. Nature Genetics, 2014, 46, 646-651.	9.4	232
8	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nature Genetics, 2011, 43, 197-203.	9.4	229
9	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
10	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
11	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Human Genetics, 2017, 136, 921-939.	1.8	209
12	Homozygous Mutations in ADAMTS10 and ADAMTS17 Cause Lenticular Myopia, Ectopia Lentis, Glaucoma, Spherophakia, and Short Stature. American Journal of Human Genetics, 2009, 85, 558-568.	2.6	204
13	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. American Journal of Human Genetics, 2011, 88, 536-547.	2.6	196
14	Treatment of retinitis pigmentosa due to MERTK mutations by ocular subretinal injection of adeno-associated virus gene vector: results of a phase I trial. Human Genetics, 2016, 135, 327-343.	1.8	195
15	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. Molecular Psychiatry, 2017, 22, 615-624.	4.1	187
16	Mutations in the RNA Granule Component TDRD7 Cause Cataract and Glaucoma. Science, 2011, 331, 1571-1576.	6.0	186
17	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	2.6	184
18	High diagnostic yield of clinical exome sequencing in Middle Eastern patients with Mendelian disorders. Human Genetics, 2015, 134, 967-980.	1.8	168

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19	SUMO1 Haploinsufficiency Leads to Cleft Lip and Palate. Science, 2006, 313, 1751-1751.	6.0	165
20	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
21	Recessive Mutations in ELOVL4 Cause Ichthyosis, Intellectual Disability, and Spastic Quadriplegia. American Journal of Human Genetics, 2011, 89, 745-750.	2.6	161
22	TLE6 mutation causes the earliest known human embryonic lethality. Genome Biology, 2015, 16, 240.	3.8	153
23	Novel CENPJ mutation causes Seckel syndrome. Journal of Medical Genetics, 2010, 47, 411-414.	1.5	149
24	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
25	Mitochondrial phenylalanyl-tRNA synthetase mutations underlie fatal infantile Alpers encephalopathy. Human Molecular Genetics, 2012, 21, 4521-4529.	1.4	143
26	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	2.6	138
27	IFT27, encoding a small GTPase component of IFT particles, is mutated in a consanguineous family with Bardet-Biedl syndrome. Human Molecular Genetics, 2014, 23, 3307-3315.	1.4	134
28	Mutation in WDR4 impairs tRNA m7G46 methylation and causes a distinct form of microcephalic primordial dwarfism. Genome Biology, 2015, 16, 210.	3.8	132
29	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
30	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	1.8	122
31	Mutations in C2orf37, Encoding a Nucleolar Protein, Cause Hypogonadism, Alopecia, Diabetes Mellitus, Mental Retardation, and Extrapyramidal Syndrome. American Journal of Human Genetics, 2008, 83, 684-691.	2.6	121
32	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	3.8	118
33	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
34	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
35	A homozygous truncating mutation in PUS3 expands the role of tRNA modification in normal cognition. Human Genetics, 2016, 135, 707-713.	1.8	112
36	In search of triallelism in Bardetâ€“Biedl syndrome. European Journal of Human Genetics, 2012, 20, 420-427.	1.4	111

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37	The genetic landscape of familial congenital hydrocephalus. Annals of Neurology, 2017, 81, 890-897.	2.8	108
38	Autozygome decoded. Genetics in Medicine, 2010, 12, 765-771.	1.1	107
39	Homozygosity mapping: One more tool in the clinical geneticist's toolbox. Genetics in Medicine, 2010, 12, 236-239.	1.1	107
40	The application of next-generation sequencing in the autozygosity mapping of human recessive diseases. Human Genetics, 2013, 132, 1197-1211.	1.8	107
41	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. American Journal of Human Genetics, 2018, 102, 696-705.	2.6	105
42	Mutations in LRPAP1 Are Associated with Severe Myopia in Humans. American Journal of Human Genetics, 2013, 93, 313-320.	2.6	104
43	NFIA Haploinsufficiency Is Associated with a CNS Malformation Syndrome and Urinary Tract Defects. PLoS Genetics, 2007, 3, e80.	1.5	100
44	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 82, 712-722.	2.6	95
45	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
46	Exaggerated follicular helper T-cell responses in patients with LRBA deficiency caused by failure of CTLA4-mediated regulation. Journal of Allergy and Clinical Immunology, 2018, 141, 1050-1059.e10.	1.5	93
47	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
48	Identification of embryonic lethal genes in humans by autozygosity mapping and exome sequencing in consanguineous families. Genome Biology, 2015, 16, 116.	3.8	91
49	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. Neuron, 2014, 84, 1240-1257.	3.8	89
50	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540.	1.8	89
51	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. Genetics in Medicine, 2016, 18, 554-562.	1.1	89
52	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	3.9	89
53	Discovery of Rare Homozygous Mutations from Studies of Consanguineous Pedigrees. Current Protocols in Human Genetics, 2012, 75, Unit6.12.	3.5	87
54	FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. American Journal of Human Genetics, 2009, 85, 414-418.	2.6	86

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55	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	1.1	85
56	Preclinical Potency and Safety Studies of an AAV2-Mediated Gene Therapy Vector for the Treatment of MERTK Associated Retinitis Pigmentosa. Human Gene Therapy Clinical Development, 2013, 24, 23-28.	3.2	84
57	Molecular autopsy in maternalâ€“fetal medicine. Genetics in Medicine, 2018, 20, 420-427.	1.1	84
58	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	13.9	84
59	Mutations in MEOX1, Encoding Mesenchyme Homeobox 1, Cause Klippel-Feil Anomaly. American Journal of Human Genetics, 2013, 92, 157-161.	2.6	82
60	Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. Genetics in Medicine, 2016, 18, 1244-1249.	1.1	82
61	Mutation in PHC1 implicates chromatin remodeling in primary microcephaly pathogenesis. Human Molecular Genetics, 2013, 22, 2200-2213.	1.4	81
62	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	9.4	81
63	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	1.1	81
64	Attitude of Saudi families affected with hemoglobinopathies towards prenatal screening and abortion and the influence of religious ruling (Fatwa). Prenatal Diagnosis, 2001, 21, 448-451.	1.1	79
65	Ciliary GenesTBC1D32/C6orf170andSCLT1are Mutated in Patients with OFD Type IX. Human Mutation, 2014, 35, 36-40.	1.1	78
66	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
67	Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. Genetics in Medicine, 2017, 19, 1144-1150.	1.1	77
68	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
69	Identification of a novelDLX5mutation in a family with autosomal recessive split hand and foot malformation. Journal of Medical Genetics, 2012, 49, 16-20.	1.5	75
70	Mutation inMPDZcauses severe congenital hydrocephalus. Journal of Medical Genetics, 2013, 50, 54-58.	1.5	75
71	3â€Methylglutaconic aciduriaâ€”lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	1.7	74
72	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

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73	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. Human Genetics, 2017, 136, 205-225.	1.8	73
74	A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. Human Mutation, 2011, 32, 573-578.	1.1	72
75	Exome sequencing reveals a novel Fanconi group defined by XRCC2 mutation: Figure 1. Journal of Medical Genetics, 2012, 49, 184-186.	1.5	72
76	Identification of novel loci for pediatric cholestatic liver disease defined by KIF12, PPM1F, USP53, LSR, and WDR83OS pathogenic variants. Genetics in Medicine, 2019, 21, 1164-1172.	1.1	71
77	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	2.6	71
78	X-linked creatine transporter defect: A report on two unrelated boys with a severe clinical phenotype. Journal of Inherited Metabolic Disease, 2006, 29, 214-219.	1.7	70
79	Deficiency of the Cytoskeletal Protein SPECC1L Leads to Oblique Facial Clefting. American Journal of Human Genetics, 2011, 89, 44-55.	2.6	70
80	POC1A Truncation Mutation Causes a Ciliopathy in Humans Characterized by Primordial Dwarfism. American Journal of Human Genetics, 2012, 91, 330-336.	2.6	70
81	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
82	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. American Journal of Human Genetics, 2018, 103, 612-620.	2.6	70
83	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	3.7	70
84	Identification of ADAMTS18 as a gene mutated in Knobloch syndrome. Journal of Medical Genetics, 2011, 48, 597-601.	1.5	68
85	Deficiency of a Retinal Dystrophy Protein, Acyl-CoA Binding Domain-containing 5 (ACBD5), Impairs Peroxisomal Î²-Oxidation of Very-long-chain Fatty Acids. Journal of Biological Chemistry, 2017, 292, 691-705.	1.6	68
86	The morbid genome of ciliopathies: an update. Genetics in Medicine, 2020, 22, 1051-1060.	1.1	68
87	A nullimorphic ERLIN2 mutation defines a complicated hereditary spastic paraplegia locus (SPG18). Neurogenetics, 2011, 12, 333-336.	0.7	67
88	Complementation of hypersensitivity to DNA interstrand crosslinking agents demonstrates thatXRCC2is a Fanconi anaemia gene. Journal of Medical Genetics, 2016, 53, 672-680.	1.5	66
89	Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. Human Genetics, 2016, 135, 1295-1298.	1.8	65
90	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	2.6	65

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91	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. American Journal of Human Genetics, 2018, 102, 858-873.	2.6	65
92	LOXL3, encoding lysyl oxidase-like 3, is mutated in a family with autosomal recessive Stickler syndrome. Human Genetics, 2015, 134, 451-453.	1.8	64
93	A novel syndrome of Klippel-Feil anomaly, myopathy, and characteristic facies is linked to a null mutation inMYO18B. Journal of Medical Genetics, 2015, 52, 400-404.	1.5	64
94	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
95	The distinct ophthalmic phenotype of Knobloch syndrome in children. British Journal of Ophthalmology, 2012, 96, 890-895.	2.1	63
96	Loss of function mutation in LARP7, chaperone of 7SK ncRNA, causes a syndrome of facial dysmorphism, intellectual disability, and primordial dwarfism. Human Mutation, 2012, 33, 1429-1434.	1.1	63
97	Immunodeficiency and EBV-induced lymphoproliferation caused by 4-1BB deficiency. Journal of Allergy and Clinical Immunology, 2019, 144, 574-583.e5.	1.5	63
98	Mutations of KIF14 cause primary microcephaly by impairing cytokinesis. Annals of Neurology, 2017, 82, 562-577.	2.8	62
99	Genetics and genomic medicine in Saudi Arabia. Molecular Genetics & Genomic Medicine, 2014, 2, 369-378.	0.6	61
100	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	2.6	61
101	Molecular characterization of retinitis pigmentosa in Saudi Arabia. Molecular Vision, 2009, 15, 2464-9.	1.1	61
102	Identification of a truncation mutation of acylglycerol kinase (AGK) gene in a novel autosomal recessive cataract locus. Human Mutation, 2012, 33, 960-962.	1.1	60
103	International perspectives on the implementation of reproductive carrier screening. Prenatal Diagnosis, 2020, 40, 301-310.	1.1	60
104	Homozygous null mutation in ODZ3 causes microphthalmia in humans. Genetics in Medicine, 2012, 14, 900-904.	1.1	59
105	Female Infertility Caused by Mutations in the Oocyte-Specific Translational Repressor PATL2. American Journal of Human Genetics, 2017, 101, 603-608.	2.6	59
106	Increasing the sensitivity of clinical exome sequencing through improved filtration strategy. Genetics in Medicine, 2017, 19, 593-598.	1.1	59
107	EROS/CYBC1 mutations: Decreased NADPH oxidase function and chronic granulomatous disease. Journal of Allergy and Clinical Immunology, 2019, 143, 782-785.e1.	1.5	59
108	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	3.8	59

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109	A genomics approach to females with infertility and recurrent pregnancy loss. Human Genetics, 2020, 139, 605-613.	1.8	59
110	A genomics approach to male infertility. Genetics in Medicine, 2020, 22, 1967-1975.	1.1	57
111	Molecular characterization of Joubert syndrome in Saudi Arabia. Human Mutation, 2012, 33, 1423-1428.	1.1	56
112	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
113	The Syndrome of Microcornea, Myopic Chorioretinal Atrophy, and Telecanthus (MMCAT) Is Caused by Mutations inADAMTS18. Human Mutation, 2013, 34, 1195-1199.	1.1	56
114	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	3.8	56
115	Genetic heterogeneity and evolutionary history of high-grade ovarian carcinoma and matched distant metastases. British Journal of Cancer, 2020, 122, 1219-1230.	2.9	56
116	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	1.1	56
117	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
118	Biometric and Molecular Characterization of Clinically Diagnosed Posterior Microphthalmos. American Journal of Ophthalmology, 2013, 155, 361-372.e7.	1.7	54
119	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	54
120	Discovery of mutations for Mendelian disorders. Human Genetics, 2016, 135, 615-623.	1.8	53
121	PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. Human Genetics, 2019, 138, 231-239.	1.8	53
122	Identification of differentially expressed proteins in the aqueous humor of primary congenital glaucoma. Experimental Eye Research, 2011, 92, 67-75.	1.2	51
123	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
124	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217.	2.6	51
125	Mutations in ASPH Cause Facial Dysmorphism, Lens Dislocation, Anterior-Segment Abnormalities, and Spontaneous Filtering Blebs, or Traboulsi Syndrome. American Journal of Human Genetics, 2014, 94, 755-759.	2.6	50
126	Severe CNS involvement in WWOX mutations: Description of five new cases. American Journal of Medical Genetics, Part A, 2015, 167, 3209-3213.	0.7	50

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127	Novel recessive BFSP2 and PITX3 mutations: Insights into mutational mechanisms from consanguineous populations. Genetics in Medicine, 2011, 13, 978-981.	1.1	49
128	Genomic analysis of pediatric cataract in Saudi Arabia reveals novel candidate disease genes. Genetics in Medicine, 2012, 14, 955-962.	1.1	49
129	Autozygosity Mapping with Exome Sequence Data. Human Mutation, 2013, 34, 50-56.	1.1	49
130	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
131	Tyrosine-Mutant AAV8 Delivery of HumanMERTKProvides Long-Term Retinal Preservation in RCS Rats. , 2012, 53, 1895.		48
132	Autozygome Sequencing Expands the Horizon of Human Knockout Research and Provides Novel Insights into Human Phenotypic Variation. PLoS Genetics, 2013, 9, e1004030.	1.5	48
133	Revisiting the morbid genome of Mendelian disorders. Genome Biology, 2016, 17, 235.	3.8	48
134	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	2.6	48
135	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. American Journal of Human Genetics, 2010, 87, 306-307.	2.6	47
136	MYSM1 is mutated in a family with transient transfusion-dependent anemia, mild thrombocytopenia, and low NK- and B-cell counts. Blood, 2013, 122, 3844-3845.	0.6	47
137	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	1.4	47
138	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	1.1	46
139	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
140	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. Human Molecular Genetics, 2019, 28, 2900-2919.	1.4	46
141	Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. American Journal of Human Genetics, 2013, 93, 555-560.	2.6	45
142	WNT1 mutation with recessive osteogenesis imperfecta and profound neurological phenotype. Journal of Medical Genetics, 2013, 50, 491-492.	1.5	45
143	On the phenotypic spectrum of serine biosynthesis defects. Journal of Inherited Metabolic Disease, 2016, 39, 373-381.	1.7	45
144	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

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145	<scp>GLI3</scp>â€related polydactyly: a review. Clinical Genetics, 2017, 92, 457-466.	1.0	45
146	A human ciliopathy reveals essential functions for NEK10 in airway mucociliary clearance. Nature Medicine, 2020, 26, 244-251.	15.2	45
147	Impact of new genomic tools on the practice of clinical genetics in consanguineous populations: the Saudi experience. Clinical Genetics, 2013, 84, 203-208.	1.0	44
148	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. American Journal of Human Genetics, 2016, 99, 228-235.	2.6	44
149	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	3.7	43
150	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	3.9	43
151	Lifting the lid on unborn lethal Mendelian phenotypes through exome sequencing. Genetics in Medicine, 2013, 15, 307-309.	1.1	42
152	Identification of a novel MKS locus defined byTMEM107mutation. Human Molecular Genetics, 2015, 24, 5211-5218.	1.4	42
153	Human knockout research: new horizons and opportunities. Trends in Genetics, 2015, 31, 108-115.	2.9	42
154	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	1.1	42
155	Mutational Spectrum ofSLC4A11in Autosomal Recessive CHED in Saudi Arabia. , 2009, 50, 4142.		41
156	NR2F1 deletion in a patient with a de novo paracentric inversion, inv(5)(q15q33.2), and syndromic deafness. American Journal of Medical Genetics, Part A, 2009, 149A, 931-938.	0.7	41
157	C2orf37 mutational spectrum in Woodhouseâ€“Sakati syndrome patients. Clinical Genetics, 2010, 78, 585-590.	1.0	41
158	Mutation of IGFBP7 Causes Upregulation of BRAF/MEK/ERK Pathway and Familial Retinal Arterial Macroaneurysms. American Journal of Human Genetics, 2011, 89, 313-319.	2.6	41
159	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessiveWNT1mutations. Journal of Medical Genetics, 2016, 53, 427-430.	1.5	41
160	Genomic Profiling of Thyroid Cancer Reveals a Role for Thyroglobulin in Metastasis. American Journal of Human Genetics, 2016, 98, 1170-1180.	2.6	41
161	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	1.1	41
162	Absence of GP130 cytokine receptor signaling causes extended StÃ¼ve-Wiedemann syndrome. Journal of Experimental Medicine, 2020, 217, .	4.2	41

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163	Molecular Characterization of Newborn Glaucoma Including a Distinct Aniridic Phenotype. Ophthalmic Genetics, 2011, 32, 138-142.	0.5	40
164	A novel syndrome of hypohidrosis and intellectual disability is linked to COG6 deficiency. Journal of Medical Genetics, 2013, 50, 431-436.	1.5	40
165	Cell-Intrinsic Adaptation Arising from Chronic Ablation of a Key Rho GTPase Regulator. Developmental Cell, 2016, 39, 28-43.	3.1	40
166	An autosomal recessiveDNASE1L3-related autoimmune disease with unusual clinical presentation mimicking systemic lupus erythematosus. Lupus, 2017, 26, 768-772.	0.8	40
167	Phenotypic and Molecular Spectrum of Aicardi-GoutiÃ¨res Syndrome: A Study of 24 Patients. Pediatric Neurology, 2018, 78, 35-40.	1.0	40
168	A novel PTF1A mutation in a patient with severe pancreatic and cerebellar involvement. Clinical Genetics, 2011, 80, 196-198.	1.0	39
169	Functional analysis of BBS3 A89V that results in non-syndromic retinal degeneration. Human Molecular Genetics, 2011, 20, 1625-1632.	1.4	38
170	GOLGA2, encoding a master regulator of golgi apparatus, is mutated in a patient with a neuromuscular disorder. Human Genetics, 2016, 135, 245-251.	1.8	38
171	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. American Journal of Human Genetics, 2017, 100, 969-977.	2.6	38
172	A null mutation in MICU2 causes abnormal mitochondrial calcium homeostasis and a severe neurodevelopmental disorder. Brain, 2017, 140, 2806-2813.	3.7	38
173	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. Clinical Genetics, 2018, 93, 1210-1222.	1.0	38
174	Tufting Enteropathy and Chronic Arthritis: A Newly Recognized Association With a Novel EpCAM Gene Mutation. Journal of Pediatric Gastroenterology and Nutrition, 2009, 49, 642-644.	0.9	37
175	Mutations in UNC80, Encoding Part of the UNC79-UNC80-NALCN Channel Complex, Cause Autosomal-Recessive Severe Infantile Encephalopathy. American Journal of Human Genetics, 2016, 98, 210-215.	2.6	37
176	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
177	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. American Journal of Human Genetics, 2019, 105, 384-394.	2.6	37
178	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
179	Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. Journal of Inherited Metabolic Disease, 2013, 36, 997-1004.	1.7	36
180	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. American Journal of Human Genetics, 2015, 97, 862-868.	2.6	36

#	Article	IF	Citations
181	GNB5 mutation causes a novel neuropsychiatric disorder featuring attention deficit hyperactivity disorder, severely impaired language development and normal cognition. Genome Biology, 2016, 17, 195.	3.8	36
182	Genetic profiling of children with advanced cholestatic liver disease. Clinical Genetics, 2017, 92, 52-61.	1.0	36
183	Characterization ofCTNSmutations in Arab patients with Cystinosis. Ophthalmic Genetics, 2009, 30, 185-189.	0.5	35
184	Neuronal ceroid lipofuscinosis caused by MFSD8 mutations: a common theme emerging. Neurogenetics, 2009, 10, 307-311.	0.7	35
185	Mutation inRAB33B, which encodes a regulator of retrograde Golgi transport, defines a second Dyggveâ€“Melchiorâ€“Clausen locus. Journal of Medical Genetics, 2012, 49, 455-461.	1.5	35
186	Mutations in C12orf57 Cause a Syndromic Form of Colobomatous Microphthalmia. American Journal of Human Genetics, 2013, 92, 387-391.	2.6	35
187	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
188	Mutation in SLC6A9 encoding a glycine transporter causes a novel form of non-ketotic hyperglycinemia in humans. Human Genetics, 2016, 135, 1263-1268.	1.8	35
189	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
190	â€˜Cone dystrophy with supranormal rod responseâ€™ in children. British Journal of Ophthalmology, 2012, 96, 422-426.	2.1	34
191	Mutation of the mitochondrial carrier SLC25A42 causes a novel form of mitochondrial myopathy in humans. Human Genetics, 2016, 135, 21-30.	1.8	34
192	Homozygous Loss-of-Function Mutations in AP1B1, Encoding Beta-1 Subunit of Adaptor-Related Protein Complex 1, Cause MEDNIK-like Syndrome. American Journal of Human Genetics, 2019, 105, 1016-1022.	2.6	34
193	Recessive Truncating Mutations in ALKBH8 Cause Intellectual Disability and Severe Impairment of Wobble Uridine Modification. American Journal of Human Genetics, 2019, 104, 1202-1209.	2.6	34
194	The landscape of early infantile epileptic encephalopathy in a consanguineous population. Seizure: the Journal of the British Epilepsy Association, 2019, 69, 154-172.	0.9	34
195	Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. American Journal of Human Genetics, 2019, 105, 1237-1253.	2.6	34
196	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	0.7	34
197	A novel Xâ€linked disorder with developmental delay and autistic features. Annals of Neurology, 2012, 71, 498-508.	2.8	33
198	Brittle Cornea Syndrome ZNF469 Mutation Carrier Phenotype and Segregation Analysis of Rare ZNF469 Variants in Familial Keratoconus. Investigative Ophthalmology and Visual Science, 2015, 56, 578-586.	3.3	33

#	Article	IF	Citations
199	<scp>WDR45B</scp>â€related intellectual disability, spastic quadriplegia, epilepsy, and cerebral hypoplasia: A consistent neurodevelopmental syndrome. Clinical Genetics, 2018, 93, 360-364.	1.0	33
200	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
201	ADAT3â€related intellectual disability: Further delineation of the phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 1142-1147.	0.7	32
202	Hyperekplexia, microcephaly and simplified gyral pattern caused by novel ASNS mutations, case report. BMC Neurology, 2016, 16, 105.	0.8	32
203	Confirming the recessive inheritance of <scp>SCN1B</scp> mutations in developmental epileptic encephalopathy. Clinical Genetics, 2017, 92, 327-331.	1.0	32
204	The human knockout phenotype of <scp>PADI6</scp> is female sterility caused by cleavage failure of their fertilized eggs. Clinical Genetics, 2017, 91, 344-345.	1.0	32
205	Clinical exome sequencing in 509 Middle Eastern families with suspected Mendelian diseases: The Qatari experience. American Journal of Medical Genetics, Part A, 2019, 179, 927-935.	0.7	32
206	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
207	Syndromic congenital sensorineural deafness, microtia and microdontia resulting from a novel homoallelic mutation in fibroblast growth factor 3 (FGF3). European Journal of Human Genetics, 2009, 17, 14-21.	1.4	31
208	Mutation of CANT1 causes Desbuquois dysplasia. American Journal of Medical Genetics, Part A, 2010, 152A, 1157-1160.	0.7	31
209	Familial spherophakia with short stature caused by a novel homozygousADAMTS17mutation. Ophthalmic Genetics, 2012, 33, 235-239.	0.5	31
210	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
211	Whole-Exome Sequencing of Matched Primary and Metastatic Papillary Thyroid Cancer. Thyroid, 2020, 30, 42-56.	2.4	31
212	DALRD3 encodes a protein mutated in epileptic encephalopathy that targets arginine tRNAs for 3-methylcytosine modification. Nature Communications, 2020, 11, 2510.	5.8	31
213	Congenital disorders of glycosylation: The Saudi experience. American Journal of Medical Genetics, Part A, 2017, 173, 2614-2621.	0.7	31
214	Positional mapping ofPRKD1,NRP1andPRDM1as novel candidate disease genes in truncus arteriosus. Journal of Medical Genetics, 2015, 52, 322-329.	1.5	30
215	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
216	Impaired telomere maintenance in Alazami syndrome patients with LARP7 deficiency. BMC Genomics, 2016, 17, 749.	1.2	30

#	Article	IF	Citations
217	Combining exome/genome sequencing with data repository analysis reveals novel geneâ€“disease associations for a wide range of genetic disorders. Genetics in Medicine, 2021, 23, 1551-1568.	1.1	30
218	Blue Sclera With and Without Corneal Fragility (Brittle Cornea Syndrome) in a Consanguineous Family Harboring ZNF469 Mutation (p.E1392X). JAMA Ophthalmology, 2010, 128, 1376.	2.6	29
219	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
220	Congenital glaucoma and CYP1B1: an old story revisited. Human Genetics, 2019, 138, 1043-1049.	1.8	29
221	A novel missense mutation in SCYL1BP1 produces geroderma osteodysplastica phenotype indistinguishable from that caused by nullimorphic mutations. American Journal of Medical Genetics, Part A, 2009, 149A, 2093-2098.	0.7	28
222	Posterior Microphthalmos as a Genetically Heterogeneous Condition That Can Be Allelic to Nanophthalmos. JAMA Ophthalmology, 2011, 129, 805.	2.6	28
223	RP1 and retinitis pigmentosa: report of novel mutations and insight into mutational mechanism. British Journal of Ophthalmology, 2012, 96, 1018-1022.	2.1	28
224	Study of Mendelian forms of Crohn's disease in Saudi Arabia reveals novel risk loci and alleles. Gut, 2014, 63, 1831-1832.	6.1	28
225	C19orf12 mutation leads to a pallido-pyramidal syndrome. Gene, 2014, 537, 352-356.	1.0	28
226	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. Hepatology, 2020, 71, 2067-2079.	3.6	28
227	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	3.7	28
228	A novel C2orf37 mutation causes the first Italian cases of Woodhouse Sakati syndrome. Clinical Genetics, 2010, 78, 594-597.	1.0	27
229	3M Syndrome: An Easily Recognizable yet Underdiagnosed Cause of Proportionate Short Stature. Journal of Pediatrics, 2012, 161, 139-145.e1.	0.9	27
230	Recessive Mutations inLEPREL1Underlie a Recognizable Lens Subluxation Phenotype. Ophthalmic Genetics, 2015, 36, 58-63.	0.5	26
231	A lethal neonatal phenotype of mitochondrial shortâ€chain enoylâ€<scp>CoA</scp> hydrataseâ€1 deficiency. Clinical Genetics, 2017, 91, 629-633.	1.0	26
232	Phenotypes of Recessive Pediatric Cataract in a Cohort of Children with Identified Homozygous Gene Mutations (An American Ophthalmological Society Thesis). Transactions of the American Ophthalmological Society, 2015, 113, T7.	1.4	26
233	The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803.	2.2	25
234	ZBTB42 mutation defines a novel lethal congenital contracture syndrome (LCCS6). Human Molecular Genetics, 2014, 23, 6584-6593.	1.4	25

#	Article	IF	Citations
235	Natural human knockouts and the era of genotype to phenotype. Genome Medicine, 2015, 7, 48.	3.6	25
236	Biallelic variants in CTU2 cause DREAMâ€PL syndrome and impair thiolation of tRNA wobble U34. Human Mutation, 2019, 40, 2108-2120.	1.1	25
237	Formation of tRNA Wobble Inosine in Humans Is Disrupted by a Millennia-Old Mutation Causing Intellectual Disability. Molecular and Cellular Biology, 2019, 39, .	1.1	25
238	A novel mutation in PRDM5 in brittle cornea syndrome. Clinical Genetics, 2012, 81, 198-199.	1.0	24
239	Expanding the phenotype of SLC25A42â€associated mitochondrial encephalomyopathy. Clinical Genetics, 2018, 93, 1097-1102.	1.0	24
240	De novo truncating variants in WHSC1 recapitulate the Wolfâ€“Hirschhorn (4p16.3 microdeletion) syndrome phenotype. Genetics in Medicine, 2019, 21, 185-188.	1.1	24
241	An intellectual disabilityâ€associated missense variant in TRMT1 impairs tRNA modification and reconstitution of enzymatic activity. Human Mutation, 2020, 41, 600-607.	1.1	24
242	Identification of a novel ZNF469 mutation in a large family with Ehlersâ€“Danlos phenotype. Gene, 2012, 511, 447-450.	1.0	23
243	NECAP1 loss of function leads to a severe infantile epileptic encephalopathy. Journal of Medical Genetics, 2014, 51, 224-228.	1.5	23
244	A null mutation in TNIK defines a novel locus for intellectual disability. Human Genetics, 2016, 135, 773-778.	1.8	23
245	Neuronal deficiency ofARV1causes an autosomal recessive epileptic encephalopathy. Human Molecular Genetics, 2016, 25, ddw157.	1.4	23
246	Expanding the spectrum of germline variants in cancer. Human Genetics, 2017, 136, 1431-1444.	1.8	23
247	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
248	Later retinal degeneration following childhood surgical aphakia in a family with recessive CRYAB mutation (p.R56W). Ophthalmic Genetics, 2010, 31, 30-36.	0.5	22
249	Childhood Cone-rod Dystrophy with Macular Cystic Degeneration from RecessiveCRB1Mutation. Ophthalmic Genetics, 2014, 35, 130-137.	0.5	22
250	Novel IFT122 mutation associated with impaired ciliogenesis and cranioectodermal dysplasia. Molecular Genetics & Genomic Medicine, 2014, 2, 103-106.	0.6	22
251	Primordial dwarfism. Current Opinion in Endocrinology, Diabetes and Obesity, 2015, 22, 55-64.	1.2	22
252	Evolution and Impact of Subclonal Mutations in Papillary Thyroid Cancer. American Journal of Human Genetics, 2019, 105, 959-973.	2.6	22

#	Article	IF	Citations
253	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	1.1	22
254	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
255	Deletion of DDB1- and CUL4- associated factor-17 (Dcaf17) gene causes spermatogenesis defects and male infertility in mice. Scientific Reports, 2018, 8, 9202.	1.6	21
256	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	1.4	21
257	New paradigms of USP53 disease: normal GGT cholestasis, BRIC, cholangiopathy, and responsiveness to rifampicin. Journal of Human Genetics, 2021, 66, 151-159.	1.1	21
258	Brittle cornea without clinically-evident extraocular findings in an adult harboring a novel homozygousZNF469mutation. Ophthalmic Genetics, 2012, 33, 257-259.	0.5	20
259	No evidence for locus heterogeneity in Knobloch syndrome. Journal of Medical Genetics, 2013, 50, 565-566.	1.5	20
260	TheRPGRIP1-related retinal phenotype in children. British Journal of Ophthalmology, 2013, 97, 760-764.	2.1	20
261	Gonadal mosaicism as a rare cause of autosomal recessive inheritance. Clinical Genetics, 2014, 85, 278-281.	1.0	20
262	A novelAPCmutation defines a second locus for Cenaniâ€“Lenz syndrome. Journal of Medical Genetics, 2015, 52, 317-321.	1.5	20
263	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. Advances in Experimental Medicine and Biology, 2017, 1031, 55-94.	0.8	20
264	Cenaniâ€“Lenz syndrome and other related syndactyly disorders due to variants in LRP4 , GREM1 / FMN1 , and APC : Insight into the pathogenesis and the relationship to polyposis through the WNT and BMP antagonistic pathways. American Journal of Medical Genetics, Part A, 2019, 179, 266-279.	0.7	20
265	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
266	Mutations in HYAL2, Encoding Hyaluronidase 2, Cause a Syndrome of Orofacial Clefting and Cor Triatriatum Sinister in Humans and Mice. PLoS Genetics, 2017, 13, e1006470.	1.5	20
267	GM2 gangliosidosis in Saudi Arabia: Multiple mutations and considerations for future carrier screening. American Journal of Medical Genetics, Part A, 2011, 155, 1281-1284.	0.7	19
268	5â€Oxoprolinase deficiency: report of the first human OPLAH mutation. Clinical Genetics, 2012, 82, 193-196.	1.0	19
269	Report of a case of Raine syndrome and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 2394-2398.	0.7	19
270	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725.	1.1	19

#	Article	IF	Citations
271	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
272	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. American Journal of Human Genetics, 2019, 104, 542-552.	2.6	19
273	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	2.6	19
274	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
275	Clinical, biochemical and molecular characterization of peroxisomal diseases in Arabs. Clinical Genetics, 2011, 79, 60-70.	1.0	18
276	Phenotype-genotype Correlation in Potential Female Carriers of X-linked Developmental Cataract (Nance-Horan Syndrome). Ophthalmic Genetics, 2012, 33, 89-95.	0.5	18
277	NPHP4 mutation is linked to cerebelloâ€oculoâ€renal syndrome and male infertility. Clinical Genetics, 2014, 85, 371-375.	1.0	18
278	SET binding factor 1 (SBF1) mutation causes Charcot-Marie-Tooth disease type 4B3. Neurology, 2014, 82, 1665-1666.	1.5	18
279	KDF1, encoding keratinocyte differentiation factor 1, is mutated in a multigenerational family with ectodermal dysplasia. Human Genetics, 2017, 136, 99-105.	1.8	18
280	Elsahyâ€“Waters syndrome is caused by biallelic mutations in CDH11. American Journal of Medical Genetics, Part A, 2018, 176, 477-482.	0.7	18
281	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	2.6	18
282	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	2.6	18
283	CNP deficiency causes severe hypomyelinating leukodystrophy in humans. Human Genetics, 2020, 139, 615-622.	1.8	18
284	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
285	Founder heterozygous P23T CRYGD mutation associated with cerulean (and coralliform) cataract in 2 Saudi families. Molecular Vision, 2009, 15, 1407-11.	1.1	18
286	Johansonâ€“Blizzard syndrome: Report of a novel mutation and severe liver involvement. American Journal of Medical Genetics, Part A, 2008, 146A, 1875-1879.	0.7	17
287	Helicoid Subretinal Fibrosis Associated With a Novel Recessive NR2E3 Mutation p.S44X. JAMA Ophthalmology, 2010, 128, 344.	2.6	17
288	Juvenile Cataract Morphology in 3 Siblings Not Yet Diagnosed with Cerebrotendinous Xanthomatosis. Ophthalmology, 2013, 120, 956-960.	2.5	17

#	Article	IF	Citations
289	Revisiting disease genes based on whole-exome sequencing in consanguineous populations. Human Genetics, 2015, 134, 1029-1034.	1.8	17
290	Further Delineation of the ALG9-CDG Phenotype. JIMD Reports, 2015, 27, 107-112.	0.7	17
291	ANKS3 is mutated in a family with autosomal recessive laterality defect. Human Genetics, 2016, 135, 1233-1239.	1.8	17
292	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
293	Phenotypic characterization of KCTD3â€related developmental epileptic encephalopathy. Clinical Genetics, 2018, 93, 1081-1086.	1.0	17
294	Identification of a novel lethal form of autosomal recessive ichthyosis caused by UDPâ€glucose ceramide glucosyltransferase deficiency. Clinical Genetics, 2018, 93, 1252-1253.	1.0	17
295	NUP214 deficiency causes severe encephalopathy and microcephaly in humans. Human Genetics, 2019, 138, 221-229.	1.8	17
296	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17
297	Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. American Journal of Human Genetics, 2020, 106, 246-255.	2.6	17
298	Lens Subluxation and Retinal Dysfunction in a Girl with HomozygousVSX2Mutation. Ophthalmic Genetics, 2015, 36, 8-13.	0.5	16
299	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
300	A novel mutation in SLC25A46 causes optic atrophy and progressive limb spasticity, with no cerebellar atrophy or axonal neuropathy. Clinical Genetics, 2017, 92, 230-231.	1.0	16
301	Warsaw breakage syndrome: Further clinical and genetic delineation. American Journal of Medical Genetics, Part A, 2018, 176, 2404-2418.	0.7	16
302	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. Human Genetics, 2020, 139, 1273-1283.	1.8	16
303	A Case with Purine Nucleoside Phosphorylase Deficiency Suffering from Late-Onset Systemic Lupus Erythematosus and Lymphoma. Journal of Clinical Immunology, 2020, 40, 833-839.	2.0	16
304	The many faces of KIF7. Human Genome Variation, 2015, 2, 15006.	0.4	16
305	ASTL is mutated in female infertility. Human Genetics, 2022, 141, 49-54.	1.8	16
306	Clinical and molecular analysis of children with central pulverulent cataract from the Arabian Peninsula. British Journal of Ophthalmology, 2012, 96, 650-655.	2.1	15

#	Article	IF	Citations
307	Excessively redundant umbilical skin as a potential early clinical feature of Morquio syndrome and <scp>FKBP14</scp>â€related Ehlersâ€“Danlos syndrome. Clinical Genetics, 2014, 86, 469-472.	1.0	15
308	Mutations of PTPN23 in developmental and epileptic encephalopathy. Human Genetics, 2017, 136, 1455-1461.	1.8	15
309	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
310	Patterns of neurological manifestations in Woodhouse-Sakati Syndrome. Parkinsonism and Related Disorders, 2019, 69, 99-103.	1.1	15
311	Biallelic variants in the small optic lobe calpain CAPN15 are associated with congenital eye anomalies, deafness and other neurodevelopmental deficits. Human Molecular Genetics, 2020, 29, 3054-3063.	1.4	15
312	Residual risk for additional recessive diseases in consanguineous couples. Genetics in Medicine, 2021, 23, 2448-2454.	1.1	15
313	Mutations in TP73 cause impaired mucociliary clearance and lissencephaly. American Journal of Human Genetics, 2021, 108, 1318-1329.	2.6	15
314	Expanding the â€œEâ€•in CHARGE. American Journal of Medical Genetics, Part A, 2008, 146A, 1890-1892.	0.7	14
315	Colorectal cancer risk is not associated with increased levels of homozygosity in Saudi Arabia. Genetics in Medicine, 2012, 14, 720-728.	1.1	14
316	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	2.6	14
317	GZF1 Mutations Expand the Genetic Heterogeneity of Larsen Syndrome. American Journal of Human Genetics, 2017, 100, 831-836.	2.6	14
318	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
319	Population structure of indigenous inhabitants of Arabia. PLoS Genetics, 2021, 17, e1009210.	1.5	14
320	Genetic and genomic analysis of classic aniridia in Saudi Arabia. Molecular Vision, 2011, 17, 708-14.	1.1	14
321	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
322	Familial juvenile glaucoma with underlying homozygous p.G61E CYP1B1 mutations. Journal of AAPOS, 2011, 15, 198-199.	0.2	13
323	Congenital glaucoma with acquired peripheral circumferential iris degeneration. Journal of AAPOS, 2013, 17, 105-107.	0.2	13
324	ARL6IP6, a susceptibility locus for ischemic stroke, is mutated in a patient with syndromic Cutis Marmorata Telangiectatica Congenita. Human Genetics, 2015, 134, 815-822.	1.8	13

#	Article	IF	Citations
325	T (brachyury) is linked to a Mendelian form of neural tube defects in humans. Human Genetics, 2015, 134, 1139-1141.	1.8	13
326	Confirming the candidacy of THOC6 in the etiology of intellectual disability. American Journal of Medical Genetics, Part A, 2016, 170, 1367-1369.	0.7	13
327	Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. Orphanet Journal of Rare Diseases, 2016, 11, 52.	1.2	13
328	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. Brain, 2020, 143, 2911-2928.	3.7	13
329	Pathogenic STX3 variants affecting the retinal and intestinal transcripts cause an early-onset severe retinal dystrophy in microvillus inclusion disease subjects. Human Genetics, 2021, 140, 1143-1156.	1.8	13
330	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Frontiers in Genetics, 2020, 11, 580484.	1.1	13
331	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. Genome Medicine, 2021, 13, 161.	3.6	13
332	Fryns syndrome with Hirschsprung disease: Support for possible neural crest involvement. American Journal of Medical Genetics, Part A, 2005, 132A, 226-230.	0.7	12
333	Ritscherâ€“Schinzel (cranioâ€cerebelloâ€cardiac, 3C) syndrome: Report of four new cases with renal involvement. American Journal of Medical Genetics, Part A, 2011, 155, 1393-1397.	0.7	12
334	Familial dorsalization of the skin of the proximal palm and the instep of the sole of the foot. Gene, 2012, 500, 216-219.	1.0	12
335	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
336	Pellagraâ€like condition is xeroderma pigmentosum/Cockayne syndrome complex and niacin confers clinical benefit. Clinical Genetics, 2015, 87, 56-61.	1.0	12
337	Autosomal recessive congenital cataract, intellectual disability phenotype linked to <scp>STX3</scp> in a consanguineous Tunisian family. Clinical Genetics, 2015, 88, 283-287.	1.0	12
338	Clinical Characterization of LRPAP1-Related Pediatric High Myopia. Ophthalmology, 2016, 123, 434-435.	2.5	12
339	MED12is recurrently mutated in Middle Eastern colorectal cancer. Gut, 2018, 67, gutjnl-2016-313334.	6.1	12
340	Mutations in known disease genes account for the majority of autosomal recessive retinal dystrophies. Clinical Genetics, 2018, 94, 554-563.	1.0	12
341	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	1.0	12
342	A novel truncating variant in ring finger protein 113A (RNF113A) confirms the association of this gene with Xâ€linked trichothiodystrophy. American Journal of Medical Genetics, Part A, 2020, 182, 513-520.	0.7	12

#	Article	IF	Citations
343	How the human genome transformed study of rare diseases. Nature, 2021, 590, 218-219.	13.7	12
344	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	0.7	12
345	Genomic medicine in the Middle East. Genome Medicine, 2021, 13, 184.	3.6	12
346	Mental retardation, growth retardation, unusual nose, and open mouth: An autosomal recessive entity. American Journal of Medical Genetics, Part A, 2010, 152A, 2160-2163.	0.7	11
347	Dorsal Dimelia: Report of Two Cases With an Emphasis On the Variation of Phenotypic Expression and a Search for Candidate Causative Genes. Journal of Hand Surgery: European Volume, 2010, 35, 715-720.	0.5	11
348	The natural history of infantile neuroaxonal dystrophy. Orphanet Journal of Rare Diseases, 2020, 15, 109.	1.2	11
349	A familial PLCB4 mutation causing auriculocondylar syndrome 2 with variable severity. European Journal of Medical Genetics, 2020, 63, 103917.	0.7	11
350	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	1.1	11
351	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
352	Corneal Decompensation in Recessive Cornea Plana. Ophthalmic Genetics, 2009, 30, 142-145.	0.5	10
353	Autozygome maps dispensable DNA and reveals potential selective bias against nullizygosity. Genetics in Medicine, 2012, 14, 515-519.	1.1	10
354	Homozygous lossâ€ofâ€function variants of TASP1 , a gene encoding an activator of the histone methyltransferases KMT2A and KMT2D, cause a syndrome of developmental delay, happy demeanor, distinctive facial features, and congenital anomalies. Human Mutation, 2019, 40, 1985-1992.	1.1	10
355	Recessive mutations in SCYL2 cause a novel syndromic form of arthrogryposis in humans. Human Genetics, 2020, 139, 513-519.	1.8	10
356	Methylation-Specific Multiplex-Ligation-Dependent Probe Amplification as a Rapid Molecular Diagnostic Tool for Pseudohypoparathyroidism Type 1b. Genetic Testing and Molecular Biomarkers, 2010, 14, 135-139.	0.3	9
357	A novel syndrome of lethal familial hyperekplexia associated with brain malformation. BMC Neurology, 2012, 12, 125.	0.8	9
358	Complete Aniridia with Central Keratopathy and Congenital Glaucoma is aCYP1B1-related Phenotype. Ophthalmic Genetics, 2014, 35, 187-189.	0.5	9
359	Congenital ptosis, scoliosis, andÂmalignant hyperthermia susceptibility in siblings with recessive RYR1 mutations. Journal of AAPOS, 2015, 19, 577-579.	0.2	9
360	RGS6: A Novel Gene Associated With Congenital Cataract, Mental Retardation, and Microcephaly in a Tunisian Family. Investigative Ophthalmology and Visual Science, 2015, 56, 1261-1266.	3.3	9

#	Article	IF	Citations
361	Intrafamilial clinical heterogeneity of CSPP1â€related ciliopathy. American Journal of Medical Genetics, Part A, 2015, 167, 2478-2480.	0.7	9
362	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 families. Molecular Genetics and Metabolism, 2016, 119, 44-49.	0.5	9
363	A mendelian form of neural tube defect caused by a de novo null variant in SMARCC1 in an identical twin. Annals of Neurology, 2018, 83, 433-436.	2.8	9
364	Clonal Evolution and Timing of Metastatic Colorectal Cancer. Cancers, 2020, 12, 2938.	1.7	9
365	Confirming <scp>TBC1D32</scp>â€related ciliopathy in humans. American Journal of Medical Genetics, Part A, 2020, 182, 1985-1987.	0.7	9
366	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	1.1	9
367	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
368	Arthrogryposis, perthes disease, and upward gaze palsy: A novel autosomal recessive syndromic form of arthrogryposis. American Journal of Medical Genetics, Part A, 2011, 155, 297-300.	0.7	8
369	CYP1B1 analysis of unilateral primary newborn glaucoma in Saudi children. Journal of AAPOS, 2012, 16, 571-572.	0.2	8
370	Expanding the clinical spectrum and allelic heterogeneity in van den Endeâ€“Gupta syndrome. Clinical Genetics, 2014, 85, 492-494.	1.0	8
371	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
372	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
373	Human knockouts ofPLA2G4Aphenocopy NSAID-induced gastrointestinal and renal toxicity. Gut, 2016, 65, 1575-1577.	6.1	8
374	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
375	<scp>MYH1</scp> is a candidate gene for recurrent rhabdomyolysis in humans. American Journal of Medical Genetics, Part A, 2021, 185, 2131-2135.	0.7	8
376	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	2.6	8
377	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	3.7	8
378	A novel syndromic form of sensory-motor polyneuropathy is linked to chromosome 22q13.31-q13.33. Clinical Genetics, 2011, 79, 193-195.	1.0	7

#	Article	IF	Citations
379	Identification of MRI1, encoding translation initiation factor eIF-2B subunit alpha/beta/delta-like protein, as a candidate locus for infantile epilepsy with severe cystic degeneration of the brain. Gene, 2013, 512, 450-452.	1.0	7
380	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 715-721.	0.7	7
381	Familial non-syndromic macular pseudocoloboma secondary to homozygousCLDN19mutation. Ophthalmic Genetics, 2018, 39, 577-583.	0.5	7
382	Rhegmatogenous Retinal Detachment in Nonsyndromic High Myopia Associated with Recessive Mutations in LRPAP1. Ophthalmology Retina, 2020, 4, 77-83.	1.2	7
383	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. Clinical Genetics, 2020, 97, 447-456.	1.0	7
384	Further delineation of <scp>HIDEA</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2999-3006.	0.7	7
385	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. Genetics in Medicine, 2020, 22, 2071-2080.	1.1	7
386	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
387	Insight into ALKBH8-related intellectual developmental disability based on the first pathogenic missense variant. Human Genetics, 2022, 141, 209-215.	1.8	7
388	A patient with a ring chromosome 2 and microdeletion of 2q detected using FISH: Further support for ?ring chromosome 2 syndrome?. American Journal of Medical Genetics, Part A, 2005, 132A, 447-449.	0.7	6
389	A syndrome of congenital hyperinsulinism and rhabdomyolysis is caused byKCNJ11mutation. Journal of Medical Genetics, 2014, 51, 271-274.	1.5	6
390	Distal acroosteolysis, poikiloderma and joint stiffness: a novel laminopathy?. European Journal of Human Genetics, 2016, 24, 1220-1222.	1.4	6
391	GWAS signals revisited using human knockouts. Genetics in Medicine, 2018, 20, 64-68.	1.1	6
392	Immunologic reconstitution following hematopoietic stem cell transplantation despite lymph node paucity in NF-Î²Bâ€“inducing kinase deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 1240-1243.e4.	1.5	6
393	A de novo TBX3 mutation presenting as dorsalization of the little fingers: A forme fruste phenotype of ulnar-mammary syndrome. European Journal of Medical Genetics, 2020, 63, 103615.	0.7	6
394	Phenotypic expansion of OTUD6Bâ€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1530-1531.	0.7	6
395	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. Brain and Development, 2021, 43, 380-388.	0.6	6
396	<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

#	Article	IF	Citations
397	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	0.7	6
398	Molecular autopsy by proxy in preconception counseling. Clinical Genetics, 2021, 100, 678-691.	1.0	6
399	Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. JCI Insight, 2022, 7, .	2.3	6
400	Bi-allelic loss-of-function variants in PPFIBP1 cause a neurodevelopmental disorder with microcephaly, epilepsy, and periventricular calcifications. American Journal of Human Genetics, 2022, 109, 1421-1435.	2.6	6
401	Vanishing white matter disease caused by EIF2B2 mutation with the presentation of an adrenoleukodystrophy phenotype. Gene, 2012, 496, 141-143.	1.0	5
402	A case of de Barsy syndrome with a severe eye phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2364-2366.	0.7	5
403	Crisponi/CISS1 syndrome: A case series. American Journal of Medical Genetics, Part A, 2016, 170, 1236-1241.	0.7	5
404	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
405	A novel ISLR2-linked autosomal recessive syndrome of congenital hydrocephalus, arthrogryposis and abdominal distension. Human Genetics, 2019, 138, 105-107.	1.8	5
406	Confirming the recessive inheritance of PERP â€related erythrokeratoderma. Clinical Genetics, 2020, 97, 661-665.	1.0	5
407	Two further cases of polyhydramnios, megalencephaly, and symptomatic epilepsy syndrome, caused by a truncating variant in <scp>STRADA</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 604-607.	0.7	5
408	Multiple Family Members With Delayed Cord Separtion and Combined Immunodeficiency With Novel Mutation in IKBKB. Frontiers in Pediatrics, 2020, 8, 9.	0.9	5
409	Further delineation of van den Endeâ€Gupta syndrome: Genetic heterogeneity and overlap with congenital heart defects and skeletal malformations syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2136-2149.	0.7	5
410	SARS-CoV-2â€“Related Acute Respiratory Distress Syndrome Uncovers a Patient with Severe Combined Immunodeficiency Disease. Journal of Clinical Immunology, 2021, 41, 1507-1510.	2.0	5
411	Confirming the involvement of PIEZO2 in the etiology of Mardenâ€“Walker syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 945-948.	0.7	5
412	A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1567-1582.	1.1	5
413	Recurrent miscarriage in a carrier of a balanced cytogenetically undetectable subtelomeric rearrangement: How many are we missing?. Prenatal Diagnosis, 2006, 26, 291-293.	1.1	4
414	Perturbation of the consensus activation site of endothelinâ€3 leads to Waardenburg syndrome type IV. American Journal of Medical Genetics, Part A, 2010, 152A, 1841-1843.	0.7	4

#	Article	IF	Citations
415	Trichorhinophalangeal syndrome: report of a novel familial TRPS1 mutation. Clinical Dysmorphology, 2010, 19, 98-100.	0.1	4
416	An autosomal recessive syndrome of severe cognitive impairment, dysmorphic facies and skeletal abnormalities maps to the long arm of chromosome 17. Clinical Genetics, 2011, 80, 489-492.	1.0	4
417	Simple and Efficient Identification of Rare Recessive Pathologically Important Sequence Variants from Next Generation Exome Sequence Data. Human Mutation, 2013, 34, 945-952.	1.1	4
418	Expanding the allelic disorders linked to TCTN1 to include Varadi syndrome (Orofaciodigital) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.7	4
419	A de novo splicing variant supports the candidacy of TLL1 in ASD pathogenesis. European Journal of Human Genetics, 2020, 28, 525-528.	1.4	4
420	A de novo mutation in FMR1 in a patient with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103763.	0.7	4
421	Bifid nose as the sole manifestation of <scp>BNAR</scp> syndrome, a <scp>FREM1</scp>â€related condition. Clinical Genetics, 2020, 98, 515-516.	1.0	4
422	Further delineation of METTL23â€associated intellectual disability. American Journal of Medical Genetics, Part A, 2020, 182, 785-791.	0.7	4
423	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
424	Successful hematopoietic stem cell transplantation in a 4-1BB deficient patient with EBV-induced lymphoproliferation. Clinical Immunology, 2021, 222, 108639.	1.4	4
425	<scp>CHEDDA</scp> syndrome is an underrecognized neurodevelopmental disorder with a highly restricted <scp>ATN1</scp> mutation spectrum. Clinical Genetics, 2021, 100, 468-477.	1.0	4
426	Progressive symmetrical erythrokeratoderma manifesting as harlequin-like ichthyosis with severe thrombocytopenia secondary to a homozygous 3-ketodihydrosphingosine reductase mutation. JAAD Case Reports, 2021, 14, 55-58.	0.4	4
427	A null founder variant in <scp>NPNT</scp>, encoding nephronectin, causes autosomal recessive renal agenesis. Clinical Genetics, 2022, 102, 61-65.	1.0	4
428	Biallelic POC1A variants cause syndromic severe insulin resistance with muscle cramps. European Journal of Endocrinology, 2022, 186, 543-552.	1.9	4
429	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. American Journal of Human Genetics, 2010, 87, 571.	2.6	3
430	Saudi genetic ophthalmology research: The local and global impact. Saudi Journal of Ophthalmology, 2010, 24, 109-110.	0.3	3
431	Study of consanguineous populations can improve the annotation of SNP databases. European Journal of Medical Genetics, 2011, 54, 118-120.	0.7	3
432	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. American Journal of Human Genetics, 2011, 88, 677.	2.6	3

#	Article	IF	Citations
433	Elevation deficiency in children with recessive RDH12-related retinopathy. Journal of AAPOS, 2015, 19, 568-570.	0.2	3
434	The alternatively spliced exon of <scp>COL5A1</scp> is mutated in autosomal recessive classical Ehlersâ€Ðanlos syndrome. Clinical Genetics, 2018, 93, 936-937.	1.0	3
435	The study of Lynch syndrome in a special population reveals a strong founder effect and an unusual mutational mechanism in familial adenomatous polyposis. Gut, 2020, 69, 2048-2049.	6.1	3
436	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
437	Mutations in phospholipase C eta-1 (PLCH1) are associated with holoprosencephaly. Journal of Medical Genetics, 2022, 59, 358-365.	1.5	3
438	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
439	Mutations in <scp>HID1</scp> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	2.8	3
440	Variants in LSM7 impair LSM complexes assembly, neurodevelopment in zebrafish and may be associated with an ultra-rare neurological disease. Human Genetics and Genomics Advances, 2021, 2, 100034.	1.0	3
441	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	1.7	3
442	<scp>PLACK</scp> syndrome is potentially treatable with intralipids. Clinical Genetics, 2021, 99, 572-576.	1.0	3
443	Homozygosity mapping: a game-changer for autosomal recessive diseases. Nature Reviews Genetics, 2021, , .	7.7	3
444	Bi-allelic variants inWNT7Bdisrupt the development of multiple organs in humans. Journal of Medical Genetics, 2023, 60, 294-300.	1.5	3
445	Trisomy 8 mosaicism in a patient with heterotaxia. Birth Defects Research Part A: Clinical and Molecular Teratology, 2005, 73, 58-60.	1.6	2
446	Smith-Lemli-Opitz syndrome in trisomy 13: How does the mix work?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2005, 73, 569-571.	1.6	2
447	Corneal enlargement without optic disk cupping in children with recessive CYP1B1 mutations. Journal of AAPOS, 2013, 17, 643-645.	0.2	2
448	Variable phenotypic expression of COG6 mutations. Journal of Medical Genetics, 2014, 51, 425.2-426.	1.5	2
449	A lethal phenotype associated with tissue plasminogen deficiency in humans. Human Genetics, 2016, 135, 1209-1211.	1.8	2
450	Joint laxity in homozygotes for severe POU1F1 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3356-3358.	0.7	2

#	Article	IF	Citations
451	Indigenous Arabs have an intermediate frequency of a Neanderthalâ€derived <scp>COVID</scp>â€19 risk haplotype compared with other world populations. Clinical Genetics, 2021, 99, 484-485.	1.0	2
452	A de novo <scp>ATXN2L</scp> variant in a child with developmental delay and macrocephaly. American Journal of Medical Genetics, Part A, 2021, 185, 949-951.	0.7	2
453	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471.	0.4	2
454	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	1.1	2
455	Recurrent spontaneous oocyte activation causes female infertility. Journal of Assisted Reproduction and Genetics, 2022, 39, 675.	1.2	2
456	A Biallelic Variant in FRA10AC1 Is Associated With Neurodevelopmental Disorder and Growth Retardation. Neurology: Genetics, 2022, 8, e200010.	0.9	2
457	Index of Suspicion. Pediatrics in Review, 2004, 25, 289-294.	0.2	1
458	FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. American Journal of Human Genetics, 2009, 85, 756.	2.6	1
459	Congenital hereditary endothelial dystrophy, not glaucoma, in a child with iris colobomas. Journal of AAPOS, 2016, 20, 370-372.	0.2	1
460	Reply to â€œan extremely severe phenotype due to WDR81 nonsense mutationsâ€• Annals of Neurology, 2017, 82, 651-651.	2.8	1
461	Vitamin B12 deficiency secondary to cobalamin F deficiency simulating dyskeratosis congenita. JAAD Case Reports, 2020, 6, 882-885.	0.4	1
462	Generation of iPSC lines (KAUSTi011-A, KAUSTi011-B) from a Saudi patient with epileptic encephalopathy carrying homozygous mutation in the GLP1R gene. Stem Cell Research, 2021, 50, 102148.	0.3	1
463	Hoarse voice in children as the presenting feature of <scp>ECM1</scp>â€related lipoid proteinosis. American Journal of Medical Genetics, Part A, 2021, 185, 3924-3925.	0.7	1
464	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. Human Genetics, 2021, 140, 1395-1401.	1.8	1
465	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 83, 425-427.	2.6	0
466	MGAT2â€CDG (CDGâ€IIa) and dysmorphism. American Journal of Medical Genetics, Part A, 2012, 158A, 2976-2976.	0.7	0
467	The journey of exome sequencing from disease mutations discovery to clinical genomics. Human Genetics, 2016, 135, 589-589.	1.8	0
468	Response to Yehia etÂal.. American Journal of Human Genetics, 2017, 100, 564-565.	2.6	0

#	Article	IF	Citations
469	Implications of mosaicism in variant interpretation: A case of a de novo homozygous NF1 variant. European Journal of Medical Genetics, 2021, 64, 104236.	0.7	0
470	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	2.6	0
471	A Diagnostic Approach for Neurogenetic Disorders in the Genome Era. , 2020, , 319-326.		0
472	Mitochondrial â€œdysmorphologyâ€•in variant classification. Human Genetics, 2022, 141, 55-64.	1.8	0
473	Hereditary hyperekplexia in Saudi Arabia. Pediatric Neurology, 2022, , .	1.0	0