Amal M Alhashem

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7022132/publications.pdf

Version: 2024-02-01

85 papers	3,344 citations	172457 29 h-index	168389 53 g-index
89 all docs	89 docs citations	89 times ranked	6905 citing authors

#	Article	IF	Citations
1	Mutations in phospholipase C eta-1 (PLCH1) are associated with holoprosencephaly. Journal of Medical Genetics, 2022, 59, 358-365.	3.2	3
2	Biallelic <scp>ZNFX1</scp> variants are associated with a spectrum of immunoâ€hematological abnormalities. Clinical Genetics, 2022, 101, 247-254.	2.0	12
3	Mitochondrial â€œdysmorphologyâ€•in variant classification. Human Genetics, 2022, 141, 55-64.	3.8	0
4	Further clinical and genetic evidence of ASC-1 complex dysfunction in congenital neuromuscular disease. European Journal of Medical Genetics, 2022, 65, 104537.	1.3	3
5	A Biallelic Variant in FRA10AC1 Is Associated With Neurodevelopmental Disorder and Growth Retardation. Neurology: Genetics, 2022, 8, e200010.	1.9	2
6	Bi-allelic loss-of-function variants in PPFIBP1 cause a neurodevelopmental disorder with microcephaly, epilepsy, and periventricular calcifications. American Journal of Human Genetics, 2022, 109, 1421-1435.	6.2	6
7	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.	1.2	7
8	Neuroimaging manifestations and genetic heterogeneity of Walker-Warburg syndrome in Saudi patients. Brain and Development, 2021, 43, 380-388.	1.1	6
9	Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. European Journal of Human Genetics, 2021, 29, 141-153.	2.8	66
10	Further delineation of <scp>SMG9</scp>â€related heart and brain malformation syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1624-1630.	1.2	3
11	Combining exome/genome sequencing with data repository analysis reveals novel geneâ€“disease associations for a wide range of genetic disorders. Genetics in Medicine, 2021, 23, 1551-1568.	2.4	30
12	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. Frontiers in Pediatrics, 2021, 9, 633385.	1.9	15
13	The genotypic and phenotypic spectrum of pycnodysostosis in Saudi Arabia: Novel variants and clinical findings. American Journal of Medical Genetics, Part A, 2021, 185, 2455-2463.	1.2	0
14	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	2.4	11
15	Hypospadias in ring X syndrome. European Journal of Medical Genetics, 2021, 64, 104225.	1.3	1
16	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.	0.8	4
17	Molecular autopsy by proxy in preconception counseling. Clinical Genetics, 2021, 100, 678-691.	2.0	6
18	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. Genome Medicine, 2021, 13, 161.	8.2	13

#	Article	IF	Citations
19	The phenotypic spectrum of dihydrolipoamide dehydrogenase deficiency in Saudi Arabia. Molecular Genetics and Metabolism Reports, 2021, 29, 100817.	1.1	1
20	A de novo splicing variant supports the candidacy of TLL1 in ASD pathogenesis. European Journal of Human Genetics, 2020, 28, 525-528.	2.8	4
21	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. Hepatology, 2020, 71, 2067-2079.	7.3	28
22	Categorized Genetic Analysis in Childhood-Onset Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, 504-514.	3.6	18
23	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. Human Genetics, 2020, 139, 1273-1283.	3.8	16
24	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	8.8	59
25	Recurrent homozygous damaging mutation in TMX2, encoding a protein disulfide isomerase, in four families with microlissencephaly. Journal of Medical Genetics, 2020, 57, 274-282.	3.2	6
26	The morbid genome of ciliopathies: an update. Genetics in Medicine, 2020, 22, 1051-1060.	2.4	68
27	Peripheral venous route for administration of ammonul infusion for treatment of acute hyperammonemia. Journal of King Abdulaziz University, Islamic Economics, 2020, 41, 98-101.	1.1	0
28	ADAMTS19 â€œassociated heart valve defects: Novel genetic variants consolidating a recognizable cardiac phenotype. Clinical Genetics, 2020, 98, 56-63.	2.0	11
29	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Frontiers in Genetics, 2020, 11, 580484.	2.3	13
30	Fructose-1,6-bisphosphatase deficiency with confirmed molecular diagnosis. Journal of King Abdulaziz University, Islamic Economics, 2020, 41, 199-202.	1.1	5
31	Molecular and clinical characteristics of very long-chain acyl-CoA dehydrogenase deficiency. Journal of King Abdulaziz University, Islamic Economics, 2020, 41, 590-596.	1.1	1
32	Epilepsy, neuropsychiatric phenotypes, neuroimaging findings, and genotype-neurophenotype correlation in 22q11.2 deletion syndrome. Journal of King Abdulaziz University, Islamic Economics, 2020, 25, 287-291.	1.1	0
33	Incidence of newborn screening disorders among 56632 infants in Central Saudi Arabia. Journal of King Abdulaziz University, Islamic Economics, 2020, 41, 703-708.	1.1	13
34	Truncating ARL6IP1 variant as the genetic cause of fatal complicated hereditary spastic paraplegia. BMC Medical Genetics, 2019, 20, 119.	2.1	14
35	Genetic, clinical and biochemical characterization of a large cohort of patients with hyaline fibromatosis syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 209.	2.7	7
36	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48

#	Article	IF	Citations
37	MDH1 deficiency is a metabolic disorder of the malateâ€“aspartate shuttle associated with early onset severe encephalopathy. Human Genetics, 2019, 138, 1247-1257.	3.8	31
38	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	6.2	184
39	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	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. Journal of Congenital Cardiology, 2019, 3, .	0.5	10
41	6-Pyruvoyltetrahydropterin Synthase Deficiency: Review and Report of 28 Arab Subjects. Pediatric Neurology, 2019, 96, 40-47.	2.1	12
42	A homozygous frameshift variant in an alternatively spliced exon of DLG5 causes hydrocephalus and renal dysplasia. Clinical Genetics, 2019, 95, 631-633.	2.0	5
43	Congenital anomalies and associated risk factors in a Saudi population: a cohort study from pregnancy to age 2 years. BMJ Open, 2019, 9, e026351.	1.9	17
44	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	2.4	85
45	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	2.0	12
46	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
47	Novel Homozygous Mutation of the AIMP1 Gene: A Milder Neuroimaging Phenotype With Preservation of the Deep White Matter. Pediatric Neurology, 2019, 91, 57-61.	2.1	7
48	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.	2.4	71
49	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
50	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 715-721.	1.2	7
51	Molecular autopsy in maternalâ€“fetal medicine. Genetics in Medicine, 2018, 20, 420-427.	2.4	84
52	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. American Journal of Human Genetics, 2018, 103, 612-620.	6.2	70
53	Genetic variants in components of the NALCNâ€“UNC80â€“UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). Human Genetics, 2018, 137, 753-768.	3.8	38
54	Embryopathy Associated With a Vitamin Therapy. Pediatric Neurology, 2018, 89, 73-74.	2.1	1

#	Article	IF	Citations
55	FARS2 deficiency; new cases, review of clinical, biochemical, and molecular spectra, and variants interpretation based on structural, functional, and evolutionary significance. Molecular Genetics and Metabolism, 2018, 125, 281-291.	1.1	28
56	Clinical profile and mutation spectrum of long QT syndrome in Saudi Arabia: The impact of consanguinity. Heart Rhythm, 2017, 14, 1191-1199.	0.7	13
57	Recessive AFG3L2 Mutation Causes Progressive Microcephaly, Early Onset Seizures, Spasticity, and Basal Ganglia Involvement. Pediatric Neurology, 2017, 71, 24-28.	2.1	19
58	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Human Genetics, 2017, 136, 921-939.	3.8	209
59	A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. Molecular Genetics and Metabolism, 2017, 121, 91-95.	1.1	68
60	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	122
61	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	2.8	291
62	Congenital disorders of glycosylation: The Saudi experience. American Journal of Medical Genetics, Part A, 2017, 173, 2614-2621.	1.2	31
63	Neuroregression, coarse features, and oligosaccharides in urines. Neurosciences, 2017, 22, 326-328.	0.1	0
64	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540.	3.8	89
65	Severe early-onset epileptic encephalopathy due to mutations in the KCNA2 gene: Expansion of the genotypic and phenotypic spectrum. European Journal of Paediatric Neurology, 2016, 20, 657-660.	1.6	33
66	Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. Human Genetics, 2016, 135, 1295-1298.	3.8	65
67	Crisponi/CISS1 syndrome: A case series. American Journal of Medical Genetics, Part A, 2016, 170, 1236-1241.	1.2	5
68	Distal acroosteolysis, poikiloderma and joint stiffness: a novel laminopathy?. European Journal of Human Genetics, 2016, 24, 1220-1222.	2.8	6
69	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. Genetics in Medicine, 2016, 18, 686-695.	2.4	55
70	Severe CNS involvement in WWOX mutations: Description of five new cases. American Journal of Medical Genetics, Part A, 2015, 167, 3209-3213.	1.2	50
71	Effect of consanguinity on birth defects in Saudi women: Results from a nested caseâ€control study. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 100-104.	1.6	28
72	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	8.8	56

#	Article	IF	Citations
73	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Reports, 2015, 10, 148-161.	6.4	375
74	Identification of a novel MKS locus defined byTMEM107mutation. Human Molecular Genetics, 2015, 24, 5211-5218.	2.9	42
75	Further Delineation of the ALG9-CDG Phenotype. JIMD Reports, 2015, 27, 107-112.	1.5	17
76	IFT27, encoding a small GTPase component of IFT particles, is mutated in a consanguineous family with Bardet-Biedl syndrome. Human Molecular Genetics, 2014, 23, 3307-3315.	2.9	134
77	Ciliary GenesTBC1D32/C6orf170andSCLT1are Mutated in Patients with OFD Type IX. Human Mutation, 2014, 35, 36-40.	2.5	78
78	A Wide Clinical Phenotype Spectrum in Patients With ATP1A2 Mutations. Journal of Child Neurology, 2014, 29, 265-268.	1.4	17
79	Neu-Laxova Syndrome, an Inborn Error of Serine Metabolism, Is Caused by Mutations in PHGDH. American Journal of Human Genetics, 2014, 94, 898-904.	6.2	93
80	Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. American Journal of Human Genetics, 2013, 93, 555-560.	6.2	45
81	Smithâ€“Lemliâ€“Opitz syndrome among Arabs. Clinical Genetics, 2012, 82, 165-172.	2.0	9
82	Munchausen syndrome by proxy mimicking as Gaucher disease. European Journal of Pediatrics, 2010, 169, 1029-1032.	2.7	9
83	Vici syndrome associated with unilateral lung hypoplasia and myopathy. American Journal of Medical Genetics, Part A, 2010, 152A, 1849-1853.	1.2	33
84	An atypical presentation of severe congenital contractures and lack of cerebellar involvement in a patient with a novel LAMA1 mutation. Journal of Biochemical and Clinical Genetics, 0, , 43-46.	0.1	1
85	De Novo Ring Chromosome 15: Molecular Cytogenetic and Clinical Characterization of First Case from Saudi Arabia. Journal of Pediatric Genetics, 0, , .	0.7	1