

# Amal M Alhashem

## List of Publications by Year in descending order

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85  
papers

3,344  
citations

172457

29  
h-index

168389

53  
g-index

89  
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89  
docs citations

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times ranked

6905  
citing authors

#	ARTICLE	IF	CITATIONS
1	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.	6.4	375
2	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. <i>European Journal of Human Genetics</i> , 2017, 25, 176-182.	2.8	291
3	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.	3.8	209
4	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.	6.2	184
5	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.	2.9	134
6	Expanding the genetic heterogeneity of intellectual disability. <i>Human Genetics</i> , 2017, 136, 1419-1429.	3.8	122
7	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.	6.2	93
8	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. <i>Human Genetics</i> , 2016, 135, 525-540.	3.8	89
9	Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , 2019, 21, 545-552.	2.4	85
10	Molecular autopsy in maternal-fetal medicine. <i>Genetics in Medicine</i> , 2018, 20, 420-427.	2.4	84
11	Autozygome and high throughput confirmation of disease genes candidacy. <i>Genetics in Medicine</i> , 2019, 21, 736-742.	2.4	81
12	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.	2.5	78
13	Identification of novel loci for pediatric cholestatic liver disease defined by <i>KIF12</i> , <i>PPM1F</i> , <i>USP53</i> , <i>LSR</i> , and <i>WDR83OS</i> pathogenic variants. <i>Genetics in Medicine</i> , 2019, 21, 1164-1172.	2.4	71
14	<i>ARL3</i> Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. <i>American Journal of Human Genetics</i> , 2018, 103, 612-620.	6.2	70
15	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.	1.1	68
16	The morbid genome of ciliopathies: an update. <i>Genetics in Medicine</i> , 2020, 22, 1051-1060.	2.4	68
17	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.	2.8	66
18	Homozygous <i>KCNMA1</i> mutation as a cause of cerebellar atrophy, developmental delay and seizures. <i>Human Genetics</i> , 2016, 135, 1295-1298.	3.8	65

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19	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. <i>Genome Biology</i> , 2020, 21, 145.	8.8	59
20	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. <i>Genome Biology</i> , 2015, 16, 293.	8.8	56
21	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. <i>Genetics in Medicine</i> , 2016, 18, 686-695.	2.4	55
22	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.	1.2	50
23	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyposis. <i>American Journal of Human Genetics</i> , 2019, 105, 689-705.	6.2	48
24	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , 2018, 20, 1609-1616.	2.4	46
25	Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. <i>American Journal of Human Genetics</i> , 2013, 93, 555-560.	6.2	45
26	Identification of a novel MKS locus defined by <i>TMEM107</i> mutation. <i>Human Molecular Genetics</i> , 2015, 24, 5211-5218.	2.9	42
27	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.	3.8	38
28	Vici syndrome associated with unilateral lung hypoplasia and myopathy. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1849-1853.	1.2	33
29	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.	1.6	33
30	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.	3.8	31
31	Congenital disorders of glycosylation: The Saudi experience. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2614-2621.	1.2	31
32	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.	2.4	30
33	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
34	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.	1.1	28
35	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. <i>Hepatology</i> , 2020, 71, 2067-2079.	7.3	28
36	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. <i>Genetics in Medicine</i> , 2019, 21, 2521-2531.	2.4	25

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37	Recessive AFG3L2 Mutation Causes Progressive Microcephaly, Early Onset Seizures, Spasticity, and Basal Ganglia Involvement. <i>Pediatric Neurology</i> , 2017, 71, 24-28.	2.1	19
38	Categorized Genetic Analysis in Childhood-Onset Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, 504-514.	3.6	18
39	A Wide Clinical Phenotype Spectrum in Patients With <i>ATP1A2</i> Mutations. <i>Journal of Child Neurology</i> , 2014, 29, 265-268.	1.4	17
40	Further Delineation of the ALG9-CDG Phenotype. <i>JIMD Reports</i> , 2015, 27, 107-112.	1.5	17
41	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.	1.9	17
42	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. <i>Human Genetics</i> , 2020, 139, 1273-1283.	3.8	16
43	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. <i>Frontiers in Pediatrics</i> , 2021, 9, 633385.	1.9	15
44	Truncating ARL6IP1 variant as the genetic cause of fatal complicated hereditary spastic paraplegia. <i>BMC Medical Genetics</i> , 2019, 20, 119.	2.1	14
45	Clinical profile and mutation spectrum of long QT syndrome in Saudi Arabia: The impact of consanguinity. <i>Heart Rhythm</i> , 2017, 14, 1191-1199.	0.7	13
46	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. <i>Frontiers in Genetics</i> , 2020, 11, 580484.	2.3	13
47	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. <i>Genome Medicine</i> , 2021, 13, 161.	8.2	13
48	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.	1.1	13
49	6-Pyruvoyltetrahydropterin Synthase Deficiency: Review and Report of 28 Arab Subjects. <i>Pediatric Neurology</i> , 2019, 96, 40-47.	2.1	12
50	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. <i>Clinical Genetics</i> , 2019, 95, 310-319.	2.0	12
51	Biallelic <i>ZNF1</i> variants are associated with a spectrum of immunohematological abnormalities. <i>Clinical Genetics</i> , 2022, 101, 247-254.	2.0	12
52	ADAMTS19-associated heart valve defects: Novel genetic variants consolidating a recognizable cardiac phenotype. <i>Clinical Genetics</i> , 2020, 98, 56-63.	2.0	11
53	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.	2.4	11
54	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

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55	Munchausen syndrome by proxy mimicking as Gaucher disease. <i>European Journal of Pediatrics</i> , 2010, 169, 1029-1032.	2.7	9
56	Smith's "Lemli" Opitz syndrome among Arabs. <i>Clinical Genetics</i> , 2012, 82, 165-172.	2.0	9
57	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 715-721.	1.2	7
58	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.	2.7	7
59	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.	2.1	7
60	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.	1.2	7
61	Distal acroosteolysis, poikiloderma and joint stiffness: a novel laminopathy?. <i>European Journal of Human Genetics</i> , 2016, 24, 1220-1222.	2.8	6
62	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.	3.2	6
63	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. <i>Brain and Development</i> , 2021, 43, 380-388.	1.1	6
64	Molecular autopsy by proxy in preconception counseling. <i>Clinical Genetics</i> , 2021, 100, 678-691.	2.0	6
65	Bi-allelic loss-of-function variants in PPFIBP1 cause a neurodevelopmental disorder with microcephaly, epilepsy, and periventricular calcifications. <i>American Journal of Human Genetics</i> , 2022, 109, 1421-1435.	6.2	6
66	Crisponi/CISS1 syndrome: A case series. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1236-1241.	1.2	5
67	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.	2.0	5
68	Fructose-1,6-bisphosphatase deficiency with confirmed molecular diagnosis. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2020, 41, 199-202.	1.1	5
69	A de novo splicing variant supports the candidacy of TLL1 in ASD pathogenesis. <i>European Journal of Human Genetics</i> , 2020, 28, 525-528.	2.8	4
70	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.	0.8	4
71	Further delineation of <i>SMC9</i> -related heart and brain malformation syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1624-1630.	1.2	3
72	Mutations in phospholipase C eta-1 ( <i>PLCH1</i> ) are associated with holoprosencephaly. <i>Journal of Medical Genetics</i> , 2022, 59, 358-365.	3.2	3

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73	Further clinical and genetic evidence of ASC-1 complex dysfunction in congenital neuromuscular disease. <i>European Journal of Medical Genetics</i> , 2022, 65, 104537.	1.3	3
74	A Biallelic Variant in <i>FRA10AC1</i> Is Associated With Neurodevelopmental Disorder and Growth Retardation. <i>Neurology: Genetics</i> , 2022, 8, e200010.	1.9	2
75	Embryopathy Associated With a Vitamin Therapy. <i>Pediatric Neurology</i> , 2018, 89, 73-74.	2.1	1
76	Hypospadias in ring X syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104225.	1.3	1
77	The phenotypic spectrum of dihydrolipoamide dehydrogenase deficiency in Saudi Arabia. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100817.	1.1	1
78	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
79	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.	1.1	1
80	De Novo Ring Chromosome 15: Molecular Cytogenetic and Clinical Characterization of First Case from Saudi Arabia. <i>Journal of Pediatric Genetics</i> , 0, , .	0.7	1
81	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.	1.1	0
82	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.	1.2	0
83	Neuroregression, coarse features, and oligosaccharides in urines. <i>Neurosciences</i> , 2017, 22, 326-328.	0.1	0
84	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.	1.1	0
85	Mitochondrial œdysmorphology in variant classification. <i>Human Genetics</i> , 2022, 141, 55-64.	3.8	0