

Amal M Alhashem

List of Publications by Year in descending order

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Version: 2024-02-01

85
papers

3,344
citations

196777

29
h-index

190340

53
g-index

89
all docs

89
docs citations

89
times ranked

7389
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in phospholipase C eta-1 (<i>PLCH1</i>) are associated with holoprosencephaly. <i>Journal of Medical Genetics</i> , 2022, 59, 358-365.	1.5	3
2	Biallelic <i>ZNFX1</i> variants are associated with a spectrum of immunohematological abnormalities. <i>Clinical Genetics</i> , 2022, 101, 247-254.	1.0	12
3	Mitochondrial œdysmorphology in variant classification. <i>Human Genetics</i> , 2022, 141, 55-64.	1.8	0
4	Further clinical and genetic evidence of ASC-1 complex dysfunction in congenital neuromuscular disease. <i>European Journal of Medical Genetics</i> , 2022, 65, 104537.	0.7	3
5	A Biallelic Variant in <i>FRA10AC1</i> Is Associated With Neurodevelopmental Disorder and Growth Retardation. <i>Neurology: Genetics</i> , 2022, 8, e200010.	0.9	2
6	Bi-allelic loss-of-function variants in <i>PPFIBP1</i> cause a neurodevelopmental disorder with microcephaly, epilepsy, and periventricular calcifications. <i>American Journal of Human Genetics</i> , 2022, 109, 1421-1435.	2.6	6
7	Survey of disorders of sex development in a large cohort of patients with diverse Mendelian phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2789-2800.	0.7	7
8	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. <i>Brain and Development</i> , 2021, 43, 380-388.	0.6	6
9	Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. <i>European Journal of Human Genetics</i> , 2021, 29, 141-153.	1.4	66
10	Further delineation of <i>SMG9</i> -related heart and brain malformation syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1624-1630.	0.7	3
11	Combining exome/genome sequencing with data repository analysis reveals novel gene-disease associations for a wide range of genetic disorders. <i>Genetics in Medicine</i> , 2021, 23, 1551-1568.	1.1	30
12	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. <i>Frontiers in Pediatrics</i> , 2021, 9, 633385.	0.9	15
13	The genotypic and phenotypic spectrum of pycnodysostosis in Saudi Arabia: Novel variants and clinical findings. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2455-2463.	0.7	0
14	Biallelic variants in <i>KARS1</i> are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. <i>Genetics in Medicine</i> , 2021, 23, 1933-1943.	1.1	11
15	Hypospadias in ring X syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104225.	0.7	1
16	Progressive symmetrical erythrokeratoderma manifesting as harlequin-like ichthyosis with severe thrombocytopenia secondary to a homozygous 3-ketodihydrosphingosine reductase mutation. <i>JAAD Case Reports</i> , 2021, 14, 55-58.	0.4	4
17	Molecular autopsy by proxy in preconception counseling. <i>Clinical Genetics</i> , 2021, 100, 678-691.	1.0	6
18	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. <i>Genome Medicine</i> , 2021, 13, 161.	3.6	13

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19	The phenotypic spectrum of dihydrolipoamide dehydrogenase deficiency in Saudi Arabia. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100817.	0.4	1
20	A de novo splicing variant supports the candidacy of TLL1 in ASD pathogenesis. <i>European Journal of Human Genetics</i> , 2020, 28, 525-528.	1.4	4
21	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. <i>Hepatology</i> , 2020, 71, 2067-2079.	3.6	28
22	Categorized Genetic Analysis in Childhood-Onset Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, 504-514.	1.6	18
23	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. <i>Human Genetics</i> , 2020, 139, 1273-1283.	1.8	16
24	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. <i>Genome Biology</i> , 2020, 21, 145.	3.8	59
25	Recurrent homozygous damaging mutation in <i>TMX2</i> , encoding a protein disulfide isomerase, in four families with microlissencephaly. <i>Journal of Medical Genetics</i> , 2020, 57, 274-282.	1.5	6
26	The morbid genome of ciliopathies: an update. <i>Genetics in Medicine</i> , 2020, 22, 1051-1060.	1.1	68
27	Peripheral venous route for administration of ammonium infusion for treatment of acute hyperammonemia. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2020, 41, 98-101.	0.5	0
28	ADAMTS19-associated heart valve defects: Novel genetic variants consolidating a recognizable cardiac phenotype. <i>Clinical Genetics</i> , 2020, 98, 56-63.	1.0	11
29	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. <i>Frontiers in Genetics</i> , 2020, 11, 580484.	1.1	13
30	Fructose-1,6-bisphosphatase deficiency with confirmed molecular diagnosis. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2020, 41, 199-202.	0.5	5
31	Molecular and clinical characteristics of very long-chain acyl-CoA dehydrogenase deficiency. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2020, 41, 590-596.	0.5	1
32	Epilepsy, neuropsychiatric phenotypes, neuroimaging findings, and genotype-neurophenotype correlation in 22q11.2 deletion syndrome. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2020, 25, 287-291.	0.5	0
33	Incidence of newborn screening disorders among 56632 infants in Central Saudi Arabia. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2020, 41, 703-708.	0.5	13
34	Truncating ARL6IP1 variant as the genetic cause of fatal complicated hereditary spastic paraplegia. <i>BMC Medical Genetics</i> , 2019, 20, 119.	2.1	14
35	Genetic, clinical and biochemical characterization of a large cohort of patients with hyaline fibromatosis syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 209.	1.2	7
36	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyposis. <i>American Journal of Human Genetics</i> , 2019, 105, 689-705.	2.6	48

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37	MDH1 deficiency is a metabolic disorder of the malate-aspartate shuttle associated with early onset severe encephalopathy. <i>Human Genetics</i> , 2019, 138, 1247-1257.	1.8	31
38	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. <i>American Journal of Human Genetics</i> , 2019, 104, 1182-1201.	2.6	184
39	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. <i>Genetics in Medicine</i> , 2019, 21, 2521-2531.	1.1	25
40	Patterns, prevalence, risk factors, and survival of newborns with congenital heart defects in a Saudi population: a three-year, cohort case-control study. <i>Journal of Congenital Cardiology</i> , 2019, 3, .	0.5	10
41	6-Pyruvoyltetrahydropterin Synthase Deficiency: Review and Report of 28 Arab Subjects. <i>Pediatric Neurology</i> , 2019, 96, 40-47.	1.0	12
42	A homozygous frameshift variant in an alternatively spliced exon of <i>DLG5</i> causes hydrocephalus and renal dysplasia. <i>Clinical Genetics</i> , 2019, 95, 631-633.	1.0	5
43	Congenital anomalies and associated risk factors in a Saudi population: a cohort study from pregnancy to age 2 years. <i>BMJ Open</i> , 2019, 9, e026351.	0.8	17
44	Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , 2019, 21, 545-552.	1.1	85
45	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. <i>Clinical Genetics</i> , 2019, 95, 310-319.	1.0	12
46	Autozygome and high throughput confirmation of disease genes candidacy. <i>Genetics in Medicine</i> , 2019, 21, 736-742.	1.1	81
47	Novel Homozygous Mutation of the AIMP1 Gene: A Milder Neuroimaging Phenotype With Preservation of the Deep White Matter. <i>Pediatric Neurology</i> , 2019, 91, 57-61.	1.0	7
48	Identification of novel loci for pediatric cholestatic liver disease defined by KIF12, PPM1F, USP53, LSR, and WDR83OS pathogenic variants. <i>Genetics in Medicine</i> , 2019, 21, 1164-1172.	1.1	71
49	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , 2018, 20, 1609-1616.	1.1	46
50	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 715-721.	0.7	7
51	Molecular autopsy in maternal-fetal medicine. <i>Genetics in Medicine</i> , 2018, 20, 420-427.	1.1	84
52	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. <i>American Journal of Human Genetics</i> , 2018, 103, 612-620.	2.6	70
53	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). <i>Human Genetics</i> , 2018, 137, 753-768.	1.8	38
54	Embryopathy Associated With a Vitamin Therapy. <i>Pediatric Neurology</i> , 2018, 89, 73-74.	1.0	1

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55	FARS2 deficiency; new cases, review of clinical, biochemical, and molecular spectra, and variants interpretation based on structural, functional, and evolutionary significance. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 281-291.	0.5	28
56	Clinical profile and mutation spectrum of long QT syndrome in Saudi Arabia: The impact of consanguinity. <i>Heart Rhythm</i> , 2017, 14, 1191-1199.	0.3	13
57	Recessive AFG3L2 Mutation Causes Progressive Microcephaly, Early Onset Seizures, Spasticity, and Basal Ganglia Involvement. <i>Pediatric Neurology</i> , 2017, 71, 24-28.	1.0	19
58	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. <i>Human Genetics</i> , 2017, 136, 921-939.	1.8	209
59	A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 91-95.	0.5	68
60	Expanding the genetic heterogeneity of intellectual disability. <i>Human Genetics</i> , 2017, 136, 1419-1429.	1.8	122
61	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. <i>European Journal of Human Genetics</i> , 2017, 25, 176-182.	1.4	291
62	Congenital disorders of glycosylation: The Saudi experience. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2614-2621.	0.7	31
63	Neuroregression, coarse features, and oligosaccharides in urines. <i>Neurosciences</i> , 2017, 22, 326-328.	0.1	0
64	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. <i>Human Genetics</i> , 2016, 135, 525-540.	1.8	89
65	Severe early-onset epileptic encephalopathy due to mutations in the KCNA2 gene: Expansion of the genotypic and phenotypic spectrum. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 657-660.	0.7	33
66	Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. <i>Human Genetics</i> , 2016, 135, 1295-1298.	1.8	65
67	Crisponi/CISS1 syndrome: A case series. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1236-1241.	0.7	5
68	Distal acroosteolysis, poikiloderma and joint stiffness: a novel laminopathy?. <i>European Journal of Human Genetics</i> , 2016, 24, 1220-1222.	1.4	6
69	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. <i>Genetics in Medicine</i> , 2016, 18, 686-695.	1.1	55
70	Severe CNS involvement in <i>WWOX</i> mutations: Description of five new cases. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3209-3213.	0.7	50
71	Effect of consanguinity on birth defects in Saudi women: Results from a nested case-control study. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2015, 103, 100-104.	1.6	28
72	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. <i>Genome Biology</i> , 2015, 16, 293.	3.8	56

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73	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. <i>Cell Reports</i> , 2015, 10, 148-161.	2.9	375
74	Identification of a novel MKS locus defined by <i>TMEM107</i> mutation. <i>Human Molecular Genetics</i> , 2015, 24, 5211-5218.	1.4	42
75	Further Delineation of the ALC9-CDG Phenotype. <i>JIMD Reports</i> , 2015, 27, 107-112.	0.7	17
76	IFT27, encoding a small GTPase component of IFT particles, is mutated in a consanguineous family with Bardet-Biedl syndrome. <i>Human Molecular Genetics</i> , 2014, 23, 3307-3315.	1.4	134
77	Ciliary Genes <i>TBC1D32</i> and <i>C6orf170</i> and <i>SCLT1</i> are Mutated in Patients with OFD Type IX. <i>Human Mutation</i> , 2014, 35, 36-40.	1.1	78
78	A Wide Clinical Phenotype Spectrum in Patients With <i>ATP1A2</i> Mutations. <i>Journal of Child Neurology</i> , 2014, 29, 265-268.	0.7	17
79	Neu-Laxova Syndrome, an Inborn Error of Serine Metabolism, Is Caused by Mutations in PHGDH. <i>American Journal of Human Genetics</i> , 2014, 94, 898-904.	2.6	93
80	Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. <i>American Journal of Human Genetics</i> , 2013, 93, 555-560.	2.6	45
81	Smith-Lemli-Opitz syndrome among Arabs. <i>Clinical Genetics</i> , 2012, 82, 165-172.	1.0	9
82	Munchausen syndrome by proxy mimicking as Gaucher disease. <i>European Journal of Pediatrics</i> , 2010, 169, 1029-1032.	1.3	9
83	Vici syndrome associated with unilateral lung hypoplasia and myopathy. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1849-1853.	0.7	33
84	An atypical presentation of severe congenital contractures and lack of cerebellar involvement in a patient with a novel LAMA1 mutation. <i>Journal of Biochemical and Clinical Genetics</i> , 0, , 43-46.	0.1	1
85	De Novo Ring Chromosome 15: Molecular Cytogenetic and Clinical Characterization of First Case from Saudi Arabia. <i>Journal of Pediatric Genetics</i> , 0, , .	0.3	1