Fowzan S Alkuraya

List of Publications by Year
in descending order

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473 papers	19,746 citations	11651 70 h-index	28297 105 g-index
516 all docs	516 docs citations	516 times ranked	26533 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.	6.4	375
2	Loss-of-function variant in DNASE1L3 causes a familial form of systemic lupus erythematosus. Nature Genetics, 2011, 43, 1186-1188.	21.4	366
3	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. Nature Genetics, 2016, 48, 1071-1076.	21.4	314
4	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
5	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	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.	2.9	232
7	Mutations in CCNO result in congenital mucociliary clearance disorder with reduced generation of multiple motile cilia. Nature Genetics, 2014, 46, 646-651.	21.4	232
8	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nature Genetics, 2011, 43, 197-203.	21.4	229
9	Autozygome-guided exome sequencing in retinal dystrophy patients reveals pathogenetic mutations and novel candidate disease genes. Genome Research, 2013, 23, 236-247.	5.5	226
10	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	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.	3.8	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.	6.2	204
13	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. American Journal of Human Genetics, 2011, 88, 536-547.	6.2	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.	3.8	195
15	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. Molecular Psychiatry, 2017, 22, 615-624.	7.9	187
16	Mutations in the RNA Granule Component TDRD7 Cause Cataract and Glaucoma. Science, 2011, 331, 1571-1576.	12.6	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.	6.2	184
18	High diagnostic yield of clinical exome sequencing in Middle Eastern patients with Mendelian disorders. Human Genetics, 2015, 134, 967-980.	3.8	168

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19	SUMO1 Haploinsufficiency Leads to Cleft Lip and Palate. Science, 2006, 313, 1751-1751.	12.6	165
20	Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes. Journal of Medical Genetics, 2012, 49, 234-241.	3.2	164
21	Recessive Mutations in ELOVL4 Cause Ichthyosis, Intellectual Disability, and Spastic Quadriplegia. American Journal of Human Genetics, 2011, 89, 745-750.	6.2	161
22	TLE6 mutation causes the earliest known human embryonic lethality. Genome Biology, 2015, 16, 240.	8.8	153
23	Novel CENPJ mutation causes Seckel syndrome. Journal of Medical Genetics, 2010, 47, 411-414.	3.2	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.	6.2	147
25	Mitochondrial phenylalanyl-tRNA synthetase mutations underlie fatal infantile Alpers encephalopathy. Human Molecular Genetics, 2012, 21, 4521-4529.	2.9	143
26	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	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.	2.9	134
28	Mutation in WDR4 impairs tRNA m7G46 methylation and causes a distinct form of microcephalic primordial dwarfism. Genome Biology, 2015, 16, 210.	8.8	132
29	Study of autosomal recessive osteogenesis imperfecta in Arabia reveals a novel locus defined byTMEM38Bmutation. Journal of Medical Genetics, 2012, 49, 630-635.	3.2	124
30	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	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.	6.2	121
32	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	8.8	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.	6.2	115
34	Mutations in EOGT Confirm the Genetic Heterogeneity of Autosomal-Recessive Adams-Oliver Syndrome. American Journal of Human Genetics, 2013, 92, 598-604.	6.2	114
35	A homozygous truncating mutation in PUS3 expands the role of tRNA modification in normal cognition. Human Genetics, 2016, 135, 707-713.	3.8	112
36	In search of triallelism in Bardetâ€“Biedl syndrome. European Journal of Human Genetics, 2012, 20, 420-427.	2.8	111

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37	The genetic landscape of familial congenital hydrocephalus. Annals of Neurology, 2017, 81, 890-897.	5.3	108
38	Autozygome decoded. Genetics in Medicine, 2010, 12, 765-771.	2.4	107
39	Homozygosity mapping: One more tool in the clinical geneticist's toolbox. Genetics in Medicine, 2010, 12, 236-239.	2.4	107
40	The application of next-generation sequencing in the autozygosity mapping of human recessive diseases. Human Genetics, 2013, 132, 1197-1211.	3.8	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.	6.2	105
42	Mutations in LRPAP1 Are Associated with Severe Myopia in Humans. American Journal of Human Genetics, 2013, 93, 313-320.	6.2	104
43	NFIA Haploinsufficiency Is Associated with a CNS Malformation Syndrome and Urinary Tract Defects. PLoS Genetics, 2007, 3, e80.	3.5	100
44	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 82, 712-722.	6.2	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.	6.2	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.	2.9	93
47	Mutation inADAT3, encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus. Journal of Medical Genetics, 2013, 50, 425-430.	3.2	91
48	Identification of embryonic lethal genes in humans by autozygosity mapping and exome sequencing in consanguineous families. Genome Biology, 2015, 16, 116.	8.8	91
49	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. Neuron, 2014, 84, 1240-1257.	8.1	89
50	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540.	3.8	89
51	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. Genetics in Medicine, 2016, 18, 554-562.	2.4	89
52	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	8.2	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.	6.2	86

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55	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	2.4	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.	3.1	84
57	Molecular autopsy in maternalâ€“fetal medicine. Genetics in Medicine, 2018, 20, 420-427.	2.4	84
58	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	27.0	84
59	Mutations in MEOX1, Encoding Mesenchyme Homeobox 1, Cause Klippel-Feil Anomaly. American Journal of Human Genetics, 2013, 92, 157-161.	6.2	82
60	Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. Genetics in Medicine, 2016, 18, 1244-1249.	2.4	82
61	Mutation in PHC1 implicates chromatin remodeling in primary microcephaly pathogenesis. Human Molecular Genetics, 2013, 22, 2200-2213.	2.9	81
62	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
63	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	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.	2.3	79
65	Ciliary GenesTBC1D32/C6orf170andSCLT1are Mutated in Patients with OFD Type IX. Human Mutation, 2014, 35, 36-40.	2.5	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.	6.2	77
67	Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. Genetics in Medicine, 2017, 19, 1144-1150.	2.4	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.	3.2	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.	3.2	75
70	Mutation inMPDZcauses severe congenital hydrocephalus. Journal of Medical Genetics, 2013, 50, 54-58.	3.2	75
71	3â€Methylglutaconic aciduriaâ€”lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	3.6	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.	6.2	74

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73	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. Human Genetics, 2017, 136, 205-225.	3.8	73
74	A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. Human Mutation, 2011, 32, 573-578.	2.5	72
75	Exome sequencing reveals a novel Fanconi group defined by XRCC2 mutation: Figure 1. Journal of Medical Genetics, 2012, 49, 184-186.	3.2	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.	2.4	71
77	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	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.	3.6	70
79	Deficiency of the Cytoskeletal Protein SPECC1L Leads to Oblique Facial Clefting. American Journal of Human Genetics, 2011, 89, 44-55.	6.2	70
80	POC1A Truncation Mutation Causes a Ciliopathy in Humans Characterized by Primordial Dwarfism. American Journal of Human Genetics, 2012, 91, 330-336.	6.2	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.	2.9	70
82	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. American Journal of Human Genetics, 2018, 103, 612-620.	6.2	70
83	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
84	Identification of ADAMTS18 as a gene mutated in Knobloch syndrome. Journal of Medical Genetics, 2011, 48, 597-601.	3.2	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.	3.4	68
86	The morbid genome of ciliopathies: an update. Genetics in Medicine, 2020, 22, 1051-1060.	2.4	68
87	A nullimorphic ERLIN2 mutation defines a complicated hereditary spastic paraplegia locus (SPG18). Neurogenetics, 2011, 12, 333-336.	1.4	67
88	Complementation of hypersensitivity to DNA interstrand crosslinking agents demonstrates thatXRCC2is a Fanconi anaemia gene. Journal of Medical Genetics, 2016, 53, 672-680.	3.2	66
89	Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. Human Genetics, 2016, 135, 1295-1298.	3.8	65
90	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	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.	6.2	65
92	LOXL3, encoding lysyl oxidase-like 3, is mutated in a family with autosomal recessive Stickler syndrome. Human Genetics, 2015, 134, 451-453.	3.8	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.	3.2	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.	1.2	63
95	The distinct ophthalmic phenotype of Knobloch syndrome in children. British Journal of Ophthalmology, 2012, 96, 890-895.	3.9	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.	2.5	63
97	Immunodeficiency and EBV-induced lymphoproliferation caused by 4-1BB deficiency. Journal of Allergy and Clinical Immunology, 2019, 144, 574-583.e5.	2.9	63
98	Mutations of KIF14 cause primary microcephaly by impairing cytokinesis. Annals of Neurology, 2017, 82, 562-577.	5.3	62
99	Genetics and genomic medicine in Saudi Arabia. Molecular Genetics & Genomic Medicine, 2014, 2, 369-378.	1.2	61
100	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	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.	2.5	60
103	International perspectives on the implementation of reproductive carrier screening. Prenatal Diagnosis, 2020, 40, 301-310.	2.3	60
104	Homozygous null mutation in ODZ3 causes microphthalmia in humans. Genetics in Medicine, 2012, 14, 900-904.	2.4	59
105	Female Infertility Caused by Mutations in the Oocyte-Specific Translational Repressor PATL2. American Journal of Human Genetics, 2017, 101, 603-608.	6.2	59
106	Increasing the sensitivity of clinical exome sequencing through improved filtration strategy. Genetics in Medicine, 2017, 19, 593-598.	2.4	59
107	EROS/CYBC1 mutations: Decreased NADPH oxidase function and chronic granulomatous disease. Journal of Allergy and Clinical Immunology, 2019, 143, 782-785.e1.	2.9	59
108	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	8.8	59

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109	A genomics approach to females with infertility and recurrent pregnancy loss. Human Genetics, 2020, 139, 605-613.	3.8	59
110	A genomics approach to male infertility. Genetics in Medicine, 2020, 22, 1967-1975.	2.4	57
111	Molecular characterization of Joubert syndrome in Saudi Arabia. Human Mutation, 2012, 33, 1423-1428.	2.5	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.	2.8	56
113	The Syndrome of Microcornea, Myopic Chorioretinal Atrophy, and Telecanthus (MMCAT) Is Caused by Mutations inADAMTS18. Human Mutation, 2013, 34, 1195-1199.	2.5	56
114	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	8.8	56
115	Genetic heterogeneity and evolutionary history of high-grade ovarian carcinoma and matched distant metastases. British Journal of Cancer, 2020, 122, 1219-1230.	6.4	56
116	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
117	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. Genetics in Medicine, 2016, 18, 686-695.	2.4	55
118	Biometric and Molecular Characterization of Clinically Diagnosed Posterior Microphthalmos. American Journal of Ophthalmology, 2013, 155, 361-372.e7.	3.3	54
119	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
120	Discovery of mutations for Mendelian disorders. Human Genetics, 2016, 135, 615-623.	3.8	53
121	PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. Human Genetics, 2019, 138, 231-239.	3.8	53
122	Identification of differentially expressed proteins in the aqueous humor of primary congenital glaucoma. Experimental Eye Research, 2011, 92, 67-75.	2.6	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.	6.2	51
124	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217.	6.2	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.	6.2	50
126	Severe CNS involvement in WWOX mutations: Description of five new cases. American Journal of Medical Genetics, Part A, 2015, 167, 3209-3213.	1.2	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.	2.4	49
128	Genomic analysis of pediatric cataract in Saudi Arabia reveals novel candidate disease genes. Genetics in Medicine, 2012, 14, 955-962.	2.4	49
129	Autozygosity Mapping with Exome Sequence Data. Human Mutation, 2013, 34, 50-56.	2.5	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.	1.2	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.	3.5	48
133	Revisiting the morbid genome of Mendelian disorders. Genome Biology, 2016, 17, 235.	8.8	48
134	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
135	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. American Journal of Human Genetics, 2010, 87, 306-307.	6.2	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.	1.4	47
137	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	2.9	47
138	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
139	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132.	6.2	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.	2.9	46
141	Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. American Journal of Human Genetics, 2013, 93, 555-560.	6.2	45
142	WNT1 mutation with recessive osteogenesis imperfecta and profound neurological phenotype. Journal of Medical Genetics, 2013, 50, 491-492.	3.2	45
143	On the phenotypic spectrum of serine biosynthesis defects. Journal of Inherited Metabolic Disease, 2016, 39, 373-381.	3.6	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.	3.8	45

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145	<scp>GLI3</scp>â€related polydactyly: a review. Clinical Genetics, 2017, 92, 457-466.	2.0	45
146	A human ciliopathy reveals essential functions for NEK10 in airway mucociliary clearance. Nature Medicine, 2020, 26, 244-251.	30.7	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.	2.0	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.	6.2	44
149	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
150	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	8.2	43
151	Lifting the lid on unborn lethal Mendelian phenotypes through exome sequencing. Genetics in Medicine, 2013, 15, 307-309.	2.4	42
152	Identification of a novel MKS locus defined byTMEM107mutation. Human Molecular Genetics, 2015, 24, 5211-5218.	2.9	42
153	Human knockout research: new horizons and opportunities. Trends in Genetics, 2015, 31, 108-115.	6.7	42
154	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	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.	1.2	41
157	C2orf37 mutational spectrum in Woodhouseâ€“Sakati syndrome patients. Clinical Genetics, 2010, 78, 585-590.	2.0	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.	6.2	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.	3.2	41
160	Genomic Profiling of Thyroid Cancer Reveals a Role for Thyroglobulin in Metastasis. American Journal of Human Genetics, 2016, 98, 1170-1180.	6.2	41
161	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
162	Absence of GP130 cytokine receptor signaling causes extended StÃ¼ve-Wiedemann syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	41

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163	Molecular Characterization of Newborn Glaucoma Including a Distinct Aniridic Phenotype. Ophthalmic Genetics, 2011, 32, 138-142.	1.2	40
164	A novel syndrome of hypohidrosis and intellectual disability is linked to COG6 deficiency. Journal of Medical Genetics, 2013, 50, 431-436.	3.2	40
165	Cell-Intrinsic Adaptation Arising from Chronic Ablation of a Key Rho GTPase Regulator. Developmental Cell, 2016, 39, 28-43.	7.0	40
166	An autosomal recessiveDNASE1L3-related autoimmune disease with unusual clinical presentation mimicking systemic lupus erythematosus. Lupus, 2017, 26, 768-772.	1.6	40
167	Phenotypic and Molecular Spectrum of Aicardi-GoutiÃ¨res Syndrome: A Study of 24 Patients. Pediatric Neurology, 2018, 78, 35-40.	2.1	40
168	A novel PTF1A mutation in a patient with severe pancreatic and cerebellar involvement. Clinical Genetics, 2011, 80, 196-198.	2.0	39
169	Functional analysis of BBS3 A89V that results in non-syndromic retinal degeneration. Human Molecular Genetics, 2011, 20, 1625-1632.	2.9	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.	3.8	38
171	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. American Journal of Human Genetics, 2017, 100, 969-977.	6.2	38
172	A null mutation in MICU2 causes abnormal mitochondrial calcium homeostasis and a severe neurodevelopmental disorder. Brain, 2017, 140, 2806-2813.	7.6	38
173	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. Clinical Genetics, 2018, 93, 1210-1222.	2.0	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.	1.8	37
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