

# Fowzan S Alkuraya

## List of Publications by Year in descending order

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473  
papers

19,746  
citations

13332

70  
h-index

32181

105  
g-index

516  
all docs

516  
docs citations

516  
times ranked

28526  
citing authors

#	ARTICLE	IF	CITATIONS
1	Bi-allelic variants in <i>WNT7B</i> disrupt the development of multiple organs in humans. <i>Journal of Medical Genetics</i> , 2023, 60, 294-300.	1.5	3
2	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	3.7	17
3	Mutations in phospholipase C eta-1 ( <i>PLCH1</i> ) are associated with holoprosencephaly. <i>Journal of Medical Genetics</i> , 2022, 59, 358-365.	1.5	3
4	ASTL is mutated in female infertility. <i>Human Genetics</i> , 2022, 141, 49-54.	1.8	16
5	Mitochondrial "œdysmorphology" in variant classification. <i>Human Genetics</i> , 2022, 141, 55-64.	1.8	0
6	Insight into ALKBH8-related intellectual developmental disability based on the first pathogenic missense variant. <i>Human Genetics</i> , 2022, 141, 209-215.	1.8	7
7	Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. <i>Human Mutation</i> , 2022, 43, 403-419.	1.1	9
8	Recurrent spontaneous oocyte activation causes female infertility. <i>Journal of Assisted Reproduction and Genetics</i> , 2022, 39, 675.	1.2	2
9	A null founder variant in <i>NPNT</i> , encoding nephronectin, causes autosomal recessive renal agenesis. <i>Clinical Genetics</i> , 2022, 102, 61-65.	1.0	4
10	THUMPD1 bi-allelic variants cause loss of tRNA acetylation and a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2022, 109, 587-600.	2.6	19
11	Biallelic <i>ADAM22</i> pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. <i>Brain</i> , 2022, 145, 2301-2312.	3.7	8
12	Biallelic <i>POC1A</i> variants cause syndromic severe insulin resistance with muscle cramps. <i>European Journal of Endocrinology</i> , 2022, 186, 543-552.	1.9	4
13	A novel <i>DPH5</i> -related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 1567-1582.	1.1	5
14	The Gene Curation Coalition: A global effort to harmonize gene "disease evidence resources. <i>Genetics in Medicine</i> , 2022, 24, 1732-1742.	1.1	56
15	Hypomorphic <i>GINS3</i> variants alter DNA replication and cause Meier-Gorlin syndrome. <i>JCI Insight</i> , 2022, 7, .	2.3	6
16	Hereditary hyperekplexia in Saudi Arabia. <i>Pediatric Neurology</i> , 2022, , .	1.0	0
17	A Biallelic Variant in <i>FRA10AC1</i> Is Associated With Neurodevelopmental Disorder and Growth Retardation. <i>Neurology: Genetics</i> , 2022, 8, e200010.	0.9	2
18	Bi-allelic loss-of-function variants in <i>PPFIBP1</i> cause a neurodevelopmental disorder with microcephaly, epilepsy, and periventricular calcifications. <i>American Journal of Human Genetics</i> , 2022, 109, 1421-1435.	2.6	6

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19	Two further cases of polyhydramnios, megalencephaly, and symptomatic epilepsy syndrome, caused by a truncating variant in <i>STRADA</i> . American Journal of Medical Genetics, Part A, 2021, 185, 604-607.	0.7	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.	0.7	7
21	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
22	Indigenous Arabs have an intermediate frequency of a Neanderthal-derived <i>COVID-19</i> risk haplotype compared with other world populations. Clinical Genetics, 2021, 99, 484-485.	1.0	2
23	Further delineation of <i>MYO18B</i> -related autosomal recessive <i>Klippel-Feil</i> syndrome with myopathy and facial dysmorphism. American Journal of Medical Genetics, Part A, 2021, 185, 370-376.	0.7	4
24	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. Brain and Development, 2021, 43, 380-388.	0.6	6
25	A de novo <i>ATXN2L</i> variant in a child with developmental delay and macrocephaly. American Journal of Medical Genetics, Part A, 2021, 185, 949-951.	0.7	2
26	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471.	0.4	2
27	Population structure of indigenous inhabitants of Arabia. PLoS Genetics, 2021, 17, e1009210.	1.5	14
28	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	2.6	71
29	Biallelic <i>UBE4A</i> loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	1.1	2
30	Successful hematopoietic stem cell transplantation in a 4-1BB deficient patient with EBV-induced lymphoproliferation. Clinical Immunology, 2021, 222, 108639.	1.4	4
31	Generation of iPSC lines (KAUSTi011-A, KAUSTi011-B) from a Saudi patient with epileptic encephalopathy carrying homozygous mutation in the <i>GLP1R</i> gene. Stem Cell Research, 2021, 50, 102148.	0.3	1
32	<i>SCUBE3</i> 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
33	How the human genome transformed study of rare diseases. Nature, 2021, 590, 218-219.	13.7	12
34	Further delineation of <i>SMG9</i> -related heart and brain malformation syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1624-1630.	0.7	3
35	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	0.7	34
36	Further delineation of van den Ende-C 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

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37	<sc><i>MYH1</i></sc> is a candidate gene for recurrent rhabdomyolysis in humans. American Journal of Medical Genetics, Part A, 2021, 185, 2131-2135.	0.7	8
38	Combining exome/genome sequencing with data repository analysis reveals novel gene-disease associations for a wide range of genetic disorders. Genetics in Medicine, 2021, 23, 1551-1568.	1.1	30
39	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> -associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	1.1	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.	1.1	22
41	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
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.	1.8	13
43	Biallelic variants in <i>HPDL</i> cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
44	Mutations in <sc><i>HID1</i></sc> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	2.8	3
45	SARS-CoV-2-Related Acute Respiratory Distress Syndrome Uncovers a Patient with Severe Combined Immunodeficiency Disease. Journal of Clinical Immunology, 2021, 41, 1507-1510.	2.0	5
46	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	13.9	84
47	Hoarse voice in children as the presenting feature of <sc><i>ECM1</i></sc>-related lipoid proteinosis. American Journal of Medical Genetics, Part A, 2021, 185, 3924-3925.	0.7	1
48	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
49	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
50	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	2.6	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.	1.1	11
52	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. Human Genetics, 2021, 140, 1395-1401.	1.8	1
53	<sc>CHEDDA</sc> syndrome is an underrecognized neurodevelopmental disorder with a highly restricted <sc><i>ATN1</i></sc> mutation spectrum. Clinical Genetics, 2021, 100, 468-477.	1.0	4
54	<sc><i>PLXNA2</i></sc> as a candidate gene in patients with intellectual disability. American Journal of Medical Genetics, Part A, 2021, 185, 3859-3865.	0.7	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. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100034.	1.0	3
56	Missense NAA20 variants impairing the NatB protein N-terminal acetyltransferase cause autosomal recessive developmental delay, intellectual disability, and microcephaly. <i>Genetics in Medicine</i> , 2021, 23, 2213-2218.	1.1	11
57	Residual risk for additional recessive diseases in consanguineous couples. <i>Genetics in Medicine</i> , 2021, 23, 2448-2454.	1.1	15
58	Implications of mosaicism in variant interpretation: A case of a de novo homozygous NF1 variant. <i>European Journal of Medical Genetics</i> , 2021, 64, 104236.	0.7	0
59	Mutations in TP73 cause impaired mucociliary clearance and lissencephaly. <i>American Journal of Human Genetics</i> , 2021, 108, 1318-1329.	2.6	15
60	Expanding the <sc><i>KIF4A</i></sc> associated phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3728-3739.	0.7	6
61	Expanding the phenotype of <sc><i>ASXL3</i></sc> related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <sc><i>ASXL3</i></sc>. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3446-3458.	0.7	12
62	Progressive symmetrical erythrokeratoderma manifesting as harlequin-like ichthyosis with severe thrombocytopenia secondary to a homozygous 3-ketodihydrosphingosine reductase mutation. <i>JAAD Case Reports</i> , 2021, 14, 55-58.	0.4	4
63	Molecular autopsy by proxy in preconception counseling. <i>Clinical Genetics</i> , 2021, 100, 678-691.	1.0	6
64	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. <i>Journal of Molecular Medicine</i> , 2021, 99, 1755-1768.	1.7	3
65	Response to Hamosh et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1809-1810.	2.6	0
66	<sc>PLACK</sc> syndrome is potentially treatable with intralipids. <i>Clinical Genetics</i> , 2021, 99, 572-576.	1.0	3
67	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. <i>Genome Medicine</i> , 2021, 13, 161.	3.6	13
68	Confirming the involvement of PIEZO2 in the etiology of Marden-Walker syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 945-948.	0.7	5
69	Homozygosity mapping: a game-changer for autosomal recessive diseases. <i>Nature Reviews Genetics</i> , 2021, , .	7.7	3
70	Genomic medicine in the Middle East. <i>Genome Medicine</i> , 2021, 13, 184.	3.6	12
71	A de novo TBX3 mutation presenting as dorsalization of the little fingers: A forme fruste phenotype of ulnar-mammary syndrome. <i>European Journal of Medical Genetics</i> , 2020, 63, 103615.	0.7	6
72	A de novo splicing variant supports the candidacy of TLL1 in ASD pathogenesis. <i>European Journal of Human Genetics</i> , 2020, 28, 525-528.	1.4	4

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73	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. <i>Hepatology</i> , 2020, 71, 2067-2079.	3.6	28
74	Whole-Exome Sequencing of Matched Primary and Metastatic Papillary Thyroid Cancer. <i>Thyroid</i> , 2020, 30, 42-56.	2.4	31
75	Rhegmatogenous Retinal Detachment in Nonsyndromic High Myopia Associated with Recessive Mutations in LRPAP1. <i>Ophthalmology Retina</i> , 2020, 4, 77-83.	1.2	7
76	A de novo mutation in FMR1 in a patient with intellectual disability. <i>European Journal of Medical Genetics</i> , 2020, 63, 103763.	0.7	4
77	An intellectual disability-associated missense variant in TRMT1 impairs tRNA modification and reconstitution of enzymatic activity. <i>Human Mutation</i> , 2020, 41, 600-607.	1.1	24
78	A novel truncating variant in ring finger protein 113A ( <i>RNF113A</i> ) confirms the association of this gene with X-linked trichothiodystrophy. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 513-520.	0.7	12
79	Phenotypic delineation of the retinal arterial macroaneurysms with supra-valvular pulmonic stenosis syndrome. <i>Clinical Genetics</i> , 2020, 97, 447-456.	1.0	7
80	Confirming the recessive inheritance of PERP-related erythrokeratoderma. <i>Clinical Genetics</i> , 2020, 97, 661-665.	1.0	5
81	International perspectives on the implementation of reproductive carrier screening. <i>Prenatal Diagnosis</i> , 2020, 40, 301-310.	1.1	60
82	Further delineation of HIDEA syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2999-3006.	0.7	7
83	Clonal Evolution and Timing of Metastatic Colorectal Cancer. <i>Cancers</i> , 2020, 12, 2938.	1.7	9
84	Recessive, Deleterious Variants in SMG8 Expand the Role of Nonsense-Mediated Decay in Developmental Disorders in Humans. <i>American Journal of Human Genetics</i> , 2020, 107, 1178-1185.	2.6	20
85	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. <i>Genetics in Medicine</i> , 2020, 22, 2071-2080.	1.1	7
86	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. <i>Brain</i> , 2020, 143, 2388-2397.	3.7	28
87	A genomics approach to male infertility. <i>Genetics in Medicine</i> , 2020, 22, 1967-1975.	1.1	57
88	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. <i>Brain</i> , 2020, 143, 2911-2928.	3.7	13
89	Vitamin B12 deficiency secondary to cobalamin F deficiency simulating dyskeratosis congenita. <i>JAAD Case Reports</i> , 2020, 6, 882-885.	0.4	1
90	Biallelic variants in the small optic lobe calpain CAPN15 are associated with congenital eye anomalies, deafness and other neurodevelopmental deficits. <i>Human Molecular Genetics</i> , 2020, 29, 3054-3063.	1.4	15

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91	Bifid nose as the sole manifestation of <i>BNAR</i> syndrome, a <i>FREM1</i> -related condition. <i>Clinical Genetics</i> , 2020, 98, 515-516.	1.0	4
92	<i>NCKAP1</i> Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. <i>American Journal of Human Genetics</i> , 2020, 107, 963-976.	2.6	18
93	Absence of GP130 cytokine receptor signaling causes extended <i>Stã¼ve-Wiedemann</i> syndrome. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	41
94	The natural history of infantile neuroaxonal dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 109.	1.2	11
95	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. <i>Human Genetics</i> , 2020, 139, 1273-1283.	1.8	16
96	<i>DALRD3</i> encodes a protein mutated in epileptic encephalopathy that targets arginine tRNAs for 3-methylcytosine modification. <i>Nature Communications</i> , 2020, 11, 2510.	5.8	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. <i>Gut</i> , 2020, 69, 2048-2049.	6.1	3
98	Biallelic <i>MFSD2A</i> variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. <i>European Journal of Human Genetics</i> , 2020, 28, 1509-1519.	1.4	21
99	Confirming <i>TBC1D32</i> -related ciliopathy in humans. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1985-1987.	0.7	9
100	Expanding the clinical and phenotypic heterogeneity associated with biallelic variants in <i>ACO2</i> . <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1013-1028.	1.7	8
101	A Case with Purine Nucleoside Phosphorylase Deficiency Suffering from Late-Onset Systemic Lupus Erythematosus and Lymphoma. <i>Journal of Clinical Immunology</i> , 2020, 40, 833-839.	2.0	16
102	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. <i>Genome Biology</i> , 2020, 21, 145.	3.8	59
103	A genomics approach to females with infertility and recurrent pregnancy loss. <i>Human Genetics</i> , 2020, 139, 605-613.	1.8	59
104	Phenotypic expansion of <i>OTUD6B</i> -related syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1530-1531.	0.7	6
105	Multiple Family Members With Delayed Cord Separation and Combined Immunodeficiency With Novel Mutation in <i>IKBKB</i> . <i>Frontiers in Pediatrics</i> , 2020, 8, 9.	0.9	5
106	<i>CNP</i> deficiency causes severe hypomyelinating leukodystrophy in humans. <i>Human Genetics</i> , 2020, 139, 615-622.	1.8	18
107	Genetic heterogeneity and evolutionary history of high-grade ovarian carcinoma and matched distant metastases. <i>British Journal of Cancer</i> , 2020, 122, 1219-1230.	2.9	56
108	Further delineation of <i>METTL23</i> -associated intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 785-791.	0.7	4

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109	The morbid genome of ciliopathies: an update. <i>Genetics in Medicine</i> , 2020, 22, 1051-1060.	1.1	68
110	Phenome-based approach identifies RIC1-linked Mendelian syndrome through zebrafish models, biobank associations and clinical studies. <i>Nature Medicine</i> , 2020, 26, 98-109.	15.2	32
111	Recessive mutations in SCYL2 cause a novel syndromic form of arthrogryposis in humans. <i>Human Genetics</i> , 2020, 139, 513-519.	1.8	10
112	A human ciliopathy reveals essential functions for NEK10 in airway mucociliary clearance. <i>Nature Medicine</i> , 2020, 26, 244-251.	15.2	45
113	A familial PLCB4 mutation causing auriculocondylar syndrome 2 with variable severity. <i>European Journal of Medical Genetics</i> , 2020, 63, 103917.	0.7	11
114	Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. <i>American Journal of Human Genetics</i> , 2020, 106, 246-255.	2.6	17
115	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. <i>Nature Communications</i> , 2020, 11, 595.	5.8	35
116	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. <i>Journal of Clinical Investigation</i> , 2020, 130, 4423-4439.	3.9	43
117	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. <i>Frontiers in Genetics</i> , 2020, 11, 580484.	1.1	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. <i>Genetics in Medicine</i> , 2019, 21, 185-188.	1.1	24
120	Biallelic variants in <i>CTU2</i> cause DREAMâ€“PL syndrome and impair thiolation of tRNA wobble U34. <i>Human Mutation</i> , 2019, 40, 2108-2120.	1.1	25
121	Formation of tRNA Wobble Inosine in Humans Is Disrupted by a Millennia-Old Mutation Causing Intellectual Disability. <i>Molecular and Cellular Biology</i> , 2019, 39, .	1.1	25
122	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. <i>American Journal of Human Genetics</i> , 2019, 105, 384-394.	2.6	37
123	Homozygous Loss-of-Function Mutations in AP1B1, Encoding Beta-1 Subunit of Adaptor-Related Protein Complex 1, Cause MEDNIK-like Syndrome. <i>American Journal of Human Genetics</i> , 2019, 105, 1016-1022.	2.6	34
124	Evolution and Impact of Subclonal Mutations in Papillary Thyroid Cancer. <i>American Journal of Human Genetics</i> , 2019, 105, 959-973.	2.6	22
125	Redefining the Etiologic Landscape of Cerebellar Malformations. <i>American Journal of Human Genetics</i> , 2019, 105, 606-615.	2.6	61
126	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. <i>American Journal of Human Genetics</i> , 2019, 105, 689-705.	2.6	48



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127	MDH1 deficiency is a metabolic disorder of the malate-aspartate shuttle associated with early onset severe encephalopathy. <i>Human Genetics</i> , 2019, 138, 1247-1257.	1.8	31
128	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. <i>Brain</i> , 2019, 142, 2948-2964.	3.7	43
129	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1543-1546.	0.7	5
130	Homozygous loss-of-function variants of <i>TASP1</i> , 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. <i>Human Mutation</i> , 2019, 40, 1985-1992.	1.1	10
131	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019, 10, 611.	1.1	14
132	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. <i>Human Molecular Genetics</i> , 2019, 28, 2900-2919.	1.4	46
133	<i>PDXK</i> mutations cause polyneuropathy responsive to pyridoxal 5-phosphate supplementation. <i>Annals of Neurology</i> , 2019, 86, 225-240.	2.8	54
134	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. <i>American Journal of Human Genetics</i> , 2019, 104, 1182-1201.	2.6	184
135	Recessive Truncating Mutations in <i>ALKBH8</i> Cause Intellectual Disability and Severe Impairment of Wobble Uracil Modification. <i>American Journal of Human Genetics</i> , 2019, 104, 1202-1209.	2.6	34
136	The landscape of early infantile epileptic encephalopathy in a consanguineous population. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 69, 154-172.	0.9	34
137	Bi-allelic Mutations in <i>FAM149B1</i> Cause Abnormal Primary Cilium and a Range of Ciliopathy Phenotypes in Humans. <i>American Journal of Human Genetics</i> , 2019, 104, 731-737.	2.6	23
138	Biallelic novel missense <i>HHAT</i> variant causes syndromic microcephaly and cerebellar vermis hypoplasia. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1053-1057.	0.7	15
139	Clinical exome sequencing in 509 Middle Eastern families with suspected Mendelian diseases: The Qatari experience. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 927-935.	0.7	32
140	Immunodeficiency and EBV-induced lymphoproliferation caused by 4-1BB deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 574-583.e5.	1.5	63
141	De Novo Variants Disrupting the HX Repeat Motif of <i>ATN1</i> Cause a Recognizable Non-Progressive Neurocognitive Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 542-552.	2.6	19
142	<i>PUS7</i> mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. <i>Human Genetics</i> , 2019, 138, 231-239.	1.8	53
143	<i>NUP214</i> deficiency causes severe encephalopathy and microcephaly in humans. <i>Human Genetics</i> , 2019, 138, 221-229.	1.8	17
144	Patterns of neurological manifestations in Woodhouse-Sakati Syndrome. <i>Parkinsonism and Related Disorders</i> , 2019, 69, 99-103.	1.1	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.	2.6	34
146	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	1.1	85
147	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	1.0	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.	0.7	20
149	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
150	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
151	A novel ISLR2-linked autosomal recessive syndrome of congenital hydrocephalus, arthrogryposis and abdominal distension. Human Genetics, 2019, 138, 105-107.	1.8	5
152	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	1.1	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.	1.1	71
154	Congenital glaucoma and CYP1B1: an old story revisited. Human Genetics, 2019, 138, 1043-1049.	1.8	29
155	<i>MED12</i> is recurrently mutated in Middle Eastern colorectal cancer. Gut, 2018, 67, gutjnl-2016-313334.	6.1	12
156	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	1.1	46
157	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. Clinical Genetics, 2018, 93, 1210-1222.	1.0	38
158	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 715-721.	0.7	7
159	Phenotypic characterization of <i>KCTD3</i> -related developmental epileptic encephalopathy. Clinical Genetics, 2018, 93, 1081-1086.	1.0	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.	1.0	17
161	A mendelian form of neural tube defect caused by a de novo null variant in <i>SMARCC1</i> in an identical twin. Annals of Neurology, 2018, 83, 433-436.	2.8	9
162	The alternatively spliced exon of <i>COL5A1</i> is mutated in autosomal recessive classical Ehlersâ€“Danlos syndrome. Clinical Genetics, 2018, 93, 936-937.	1.0	3

#	ARTICLE	IF	CITATIONS
163	Expanding the phenotype of <i>SLC25A42</i> -associated mitochondrial encephalomyopathy. <i>Clinical Genetics</i> , 2018, 93, 1097-1102.	1.0	24
164	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogyrosis. <i>American Journal of Human Genetics</i> , 2018, 102, 116-132.	2.6	46
165	Elsahy "Waters syndrome is caused by biallelic mutations in <i>CDH11</i> . <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 477-482.	0.7	18
166	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 696-705.	2.6	105
167	GWAS signals revisited using human knockouts. <i>Genetics in Medicine</i> , 2018, 20, 64-68.	1.1	6
168	<i>WDR45B</i> -related intellectual disability, spastic quadriplegia, epilepsy, and cerebral hypoplasia: A consistent neurodevelopmental syndrome. <i>Clinical Genetics</i> , 2018, 93, 360-364.	1.0	33
169	Exaggerated follicular helper T-cell responses in patients with LRBA deficiency caused by failure of CTLA4-mediated regulation. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1050-1059.e10.	1.5	93
170	Molecular autopsy in maternal "fetal medicine. <i>Genetics in Medicine</i> , 2018, 20, 420-427.	1.1	84
171	Phenotypic and Molecular Spectrum of Aicardi-GoutiÃres Syndrome: A Study of 24 Patients. <i>Pediatric Neurology</i> , 2018, 78, 35-40.	1.0	40
172	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2018, 128, 4313-4328.	3.9	89
173	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 948-967.	2.6	18
174	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. <i>American Journal of Human Genetics</i> , 2018, 103, 612-620.	2.6	70
175	Warsaw breakage syndrome: Further clinical and genetic delineation. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2404-2418.	0.7	16
176	Mutations in known disease genes account for the majority of autosomal recessive retinal dystrophies. <i>Clinical Genetics</i> , 2018, 94, 554-563.	1.0	12
177	Familial non-syndromic macular pseudocoloboma secondary to homozygous <i>CLDN19</i> mutation. <i>Ophthalmic Genetics</i> , 2018, 39, 577-583.	0.5	7
178	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. <i>American Journal of Human Genetics</i> , 2018, 103, 221-231.	2.6	65
179	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 858-873.	2.6	65
180	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. <i>Brain</i> , 2018, 141, 1934-1945.	3.7	70

#	ARTICLE	IF	CITATIONS
181	Further delineation of Malan syndrome. <i>Human Mutation</i> , 2018, 39, 1226-1237.	1.1	42
182	Deletion of DDB1- and CUL4- associated factor-17 (Dcaf17) gene causes spermatogenesis defects and male infertility in mice. <i>Scientific Reports</i> , 2018, 8, 9202.	1.6	21
183	Confirming the recessive inheritance of <i>SCN1B</i> mutations in developmental epileptic encephalopathy. <i>Clinical Genetics</i> , 2017, 92, 327-331.	1.0	32
184	<i>GLI3</i> -related polydactyly: a review. <i>Clinical Genetics</i> , 2017, 92, 457-466.	1.0	45
185	Response to Yehia et al.. <i>American Journal of Human Genetics</i> , 2017, 100, 564-565.	2.6	0
186	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. <i>Nature Genetics</i> , 2017, 49, 537-549.	9.4	81
187	PLAA Mutations Cause a Lethal Infantile Epileptic Encephalopathy by Disrupting Ubiquitin-Mediated Endolysosomal Degradation of Synaptic Proteins. <i>American Journal of Human Genetics</i> , 2017, 100, 706-724.	2.6	37
188	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , 2017, 100, 695-705.	2.6	305
189	GZF1 Mutations Expand the Genetic Heterogeneity of Larsen Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 831-836.	2.6	14
190	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. <i>American Journal of Human Genetics</i> , 2017, 101, 23-36.	2.6	74
191	The genetic landscape of familial congenital hydrocephalus. <i>Annals of Neurology</i> , 2017, 81, 890-897.	2.8	108
192	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. <i>American Journal of Human Genetics</i> , 2017, 100, 969-977.	2.6	38
193	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. <i>Human Genetics</i> , 2017, 136, 921-939.	1.8	209
194	A novel mutation in <i>SLC25A46</i> causes optic atrophy and progressive limb spasticity, with no cerebellar atrophy or axonal neuropathy. <i>Clinical Genetics</i> , 2017, 92, 230-231.	1.0	16
195	Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. <i>Genetics in Medicine</i> , 2017, 19, 1144-1150.	1.1	77
196	Genetic profiling of children with advanced cholestatic liver disease. <i>Clinical Genetics</i> , 2017, 92, 52-61.	1.0	36
197	Deficiency of a Retinal Dystrophy Protein, Acyl-CoA Binding Domain-containing 5 (ACBD5), Impairs Peroxisomal $\beta$ -Oxidation of Very-long-chain Fatty Acids. <i>Journal of Biological Chemistry</i> , 2017, 292, 691-705.	1.6	68
198	Expanding the spectrum of germline variants in cancer. <i>Human Genetics</i> , 2017, 136, 1431-1444.	1.8	23

#	ARTICLE	IF	CITATIONS
199	Mutations of PTPN23 in developmental and epileptic encephalopathy. <i>Human Genetics</i> , 2017, 136, 1455-1461.	1.8	15
200	Female Infertility Caused by Mutations in the Oocyte-Specific Translational Repressor PATL2. <i>American Journal of Human Genetics</i> , 2017, 101, 603-608.	2.6	59
201	Reply to "œan extremely severe phenotype due to WDR81 nonsense mutations". <i>Annals of Neurology</i> , 2017, 82, 651-651.	2.8	1
202	A null mutation in MICU2 causes abnormal mitochondrial calcium homeostasis and a severe neurodevelopmental disorder. <i>Brain</i> , 2017, 140, 2806-2813.	3.7	38
203	Mutations of <i>KIF14</i> cause primary microcephaly by impairing cytokinesis. <i>Annals of Neurology</i> , 2017, 82, 562-577.	2.8	62
204	Molecular and clinical spectra of FBXL4 deficiency. <i>Human Mutation</i> , 2017, 38, 1649-1659.	1.1	41
205	Expanding the genetic heterogeneity of intellectual disability. <i>Human Genetics</i> , 2017, 136, 1419-1429.	1.8	122
206	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. <i>American Journal of Human Genetics</i> , 2017, 101, 206-217.	2.6	51
207	Expanding the allelic disorders linked to <i>TCTN1</i> to include Varadi syndrome (Orofaciodigital) Tj ETQq1 1 0.784314 rgBT <sub>4</sub> /Overlo 0.7	0.7	
208	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 55-94.	0.8	20
209	KDF1, encoding keratinocyte differentiation factor 1, is mutated in a multigenerational family with ectodermal dysplasia. <i>Human Genetics</i> , 2017, 136, 99-105.	1.8	18
210	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	9.4	251
211	A lethal neonatal phenotype of mitochondrial short-chain enoyl-CoA hydratase deficiency. <i>Clinical Genetics</i> , 2017, 91, 629-633.	1.0	26
212	An autosomal recessive <i>DNASE1L3</i> -related autoimmune disease with unusual clinical presentation mimicking systemic lupus erythematosus. <i>Lupus</i> , 2017, 26, 768-772.	0.8	40
213	Increasing the sensitivity of clinical exome sequencing through improved filtration strategy. <i>Genetics in Medicine</i> , 2017, 19, 593-598.	1.1	59
214	The human knockout phenotype of <i>PADI6</i> is female sterility caused by cleavage failure of their fertilized eggs. <i>Clinical Genetics</i> , 2017, 91, 344-345.	1.0	32
215	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. <i>Molecular Psychiatry</i> , 2017, 22, 615-624.	4.1	187
216	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. <i>Human Genetics</i> , 2017, 136, 205-225.	1.8	73

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217	A novel mechanism for variable phenotypic expressivity in Mendelian diseases uncovered by an AU-rich element (ARE)-creating mutation. <i>Genome Biology</i> , 2017, 18, 144.	3.8	19
218	Congenital disorders of glycosylation: The Saudi experience. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2614-2621.	0.7	31
219	Mutations in <i>HYAL2</i> , Encoding Hyaluronidase 2, Cause a Syndrome of Orofacial Clefting and Cor Triatriatum Sinister in Humans and Mice. <i>PLoS Genetics</i> , 2017, 13, e1006470.	1.5	20
220	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit <i>TSEN15</i> Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. <i>American Journal of Human Genetics</i> , 2016, 99, 228-235.	2.6	44
221	Gonadal mosaicism for <i>ACTA1</i> gene masquerading as autosomal recessive nemaline myopathy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2219-2221.	0.7	8
222	<i>ADAT3</i> -related intellectual disability: Further delineation of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1142-1147.	0.7	32
223	Congenital hereditary endothelial dystrophy, not glaucoma, in a child with iris colobomas. <i>Journal of AAPOS</i> , 2016, 20, 370-372.	0.2	1
224	Epileptic encephalopathy with continuous spike-and-wave during sleep maps to a homozygous truncating mutation in AMPA receptor component <i>FRRS1L</i> . <i>Clinical Genetics</i> , 2016, 90, 282-283.	1.0	16
225	Confirming the candidacy of <i>THOC6</i> in the etiology of intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1367-1369.	0.7	13
226	Novel copy number variants and major limb reduction malformation: Report of three cases. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1245-1250.	0.7	8
227	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessive <i>WNT1</i> mutations. <i>Journal of Medical Genetics</i> , 2016, 53, 427-430.	1.5	41
228	Characterizing the morbid genome of ciliopathies. <i>Genome Biology</i> , 2016, 17, 242.	3.8	118
229	Revisiting the morbid genome of Mendelian disorders. <i>Genome Biology</i> , 2016, 17, 235.	3.8	48
230	Complementation of hypersensitivity to DNA interstrand crosslinking agents demonstrates that <i>XRCC2</i> is a Fanconi anaemia gene. <i>Journal of Medical Genetics</i> , 2016, 53, 672-680.	1.5	66
231	Hyperekplexia, microcephaly and simplified gyral pattern caused by novel <i>ASNS</i> mutations, case report. <i>BMC Neurology</i> , 2016, 16, 105.	0.8	32
232	<i>GNB5</i> mutation causes a novel neuropsychiatric disorder featuring attention deficit hyperactivity disorder, severely impaired language development and normal cognition. <i>Genome Biology</i> , 2016, 17, 195.	3.8	36
233	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. <i>Human Genetics</i> , 2016, 135, 525-540.	1.8	89
234	On the phenotypic spectrum of serine biosynthesis defects. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 373-381.	1.7	45

#	ARTICLE	IF	CITATIONS
235	Genomic Profiling of Thyroid Cancer Reveals a Role for Thyroglobulin in Metastasis. American Journal of Human Genetics, 2016, 98, 1170-1180.	2.6	41
236	Discovery of mutations for Mendelian disorders. Human Genetics, 2016, 135, 615-623.	1.8	53
237	A homozygous truncating mutation in PUS3 expands the role of tRNA modification in normal cognition. Human Genetics, 2016, 135, 707-713.	1.8	112
238	A null mutation in TNIK defines a novel locus for intellectual disability. Human Genetics, 2016, 135, 773-778.	1.8	23
239	Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. Genetics in Medicine, 2016, 18, 1244-1249.	1.1	82
240	Cell-Intrinsic Adaptation Arising from Chronic Ablation of a Key Rho GTPase Regulator. Developmental Cell, 2016, 39, 28-43.	3.1	40
241	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
242	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
243	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
244	ANKS3 is mutated in a family with autosomal recessive laterality defect. Human Genetics, 2016, 135, 1233-1239.	1.8	17
245	A lethal phenotype associated with tissue plasminogen deficiency in humans. Human Genetics, 2016, 135, 1209-1211.	1.8	2
246	The syndrome dysmorphic facies, renal agenesis, ambiguous genitalia, microcephaly, polydactyly and lissencephaly (DREAMâ€œPL): Report of two additional patients. American Journal of Medical Genetics, Part A, 2016, 170, 3222-3226.	0.7	17
247	Joint laxity in homozygotes for severe <i>POU1F1</i> mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3356-3358.	0.7	2
248	Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. Human Genetics, 2016, 135, 1295-1298.	1.8	65
249	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. Nature Genetics, 2016, 48, 1071-1076.	9.4	314
250	Human knockouts of PLA2G4A phenocopy NSAID-induced gastrointestinal and renal toxicity. Gut, 2016, 65, 1575-1577.	6.1	8
251	Impaired telomere maintenance in Alazami syndrome patients with LARP7 deficiency. BMC Genomics, 2016, 17, 749.	1.2	30
252	The journey of exome sequencing from disease mutations discovery to clinical genomics. Human Genetics, 2016, 135, 589-589.	1.8	0

#	ARTICLE	IF	CITATIONS
253	Crisponi/CISS1 syndrome: A case series. American Journal of Medical Genetics, Part A, 2016, 170, 1236-1241.	0.7	5
254	Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. Orphanet Journal of Rare Diseases, 2016, 11, 52.	1.2	13
255	Neuronal deficiency of <i>ARV1</i> causes an autosomal recessive epileptic encephalopathy. Human Molecular Genetics, 2016, 25, ddw157.	1.4	23
256	Distal acroosteolysis, poikiloderma and joint stiffness: a novel laminopathy?. European Journal of Human Genetics, 2016, 24, 1220-1222.	1.4	6
257	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
258	Mutations in <i>SMG9</i> , 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
259	Clinical Characterization of <i>LRPAP1</i> -Related Pediatric High Myopia. Ophthalmology, 2016, 123, 434-435.	2.5	12
260	Treatment of retinitis pigmentosa due to <i>MERTK</i> 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
261	Exome-based case-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
262	<i>GOLGA2</i> , 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
263	Mutations in <i>UNC80</i> , Encoding Part of the <i>UNC79-UNC80-NALCN</i> Channel Complex, Cause Autosomal-Recessive Severe Infantile Encephalopathy. American Journal of Human Genetics, 2016, 98, 210-215.	2.6	37
264	Mutation of the mitochondrial carrier <i>SLC25A42</i> causes a novel form of mitochondrial myopathy in humans. Human Genetics, 2016, 135, 21-30.	1.8	34
265	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. Genetics in Medicine, 2016, 18, 554-562.	1.1	89
266	Elevation deficiency in children with recessive <i>RDH12</i> -related retinopathy. Journal of AAPOS, 2015, 19, 568-570.	0.2	3
267	Matching Two Independent Cohorts Validates <i>DPH1</i> as a Gene Responsible for Autosomal Recessive Intellectual Disability with Short Stature, Craniofacial, and Ectodermal Anomalies. Human Mutation, 2015, 36, 1015-1019.	1.1	32
268	Natural human knockouts and the era of genotype to phenotype. Genome Medicine, 2015, 7, 48.	3.6	25
269	Identification of embryonic lethal genes in humans by autozygosity mapping and exome sequencing in consanguineous families. Genome Biology, 2015, 16, 116.	3.8	91
270	Severe CNS involvement in <i>WWOX</i> mutations: Description of five new cases. American Journal of Medical Genetics, Part A, 2015, 167, 3209-3213.	0.7	50



#	ARTICLE	IF	CITATIONS
271	A founder CEP120 mutation in Jeune asphyxiating thoracic dystrophy expands the role of centriolar proteins in skeletal ciliopathies. <i>Human Molecular Genetics</i> , 2015, 24, 1410-1419.	1.4	70
272	Congenital ptosis, scoliosis, and malignant hyperthermia susceptibility in siblings with recessive RYR1 mutations. <i>Journal of AAPOS</i> , 2015, 19, 577-579.	0.2	9
273	TLE6 mutation causes the earliest known human embryonic lethality. <i>Genome Biology</i> , 2015, 16, 240.	3.8	153
274	Mutation in WDR4 impairs tRNA m7G46 methylation and causes a distinct form of microcephalic primordial dwarfism. <i>Genome Biology</i> , 2015, 16, 210.	3.8	132
275	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. <i>Genome Biology</i> , 2015, 16, 293.	3.8	56
276	Recessive Mutations in <i>LEPREL1</i> Underlie a Recognizable Lens Subluxation Phenotype. <i>Ophthalmic Genetics</i> , 2015, 36, 58-63.	0.5	26
277	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. <i>Cell Reports</i> , 2015, 10, 148-161.	2.9	375
278	RGS6: A Novel Gene Associated With Congenital Cataract, Mental Retardation, and Microcephaly in a Tunisian Family. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 1261-1266.	3.3	9
279	LOXL3, encoding lysyl oxidase-like 3, is mutated in a family with autosomal recessive Stickler syndrome. <i>Human Genetics</i> , 2015, 134, 451-453.	1.8	64
280	Brittle Cornea Syndrome ZNF469 Mutation Carrier Phenotype and Segregation Analysis of Rare ZNF469 Variants in Familial Keratoconus. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 578-586.	3.3	33
281	Pellagra-like condition is xeroderma pigmentosum/Cockayne syndrome complex and niacin confers clinical benefit. <i>Clinical Genetics</i> , 2015, 87, 56-61.	1.0	12
282	A novel APC mutation defines a second locus for Cenani's Lenz syndrome. <i>Journal of Medical Genetics</i> , 2015, 52, 317-321.	1.5	20
283	High diagnostic yield of clinical exome sequencing in Middle Eastern patients with Mendelian disorders. <i>Human Genetics</i> , 2015, 134, 967-980.	1.8	168
284	Lens Subluxation and Retinal Dysfunction in a Girl with Homozygous <i>VSX2</i> Mutation. <i>Ophthalmic Genetics</i> , 2015, 36, 8-13.	0.5	16
285	Revisiting disease genes based on whole-exome sequencing in consanguineous populations. <i>Human Genetics</i> , 2015, 134, 1029-1034.	1.8	17
286	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. <i>Nature Cell Biology</i> , 2015, 17, 1074-1087.	4.6	215
287	ARL6IP6, a susceptibility locus for ischemic stroke, is mutated in a patient with syndromic Cutis Marmorata Telangiectatica Congenita. <i>Human Genetics</i> , 2015, 134, 815-822.	1.8	13
288	Positional mapping of <i>PRKD1</i> , <i>NRP1</i> and <i>PRDM1</i> as novel candidate disease genes in truncus arteriosus. <i>Journal of Medical Genetics</i> , 2015, 52, 322-329.	1.5	30

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289	Report of a case of Raine syndrome and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 2394-2398.	0.7	19
290	A novel syndrome of Klippel-Feil anomaly, myopathy, and characteristic facies is linked to a null mutation in <i>MYO18B</i> . Journal of Medical Genetics, 2015, 52, 400-404.	1.5	64
291	Primordial dwarfism. Current Opinion in Endocrinology, Diabetes and Obesity, 2015, 22, 55-64.	1.2	22
292	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725.	1.1	19
293	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by <i>RSPRY1</i> Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	2.6	14
294	T (brachyury) is linked to a Mendelian form of neural tube defects in humans. Human Genetics, 2015, 134, 1139-1141.	1.8	13
295	Intrafamilial clinical heterogeneity of <i>CSPP1</i> -related ciliopathy. American Journal of Medical Genetics, Part A, 2015, 167, 2478-2480.	0.7	9
296	Identification of a novel MKS locus defined by <i>TMEM107</i> mutation. Human Molecular Genetics, 2015, 24, 5211-5218.	1.4	42
297	<i>RTTN</i> Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. American Journal of Human Genetics, 2015, 97, 862-868.	2.6	36
298	Further Delineation of the ALG9-CDG Phenotype. JIMD Reports, 2015, 27, 107-112.	0.7	17
299	Human knockout research: new horizons and opportunities. Trends in Genetics, 2015, 31, 108-115.	2.9	42
300	Autosomal recessive congenital cataract, intellectual disability phenotype linked to <i>STX3</i> in a consanguineous Tunisian family. Clinical Genetics, 2015, 88, 283-287.	1.0	12
301	The many faces of <i>KIF7</i> . Human Genome Variation, 2015, 2, 15006.	0.4	16
302	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
303	A syndrome of congenital hyperinsulinism and rhabdomyolysis is caused by <i>KCNJ11</i> mutation. Journal of Medical Genetics, 2014, 51, 271-274.	1.5	6
304	Variable phenotypic expression of <i>COG6</i> mutations. Journal of Medical Genetics, 2014, 51, 425.2-426.	1.5	2
305	Complete Aniridia with Central Keratopathy and Congenital Glaucoma is a <i>CYP11B1</i> -related Phenotype. Ophthalmic Genetics, 2014, 35, 187-189.	0.5	9
306	<i>METTL23</i> , a transcriptional partner of <i>GABPA</i> , is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	1.4	47

#	ARTICLE	IF	CITATIONS
307	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. <i>Neuron</i> , 2014, 84, 1240-1257.	3.8	89
308	Expanding the clinical spectrum and allelic heterogeneity in van den Endeâ€“Gupta syndrome. <i>Clinical Genetics</i> , 2014, 85, 492-494.	1.0	8
309	IFT27, encoding a small GTPase component of IFT particles, is mutated in a consanguineous family with Bardet-Biedl syndrome. <i>Human Molecular Genetics</i> , 2014, 23, 3307-3315.	1.4	134
310	<i>NPHP4</i> mutation is linked to cerebelloâ€“oculoâ€“renal syndrome and male infertility. <i>Clinical Genetics</i> , 2014, 85, 371-375.	1.0	18
311	Excessively redundant umbilical skin as a potential early clinical feature of Morquio syndrome and <i>FKBP14</i> -related Ehlersâ€“Danlos syndrome. <i>Clinical Genetics</i> , 2014, 86, 469-472.	1.0	15
312	Genetics and genomic medicine in Saudi Arabia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2014, 2, 369-378.	0.6	61
313	Ciliary Genes <i>TBC1D32</i> and <i>C6orf170</i> and <i>SCLT1</i> are Mutated in Patients with OFD Type IX. <i>Human Mutation</i> , 2014, 35, 36-40.	1.1	78
314	Mutation in <i>PLK4</i> , encoding a master regulator of centriole formation, defines a novel locus for primordial dwarfism. <i>Journal of Medical Genetics</i> , 2014, 51, 814-816.	1.5	35
315	<i>NECAP1</i> loss of function leads to a severe infantile epileptic encephalopathy. <i>Journal of Medical Genetics</i> , 2014, 51, 224-228.	1.5	23
316	SET binding factor 1 ( <i>SBF1</i> ) mutation causes Charcot-Marie-Tooth disease type 4B3. <i>Neurology</i> , 2014, 82, 1665-1666.	1.5	18
317	Study of Mendelian forms of Crohn's disease in Saudi Arabia reveals novel risk loci and alleles. <i>Gut</i> , 2014, 63, 1831-1832.	6.1	28
318	<i>ZBTB42</i> mutation defines a novel lethal congenital contracture syndrome (LCCS6). <i>Human Molecular Genetics</i> , 2014, 23, 6584-6593.	1.4	25
319	Childhood Cone-rod Dystrophy with Macular Cystic Degeneration from Recessive <i>CRB1</i> Mutation. <i>Ophthalmic Genetics</i> , 2014, 35, 130-137.	0.5	22
320	Mutations in <i>CSPP1</i> , Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. <i>American Journal of Human Genetics</i> , 2014, 94, 73-79.	2.6	77
321	Gonadal mosaicism as a rare cause of autosomal recessive inheritance. <i>Clinical Genetics</i> , 2014, 85, 278-281.	1.0	20
322	Mutations in <i>CCNO</i> result in congenital mucociliary clearance disorder with reduced generation of multiple motile cilia. <i>Nature Genetics</i> , 2014, 46, 646-651.	9.4	232
323	Identification of three novel <i>ECEL1</i> mutations in three families with distal arthrogyrosis type 5D. <i>Clinical Genetics</i> , 2014, 85, 568-572.	1.0	29
324	Novel <i>IFT122</i> mutation associated with impaired ciliogenesis and cranioectodermal dysplasia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2014, 2, 103-106.	0.6	22

#	ARTICLE	IF	CITATIONS
325	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
326	C19orf12 mutation leads to a pallido-pyramidal syndrome. Gene, 2014, 537, 352-356.	1.0	28
327	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
328	Mutations in LRPAP1 Are Associated with Severe Myopia in Humans. American Journal of Human Genetics, 2013, 93, 313-320.	2.6	104
329	“Methylglutaconic aciduria” lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	1.7	74
330	The application of next-generation sequencing in the autozygosity mapping of human recessive diseases. Human Genetics, 2013, 132, 1197-1211.	1.8	107
331	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
332	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
333	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	2.6	138
334	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
335	Corneal enlargement without optic disk cupping in children with recessive CYP1B1 mutations. Journal of AAPOS, 2013, 17, 643-645.	0.2	2
336	Biometric and Molecular Characterization of Clinically Diagnosed Posterior Microphthalmos. American Journal of Ophthalmology, 2013, 155, 361-372.e7.	1.7	54
337	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
338	Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. American Journal of Human Genetics, 2013, 93, 555-560.	2.6	45
339	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
340	Mutations in MEOX1, Encoding Mesenchyme Homeobox 1, Cause Klippel-Feil Anomaly. American Journal of Human Genetics, 2013, 92, 157-161.	2.6	82
341	Autozygosity Mapping with Exome Sequence Data. Human Mutation, 2013, 34, 50-56.	1.1	49
342	Congenital glaucoma with acquired peripheral circumferential iris degeneration. Journal of AAPOS, 2013, 17, 105-107.	0.2	13

#	ARTICLE	IF	CITATIONS
343	Mutations in C12orf57 Cause a Syndromic Form of Colobomatous Microphthalmia. <i>American Journal of Human Genetics</i> , 2013, 92, 387-391.	2.6	35
344	Juvenile Cataract Morphology in 3 Siblings Not Yet Diagnosed with Cerebrotendinous Xanthomatosis. <i>Ophthalmology</i> , 2013, 120, 956-960.	2.5	17
345	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. <i>Gene</i> , 2013, 512, 450-452.	1.0	7
346	Mutation in <i>ADAT3</i> , encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus. <i>Journal of Medical Genetics</i> , 2013, 50, 425-430.	1.5	91
347	Genomic analysis of Meckel-Gruber syndrome in Arabs reveals marked genetic heterogeneity and novel candidate genes. <i>European Journal of Human Genetics</i> , 2013, 21, 762-768.	1.4	56
348	Preclinical Potency and Safety Studies of an AAV2-Mediated Gene Therapy Vector for the Treatment of <i>MERTK</i> Associated Retinitis Pigmentosa. <i>Human Gene Therapy Clinical Development</i> , 2013, 24, 23-28.	3.2	84
349	Autozygome Sequencing Expands the Horizon of Human Knockout Research and Provides Novel Insights into Human Phenotypic Variation. <i>PLoS Genetics</i> , 2013, 9, e1004030.	1.5	48
350	A novel syndrome of hypohidrosis and intellectual disability is linked to COG6 deficiency. <i>Journal of Medical Genetics</i> , 2013, 50, 431-436.	1.5	40
351	No evidence for locus heterogeneity in Knobloch syndrome. <i>Journal of Medical Genetics</i> , 2013, 50, 565-566.	1.5	20
352	Autozygome-guided exome sequencing in retinal dystrophy patients reveals pathogenetic mutations and novel candidate disease genes. <i>Genome Research</i> , 2013, 23, 236-247.	2.4	226
353	The <i>RPGRI1</i> -related retinal phenotype in children. <i>British Journal of Ophthalmology</i> , 2013, 97, 760-764.	2.1	20
354	Impact of new genomic tools on the practice of clinical genetics in consanguineous populations: the Saudi experience. <i>Clinical Genetics</i> , 2013, 84, 203-208.	1.0	44
355	Mutation in <i>PHC1</i> implicates chromatin remodeling in primary microcephaly pathogenesis. <i>Human Molecular Genetics</i> , 2013, 22, 2200-2213.	1.4	81
356	The Syndrome of Microcornea, Myopic Chorioretinal Atrophy, and Telecanthus (MMCAT) Is Caused by Mutations in <i>ADAMTS18</i> . <i>Human Mutation</i> , 2013, 34, 1195-1199.	1.1	56
357	<i>WNT1</i> mutation with recessive osteogenesis imperfecta and profound neurological phenotype. <i>Journal of Medical Genetics</i> , 2013, 50, 491-492.	1.5	45
358	Lifting the lid on unborn lethal Mendelian phenotypes through exome sequencing. <i>Genetics in Medicine</i> , 2013, 15, 307-309.	1.1	42
359	The syndrome of deafness-dystonia: Clinical and genetic heterogeneity. <i>Movement Disorders</i> , 2013, 28, 795-803.	2.2	25
360	Mutation in <i>MPDZ</i> causes severe congenital hydrocephalus. <i>Journal of Medical Genetics</i> , 2013, 50, 54-58.	1.5	75

#	ARTICLE	IF	CITATIONS
361	Mutation in <i>RAB33B</i> , which encodes a regulator of retrograde Golgi transport, defines a second Dyggve-Melchior-Clausen locus. <i>Journal of Medical Genetics</i> , 2012, 49, 455-461.	1.5	35
362	Clinical and molecular analysis of children with central pulverulent cataract from the Arabian Peninsula. <i>British Journal of Ophthalmology</i> , 2012, 96, 650-655.	2.1	15
363	“Cone dystrophy with supranormal rod response”™ in children. <i>British Journal of Ophthalmology</i> , 2012, 96, 422-426.	2.1	34
364	Exome sequencing reveals a novel Fanconi group defined by XRCC2 mutation: Figure 1. <i>Journal of Medical Genetics</i> , 2012, 49, 184-186.	1.5	72
365	Autozygome maps dispensable DNA and reveals potential selective bias against nullizygosity. <i>Genetics in Medicine</i> , 2012, 14, 515-519.	1.1	10
366	Colorectal cancer risk is not associated with increased levels of homozygosity in Saudi Arabia. <i>Genetics in Medicine</i> , 2012, 14, 720-728.	1.1	14
367	Familial spherophakia with short stature caused by a novel homozygous <i>ADAMTS17</i> mutation. <i>Ophthalmic Genetics</i> , 2012, 33, 235-239.	0.5	31
368	Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes. <i>Journal of Medical Genetics</i> , 2012, 49, 234-241.	1.5	164
369	Discovery of Rare Homozygous Mutations from Studies of Consanguineous Pedigrees. <i>Current Protocols in Human Genetics</i> , 2012, 75, Unit6.12.	3.5	87
370	Study of autosomal recessive osteogenesis imperfecta in Arabia reveals a novel locus defined by <i>TMEM38B</i> mutation. <i>Journal of Medical Genetics</i> , 2012, 49, 630-635.	1.5	124
371	Homozygous null mutation in <i>ODZ3</i> causes microphthalmia in humans. <i>Genetics in Medicine</i> , 2012, 14, 900-904.	1.1	59
372	The distinct ophthalmic phenotype of Knobloch syndrome in children. <i>British Journal of Ophthalmology</i> , 2012, 96, 890-895.	2.1	63
373	In search of triallelism in Bardet-Biedl syndrome. <i>European Journal of Human Genetics</i> , 2012, 20, 420-427.	1.4	111
374	Mitochondrial phenylalanyl-tRNA synthetase mutations underlie fatal infantile Alpers encephalopathy. <i>Human Molecular Genetics</i> , 2012, 21, 4521-4529.	1.4	143
375	RP1 and retinitis pigmentosa: report of novel mutations and insight into mutational mechanism. <i>British Journal of Ophthalmology</i> , 2012, 96, 1018-1022.	2.1	28
376	Brittle cornea without clinically-evident extraocular findings in an adult harboring a novel homozygous <i>ZNF469</i> mutation. <i>Ophthalmic Genetics</i> , 2012, 33, 257-259.	0.5	20
377	Identification of a novel <i>DLX5</i> mutation in a family with autosomal recessive split hand and foot malformation. <i>Journal of Medical Genetics</i> , 2012, 49, 16-20.	1.5	75
378	Phenotype-genotype Correlation in Potential Female Carriers of X-linked Developmental Cataract (Nance-Horan Syndrome). <i>Ophthalmic Genetics</i> , 2012, 33, 89-95.	0.5	18

#	ARTICLE	IF	CITATIONS
379	Genomic analysis of pediatric cataract in Saudi Arabia reveals novel candidate disease genes. <i>Genetics in Medicine</i> , 2012, 14, 955-962.	1.1	49
380	Vanishing white matter disease caused by EIF2B2 mutation with the presentation of an adrenoleukodystrophy phenotype. <i>Gene</i> , 2012, 496, 141-143.	1.0	5
381	Familial dorsalization of the skin of the proximal palm and the instep of the sole of the foot. <i>Gene</i> , 2012, 500, 216-219.	1.0	12
382	A case of de Barsy syndrome with a severe eye phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2364-2366.	0.7	5
383	Loss of function mutation in LARP7, chaperone of 7SK ncRNA, causes a syndrome of facial dysmorphism, intellectual disability, and primordial dwarfism. <i>Human Mutation</i> , 2012, 33, 1429-1434.	1.1	63
384	A novel syndrome of lethal familial hyperekplexia associated with brain malformation. <i>BMC Neurology</i> , 2012, 12, 125.	0.8	9
385	CYP1B1 analysis of unilateral primary newborn glaucoma in Saudi children. <i>Journal of AAPOS</i> , 2012, 16, 571-572.	0.2	8
386	LPS-responsive beige-like anchor (LRBA) gene mutation in a family with inflammatory bowel disease and combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 130, 481-488.e2.	1.5	232
387	POC1A Truncation Mutation Causes a Ciliopathy in Humans Characterized by Primordial Dwarfism. <i>American Journal of Human Genetics</i> , 2012, 91, 330-336.	2.6	70
388	Identification of a novel ZNF469 mutation in a large family with Ehlers-Danlos phenotype. <i>Gene</i> , 2012, 511, 447-450.	1.0	23
389	Tyrosine-Mutant AAV8 Delivery of Human <i>MERTK</i> Provides Long-Term Retinal Preservation in RCS Rats. , 2012, 53, 1895.		48
390	Identification of a truncation mutation of acylglycerol kinase (AGK) gene in a novel autosomal recessive cataract locus. <i>Human Mutation</i> , 2012, 33, 960-962.	1.1	60
391	Molecular characterization of Joubert syndrome in Saudi Arabia. <i>Human Mutation</i> , 2012, 33, 1423-1428.	1.1	56
392	MGAT2-related CDG (CDG-IIa) and dysmorphism. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2976-2976.	0.7	0
393	A novel X-linked disorder with developmental delay and autistic features. <i>Annals of Neurology</i> , 2012, 71, 498-508.	2.8	33
394	A novel mutation in <i>PRDM5</i> in brittle cornea syndrome. <i>Clinical Genetics</i> , 2012, 81, 198-199.	1.0	24
395	3M Syndrome: An Easily Recognizable yet Underdiagnosed Cause of Proportionate Short Stature. <i>Journal of Pediatrics</i> , 2012, 161, 139-145.e1.	0.9	27
396	Oxoprolinase deficiency: report of the first human <i>OPLAH</i> mutation. <i>Clinical Genetics</i> , 2012, 82, 193-196.	1.0	19

#	ARTICLE	IF	CITATIONS
397	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
398	Novel recessive BFSP2 and PITX3 mutations: Insights into mutational mechanisms from consanguineous populations. Genetics in Medicine, 2011, 13, 978-981.	1.1	49
399	Identification of differentially expressed proteins in the aqueous humor of primary congenital glaucoma. Experimental Eye Research, 2011, 92, 67-75.	1.2	51
400	Study of consanguineous populations can improve the annotation of SNP databases. European Journal of Medical Genetics, 2011, 54, 118-120.	0.7	3
401	Familial juvenile glaucoma with underlying homozygous p.G61E CYP1B1 mutations. Journal of AAPOS, 2011, 15, 198-199.	0.2	13
402	Posterior Microphthalmos as a Genetically Heterogeneous Condition That Can Be Allelic to Nanophthalmos. JAMA Ophthalmology, 2011, 129, 805.	2.6	28
403	Clinical, biochemical and molecular characterization of peroxisomal diseases in Arabs. Clinical Genetics, 2011, 79, 60-70.	1.0	18
404	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
405	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
406	A novel <i>PTF1A</i> mutation in a patient with severe pancreatic and cerebellar involvement. Clinical Genetics, 2011, 80, 196-198.	1.0	39
407	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nature Genetics, 2011, 43, 197-203.	9.4	229
408	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. American Journal of Human Genetics, 2011, 88, 536-547.	2.6	196
409	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. American Journal of Human Genetics, 2011, 88, 677.	2.6	3
410	Deficiency of the Cytoskeletal Protein SPECC1L Leads to Oblique Facial Clefting. American Journal of Human Genetics, 2011, 89, 44-55.	2.6	70
411	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
412	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
413	Recessive Mutations in ELOVL4 Cause Ichthyosis, Intellectual Disability, and Spastic Quadriplegia. American Journal of Human Genetics, 2011, 89, 745-750.	2.6	161
414	A nullimorphic ERLIN2 mutation defines a complicated hereditary spastic paraplegia locus (SPG18). Neurogenetics, 2011, 12, 333-336.	0.7	67



#	ARTICLE	IF	CITATIONS
415	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
416	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
417	Ritscherâ€œSchinzel (cranioâ€œcerebelloâ€œcardiac, 3C) syndrome: Report of four new cases with renal involvement. American Journal of Medical Genetics, Part A, 2011, 155, 1393-1397.	0.7	12
418	Mutations in <i>FKBP10</i> cause both Bruck syndrome and isolated osteogenesis imperfecta in humans. American Journal of Medical Genetics, Part A, 2011, 155, 1448-1452.	0.7	63
419	A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. Human Mutation, 2011, 32, 573-578.	1.1	72
420	Functional analysis of BBS3 A89V that results in non-syndromic retinal degeneration. Human Molecular Genetics, 2011, 20, 1625-1632.	1.4	38
421	Loss-of-function variant in DNASE1L3 causes a familial form of systemic lupus erythematosus. Nature Genetics, 2011, 43, 1186-1188.	9.4	366
422	Molecular Characterization of Newborn Glaucoma Including a Distinct Aniridic Phenotype. Ophthalmic Genetics, 2011, 32, 138-142.	0.5	40
423	Mutations in the RNA Granule Component TDRD7 Cause Cataract and Glaucoma. Science, 2011, 331, 1571-1576.	6.0	186
424	Identification of ADAMTS18 as a gene mutated in Knobloch syndrome. Journal of Medical Genetics, 2011, 48, 597-601.	1.5	68
425	Genetic and genomic analysis of classic aniridia in Saudi Arabia. Molecular Vision, 2011, 17, 708-14.	1.1	14
426	Helicoid Subretinal Fibrosis Associated With a Novel Recessive NR2E3 Mutation p.S44X. JAMA Ophthalmology, 2010, 128, 344.	2.6	17
427	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. American Journal of Human Genetics, 2010, 87, 306-307.	2.6	47
428	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. American Journal of Human Genetics, 2010, 87, 571.	2.6	3
429	Perturbation of the consensus activation site of endothelinâ€œ3 leads to Waardenburg syndrome type IV. American Journal of Medical Genetics, Part A, 2010, 152A, 1841-1843.	0.7	4
430	Mutation of <i>CANT1</i> causes Desbuquois dysplasia. American Journal of Medical Genetics, Part A, 2010, 152A, 1157-1160.	0.7	31
431	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
432	<i>C2orf37</i> mutational spectrum in Woodhouseâ€œSakati syndrome patients. Clinical Genetics, 2010, 78, 585-590.	1.0	41

#	ARTICLE	IF	CITATIONS
433	Dorsal Dimelia: Report of Two Cases With an Emphasis On the Variation of Phenotypic Expression and a Search for Candidate Causative Genes. <i>Journal of Hand Surgery: European Volume</i> , 2010, 35, 715-720.	0.5	11
434	Trichorhinophalangeal syndrome: report of a novel familial TRPS1 mutation. <i>Clinical Dysmorphology</i> , 2010, 19, 98-100.	0.1	4
435	Blue Sclera With and Without Corneal Fragility (Brittle Cornea Syndrome) in a Consanguineous Family Harboring ZNF469 Mutation (p.E1392X). <i>JAMA Ophthalmology</i> , 2010, 128, 1376.	2.6	29
436	Methylation-Specific Multiplex-Ligation-Dependent Probe Amplification as a Rapid Molecular Diagnostic Tool for Pseudohypoparathyroidism Type 1b. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 135-139.	0.3	9
437	Clinical and molecular characterisation of Bardet-Biedl syndrome in consanguineous populations: the power of homozygosity mapping. <i>Journal of Medical Genetics</i> , 2010, 47, 236-241.	1.5	76
438	Autozygome decoded. <i>Genetics in Medicine</i> , 2010, 12, 765-771.	1.1	107
439	Saudi genetic ophthalmology research: The local and global impact. <i>Saudi Journal of Ophthalmology</i> , 2010, 24, 109-110.	0.3	3
440	Later retinal degeneration following childhood surgical aphakia in a family with recessive CRYAB mutation (p.R56W). <i>Ophthalmic Genetics</i> , 2010, 31, 30-36.	0.5	22
441	A novel <i>C2orf37</i> mutation causes the first Italian cases of Woodhouse Sakati syndrome. <i>Clinical Genetics</i> , 2010, 78, 594-597.	1.0	27
442	Homozygosity mapping: One more tool in the clinical geneticist's toolbox. <i>Genetics in Medicine</i> , 2010, 12, 236-239.	1.1	107
443	Novel CENPJ mutation causes Seckel syndrome. <i>Journal of Medical Genetics</i> , 2010, 47, 411-414.	1.5	149
444	Mutational Spectrum of <i>SLC4A11</i> in Autosomal Recessive CHED in Saudi Arabia. , 2009, 50, 4142.		41
445	Corneal Decompensation in Recessive Cornea Plana. <i>Ophthalmic Genetics</i> , 2009, 30, 142-145.	0.5	10
446	Characterization of <i>CTNS</i> mutations in Arab patients with Cystinosis. <i>Ophthalmic Genetics</i> , 2009, 30, 185-189.	0.5	35
447	Allelic heterogeneity in inbred populations: The Saudi experience with Alström syndrome as an illustrative example. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 662-665.	0.7	48
448	<i>NR2F1</i> deletion in a patient with a de novo paracentric inversion, inv(5)(q15q33.2), and syndromic deafness. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 931-938.	0.7	41
449	Zellweger syndrome caused by PEX13 deficiency: Report of two novel mutations. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1219-1223.	0.7	13
450	A novel missense mutation in <i>SCYL1BP1</i> produces geroderma osteodysplastica phenotype indistinguishable from that caused by nullimorphic mutations. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2093-2098.	0.7	28

#	ARTICLE	IF	CITATIONS
451	Neuronal ceroid lipofuscinosis caused by MFSD8 mutations: a common theme emerging. <i>Neurogenetics</i> , 2009, 10, 307-311.	0.7	35
452	Syndromic congenital sensorineural deafness, microtia and microdontia resulting from a novel homoallelic mutation in fibroblast growth factor 3 (FGF3). <i>European Journal of Human Genetics</i> , 2009, 17, 14-21.	1.4	31
453	FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. <i>American Journal of Human Genetics</i> , 2009, 85, 414-418.	2.6	86
454	Homozygous Mutations in ADAMTS10 and ADAMTS17 Cause Lenticular Myopia, Ectopia Lentis, Glaucoma, Spherophakia, and Short Stature. <i>American Journal of Human Genetics</i> , 2009, 85, 558-568.	2.6	204
455	FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. <i>American Journal of Human Genetics</i> , 2009, 85, 756.	2.6	1
456	Tufting Enteropathy and Chronic Arthritis: A Newly Recognized Association With a Novel <i>EpCAM</i> Gene Mutation. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2009, 49, 642-644.	0.9	37
457	Founder heterozygous P23T CRYGD mutation associated with cerulean (and coralliform) cataract in 2 Saudi families. <i>Molecular Vision</i> , 2009, 15, 1407-11.	1.1	18
458	Molecular characterization of retinitis pigmentosa in Saudi Arabia. <i>Molecular Vision</i> , 2009, 15, 2464-9.	1.1	61
459	Expanding the <i>in CHARGE</i> . <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1890-1892.	0.7	14
460	Johanson's "Blizzard syndrome: Report of a novel mutation and severe liver involvement. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1875-1879.	0.7	17
461	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. <i>American Journal of Human Genetics</i> , 2008, 82, 712-722.	2.6	95
462	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. <i>American Journal of Human Genetics</i> , 2008, 83, 425-427.	2.6	0
463	Mutations in C2orf37, Encoding a Nucleolar Protein, Cause Hypogonadism, Alopecia, Diabetes Mellitus, Mental Retardation, and Extrapryamidal Syndrome. <i>American Journal of Human Genetics</i> , 2008, 83, 684-691.	2.6	121
464	NFIA Haploinsufficiency Is Associated with a CNS Malformation Syndrome and Urinary Tract Defects. <i>PLoS Genetics</i> , 2007, 3, e80.	1.5	100
465	SUMO1 Haploinsufficiency Leads to Cleft Lip and Palate. <i>Science</i> , 2006, 313, 1751-1751.	6.0	165
466	Recurrent miscarriage in a carrier of a balanced cytogenetically undetectable subtelomeric rearrangement: How many are we missing?. <i>Prenatal Diagnosis</i> , 2006, 26, 291-293.	1.1	4
467	X-linked creatine transporter defect: A report on two unrelated boys with a severe clinical phenotype. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 214-219.	1.7	70
468	Trisomy 8 mosaicism in a patient with heterotaxia. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2005, 73, 58-60.	1.6	2

#	ARTICLE	IF	CITATIONS
469	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
470	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
471	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
472	Index of Suspicion. Pediatrics in Review, 2004, 25, 289-294.	0.2	1
473	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