Fowzan S Alkuraya

List of Publications by Year in Descending Order

460 papers	14,022 citations	61 h-index	92 g-index
516 ext. papers	17,417 ext. citations	7.1 avg, IF	6.55 L-index

#	Paper	IF	Citations
460	Recurrent spontaneous oocyte activation causes female infertility.. *Journal of Assisted Reproduction and Genetics*, **2022**, 39, 675	3.4	2
459	The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. *Genetics in Medicine*, **2022**,	8.1	4
458	Mitochondrial "dysmorphology" in variant classification. *Human Genetics*, **2021**, 1	6.3	
457	Insight into ALKBH8-related intellectual developmental disability based on the first pathogenic missense variant. *Human Genetics*, **2021**, 1	6.3	2
456	Genomic medicine in the Middle East. *Genome Medicine*, **2021**, 13, 184	14.4	4
455	ASTL is mutated in female infertility. *Human Genetics*, **2021**, 1	6.3	0
454	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. *Genome Medicine* , **2021**, 13, 161	14.4	3
453	Confirming the involvement of PIEZO2 in the etiology of Marden-Walker syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 945-948	2.5	1
452	Two further cases of polyhydramnios, megalencephaly, and symptomatic epilepsy syndrome, caused by a truncating variant in STRADA. *American Journal of Medical Genetics, Part A*, **2021**, 185, 604-607	2.5	2
451	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	2.5	3
450	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	2.5	0
449	Further delineation of van den Ende-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	2.5	0
448	MYH1 is a candidate gene for recurrent rhabdomyolysis in humans. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2131-2135	2.5	1
447	Mutations in phospholipase C eta-1 () are associated with holoprosencephaly. *Journal of Medical Genetics*, **2021**,	5.8	1
446	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	8.1	6
445	Clinical, neuroimaging, and molecular spectrum of TECPR2-associated hereditary sensory and autonomic neuropathy with intellectual disability. *Human Mutation*, **2021**, 42, 762-776	4.7	5
444	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	8.1	6

443	Genetic testing results of children suspected to have Stickler syndrome type collagenopathy after ocular examination. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1628	2.3	0
442	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	6.3	0
441	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. *Brain*, **2021**, 144, 1422-1434	11.2	7
440	Mutations in HID1 Cause Syndromic Infantile Encephalopathy and Hypopituitarism. *Annals of Neurology*, **2021**, 90, 143-158	9.4	1
439	SARS-CoV-2-Related Acute Respiratory Distress Syndrome Uncovers a Patient with Severe Combined Immunodeficiency Disease. *Journal of Clinical Immunology*, **2021**, 41, 1507-1510	5.7	1
438	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. *New England Journal of Medicine*, **2021**, 384, 2406-2417	59.2	16
437	Hoarse voice in children as the presenting feature of ECM1-related lipoid proteinosis. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3924-3925	2.5	0
436	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. *American Journal of Human Genetics*, **2021**, 108, 1069-1082	11	4
435	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. *Genetics in Medicine*, **2021**, 23, 1901-1911	8.1	1
434	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. *American Journal of Human Genetics*, **2021**, 108, 1095-1114	11	1
433	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. *Genetics in Medicine*, **2021**, 23, 1933-1943	8.1	2
432	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. *Human Genetics*, **2021**, 140, 1395-1401	6.3	
431	CHEDDA syndrome is an underrecognized neurodevelopmental disorder with a highly restricted ATN1 mutation spectrum. *Clinical Genetics*, **2021**, 100, 468-477	4	2
430	PLXNA2 as a candidate gene in patients with intellectual disability. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3859-3865	2.5	2
429	New paradigms of USP53 disease: normal GGT cholestasis, BRIC, cholangiopathy, and responsiveness to rifampicin. *Journal of Human Genetics*, **2021**, 66, 151-159	4.3	12
428	Indigenous Arabs have an intermediate frequency of a Neanderthal-derived COVID-19 risk haplotype compared with other world populations. *Clinical Genetics*, **2021**, 99, 484-485	4	1
427	Further delineation of MYO18B-related autosomal recessive Klippel-Feil syndrome with myopathy and facial dysmorphism. *American Journal of Medical Genetics, Part A*, **2021**, 185, 370-376	2.5	1
426	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. *Brain and Development*, **2021**, 43, 380-388	2.2	2

425	A de novo ATXN2L variant in a child with developmental delay and macrocephaly. *American Journal of Medical Genetics, Part A*, **2021**, 185, 949-951	2.5	2
424	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. *Kidney International Reports*, **2021**, 6, 460-471	4.1	2
423	Population structure of indigenous inhabitants of Arabia. *PLoS Genetics*, **2021**, 17, e1009210	6	5
422	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19
421	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. *Genetics in Medicine*, **2021**, 23, 661-668	8.1	1
420	Successful hematopoietic stem cell transplantation in a 4-1BB deficient patient with EBV-induced lymphoproliferation. *Clinical Immunology*, **2021**, 222, 108639	9	2
419	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. *Brain*, **2021**,	11.2	2
418	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	11	8
417	Further delineation of SMG9-related heart and brain malformation syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1624-1630	2.5	3
416	Variants in 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	0.8	1
415	Missense NAA20 variants impairing the NatB protein N-terminal acetyltransferase cause autosomal recessive developmental delay, intellectual disability, and microcephaly. *Genetics in Medicine*, **2021**, 23, 2213-2218	8.1	1
414	Residual risk for additional recessive diseases in consanguineous couples. *Genetics in Medicine*, **2021**, 23, 2448-2454	8.1	2
413	Implications of mosaicism in variant interpretation: A case of a de novo homozygous NF1 variant. *European Journal of Medical Genetics*, **2021**, 64, 104236	2.6	
412	Mutations in TP73 cause impaired mucociliary clearance and lissencephaly. *American Journal of Human Genetics*, **2021**, 108, 1318-1329	11	4
411	Expanding the KIF4A-associated phenotype. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3728-3739	2.5	0
410	Expanding the phenotype of ASXL3-related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in ASXL3. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3446-3458	2.5	3
409	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	1.4	0
408	Molecular autopsy by proxy in preconception counseling. *Clinical Genetics*, **2021**, 100, 678-691	4	2

407	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. *Journal of Molecular Medicine*, **2021**, 99, 1755-1768	5.5	0
406	Response to Hamosh et al. *American Journal of Human Genetics*, **2021**, 108, 1809-1810	11	
405	PLACK syndrome is potentially treatable with intralipids. *Clinical Genetics*, **2021**, 99, 572-576	4	
404	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. *American Journal of Human Genetics*, **2020**, 107, 963-976	11	4
403	Absence of GP130 cytokine receptor signaling causes extended Stüve-Wiedemann syndrome. *Journal of Experimental Medicine*, **2020**, 217,	16.6	27
402	The natural history of infantile neuroaxonal dystrophy. *Orphanet Journal of Rare Diseases*, **2020**, 15, 109	4.2	5
401	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. *Human Genetics*, **2020**, 139, 1273-1283	6.3	9
400	DALRD3 encodes a protein mutated in epileptic encephalopathy that targets arginine tRNAs for 3-methylcytosine modification. *Nature Communications*, **2020**, 11, 2510	17.4	15
399	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	19.2	0
398	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. *European Journal of Human Genetics*, **2020**, 28, 1509-1519	5.3	11
397	Confirming TBC1D32-related ciliopathy in humans. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1985-1987	2.5	5
396	Expanding the clinical and phenotypic heterogeneity associated with biallelic variants in ACO2. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1013-1028	5.3	3
395	A Case with Purine Nucleoside Phosphorylase Deficiency Suffering from Late-Onset Systemic Lupus Erythematosus and Lymphoma. *Journal of Clinical Immunology*, **2020**, 40, 833-839	5.7	5
394	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. *Genome Biology*, **2020**, 21, 145	18.3	18
393	A genomics approach to females with infertility and recurrent pregnancy loss. *Human Genetics*, **2020**, 139, 605-613	6.3	33
392	Phenotypic expansion of OTUD6B-related syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1530-1531	2.5	5
391	Multiple Family Members With Delayed Cord Separtion and Combined Immunodeficiency With Novel Mutation in. *Frontiers in Pediatrics*, **2020**, 8, 9	3.4	4
390	CNP deficiency causes severe hypomyelinating leukodystrophy in humans. *Human Genetics*, **2020**, 139, 615-622	6.3	5

389	Genetic heterogeneity and evolutionary history of high-grade ovarian carcinoma and matched distant metastases. *British Journal of Cancer*, **2020**, 122, 1219-1230	8.7	17
388	Further delineation of METTL23-associated intellectual disability. *American Journal of Medical Genetics, Part A*, **2020**, 182, 785-791	2.5	2
387	The morbid genome of ciliopathies: an update. *Genetics in Medicine*, **2020**, 22, 1051-1060	8.1	30
386	Phenome-based approach identifies RIC1-linked Mendelian syndrome through zebrafish models, biobank associations and clinical studies. *Nature Medicine*, **2020**, 26, 98-109	50.5	16
385	Recessive mutations in SCYL2 cause a novel syndromic form of arthrogryposis in humans. *Human Genetics*, **2020**, 139, 513-519	6.3	2
384	A human ciliopathy reveals essential functions for NEK10 in airway mucociliary clearance. *Nature Medicine*, **2020**, 26, 244-251	50.5	22
383	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. *Journal of Clinical Investigation*, **2020**, 130, 4423-4439	15.9	19
382	A Diagnostic Approach for Neurogenetic Disorders in the Genome Era **2020**, 319-326		
381	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. *Frontiers in Genetics*, **2020**, 11, 580484	4.5	4
380	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	11	6
379	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. *Nature Communications*, **2020**, 11, 595	17.4	18
378	Whole-Exome Sequencing of Matched Primary and Metastatic Papillary Thyroid Cancer. *Thyroid*, **2020**, 30, 42-56	6.2	18
377	Rhegmatogenous Retinal Detachment in Nonsyndromic High Myopia Associated with Recessive Mutations in LRPAP1. *Ophthalmology Retina*, **2020**, 4, 77-83	3.8	3
376	A de novo mutation in FMR1 in a patient with intellectual disability. *European Journal of Medical Genetics*, **2020**, 63, 103763	2.6	3
375	An intellectual disability-associated missense variant in TRMT1 impairs tRNA modification and reconstitution of enzymatic activity. *Human Mutation*, **2020**, 41, 600-607	4.7	11
374	A novel truncating variant in ring finger protein 113A (RNF113A) confirms the association of this gene with X-linked trichothiodystrophy. *American Journal of Medical Genetics, Part A*, **2020**, 182, 513-520	2.5	3
373	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. *Clinical Genetics*, **2020**, 97, 447-456	4	4
372	Confirming the recessive inheritance of PERP-related erythrokeratoderma. *Clinical Genetics*, **2020**, 97, 661-665	4	1

371	International perspectives on the implementation of reproductive carrier screening. *Prenatal Diagnosis*, **2020**, 40, 301-310	3.2	29
370	Further delineation of HIDEA syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2999-3006	2.5	1
369	Clonal Evolution and Timing of Metastatic Colorectal Cancer. *Cancers*, **2020**, 12,	6.6	3
368	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	11	10
367	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. *Genetics in Medicine*, **2020**, 22, 2071-2080	8.1	0
366	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. *Brain*, **2020**, 143, 2388-2397	11.2	10
365	A genomics approach to male infertility. *Genetics in Medicine*, **2020**, 22, 1967-1975	8.1	16
364	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. *Brain*, **2020**, 143, 2911-2928	11.2	6
363	Vitamin B12 deficiency secondary to cobalamin F deficiency simulating dyskeratosis congenita. *JAAD Case Reports*, **2020**, 6, 882-885	1.4	0
362	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	5.6	8
361	Bifid nose as the sole manifestation of BNAR syndrome, a FREM1-related condition. *Clinical Genetics*, **2020**, 98, 515-516	4	0
360	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	2.6	3
359	A de novo splicing variant supports the candidacy of TLL1 in ASD pathogenesis. *European Journal of Human Genetics*, **2020**, 28, 525-528	5.3	2
358	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. *Hepatology*, **2020**, 71, 2067-2079	11.2	13
357	A familial PLCB4 mutation causing auriculocondylar syndrome 2 with variable severity. *European Journal of Medical Genetics*, **2020**, 63, 103917	2.6	5
356	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*, **2020**, 50, 102148	1.6	
355	Redefining the Etiologic Landscape of Cerebellar Malformations. *American Journal of Human Genetics*, **2019**, 105, 606-615	11	40
354	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. *American Journal of Human Genetics*, **2019**, 105, 689-705	11	22

353	MDH1 deficiency is a metabolic disorder of the malate-aspartate shuttle associated with early onset severe encephalopathy. *Human Genetics*, **2019**, 138, 1247-1257	6.3	15
352	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. *Brain*, **2019**, 142, 2948-2964	11.2	13
351	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1543-1546	2.5	3
350	Homozygous loss-of-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	4.7	6
349	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611	4.5	7
348	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. *Human Molecular Genetics*, **2019**, 28, 2900-2919	5.6	23
347	PDXK mutations cause polyneuropathy responsive to pyridoxal 5′-phosphate supplementation. *Annals of Neurology*, **2019**, 86, 225-240	9.4	18
346	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. *American Journal of Human Genetics*, **2019**, 104, 1182-1201	11	95
345	Recessive Truncating Mutations in ALKBH8 Cause Intellectual Disability and Severe Impairment of Wobble Uridine Modification. *American Journal of Human Genetics*, **2019**, 104, 1202-1209	11	18
344	The landscape of early infantile epileptic encephalopathy in a consanguineous population. *Seizure: the Journal of the British Epilepsy Association*, **2019**, 69, 154-172	3.2	17
343	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	11	6
342	Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellar-vermis hypoplasia. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1053-1057	2.5	8
341	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	2.5	14
340	Immunodeficiency and EBV-induced lymphoproliferation caused by 4-1BB deficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 144, 574-583.e5	11.5	41
339	De novo truncating variants in WHSC1 recapitulate the Wolf-Hirschhorn (4p16.3 microdeletion) syndrome phenotype. *Genetics in Medicine*, **2019**, 21, 185-188	8.1	15
338	Biallelic variants in CTU2 cause DREAM-PL syndrome and impair thiolation of tRNA wobble U34. *Human Mutation*, **2019**, 40, 2108-2120	4.7	12
337	Formation of tRNA Wobble Inosine in Humans Is Disrupted by a Millennia-Old Mutation Causing Intellectual Disability. *Molecular and Cellular Biology*, **2019**, 39,	4.8	15
336	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	11	29

335	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	11	15
334	Evolution and Impact of Subclonal Mutations in Papillary Thyroid Cancer. *American Journal of Human Genetics*, **2019**, 105, 959-973	11	11
333	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	11	11
332	PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. *Human Genetics*, **2019**, 138, 231-239	6.3	25
331	NUP214 deficiency causes severe encephalopathy and microcephaly in humans. *Human Genetics*, **2019**, 138, 221-229	6.3	9
330	Patterns of neurological manifestations in Woodhouse-Sakati Syndrome. *Parkinsonism and Related Disorders*, **2019**, 69, 99-103	3.6	6
329	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	11	17
328	Genomic and phenotypic delineation of congenital microcephaly. *Genetics in Medicine*, **2019**, 21, 545-552	8.1	55
327	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. *Clinical Genetics*, **2019**, 95, 310-319	4	8
326	Cenani-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	2.5	14
325	Immunologic reconstitution following hematopoietic stem cell transplantation despite lymph node paucity in NF-**B**-inducing kinase deficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 1240-1243.e4	11.5	5
324	EROS/CYBC1 mutations: Decreased NADPH oxidase function and chronic granulomatous disease. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 782-785.e1	11.5	38
323	A novel ISLR2-linked autosomal recessive syndrome of congenital hydrocephalus, arthrogryposis and abdominal distension. *Human Genetics*, **2019**, 138, 105-107	6.3	1
322	Autozygome and high throughput confirmation of disease genes candidacy. *Genetics in Medicine*, **2019**, 21, 736-742	8.1	51
321	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	8.1	41
320	Congenital glaucoma and CYP1B1: an old story revisited. *Human Genetics*, **2019**, 138, 1043-1049	6.3	21
319	is recurrently mutated in Middle Eastern colorectal cancer. *Gut*, **2018**, 67, 663-671	19.2	9
318	Expanding the phenome and variome of skeletal dysplasia. *Genetics in Medicine*, **2018**, 20, 1609-1616	8.1	20

317	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. *Clinical Genetics*, **2018**, 93, 1210-1222	4	25
316	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. *American Journal of Medical Genetics, Part A*, **2018**, 176, 715-721	2.5	3
315	Phenotypic characterization of KCTD3-related developmental epileptic encephalopathy. *Clinical Genetics*, **2018**, 93, 1081-1086	4	5
314	Identification of a novel lethal form of autosomal recessive ichthyosis caused by UDP-glucose ceramide glucosyltransferase deficiency. *Clinical Genetics*, **2018**, 93, 1252-1253	4	8
313	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	9.4	4
312	The alternatively spliced exon of COL5A1 is mutated in autosomal recessive classical Ehlers-Danlos syndrome. *Clinical Genetics*, **2018**, 93, 936-937	4	3
311	Expanding the phenotype of SLC25A42-associated mitochondrial encephalomyopathy. *Clinical Genetics*, **2018**, 93, 1097-1102	4	20
310	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. *American Journal of Human Genetics*, **2018**, 102, 116-132	11	32
309	Elsahy-Waters syndrome is caused by biallelic mutations in CDH11. *American Journal of Medical Genetics, Part A*, **2018**, 176, 477-482	2.5	9
308	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. *American Journal of Human Genetics*, **2018**, 102, 696-705	11	65
307	GWAS signals revisited using human knockouts. *Genetics in Medicine*, **2018**, 20, 64-68	8.1	5
306	WDR45B-related intellectual disability, spastic quadriplegia, epilepsy, and cerebral hypoplasia: A consistent neurodevelopmental syndrome. *Clinical Genetics*, **2018**, 93, 360-364	4	23
305	Exaggerated follicular helper T-cell responses in patients with LRBA deficiency caused by failure of CTLA4-mediated regulation. *Journal of Allergy and Clinical Immunology*, **2018**, 141, 1050-1059.e10	11.5	58
304	Molecular autopsy in maternal-fetal medicine. *Genetics in Medicine*, **2018**, 20, 420-427	8.1	57
303	Phenotypic and Molecular Spectrum of Aicardi-Goutières Syndrome: A Study of 24 Patients. *Pediatric Neurology*, **2018**, 78, 35-40	2.9	28
302	Mutations in known disease genes account for the majority of autosomal recessive retinal dystrophies. *Clinical Genetics*, **2018**, 94, 554-563	4	7
301	Familial non-syndromic macular pseudocoloboma secondary to homozygous CLDN19 mutation. *Ophthalmic Genetics*, **2018**, 39, 577-583	1.2	6
300	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. *American Journal of Human Genetics*, **2018**, 103, 221-231	11	35

299	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. *American Journal of Human Genetics*, **2018**, 102, 858-873	11	37
298	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. *Brain*, **2018**, 141, 1934-1945	11.2	34
297	Further delineation of Malan syndrome. *Human Mutation*, **2018**, 39, 1226-1237	4.7	24
296	Deletion of DDB1- and CUL4- associated factor-17 (Dcaf17) gene causes spermatogenesis defects and male infertility in mice. *Scientific Reports*, **2018**, 8, 9202	4.9	10
295	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. *Journal of Clinical Investigation*, **2018**, 128, 4313-4328	15.9	49
294	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. *American Journal of Human Genetics*, **2018**, 103, 948-967	11	8
293	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. *American Journal of Human Genetics*, **2018**, 103, 612-620	11	41
292	Warsaw breakage syndrome: Further clinical and genetic delineation. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2404-2418	2.5	13
291	Confirming the recessive inheritance of SCN1B mutations in developmental epileptic encephalopathy. *Clinical Genetics*, **2017**, 92, 327-331	4	22
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