Fowzan S Alkuraya

List of Publications by Citations

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	Accelerating novel candidate gene discovery in neurogenetic disorders via whole-exome sequencing of prescreened multiplex consanguineous families. *Cell Reports*, **2015**, 10, 148-61	10.6	262
459	Loss-of-function variant in DNASE1L3 causes a familial form of systemic lupus erythematosus. *Nature Genetics*, **2011**, 43, 1186-8	36.3	259
458	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. *American Journal of Human Genetics*, **2017**, 100, 695-705	11	200
457	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. *Nature Genetics*, **2016**, 48, 1071-6	36.3	192
456	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. *Nature Genetics*, **2011**, 43, 197-203	36.3	190
455	Autozygome-guided exome sequencing in retinal dystrophy patients reveals pathogenetic mutations and novel candidate disease genes. *Genome Research*, **2013**, 23, 236-47	9.7	182
454	LPS-responsive beige-like anchor (LRBA) gene mutation in a family with inflammatory bowel disease and combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2012**, 130, 481-8.e2	11.5	181
453	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nature Genetics*, **2017**, 49, 36-45	36.3	172
452	Mutations in CCNO result in congenital mucociliary clearance disorder with reduced generation of multiple motile cilia. *Nature Genetics*, **2014**, 46, 646-51	36.3	166
451	Homozygous mutations in ADAMTS10 and ADAMTS17 cause lenticular myopia, ectopia lentis, glaucoma, spherophakia, and short stature. *American Journal of Human Genetics*, **2009**, 85, 558-68	11	166
450	Human mutations in NDE1 cause extreme microcephaly with lissencephaly [corrected]. *American Journal of Human Genetics*, **2011**, 88, 536-47	11	165
449	Treatment of retinitis pigmentosa due to MERTK mutations by ocular subretinal injection of adeno-associated virus gene vector: results of a phase I trial. *Human Genetics*, **2016**, 135, 327-43	6.3	156
448	Mutations in the RNA granule component TDRD7 cause cataract and glaucoma. *Science*, **2011**, 331, 1571-6	33.3	145
447	SUMO1 haploinsufficiency leads to cleft lip and palate. *Science*, **2006**, 313, 1751	33.3	144
446	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. *Nature Cell Biology*, **2015**, 17, 1074-1087	23.4	140
445	Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes. *Journal of Medical Genetics*, **2012**, 49, 234-41	5.8	136
444	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. *Human Genetics*, **2017**, 136, 921-939	6.3	132

443	Recessive mutations in ELOVL4 cause ichthyosis, intellectual disability, and spastic quadriplegia. *American Journal of Human Genetics*, **2011**, 89, 745-50	11	130
442	Novel CENPJ mutation causes Seckel syndrome. *Journal of Medical Genetics*, **2010**, 47, 411-4	5.8	129
441	High diagnostic yield of clinical exome sequencing in Middle Eastern patients with Mendelian disorders. *Human Genetics*, **2015**, 134, 967-80	6.3	127
440	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. *Molecular Psychiatry*, **2017**, 22, 615-624	15.1	123
439	Mitochondrial phenylalanyl-tRNA synthetase mutations underlie fatal infantile Alpers encephalopathy. *Human Molecular Genetics*, **2012**, 21, 4521-9	5.6	123
438	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, 1108-17	11	120
437	TLE6 mutation causes the earliest known human embryonic lethality. *Genome Biology*, **2015**, 16, 240	18.3	108
436	Mutations in FBXL4, encoding a mitochondrial protein, cause early-onset mitochondrial encephalomyopathy. *American Journal of Human Genetics*, **2013**, 93, 482-95	11	106
435	Study of autosomal recessive osteogenesis imperfecta in Arabia reveals a novel locus defined by TMEM38B mutation. *Journal of Medical Genetics*, **2012**, 49, 630-5	5.8	105
434	IFT27, encoding a small GTPase component of IFT particles, is mutated in a consanguineous family with Bardet-Biedl syndrome. *Human Molecular Genetics*, **2014**, 23, 3307-15	5.6	103
433	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, 328-33	11	101
432	Mutations in C2orf37, encoding a nucleolar protein, cause hypogonadism, alopecia, diabetes mellitus, mental retardation, and extrapyramidal syndrome. *American Journal of Human Genetics*, **2008**, 83, 684-91	11	100
431	The application of next-generation sequencing in the autozygosity mapping of human recessive diseases. *Human Genetics*, **2013**, 132, 1197-211	6.3	98
430	Homozygosity mapping: one more tool in the clinical geneticist toolbox. *Genetics in Medicine*, **2010**, 12, 236-9	8.1	98
429	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
428	Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. *American Journal of Human Genetics*, **2013**, 92, 598-604	11	90
427	Characterizing the morbid genome of ciliopathies. *Genome Biology*, **2016**, 17, 242	18.3	89
426	In search of triallelism in Bardet-Biedl syndrome. *European Journal of Human Genetics*, **2012**, 20, 420-7	5.3	88

425	Characterization of apparently balanced chromosomal rearrangements from the developmental genome anatomy project. *American Journal of Human Genetics*, **2008**, 82, 712-22	11	84
424	Autozygome decoded. *Genetics in Medicine*, **2010**, 12, 765-71	8.1	83
423	The genetic landscape of familial congenital hydrocephalus. *Annals of Neurology*, **2017**, 81, 890-897	9.4	80
422	NFIA haploinsufficiency is associated with a CNS malformation syndrome and urinary tract defects. *PLoS Genetics*, **2007**, 3, e80	6	80
421	FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. *American Journal of Human Genetics*, **2009**, 85, 756	11	78
420	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. *American Journal of Human Genetics*, **2008**, 83, 425-427	11	78
419	A homozygous truncating mutation in PUS3 expands the role of tRNA modification in normal cognition. *Human Genetics*, **2016**, 135, 707-13	6.3	78
418	Mutation in WDR4 impairs tRNA m(7)G46 methylation and causes a distinct form of microcephalic primordial dwarfism. *Genome Biology*, **2015**, 16, 210	18.3	77
417	Discovery of rare homozygous mutations from studies of consanguineous pedigrees. *Current Protocols in Human Genetics*, **2012**, Chapter 6, Unit6.12	3.2	77
416	Preclinical potency and safety studies of an AAV2-mediated gene therapy vector for the treatment of MERTK associated retinitis pigmentosa. *Human Gene Therapy Clinical Development*, **2013**, 24, 23-8	3.2	75
415	Expanding the genetic heterogeneity of intellectual disability. *Human Genetics*, **2017**, 136, 1419-1429	6.3	74
414	FREM1 mutations cause bifid nose, renal agenesis, and anorectal malformations syndrome. *American Journal of Human Genetics*, **2009**, 85, 414-8	11	74
413	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. *Genetics in Medicine*, **2016**, 18, 554-62	8.1	73
412	Mutations in LRPAP1 are associated with severe myopia in humans. *American Journal of Human Genetics*, **2013**, 93, 313-20	11	72
411	Mutation in PHC1 implicates chromatin remodeling in primary microcephaly pathogenesis. *Human Molecular Genetics*, **2013**, 22, 2200-13	5.6	72
410	Mutation in ADAT3, encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus. *Journal of Medical Genetics*, **2013**, 50, 425-30	5.8	71
409	Neu-Laxova syndrome, an inborn error of serine metabolism, is caused by mutations in PHGDH. *American Journal of Human Genetics*, **2014**, 94, 898-904	11	68
408	Identification of embryonic lethal genes in humans by autozygosity mapping and exome sequencing in consanguineous families. *Genome Biology*, **2015**, 16, 116	18.3	67

407	Exome sequencing reveals a novel Fanconi group defined by XRCC2 mutation. *Journal of Medical Genetics*, **2012**, 49, 184-6	5.8	67
406	Attitude of Saudi families affected with hemoglobinopathies towards prenatal screening and abortion and the influence of religious ruling (Fatwa). *Prenatal Diagnosis*, **2001**, 21, 448-51	3.2	67
405	3-Methylglutaconic aciduria--lessons from 50 genes and 977 patients. *Journal of Inherited Metabolic Disease*, **2013**, 36, 913-21	5.4	66
404	Identification of a novel DLX5 mutation in a family with autosomal recessive split hand and foot malformation. *Journal of Medical Genetics*, **2012**, 49, 16-20	5.8	66
403	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
402	Clinical and molecular characterisation of Bardet-Biedl syndrome in consanguineous populations: the power of homozygosity mapping. *Journal of Medical Genetics*, **2010**, 47, 236-41	5.8	65
401	Katanin p80 regulates human cortical development by limiting centriole and cilia number. *Neuron*, **2014**, 84, 1240-57	13.9	63
400	Mutations in CSPP1, encoding a core centrosomal protein, cause a range of ciliopathy phenotypes in humans. *American Journal of Human Genetics*, **2014**, 94, 73-9	11	63
399	Mutations in MEOX1, encoding mesenchyme homeobox 1, cause Klippel-Feil anomaly. *American Journal of Human Genetics*, **2013**, 92, 157-61	11	61
398	POC1A truncation mutation causes a ciliopathy in humans characterized by primordial dwarfism. *American Journal of Human Genetics*, **2012**, 91, 330-6	11	61
397	A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. *Human Mutation*, **2011**, 32, 573-8	4.7	61
396	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. *Human Genetics*, **2016**, 135, 525-540	6.3	61
395	Identification of ADAMTS18 as a gene mutated in Knobloch syndrome. *Journal of Medical Genetics*, **2011**, 48, 597-601	5.8	59
394	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
393	Ciliary genes TBC1D32/C6orf170 and SCLT1 are mutated in patients with OFD type IX. *Human Mutation*, **2014**, 35, 36-40	4.7	58
392	Complementation of hypersensitivity to DNA interstrand crosslinking agents demonstrates that XRCC2 is a Fanconi anaemia gene. *Journal of Medical Genetics*, **2016**, 53, 672-680	5.8	58
391	Molecular autopsy in maternal-fetal medicine. *Genetics in Medicine*, **2018**, 20, 420-427	8.1	57
390	Molecular characterization of retinitis pigmentosa in Saudi Arabia. *Molecular Vision*, **2009**, 15, 2464-9	2.3	57

389	Genomic and phenotypic delineation of congenital microcephaly. *Genetics in Medicine*, **2019**, 21, 545-552	8.1	55
388	A novel syndrome of Klippel-Feil anomaly, myopathy, and characteristic facies is linked to a null mutation in MYO18B. *Journal of Medical Genetics*, **2015**, 52, 400-4	5.8	54
387	A founder CEP120 mutation in Jeune asphyxiating thoracic dystrophy expands the role of centriolar proteins in skeletal ciliopathies. *Human Molecular Genetics*, **2015**, 24, 1410-9	5.6	54
386	Mutation in MPDZ causes severe congenital hydrocephalus. *Journal of Medical Genetics*, **2013**, 50, 54-8	5.8	54
385	A nullimorphic ERLIN2 mutation defines a complicated hereditary spastic paraplegia locus (SPG18). *Neurogenetics*, **2011**, 12, 333-6	3	54
384	Loss of function mutation in LARP7, chaperone of 7SK ncRNA, causes a syndrome of facial dysmorphism, intellectual disability, and primordial dwarfism. *Human Mutation*, **2012**, 33, 1429-34	4.7	53
383	Mutations in FKBP10 cause both Bruck syndrome and isolated osteogenesis imperfecta in humans. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1448-52	2.5	53
382	The distinct ophthalmic phenotype of Knobloch syndrome in children. *British Journal of Ophthalmology*, **2012**, 96, 890-5	5.5	53
381	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. *Nature Genetics*, **2017**, 49, 537-549	36.3	52
380	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. *American Journal of Human Genetics*, **2017**, 101, 23-36	11	52
379	X-linked creatine transporter defect: a report on two unrelated boys with a severe clinical phenotype. *Journal of Inherited Metabolic Disease*, **2006**, 29, 214-9	5.4	52
378	Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. *Genetics in Medicine*, **2016**, 18, 1244-1249	8.1	52
377	Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. *Genetics in Medicine*, **2017**, 19, 1144-1150	8.1	51
376	Deficiency of a Retinal Dystrophy Protein, Acyl-CoA Binding Domain-containing 5 (ACBD5), Impairs Peroxisomal β-Oxidation of Very-long-chain Fatty Acids. *Journal of Biological Chemistry*, **2017**, 292, 691-705	5.4	51
375	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. *Human Genetics*, **2017**, 136, 205-225	6.3	51
374	Genetics and genomic medicine in Saudi Arabia. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 369-78	2.3	51
373	Genomic analysis of Meckel-Gruber syndrome in Arabs reveals marked genetic heterogeneity and novel candidate genes. *European Journal of Human Genetics*, **2013**, 21, 762-8	5.3	51
372	Autozygome and high throughput confirmation of disease genes candidacy. *Genetics in Medicine*, **2019**, 21, 736-742	8.1	51

371	LOXL3, encoding lysyl oxidase-like 3, is mutated in a family with autosomal recessive Stickler syndrome. *Human Genetics*, **2015**, 134, 451-3	6.3	50
370	Identification of a truncation mutation of acylglycerol kinase (AGK) gene in a novel autosomal recessive cataract locus. *Human Mutation*, **2012**, 33, 960-2	4.7	50
369	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. *Journal of Clinical Investigation*, **2018**, 128, 4313-4328	15.9	49
368	Mutations of KIF14 cause primary microcephaly by impairing cytokinesis. *Annals of Neurology*, **2017**, 82, 562-577	9.4	48
367	Homozygous null mutation in ODZ3 causes microphthalmia in humans. *Genetics in Medicine*, **2012**, 14, 900-4	8.1	48
366	Identification of differentially expressed proteins in the aqueous humor of primary congenital glaucoma. *Experimental Eye Research*, **2011**, 92, 67-75	3.7	47
365	Molecular characterization of Joubert syndrome in Saudi Arabia. *Human Mutation*, **2012**, 33, 1423-8	4.7	46
364	Deficiency of the cytoskeletal protein SPECC1L leads to oblique facial clefting. *American Journal of Human Genetics*, **2011**, 89, 44-55	11	46
363	Tyrosine-mutant AAV8 delivery of human MERTK provides long-term retinal preservation in RCS rats **2012**, 53, 1895-904		44
362	Discovery of mutations for Mendelian disorders. *Human Genetics*, **2016**, 135, 615-23	6.3	44
361	Genomic analysis of pediatric cataract in Saudi Arabia reveals novel candidate disease genes. *Genetics in Medicine*, **2012**, 14, 955-62	8.1	43
360	FKBP10 and Bruck syndrome: phenotypic heterogeneity or call for reclassification?. *American Journal of Human Genetics*, **2010**, 87, 306-7; author reply 308	11	42
359	Immunodeficiency and EBV-induced lymphoproliferation caused by 4-1BB deficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 144, 574-583.e5	11.5	41
358	Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. *Human Genetics*, **2016**, 135, 1295-1298	6.3	41
357	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, 755-9	11	41
356	Increasing the sensitivity of clinical exome sequencing through improved filtration strategy. *Genetics in Medicine*, **2017**, 19, 593-598	8.1	41
355	The syndrome of microcornea, myopic chorioretinal atrophy, and telecanthus (MMCAT) is caused by mutations in ADAMTS18. *Human Mutation*, **2013**, 34, 1195-9	4.7	41
354	Allelic heterogeneity in inbred populations: the Saudi experience with Alstrm syndrome as an illustrative example. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 662-5	2.5	41

353	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
352	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. *American Journal of Human Genetics*, **2018**, 103, 612-620	11	41
351	Redefining the Etiologic Landscape of Cerebellar Malformations. *American Journal of Human Genetics*, **2019**, 105, 606-615	11	40
350	Biometric and molecular characterization of clinically diagnosed posterior microphthalmos. *American Journal of Ophthalmology*, **2013**, 155, 361-372.e7	4.9	40
349	Mutations in SMG9, Encoding an Essential Component of Nonsense-Mediated Decay Machinery, Cause a Multiple Congenital Anomaly Syndrome in Humans and Mice. *American Journal of Human Genetics*, **2016**, 98, 643-52	11	39
348	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 206-217	11	38
347	Lifting the lid on unborn lethal Mendelian phenotypes through exome sequencing. *Genetics in Medicine*, **2013**, 15, 307-9	8.1	38
346	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
345	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
344	Mutations in DDX59 implicate RNA helicase in the pathogenesis of orofaciodigital syndrome. *American Journal of Human Genetics*, **2013**, 93, 555-60	11	37
343	Novel recessive BFSP2 and PITX3 mutations: insights into mutational mechanisms from consanguineous populations. *Genetics in Medicine*, **2011**, 13, 978-81	8.1	37
342	Genomic Profiling of Thyroid Cancer Reveals a Role for Thyroglobulin in Metastasis. *American Journal of Human Genetics*, **2016**, 98, 1170-1180	11	37
341	Autozygome sequencing expands the horizon of human knockout research and provides novel insights into human phenotypic variation. *PLoS Genetics*, **2013**, 9, e1004030	6	36
340	WNT1 mutation with recessive osteogenesis imperfecta and profound neurological phenotype. *Journal of Medical Genetics*, **2013**, 50, 491-2	5.8	36
339	Tufting enteropathy and chronic arthritis: a newly recognized association with a novel EpCAM gene mutation. *Journal of Pediatric Gastroenterology and Nutrition*, **2009**, 49, 642-4	2.8	36
338	GLI3-related polydactyly: a review. *Clinical Genetics*, **2017**, 92, 457-466	4	35
337	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. *Genetics in Medicine*, **2016**, 18, 686-95	8.1	35
336	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. *American Journal of Human Genetics*, **2018**, 103, 221-231	11	35

335	A novel syndrome of hypohidrosis and intellectual disability is linked to COG6 deficiency. *Journal of Medical Genetics*, **2013**, 50, 431-6	5.8	35
334	Mutation of IGFBP7 causes upregulation of BRAF/MEK/ERK pathway and familial retinal arterial macroaneurysms. *American Journal of Human Genetics*, **2011**, 89, 313-9	11	35
333	Mutational spectrum of SLC4A11 in autosomal recessive CHED in Saudi Arabia **2009**, 50, 4142-5		35
332	Female Infertility Caused by Mutations in the Oocyte-Specific Translational Repressor PATL2. *American Journal of Human Genetics*, **2017**, 101, 603-608	11	34
331	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. *Brain*, **2018**, 141, 1934-1945	11.2	34
330	Autozygosity mapping with exome sequence data. *Human Mutation*, **2013**, 34, 50-6	4.7	34
329	A genomics approach to females with infertility and recurrent pregnancy loss. *Human Genetics*, **2020**, 139, 605-613	6.3	33
328	Impact of new genomic tools on the practice of clinical genetics in consanguineous populations: the Saudi experience. *Clinical Genetics*, **2013**, 84, 203-8	4	33
327	NR2F1 deletion in a patient with a de novo paracentric inversion, inv(5)(q15q33.2), and syndromic deafness. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 931-8	2.5	33
326	Revisiting the morbid genome of Mendelian disorders. *Genome Biology*, **2016**, 17, 235	18.3	33
325	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. *American Journal of Human Genetics*, **2018**, 102, 116-132	11	32
324	MYSM1 is mutated in a family with transient transfusion-dependent anemia, mild thrombocytopenia, and low NK- and B-cell counts. *Blood*, **2013**, 122, 3844-5	2.2	32
323	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. *Genome Biology*, **2015**, 16, 293	18.3	32
322	A novel PTF1A mutation in a patient with severe pancreatic and cerebellar involvement. *Clinical Genetics*, **2011**, 80, 196-8	4	32
321	Cone dystrophy with supranormal rod response in children. *British Journal of Ophthalmology*, **2012**, 96, 422-6	5.5	32
320	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	6.3	31
319	Mutation in PLK4, encoding a master regulator of centriole formation, defines a novel locus for primordial dwarfism. *Journal of Medical Genetics*, **2014**, 51, 814-6	5.8	31
318	C2orf37 mutational spectrum in Woodhouse-Sakati syndrome patients. *Clinical Genetics*, **2010**, 78, 585-90	4	31

317	Functional analysis of BBS3 A89V that results in non-syndromic retinal degeneration. *Human Molecular Genetics*, **2011**, 20, 1625-32	5.6	31
316	Molecular characterization of newborn glaucoma including a distinct aniridic phenotype. *Ophthalmic Genetics*, **2011**, 32, 138-42	1.2	31
315	Human knockout research: new horizons and opportunities. *Trends in Genetics*, **2015**, 31, 108-15	8.5	30
314	The morbid genome of ciliopathies: an update. *Genetics in Medicine*, **2020**, 22, 1051-1060	8.1	30
313	Mutations in c12orf57 cause a syndromic form of colobomatous microphthalmia. *American Journal of Human Genetics*, **2013**, 92, 387-91	11	30
312	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessive WNT1 mutations. *Journal of Medical Genetics*, **2016**, 53, 427-30	5.8	30
311	On the phenotypic spectrum of serine biosynthesis defects. *Journal of Inherited Metabolic Disease*, **2016**, 39, 373-381	5.4	30
310	Identification of a novel MKS locus defined by TMEM107 mutation. *Human Molecular Genetics*, **2015**, 24, 5211-8	5.6	29
309	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. *American Journal of Human Genetics*, **2015**, 97, 862-8	11	29
308	Mutations in UNC80, Encoding Part of the UNC79-UNC80-NALCN Channel Complex, Cause Autosomal-Recessive Severe Infantile Encephalopathy. *American Journal of Human Genetics*, **2016**, 98, 210-5	11	29
307	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
306	Molecular and clinical spectra of FBXL4 deficiency. *Human Mutation*, **2017**, 38, 1649-1659	4.7	29
305	Characterization of CTNS mutations in Arab patients with cystinosis. *Ophthalmic Genetics*, **2009**, 30, 185-9	1.2	29
304	Mutation in RAB33B, which encodes a regulator of retrograde Golgi transport, defines a second Dyggve--Melchior--Clausen locus. *Journal of Medical Genetics*, **2012**, 49, 455-61	5.8	29
303	Mutation of CANT1 causes Desbuquois dysplasia. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1157-60	2.5	29
302	International perspectives on the implementation of reproductive carrier screening. *Prenatal Diagnosis*, **2020**, 40, 301-310	3.2	29
301	Cell-Intrinsic Adaptation Arising from Chronic Ablation of a Key Rho GTPase Regulator. *Developmental Cell*, **2016**, 39, 28-43	10.2	29
300	Brittle cornea syndrome ZNF469 mutation carrier phenotype and segregation analysis of rare ZNF469 variants in familial keratoconus. *Investigative Ophthalmology and Visual Science*, **2015**, 56, 578-86		28

299	Phenotypic and Molecular Spectrum of Aicardi-Goutières Syndrome: A Study of 24 Patients. *Pediatric Neurology*, **2018**, 78, 35-40	2.9	28
298	Absence of GP130 cytokine receptor signaling causes extended Stüve-Wiedemann syndrome. *Journal of Experimental Medicine*, **2020**, 217,	16.6	27
297	METTL23, a transcriptional partner of GABPA, is essential for human cognition. *Human Molecular Genetics*, **2014**, 23, 3456-66	5.6	27
296	Mutation of the mitochondrial carrier SLC25A42 causes a novel form of mitochondrial myopathy in humans. *Human Genetics*, **2016**, 135, 21-30	6.3	26
295	Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. *Journal of Inherited Metabolic Disease*, **2013**, 36, 997-1004	5.4	26
294	A novel missense mutation in SCYL1BP1 produces geroderma osteodysplastica phenotype indistinguishable from that caused by nullimorphic mutations. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2093-8	2.5	26
293	Neuronal ceroid lipofuscinosis caused by MFSD8 mutations: a common theme emerging. *Neurogenetics*, **2009**, 10, 307-11	3	26
292	RP1 and retinitis pigmentosa: report of novel mutations and insight into mutational mechanism. *British Journal of Ophthalmology*, **2012**, 96, 1018-22	5.5	26
291	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. *American Journal of Human Genetics*, **2016**, 99, 228-35	11	26
290	Mutations in CIT, encoding citron rho-interacting serine/threonine kinase, cause severe primary microcephaly in humans. *Human Genetics*, **2016**, 135, 1191-7	6.3	26
289	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. *American Journal of Human Genetics*, **2017**, 100, 969-977	11	25
288	A null mutation in MICU2 causes abnormal mitochondrial calcium homeostasis and a severe neurodevelopmental disorder. *Brain*, **2017**, 140, 2806-2813	11.2	25
287	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. *Clinical Genetics*, **2018**, 93, 1210-1222	4	25
286	Impaired telomere maintenance in Alazami syndrome patients with LARP7 deficiency. *BMC Genomics*, **2016**, 17, 749	4.5	25
285	C19orf12 mutation leads to a pallido-pyramidal syndrome. *Gene*, **2014**, 537, 352-6	3.8	25
284	Matching two independent cohorts validates DPH1 as a gene responsible for autosomal recessive intellectual disability with short stature, craniofacial, and ectodermal anomalies. *Human Mutation*, **2015**, 36, 1015-9	4.7	25
283	Severe CNS involvement in WWOX mutations: Description of five new cases. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3209-13	2.5	25
282	A novel X-linked disorder with developmental delay and autistic features. *Annals of Neurology*, **2012**, 71, 498-508	9.4	25

281	Syndromic congenital sensorineural deafness, microtia and microdontia resulting from a novel homoallelic mutation in fibroblast growth factor 3 (FGF3). *European Journal of Human Genetics*, **2009**, 17, 14-21	5.3	25
280	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		25
279	GNB5 mutation causes a novel neuropsychiatric disorder featuring attention deficit hyperactivity disorder, severely impaired language development and normal cognition. *Genome Biology*, **2016**, 17, 195	18.3	25
278	PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. *Human Genetics*, **2019**, 138, 231-239	6.3	25
277	Further delineation of Malan syndrome. *Human Mutation*, **2018**, 39, 1226-1237	4.7	24
276	An autosomal recessive DNASE1L3-related autoimmune disease with unusual clinical presentation mimicking systemic lupus erythematosus. *Lupus*, **2017**, 26, 768-772	2.6	24
275	Blue sclera with and without corneal fragility (brittle cornea syndrome) in a consanguineous family harboring ZNF469 mutation (p.E1392X). *JAMA Ophthalmology*, **2010**, 128, 1376-9		24
274	Genetic profiling of children with advanced cholestatic liver disease. *Clinical Genetics*, **2017**, 92, 52-61	4	23
273	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
272	Positional mapping of PRKD1, NRP1 and PRDM1 as novel candidate disease genes in truncus arteriosus. *Journal of Medical Genetics*, **2015**, 52, 322-9	5.8	23
271	WDR45B-related intellectual disability, spastic quadriplegia, epilepsy, and cerebral hypoplasia: A consistent neurodevelopmental syndrome. *Clinical Genetics*, **2018**, 93, 360-364	4	23
270	GOLGA2, encoding a master regulator of golgi apparatus, is mutated in a patient with a neuromuscular disorder. *Human Genetics*, **2016**, 135, 245-251	6.3	23
269	The human knockout phenotype of PADI6 is female sterility caused by cleavage failure of their fertilized eggs. *Clinical Genetics*, **2017**, 91, 344-345	4	23
268	NECAP1 loss of function leads to a severe infantile epileptic encephalopathy. *Journal of Medical Genetics*, **2014**, 51, 224-8	5.8	23
267	Study of Mendelian forms of Crohn's disease in Saudi Arabia reveals novel risk loci and alleles. *Gut*, **2014**, 63, 1831-2	19.2	23
266	Posterior microphthalmos as a genetically heterogeneous condition that can be allelic to nanophthalmos. *JAMA Ophthalmology*, **2011**, 129, 805-7		23
265	Familial spherophakia with short stature caused by a novel homozygous ADAMTS17 mutation. *Ophthalmic Genetics*, **2012**, 33, 235-9	1.2	23
264	ADAT3-related intellectual disability: Further delineation of the phenotype. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1142-7	2.5	23

263	Mutation in SLC6A9 encoding a glycine transporter causes a novel form of non-ketotic hyperglycinemia in humans. *Human Genetics*, **2016**, 135, 1263-1268	6.3	23
262	Congenital disorders of glycosylation: The Saudi experience. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2614-2621	2.5	23
261	Confirming the recessive inheritance of SCN1B mutations in developmental epileptic encephalopathy. *Clinical Genetics*, **2017**, 92, 327-331	4	22
260	PLAA Mutations Cause a Lethal Infantile Epileptic Encephalopathy by Disrupting Ubiquitin-Mediated Endolysosomal Degradation of Synaptic Proteins. *American Journal of Human Genetics*, **2017**, 100, 706-724	11	22
259	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. *American Journal of Human Genetics*, **2019**, 105, 689-705	11	22
258	A human ciliopathy reveals essential functions for NEK10 in airway mucociliary clearance. *Nature Medicine*, **2020**, 26, 244-251	50.5	22
257	Identification of three novel ECEL1 mutations in three families with distal arthrogryposis type 5D. *Clinical Genetics*, **2014**, 85, 568-72	4	22
256	3M syndrome: an easily recognizable yet underdiagnosed cause of proportionate short stature. *Journal of Pediatrics*, **2012**, 161, 139-45.e1	3.6	22
255	A novel mutation in PRDM5 in brittle cornea syndrome. *Clinical Genetics*, **2012**, 81, 198-9	4	21
254	The syndrome of deafness-dystonia: clinical and genetic heterogeneity. *Movement Disorders*, **2013**, 28, 795-803	7	21
253	Later retinal degeneration following childhood surgical aphakia in a family with recessive CRYAB mutation (p.R56W). *Ophthalmic Genetics*, **2010**, 31, 30-6	1.2	21
252	Hyperekplexia, microcephaly and simplified gyral pattern caused by novel ASNS mutations, case report. *BMC Neurology*, **2016**, 16, 105	3.1	21
251	Congenital glaucoma and CYP1B1: an old story revisited. *Human Genetics*, **2019**, 138, 1043-1049	6.3	21
250	Primordial dwarfism: an update. *Current Opinion in Endocrinology, Diabetes and Obesity*, **2015**, 22, 55-64	4	20
249	Expanding the phenome and variome of skeletal dysplasia. *Genetics in Medicine*, **2018**, 20, 1609-1616	8.1	20
248	Expanding the phenotype of SLC25A42-associated mitochondrial encephalomyopathy. *Clinical Genetics*, **2018**, 93, 1097-1102	4	20
247	Neuronal deficiency of ARV1 causes an autosomal recessive epileptic encephalopathy. *Human Molecular Genetics*, **2016**, 25, 3042-3054	5.6	20
246	Recessive mutations in LEPREL1 underlie a recognizable lens subluxation phenotype. *Ophthalmic Genetics*, **2015**, 36, 58-63	1.2	20

245	Identification of a novel ZNF469 mutation in a large family with Ehlers-Danlos phenotype. *Gene*, **2012**, 511, 447-50	3.8	20
244	A novel APC mutation defines a second locus for Cenani-Lenz syndrome. *Journal of Medical Genetics* , **2015**, 52, 317-21	5.8	19
243	A lethal neonatal phenotype of mitochondrial short-chain enoyl-CoA hydratase-1 deficiency. *Clinical Genetics*, **2017**, 91, 629-633	4	19
242	A novel C2orf37 mutation causes the first Italian cases of Woodhouse Sakati syndrome. *Clinical Genetics*, **2010**, 78, 594-7	4	19
241	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. *Journal of Clinical Investigation*, **2020**, 130, 4423-4439	15.9	19
240	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19
239	PDXK mutations cause polyneuropathy responsive to pyridoxal 5Ꝙphosphate supplementation. *Annals of Neurology*, **2019**, 86, 225-240	9.4	18
238	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
237	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. *Genome Biology*, **2020**, 21, 145	18.3	18
236	Childhood cone-rod dystrophy with macular cystic degeneration from recessive CRB1 mutation. *Ophthalmic Genetics*, **2014**, 35, 130-7	1.2	18
235	The RPGRIP1-related retinal phenotype in children. *British Journal of Ophthalmology*, **2013**, 97, 760-4	5.5	18
234	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. *Nature Communications*, **2020**, 11, 595	17.4	18
233	Whole-Exome Sequencing of Matched Primary and Metastatic Papillary Thyroid Cancer. *Thyroid*, **2020**, 30, 42-56	6.2	18
232	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
231	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
230	Novel IFT122 mutation associated with impaired ciliogenesis and cranioectodermal dysplasia. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 103-6	2.3	17
229	Natural human knockouts and the era of genotype to phenotype. *Genome Medicine*, **2015**, 7, 48	14.4	17
228	ZBTB42 mutation defines a novel lethal congenital contracture syndrome (LCCS6). *Human Molecular Genetics*, **2014**, 23, 6584-93	5.6	17

227	Gonadal mosaicism as a rare cause of autosomal recessive inheritance. *Clinical Genetics*, **2014**, 85, 278-814		17
226	No evidence for locus heterogeneity in Knobloch syndrome. *Journal of Medical Genetics*, **2013**, 50, 565-6	5.8	17
225	Clinical, biochemical and molecular characterization of peroxisomal diseases in Arabs. *Clinical Genetics*, **2011**, 79, 60-70	4	17
224	Founder heterozygous P23T CRYGD mutation associated with cerulean (and coralliform) cataract in 2 Saudi families. *Molecular Vision*, **2009**, 15, 1407-11	2.3	17
223	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
222	Report of a case of Raine syndrome and literature review. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2394-8	2.5	16
221	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. *Genetics in Medicine*, **2015**, 17, 719-25	8.1	16
220	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
219	SET binding factor 1 (SBF1) mutation causes Charcot-Marie-tooth disease type 4B3. *Neurology*, **2014**, 82, 1665-6	6.5	16
218	GM2 gangliosidosis in Saudi Arabia: multiple mutations and considerations for future carrier screening. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1281-4	2.5	16
217	A genomics approach to male infertility. *Genetics in Medicine*, **2020**, 22, 1967-1975	8.1	16
216	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. *New England Journal of Medicine*, **2021**, 384, 2406-2417	59.2	16
215	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
214	DALRD3 encodes a protein mutated in epileptic encephalopathy that targets arginine tRNAs for 3-methylcytosine modification. *Nature Communications*, **2020**, 11, 2510	17.4	15
213	A novel mechanism for variable phenotypic expressivity in Mendelian diseases uncovered by an AU-rich element (ARE)-creating mutation. *Genome Biology*, **2017**, 18, 144	18.3	15
212	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
211	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
210	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

209	Brittle cornea without clinically-evident extraocular findings in an adult harboring a novel homozygous ZNF469 mutation. *Ophthalmic Genetics*, **2012**, 33, 257-9	1.2	15
208	Expanding the spectrum of germline variants in cancer. *Human Genetics*, **2017**, 136, 1431-1444	6.3	14
207	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
206	Further Delineation of the ALG9-CDG Phenotype. *JIMD Reports*, **2016**, 27, 107-12	1.9	14
205	5-Oxoprolinase deficiency: report of the first human OPLAH mutation. *Clinical Genetics*, **2012**, 82, 193-6	4	14
204	Clinical and molecular analysis of children with central pulverulent cataract from the Arabian Peninsula. *British Journal of Ophthalmology*, **2012**, 96, 650-5	5.5	14
203	Helicoid subretinal fibrosis associated with a novel recessive NR2E3 mutation p.S44X. *JAMA Ophthalmology*, **2010**, 128, 344-8		14
202	A null mutation in TNIK defines a novel locus for intellectual disability. *Human Genetics*, **2016**, 135, 773-86	6.3	14
201	ANKS3 is mutated in a family with autosomal recessive laterality defect. *Human Genetics*, **2016**, 135, 1233-1239	6.3	14
200	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
199	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. *Brain*, **2019**, 142, 2948-2964	11.2	13
198	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. *Advances in Experimental Medicine and Biology*, **2017**, 1031, 55-94	3.6	13
197	NPHP4 mutation is linked to cerebello-oculo-renal syndrome and male infertility. *Clinical Genetics*, **2014**, 85, 371-5	4	13
196	Johanson-Blizzard syndrome: report of a novel mutation and severe liver involvement. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1875-9	2.5	13
195	Genetic and genomic analysis of classic aniridia in Saudi Arabia. *Molecular Vision*, **2011**, 17, 708-14	2.3	13
194	Epileptic encephalopathy with continuous spike-and-wave during sleep maps to a homozygous truncating mutation in AMPA receptor component FRRS1L. *Clinical Genetics*, **2016**, 90, 282-3	4	13
193	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. *Hepatology*, **2020**, 71, 2067-2079	11.2	13
192	Warsaw breakage syndrome: Further clinical and genetic delineation. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2404-2418	2.5	13

191	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. *American Journal of Human Genetics*, **2015**, 97, 608-15	11	12
190	The syndrome dysmorphic facies, renal agenesis, ambiguous genitalia, microcephaly, polydactyly and lissencephaly (DREAM-PL): Report of two additional patients. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3222-3226	2.5	12
189	Biallelic variants in CTU2 cause DREAM-PL syndrome and impair thiolation of tRNA wobble U34. *Human Mutation*, **2019**, 40, 2108-2120	4.7	12
188	Congenital glaucoma with acquired peripheral circumferential iris degeneration. *Journal of AAPOS*, **2013**, 17, 105-7	1.3	12
187	Juvenile cataract morphology in 3 siblings not yet diagnosed with cerebrotendinous xanthomatosis. *Ophthalmology*, **2013**, 120, 956-60	7.3	12
186	KDF1, encoding keratinocyte differentiation factor 1, is mutated in a multigenerational family with ectodermal dysplasia. *Human Genetics*, **2017**, 136, 99-105	6.3	12
185	Excessively redundant umbilical skin as a potential early clinical feature of Morquio syndrome and FKBP14-related Ehlers-Danlos syndrome. *Clinical Genetics*, **2014**, 86, 469-72	4	12
184	Familial juvenile glaucoma with underlying homozygous p.G61E CYP1B1 mutations. *Journal of AAPOS*, **2011**, 15, 198-9	1.3	12
183	Colorectal cancer risk is not associated with increased levels of homozygosity in Saudi Arabia. *Genetics in Medicine*, **2013**, 14, 720-728	8.1	12
182	Expanding the "E" in CHARGE. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1890-2	2.5	12
181	The many faces of KIF7. *Human Genome Variation*, **2015**, 2, 15006	1.8	12
180	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
179	A novel mutation in SLC25A46 causes optic atrophy and progressive limb spasticity, with no cerebellar atrophy or axonal neuropathy. *Clinical Genetics*, **2017**, 92, 230-231	4	11
178	Lens subluxation and retinal dysfunction in a girl with homozygous VSX2 mutation. *Ophthalmic Genetics*, **2015**, 36, 8-13	1.2	11
177	ARL6IP6, a susceptibility locus for ischemic stroke, is mutated in a patient with syndromic Cutis Marmorata Telangiectatica Congenita. *Human Genetics*, **2015**, 134, 815-22	6.3	11
176	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
175	Evolution and Impact of Subclonal Mutations in Papillary Thyroid Cancer. *American Journal of Human Genetics*, **2019**, 105, 959-973	11	11
174	Phenotype-genotype correlation in potential female carriers of X-linked developmental cataract (Nance-Horan syndrome). *Ophthalmic Genetics*, **2012**, 33, 89-95	1.2	11

173	Familial dorsalization of the skin of the proximal palm and the instep of the sole of the foot. *Gene*, **2012**, 500, 216-9	3.8	11
172	Dorsal dimelia: report of two cases with an emphasis on the variation of phenotypic expression and a search for candidate causative genes. *Journal of Hand Surgery: European Volume*, **2010**, 35, 715-20	1.4	11
171	Zellweger syndrome caused by PEX13 deficiency: report of two novel mutations. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1219-23	2.5	11
170	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
169	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
168	Pellagra-like condition is xeroderma pigmentosum/Cockayne syndrome complex and niacin confers clinical benefit. *Clinical Genetics*, **2015**, 87, 56-61	4	10
167	Revisiting disease genes based on whole-exome sequencing in consanguineous populations. *Human Genetics*, **2015**, 134, 1029-34	6.3	10
166	Autosomal recessive congenital cataract, intellectual disability phenotype linked to STX3 in a consanguineous Tunisian family. *Clinical Genetics*, **2015**, 88, 283-7	4	10
165	Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. *Orphanet Journal of Rare Diseases*, **2016**, 11, 52	4.2	10
164	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
163	Congenital disorder of glycosylation IIa: the trouble with diagnosing a dysmorphic inborn error of metabolism. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 245-6	2.5	10
162	Corneal decompensation in recessive cornea plana. *Ophthalmic Genetics*, **2009**, 30, 142-5	1.2	10
161	Autozygome maps dispensable DNA and reveals potential selective bias against nullizygosity. *Genetics in Medicine*, **2012**, 14, 515-9	8.1	10
160	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
159	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. *Brain*, **2020**, 143, 2388-2397	11.2	10
158	is recurrently mutated in Middle Eastern colorectal cancer. *Gut*, **2018**, 67, 663-671	19.2	9
157	GZF1 Mutations Expand the Genetic Heterogeneity of Larsen Syndrome. *American Journal of Human Genetics*, **2017**, 100, 831-836	11	9
156	T (brachyury) is linked to a Mendelian form of neural tube defects in humans. *Human Genetics*, **2015**, 134, 1139-41	6.3	9

155	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. *Human Genetics*, **2020**, 139, 1273-1283	6.3	9
154	Elsahy-Waters syndrome is caused by biallelic mutations in CDH11. *American Journal of Medical Genetics, Part A*, **2018**, 176, 477-482	2.5	9
153	Clinical Characterization of LRPAP1-Related Pediatric High Myopia. *Ophthalmology*, **2016**, 123, 434-435	7.3	9
152	Fryns syndrome with Hirschsprung disease: support for possible neural crest involvement. *American Journal of Medical Genetics, Part A*, **2005**, 132A, 226-30	2.5	9
151	Mutations in HYAL2, Encoding Hyaluronidase 2, Cause a Syndrome of Orofacial Clefting and Cor Triatriatum Sinister in Humans and Mice. *PLoS Genetics*, **2017**, 13, e1006470	6	9
150	Confirming the candidacy of THOC6 in the etiology of intellectual disability. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1367-9	2.5	9
149	NUP214 deficiency causes severe encephalopathy and microcephaly in humans. *Human Genetics*, **2019**, 138, 221-229	6.3	9
148	Mutations of PTPN23 in developmental and epileptic encephalopathy. *Human Genetics*, **2017**, 136, 1455-1461	6.3	8
147	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
146	Intrafamilial clinical heterogeneity of CSPP1-related ciliopathy. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2478-80	2.5	8
145	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
144	Ritscher-Schinzel (cranio-cerebello-cardiac, 3C) syndrome: report of four new cases with renal involvement. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1393-7	2.5	8
143	Mental retardation, growth retardation, unusual nose, and open mouth: an autosomal recessive entity. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2160-3	2.5	8
142	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
141	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. *Clinical Genetics*, **2019**, 95, 310-319	4	8
140	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
139	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
138	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611	4.5	7

137	Mutations in known disease genes account for the majority of autosomal recessive retinal dystrophies. *Clinical Genetics*, **2018**, 94, 554-563	4	7
136	Congenital ptosis, scoliosis, and malignant hyperthermia susceptibility in siblings with recessive RYR1 mutations. *Journal of AAPOS*, **2015**, 19, 577-9	1.3	7
135	Expanding the clinical spectrum and allelic heterogeneity in van den Ende-Gupta syndrome. *Clinical Genetics*, **2014**, 85, 492-4	4	7
134	A novel syndrome of lethal familial hyperekplexia associated with brain malformation. *BMC Neurology*, **2012**, 12, 125	3.1	7
133	Methylation-specific multiplex-ligation-dependent probe amplification as a rapid molecular diagnostic tool for pseudohypoparathyroidism type 1b. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 135-9	1.6	7
132	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. *Brain*, **2021**, 144, 1422-1434	11.2	7
131	Novel copy number variants and major limb reduction malformation: Report of three cases. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1245-50	2.5	7
130	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
129	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
128	Familial non-syndromic macular pseudocoloboma secondary to homozygous CLDN19 mutation. *Ophthalmic Genetics*, **2018**, 39, 577-583	1.2	6
127	RGS6: a novel gene associated with congenital cataract, mental retardation, and microcephaly in a Tunisian family. *Investigative Ophthalmology and Visual Science*, **2014**, 56, 1261-6		6
126	A novel syndromic form of sensory-motor polyneuropathy is linked to chromosome 22q13.31-q13.33. *Clinical Genetics*, **2011**, 79, 193-5	4	6
125	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
124	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. *Brain*, **2020**, 143, 2911-2928	11.2	6
123	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
122	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
121	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 families. *Molecular Genetics and Metabolism*, **2016**, 119, 44-9	3.7	6
120	Patterns of neurological manifestations in Woodhouse-Sakati Syndrome. *Parkinsonism and Related Disorders*, **2019**, 69, 99-103	3.6	6

119	The natural history of infantile neuroaxonal dystrophy. *Orphanet Journal of Rare Diseases*, **2020**, 15, 109	4.2	5
118	Confirming TBC1D32-related ciliopathy in humans. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1985-1987	2.5	5
117	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
116	Phenotypic expansion of OTUD6B-related syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1530-1531	2.5	5
115	CNP deficiency causes severe hypomyelinating leukodystrophy in humans. *Human Genetics*, **2020**, 139, 615-622	6.3	5
114	Phenotypic characterization of KCTD3-related developmental epileptic encephalopathy. *Clinical Genetics*, **2018**, 93, 1081-1086	4	5
113	GWAS signals revisited using human knockouts. *Genetics in Medicine*, **2018**, 20, 64-68	8.1	5
112	Human knockouts of PLA2G4A phenocopy NSAID-induced gastrointestinal and renal toxicity. *Gut*, **2016**, 65, 1575-7	19.2	5
111	A syndrome of congenital hyperinsulinism and rhabdomyolysis is caused by KCNJ11 mutation. *Journal of Medical Genetics*, **2014**, 51, 271-4	5.8	5
110	A case of de Barsy syndrome with a severe eye phenotype. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2364-6	2.5	5
109	CYP1B1 analysis of unilateral primary newborn glaucoma in Saudi children. *Journal of AAPOS*, **2012**, 16, 571-2	1.3	5
108	Identification of MRI1, encoding translation initiation factor eIF-2B subunit alpha/beta/delta-like protein, as a candidate locus for infantile epilepsy with severe cystic degeneration of the brain. *Gene*, **2013**, 512, 450-2	3.8	5
107	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, 447-9	2.5	5
106	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
105	Immunologic reconstitution following hematopoietic stem cell transplantation despite lymph node paucity in NF-κB-inducing kinase deficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 1240-1243.e4	11.5	5
104	A familial PLCB4 mutation causing auriculocondylar syndrome 2 with variable severity. *European Journal of Medical Genetics*, **2020**, 63, 103917	2.6	5
103	Population structure of indigenous inhabitants of Arabia. *PLoS Genetics*, **2021**, 17, e1009210	6	5
102	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. *American Journal of Human Genetics*, **2020**, 107, 963-976	11	4

101	Multiple Family Members With Delayed Cord Separtion and Combined Immunodeficiency With Novel Mutation in. *Frontiers in Pediatrics*, **2020**, 8, 9	3.4	4
100	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
99	Crisponi/CISS1 syndrome: A case series. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1236-41	2.5	4
98	Distal acroosteolysis, poikiloderma and joint stiffness: a novel laminopathy?. *European Journal of Human Genetics*, **2016**, 24, 1220-2	5.3	4
97	Complete aniridia with central keratopathy and congenital glaucoma is a CYP1B1-related phenotype. *Ophthalmic Genetics*, **2014**, 35, 187-9	1.2	4
96	Vanishing white matter disease caused by EIF2B2 mutation with the presentation of an adrenoleukodystrophy phenotype. *Gene*, **2012**, 496, 141-3	3.8	4
95	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, 489-92	4	4
94	Arthrogryposis, perthes disease, and upward gaze palsy: a novel autosomal recessive syndromic form of arthrogryposis. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 297-300	2.5	4
93	Genomic medicine in the Middle East. *Genome Medicine*, **2021**, 13, 184	14.4	4
92	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. *Frontiers in Genetics*, **2020**, 11, 580484	4.5	4
91	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. *Clinical Genetics*, **2020**, 97, 447-456	4	4
90	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. *American Journal of Human Genetics*, **2021**, 108, 1069-1082	11	4
89	Gonadal mosaicism for ACTA1 gene masquerading as autosomal recessive nemaline myopathy. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2219-21	2.5	4
88	Mutations in TP73 cause impaired mucociliary clearance and lissencephaly. *American Journal of Human Genetics*, **2021**, 108, 1318-1329	11	4
87	The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. *Genetics in Medicine*, **2022**,	8.1	4
86	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
85	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
84	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

83	The alternatively spliced exon of COL5A1 is mutated in autosomal recessive classical Ehlers-Danlos syndrome. *Clinical Genetics*, **2018**, 93, 936-937	4	3
82	Elevation deficiency in children with recessive RDH12-related retinopathy. *Journal of AAPOS*, **2015**, 19, 568-70	1.3	3
81	Trichorhinophalangeal syndrome: Report of a novel familial TRPS1 mutation. *Clinical Dysmorphology*, **2010**, 19, 98-100	0.9	3
80	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. *American Journal of Human Genetics*, **2010**, 87, 571	11	3
79	Perturbation of the consensus activation site of endothelin-3 leads to Waardenburg syndrome type IV. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1841-3	2.5	3
78	Recurrent miscarriage in a carrier of a balanced cytogenetically undetectable subtelomeric rearrangement: how many are we missing?. *Prenatal Diagnosis*, **2006**, 26, 291-3	3.2	3
77	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. *Genome Medicine* , **2021**, 13, 161	14.4	3
76	Rhegmatogenous Retinal Detachment in Nonsyndromic High Myopia Associated with Recessive Mutations in LRPAP1. *Ophthalmology Retina*, **2020**, 4, 77-83	3.8	3
75	A de novo mutation in FMR1 in a patient with intellectual disability. *European Journal of Medical Genetics*, **2020**, 63, 103763	2.6	3
74	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
73	Clonal Evolution and Timing of Metastatic Colorectal Cancer. *Cancers*, **2020**, 12,	6.6	3
72	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
71	A de novo TBX3 mutation presenting as dorsalization of the little fingers: A forme fruste phenotype of ulnar-mammary syndrome. *European Journal of Medical Genetics*, **2020**, 63, 103615	2.6	3
70	Further delineation of SMG9-related heart and brain malformation syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1624-1630	2.5	3
69	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
68	Further delineation of METTL23-associated intellectual disability. *American Journal of Medical Genetics, Part A*, **2020**, 182, 785-791	2.5	2
67	Recessive mutations in SCYL2 cause a novel syndromic form of arthrogryposis in humans. *Human Genetics*, **2020**, 139, 513-519	6.3	2
66	A lethal phenotype associated with tissue plasminogen deficiency in humans. *Human Genetics*, **2016** , 135, 1209-11	6.3	2

65	Joint laxity in homozygotes for severe POU1F1 mutations. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3356-3358	2.5	2
64	Corneal enlargement without optic disk cupping in children with recessive CYP1B1 mutations. *Journal of AAPOS*, **2013**, 17, 643-5	1.3	2
63	Expanding the allelic disorders linked to TCTN1 to include Varadi syndrome (Orofaciodigital syndrome type VI). *American Journal of Medical Genetics, Part A*, **2017**, 173, 2439-2441	2.5	2
62	Variable phenotypic expression of COG6 mutations. *Journal of Medical Genetics*, **2014**, 51, 425-6	5.8	2
61	Study of consanguineous populations can improve the annotation of SNP databases. *European Journal of Medical Genetics*, **2011**, 54, 118-20	2.6	2
60	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. *American Journal of Human Genetics*, **2011**, 88, 677	11	2
59	Saudi genetic ophthalmology research: The local and global impact. *Saudi Journal of Ophthalmology* , **2010**, 24, 109-10	0.9	2
58	Smith-Lemli-Opitz syndrome in trisomy 13: how does the mix work?. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2005**, 73, 569-71		2
57	Insight into ALKBH8-related intellectual developmental disability based on the first pathogenic missense variant. *Human Genetics*, **2021**, 1	6.3	2
56	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
55	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
54	CHEDDA syndrome is an underrecognized neurodevelopmental disorder with a highly restricted ATN1 mutation spectrum. *Clinical Genetics*, **2021**, 100, 468-477	4	2
53	PLXNA2 as a candidate gene in patients with intellectual disability. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3859-3865	2.5	2
52	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
51	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. *Brain and Development*, **2021**, 43, 380-388	2.2	2
50	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
49	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. *Kidney International Reports*, **2021**, 6, 460-471	4.1	2
48	Successful hematopoietic stem cell transplantation in a 4-1BB deficient patient with EBV-induced lymphoproliferation. *Clinical Immunology*, **2021**, 222, 108639	9	2

47	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. *Brain*, **2021**,	11.2	2
46	Residual risk for additional recessive diseases in consanguineous couples. *Genetics in Medicine*, **2021**, 23, 2448-2454	8.1	2
45	Molecular autopsy by proxy in preconception counseling. *Clinical Genetics*, **2021**, 100, 678-691	4	2
44	Recurrent spontaneous oocyte activation causes female infertility.. *Journal of Assisted Reproduction and Genetics*, **2022**, 39, 675	3.4	2
43	Reply to "an extremely severe phenotype due to WDR81 nonsense mutations". *Annals of Neurology* , **2017**, 82, 651	9.4	1
42	Simple and efficient identification of rare recessive pathologically important sequence variants from next generation exome sequence data. *Human Mutation*, **2013**, 34, 945-52	4.7	1
41	Index of suspicion. *Pediatrics in Review*, **2004**, 25, 289-94	1.1	1
40	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
39	An RNA-seq quantification method for analysis of transcriptional aberrations		1
38	Confirming the recessive inheritance of PERP-related erythrokeratoderma. *Clinical Genetics*, **2020**, 97, 661-665	4	1
37	Further delineation of HIDEA syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2999-3006	2.5	1
36	MYH1 is a candidate gene for recurrent rhabdomyolysis in humans. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2131-2135	2.5	1
35	Mutations in phospholipase C eta-1 () are associated with holoprosencephaly. *Journal of Medical Genetics*, **2021**,	5.8	1
34	Mutations in HID1 Cause Syndromic Infantile Encephalopathy and Hypopituitarism. *Annals of Neurology*, **2021**, 90, 143-158	9.4	1
33	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
32	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
31	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. *American Journal of Human Genetics*, **2021**, 108, 1095-1114	11	1
30	Congenital hereditary endothelial dystrophy, not glaucoma, in a child with iris colobomas. *Journal of AAPOS*, **2016**, 20, 370-2	1.3	1

29	A novel ISLR2-linked autosomal recessive syndrome of congenital hydrocephalus, arthrogryposis and abdominal distension. *Human Genetics*, **2019**, 138, 105-107	6.3	1
28	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
27	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
26	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. *Genetics in Medicine*, **2021**, 23, 661-668	8.1	1
25	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
24	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
23	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
22	Trisomy 8 mosaicism in a patient with heterotaxia. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2005**, 73, 58-60		0
21	ASTL is mutated in female infertility. *Human Genetics*, **2021**, 1	6.3	0
20	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. *Genetics in Medicine*, **2020**, 22, 2071-2080	8.1	0
19	Vitamin B12 deficiency secondary to cobalamin F deficiency simulating dyskeratosis congenita. *JAAD Case Reports*, **2020**, 6, 882-885	1.4	0
18	Bifid nose as the sole manifestation of BNAR syndrome, a FREM1-related condition. *Clinical Genetics*, **2020**, 98, 515-516	4	0
17	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
16	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
15	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
14	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
13	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
12	Expanding the KIF4A-associated phenotype. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3728-3739	2.5	0

List of Publications

11	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
10	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. *Journal of Molecular Medicine*, **2021**, 99, 1755-1768	5.5	0
9	Response to Yehia et al. *American Journal of Human Genetics*, **2017**, 100, 564-565	11	
8	MGAT2-CDG (CDG-IIa) and dysmorphism **2012**, 158A, 2976-2976		
7	A Diagnostic Approach for Neurogenetic Disorders in the Genome Era **2020**, 319-326		
6	Mitochondrial "dysmorphology" in variant classification. *Human Genetics*, **2021**, 1	6.3	
5	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. *Human Genetics*, **2021**, 140, 1395-1401	6.3	
4	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	
3	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	
2	Response to Hamosh et al. *American Journal of Human Genetics*, **2021**, 108, 1809-1810	11	
1	PLACK syndrome is potentially treatable with intralipids. *Clinical Genetics*, **2021**, 99, 572-576	4	