Fowzan S Alkuraya

List of Publications by Year
in descending order

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Version: 2024-02-01

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	Bi-allelic variants inWNT7Bdisrupt the development of multiple organs in humans. Journal of Medical Genetics, 2023, 60, 294-300.	3.2	3
2	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
3	Mutations in phospholipase C eta-1 (PLCH1) are associated with holoprosencephaly. Journal of Medical Genetics, 2022, 59, 358-365.	3.2	3
4	ASTL is mutated in female infertility. Human Genetics, 2022, 141, 49-54.	3.8	16
5	Mitochondrial â€œdysmorphologyâ€•in variant classification. Human Genetics, 2022, 141, 55-64.	3.8	0
6	Insight into ALKBH8-related intellectual developmental disability based on the first pathogenic missense variant. Human Genetics, 2022, 141, 209-215.	3.8	7
7	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.	2.5	9
8	Recurrent spontaneous oocyte activation causes female infertility. Journal of Assisted Reproduction and Genetics, 2022, 39, 675.	2.5	2
9	A null founder variant in <scp>NPNT</scp>, encoding nephronectin, causes autosomal recessive renal agenesis. Clinical Genetics, 2022, 102, 61-65.	2.0	4
10	THUMPD1 bi-allelic variants cause loss of tRNA acetylation and a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 587-600.	6.2	19
11	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	7.6	8
12	Biallelic POC1A variants cause syndromic severe insulin resistance with muscle cramps. European Journal of Endocrinology, 2022, 186, 543-552.	3.7	4
13	A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1567-1582.	2.4	5
14	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
15	Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. JCI Insight, 2022, 7, .	5.0	6
16	Hereditary hyperekplexia in Saudi Arabia. Pediatric Neurology, 2022, , .	2.1	0
17	A Biallelic Variant in FRA10AC1 Is Associated With Neurodevelopmental Disorder and Growth Retardation. Neurology: Genetics, 2022, 8, e200010.	1.9	2
18	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.	6.2	6

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19	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.	1.2	5
20	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.	1.2	7
21	New paradigms of USP53 disease: normal GGT cholestasis, BRIC, cholangiopathy, and responsiveness to rifampicin. Journal of Human Genetics, 2021, 66, 151-159.	2.3	21
22	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.	2.0	2
23	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.	1.2	4
24	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. Brain and Development, 2021, 43, 380-388.	1.1	6
25	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.	1.2	2
26	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471.	0.8	2
27	Population structure of indigenous inhabitants of Arabia. PLoS Genetics, 2021, 17, e1009210.	3.5	14
28	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
29	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	2.4	2
30	Successful hematopoietic stem cell transplantation in a 4-1BB deficient patient with EBV-induced lymphoproliferation. Clinical Immunology, 2021, 222, 108639.	3.2	4
31	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.	0.7	1
32	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.	6.2	37
33	How the human genome transformed study of rare diseases. Nature, 2021, 590, 218-219.	27.8	12
34	Further delineation of <scp>SMG9</scp>â€related heart and brain malformation syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1624-1630.	1.2	3
35	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.	1.2	34
36	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.	1.2	5

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37	<scp>MYH1</scp> is a candidate gene for recurrent rhabdomyolysis in humans. American Journal of Medical Genetics, Part A, 2021, 185, 2131-2135.	1.2	8
38	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.	2.4	30
39	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18
40	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.	2.4	22
41	Genetic testing results of children suspected to have Stickler syndrome type collagenopathy after ocular examination. Molecular Genetics & Genomic Medicine, 2021, 9, e1628.	1.2	3
42	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.	3.8	13
43	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
44	Mutations in <scp>HID1</scp> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	5.3	3
45	SARS-CoV-2â€“Related Acute Respiratory Distress Syndrome Uncovers a Patient with Severe Combined Immunodeficiency Disease. Journal of Clinical Immunology, 2021, 41, 1507-1510.	3.8	5
46	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	27.0	84
47	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.	1.2	1
48	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
49	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	2.4	9
50	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
51	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	2.4	11
52	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. Human Genetics, 2021, 140, 1395-1401.	3.8	1
53	<scp>CHEDDA</scp> syndrome is an underrecognized neurodevelopmental disorder with a highly restricted <scp>ATN1</scp> mutation spectrum. Clinical Genetics, 2021, 100, 468-477.	2.0	4
54	<scp>PLXNA2</scp> as a candidate gene in patients with intellectual disability. American Journal of Medical Genetics, Part A, 2021, 185, 3859-3865.	1.2	6

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55	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.	1.7	3
56	Missense NAA20 variantsimpairing the NatB protein N-terminal acetyltransferase cause autosomal recessivedevelopmental delay, intellectual disability, and microcephaly. Genetics in Medicine, 2021, 23, 2213-2218.	2.4	11
57	Residual risk for additional recessive diseases in consanguineous couples. Genetics in Medicine, 2021, 23, 2448-2454.	2.4	15
58	Implications of mosaicism in variant interpretation: A case of a de novo homozygous NF1 variant. European Journal of Medical Genetics, 2021, 64, 104236.	1.3	0
59	Mutations in TP73 cause impaired mucociliary clearance and lissencephaly. American Journal of Human Genetics, 2021, 108, 1318-1329.	6.2	15
60	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	1.2	6
61	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.	1.2	12
62	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.	0.8	4
63	Molecular autopsy by proxy in preconception counseling. Clinical Genetics, 2021, 100, 678-691.	2.0	6
64	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	3.9	3
65	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
66	<scp>PLACK</scp> syndrome is potentially treatable with intralipids. Clinical Genetics, 2021, 99, 572-576.	2.0	3
67	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. Genome Medicine, 2021, 13, 161.	8.2	13
68	Confirming the involvement of PIEZO2 in the etiology of Mardenâ€“Walker syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 945-948.	1.2	5
69	Homozygosity mapping: a game-changer for autosomal recessive diseases. Nature Reviews Genetics, 2021, , .	16.3	3
70	Genomic medicine in the Middle East. Genome Medicine, 2021, 13, 184.	8.2	12
71	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.	1.3	6
72	A de novo splicing variant supports the candidacy of TLL1 in ASD pathogenesis. European Journal of Human Genetics, 2020, 28, 525-528.	2.8	4

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73	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. Hepatology, 2020, 71, 2067-2079.	7.3	28
74	Whole-Exome Sequencing of Matched Primary and Metastatic Papillary Thyroid Cancer. Thyroid, 2020, 30, 42-56.	4.5	31
75	Rhegmatogenous Retinal Detachment in Nonsyndromic High Myopia Associated with Recessive Mutations in LRPAP1. Ophthalmology Retina, 2020, 4, 77-83.	2.4	7
76	A de novo mutation in FMR1 in a patient with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103763.	1.3	4
77	An intellectual disabilityâ€associated missense variant in TRMT1 impairs tRNA modification and reconstitution of enzymatic activity. Human Mutation, 2020, 41, 600-607.	2.5	24
78	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.	1.2	12
79	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. Clinical Genetics, 2020, 97, 447-456.	2.0	7
80	Confirming the recessive inheritance of PERP â€related erythrokeratoderma. Clinical Genetics, 2020, 97, 661-665.	2.0	5
81	International perspectives on the implementation of reproductive carrier screening. Prenatal Diagnosis, 2020, 40, 301-310.	2.3	60
82	Further delineation of <scp>HIDEA</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2999-3006.	1.2	7
83	Clonal Evolution and Timing of Metastatic Colorectal Cancer. Cancers, 2020, 12, 2938.	3.7	9
84	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.	6.2	20
85	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. Genetics in Medicine, 2020, 22, 2071-2080.	2.4	7
86	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
87	A genomics approach to male infertility. Genetics in Medicine, 2020, 22, 1967-1975.	2.4	57
88	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. Brain, 2020, 143, 2911-2928.	7.6	13
89	Vitamin B12 deficiency secondary to cobalamin F deficiency simulating dyskeratosis congenita. JAAD Case Reports, 2020, 6, 882-885.	0.8	1
90	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.	2.9	15

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91	Bifid nose as the sole manifestation of <scp>BNAR</scp> syndrome, a <scp>FREM1</scp>â€related condition. Clinical Genetics, 2020, 98, 515-516.	2.0	4
92	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	6.2	18
93	Absence of GP130 cytokine receptor signaling causes extended StÃ¼ve-Wiedemann syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	41
94	The natural history of infantile neuroaxonal dystrophy. Orphanet Journal of Rare Diseases, 2020, 15, 109.	2.7	11
95	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. Human Genetics, 2020, 139, 1273-1283.	3.8	16
96	DALRD3 encodes a protein mutated in epileptic encephalopathy that targets arginine tRNAs for 3-methylcytosine modification. Nature Communications, 2020, 11, 2510.	12.8	31
97	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.	12.1	3
98	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
99	Confirming <scp>TBC1D32</scp>â€related ciliopathy in humans. American Journal of Medical Genetics, Part A, 2020, 182, 1985-1987.	1.2	9
100	Expanding the clinical and phenotypic heterogeneity associated with biallelic variants in ACO2. Annals of Clinical and Translational Neurology, 2020, 7, 1013-1028.	3.7	8
101	A Case with Purine Nucleoside Phosphorylase Deficiency Suffering from Late-Onset Systemic Lupus Erythematosus and Lymphoma. Journal of Clinical Immunology, 2020, 40, 833-839.	3.8	16
102	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	8.8	59
103	A genomics approach to females with infertility and recurrent pregnancy loss. Human Genetics, 2020, 139, 605-613.	3.8	59
104	Phenotypic expansion of OTUD6Bâ€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1530-1531.	1.2	6
105	Multiple Family Members With Delayed Cord Separtion and Combined Immunodeficiency With Novel Mutation in IKBKB. Frontiers in Pediatrics, 2020, 8, 9.	1.9	5
106	CNP deficiency causes severe hypomyelinating leukodystrophy in humans. Human Genetics, 2020, 139, 615-622.	3.8	18
107	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
108	Further delineation of METTL23â€associated intellectual disability. American Journal of Medical Genetics, Part A, 2020, 182, 785-791.	1.2	4

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109	The morbid genome of ciliopathies: an update. Genetics in Medicine, 2020, 22, 1051-1060.	2.4	68
110	Phenome-based approach identifies RIC1-linked Mendelian syndrome through zebrafish models, biobank associations and clinical studies. Nature Medicine, 2020, 26, 98-109.	30.7	32
111	Recessive mutations in SCYL2 cause a novel syndromic form of arthrogryposis in humans. Human Genetics, 2020, 139, 513-519.	3.8	10
112	A human ciliopathy reveals essential functions for NEK10 in airway mucociliary clearance. Nature Medicine, 2020, 26, 244-251.	30.7	45
113	A familial PLCB4 mutation causing auriculocondylar syndrome 2 with variable severity. European Journal of Medical Genetics, 2020, 63, 103917.	1.3	11
114	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.	6.2	17
115	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
116	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	8.2	43
117	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Frontiers in Genetics, 2020, 11, 580484.	2.3	13
118	A Diagnostic Approach for Neurogenetic Disorders in the Genome Era. , 2020, , 319-326.		0
119	De novo truncating variants in WHSC1 recapitulate the Wolfâ€“Hirschhorn (4p16.3 microdeletion) syndrome phenotype. Genetics in Medicine, 2019, 21, 185-188.	2.4	24
120	Biallelic variants in CTU2 cause DREAMâ€PL syndrome and impair thiolation of tRNA wobble U34. Human Mutation, 2019, 40, 2108-2120.	2.5	25
121	Formation of tRNA Wobble Inosine in Humans Is Disrupted by a Millennia-Old Mutation Causing Intellectual Disability. Molecular and Cellular Biology, 2019, 39, .	2.3	25
122	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.	6.2	37
123	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.	6.2	34
124	Evolution and Impact of Subclonal Mutations in Papillary Thyroid Cancer. American Journal of Human Genetics, 2019, 105, 959-973.	6.2	22
125	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	61
126	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

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127	MDH1 deficiency is a metabolic disorder of the malateâ€“aspartate shuttle associated with early onset severe encephalopathy. Human Genetics, 2019, 138, 1247-1257.	3.8	31
128	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
129	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546.	1.2	5
130	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.	2.5	10
131	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
132	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
133	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
134	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
135	Recessive Truncating Mutations in ALKBH8 Cause Intellectual Disability and Severe Impairment of Wobble Uridine Modification. American Journal of Human Genetics, 2019, 104, 1202-1209.	6.2	34
136	The landscape of early infantile epileptic encephalopathy in a consanguineous population. Seizure: the Journal of the British Epilepsy Association, 2019, 69, 154-172.	2.0	34
137	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.	6.2	23
138	Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellarâ€vermis hypoplasia. American Journal of Medical Genetics, Part A, 2019, 179, 1053-1057.	1.2	15
139	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.	1.2	32
140	Immunodeficiency and EBV-induced lymphoproliferation caused by 4-1BB deficiency. Journal of Allergy and Clinical Immunology, 2019, 144, 574-583.e5.	2.9	63
141	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.	6.2	19
142	PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. Human Genetics, 2019, 138, 231-239.	3.8	53
143	NUP214 deficiency causes severe encephalopathy and microcephaly in humans. Human Genetics, 2019, 138, 221-229.	3.8	17
144	Patterns of neurological manifestations in Woodhouse-Sakati Syndrome. Parkinsonism and Related Disorders, 2019, 69, 99-103.	2.2	15

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145	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.	6.2	34
146	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	2.4	85
147	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	2.0	12
148	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.	1.2	20
149	Immunologic reconstitution following hematopoietic stem cell transplantation despite lymph node paucity in NF-Î°Bâ€“inducing kinase deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 1240-1243.e4.	2.9	6
150	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
151	A novel ISLR2-linked autosomal recessive syndrome of congenital hydrocephalus, arthrogryposis and abdominal distension. Human Genetics, 2019, 138, 105-107.	3.8	5
152	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
153	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
154	Congenital glaucoma and CYP1B1: an old story revisited. Human Genetics, 2019, 138, 1043-1049.	3.8	29
155	MED12is recurrently mutated in Middle Eastern colorectal cancer. Gut, 2018, 67, gutjnl-2016-313334.	12.1	12
156	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
157	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. Clinical Genetics, 2018, 93, 1210-1222.	2.0	38
158	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 715-721.	1.2	7
159	Phenotypic characterization of KCTD3â€related developmental epileptic encephalopathy. Clinical Genetics, 2018, 93, 1081-1086.	2.0	17
160	Identification of a novel lethal form of autosomal recessive ichthyosis caused by UDPâ€glucose ceramide glucosyltransferase deficiency. Clinical Genetics, 2018, 93, 1252-1253.	2.0	17
161	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.	5.3	9
162	The alternatively spliced exon of <scp>COL5A1</scp> is mutated in autosomal recessive classical Ehlersâ€Ðanlos syndrome. Clinical Genetics, 2018, 93, 936-937.	2.0	3

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163	Expanding the phenotype of SLC25A42â€associated mitochondrial encephalomyopathy. Clinical Genetics, 2018, 93, 1097-1102.	2.0	24
164	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
165	Elsahyâ€“Waters syndrome is caused by biallelic mutations in CDH11. American Journal of Medical Genetics, Part A, 2018, 176, 477-482.	1.2	18
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