

Mohamed S Abdel-Hamid

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

48 papers	381 citations	10 h-index	18 g-index
51 ext. papers	540 ext. citations	3.4 avg, IF	3.29 L-index

#	Paper	IF	Citations
48	Mutation in WDR4 impairs tRNA m(7)G46 methylation and causes a distinct form of microcephalic primordial dwarfism. <i>Genome Biology</i> , 2015 , 16, 210	18.3	77
47	Mutations in ADAR1, IFIH1, and RNASEH2B presenting as spastic paraplegia. <i>Neuropediatrics</i> , 2014 , 45, 386-93	1.6	53
46	Identification of NR5A1 Mutations and Possible Digenic Inheritance in 46,XY Gonadal Dysgenesis. <i>Sexual Development</i> , 2016 , 10, 147-51	1.6	27
45	PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. <i>Annals of Neurology</i> , 2016 , 80, 59-70	9.4	25
44	Molecular and phenotypic spectrum of ASPM-related primary microcephaly: Identification of eight novel mutations. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2133-40	2.5	22
43	De Novo Mutation in ABCC9 Causes Hypertrichosis Acromegaloid Facial Features Disorder. <i>Pediatric Dermatology</i> , 2016 , 33, e109-13	1.9	17
42	Genetic polymorphisms of human cytochrome P450 CYP1A1 in an Egyptian population and tobacco-induced lung cancer. <i>Genes and Environment</i> , 2017 , 39, 7	2.8	15
41	APOA2 Polymorphism in Relation to Obesity and Lipid Metabolism. <i>Cholesterol</i> , 2013 , 2013, 289481		14
40	Band-like calcification with simplified gyration and polymicrogyria: report of 10 new families and identification of five novel OCLN mutations. <i>Journal of Human Genetics</i> , 2017 , 62, 553-559	4.3	13
39	Aromatase Deficiency due to a Homozygous CYP19A1 Mutation in a 46,XX Egyptian Patient with Ambiguous Genitalia. <i>Sexual Development</i> , 2017 , 11, 275-279	1.6	12
38	Evaluating the association of APOA2 polymorphism with insulin resistance in adolescents. <i>Meta Gene</i> , 2014 , 2, 366-73	0.7	8
37	Aicardi-Goutières syndrome: unusual neuro-radiological manifestations. <i>Metabolic Brain Disease</i> , 2017 , 32, 679-683	3.9	7
36	Clinical and molecular characterization of seven Egyptian families with autosomal recessive robinow syndrome: Identification of four novel ROR2 gene mutations. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 3054-61	2.5	7
35	Microcephalic osteodysplastic primordial dwarfism type II: Additional nine patients with implications on phenotype and genotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1407-1420	2.5	6
34	Micro and Martsolf syndromes in 34 new patients: Refining the phenotypic spectrum and further molecular insights. <i>Clinical Genetics</i> , 2020 , 98, 445-456	4	6
33	Phenotypic spectrum of NDE1-related disorders: from microlissencephaly to microhydranencephaly. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 494-497	2.5	6
32	Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia type-2. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1190-1194	2.5	5

31	Genetic mutation in Egyptian children with steroid-resistant nephrotic syndrome. <i>Journal of the Formosan Medical Association</i> , 2018 , 117, 48-53	3.2	5
30	Phenotypic and mutational spectrum of thirty-five patients with Sjögren-Larsson syndrome: identification of eleven novel ALDH3A2 mutations and founder effects. <i>Journal of Human Genetics</i> , 2019 , 64, 859-865	4.3	5
29	Long-term survival in microcephalic osteodysplastic primordial dwarfism type I: Evaluation of an 18-year-old male with g.55G>A homozygous mutation in RNU4ATAC. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 277-82	2.5	5
28	GAP0 syndrome in seven new patients: Identification of five novel ANTXR1 mutations including the first large intragenic deletion. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 237-242	2.5	5
27	Advances in genomic diagnosis of a large cohort of Egyptian patients with disorders of sex development. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1666-1677	2.5	4
26	Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. <i>Metabolic Brain Disease</i> , 2016 , 31, 1171-9	3.9	4
25	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. <i>Neuron</i> , 2021 , 109, 241-256.e9	13.9	4
24	Novel AMH and AMHR2 Mutations in Two Egyptian Families with Persistent Müllerian Duct Syndrome. <i>Sexual Development</i> , 2017 , 11, 29-33	1.6	3
23	KBG syndrome in two patients from Egypt. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 13092-1312	3.12	3
22	A novel mutation (c.2735_2736delTC) in the androgen receptor gene in 46,XY females with complete androgen insensitivity syndrome in an Egyptian family. <i>Hormone Research in Paediatrics</i> , 2014 , 82, 411-4	3.3	3
21	Further delineation of the oculoauricular syndrome phenotype: A new family with a novel truncating HMX1 mutation. <i>Ophthalmic Genetics</i> , 2018 , 39, 215-220	1.2	3
20	Two new families with enamel renal syndrome: A novel FAM20A gene mutation and review of literature. <i>European Journal of Medical Genetics</i> , 2020 , 63, 104045	2.6	2
19	Raine syndrome: Prenatal diagnosis based on recognizable fetal facial features and characteristic intracranial calcification. <i>Prenatal Diagnosis</i> , 2020 , 40, 1578-1597	3.2	2
18	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2021 ,	11.2	2
17	OTUD6B-associated intellectual disability: novel variants and genetic exclusion of retinal degeneration as part of a refined phenotype. <i>Journal of Human Genetics</i> , 2021 ,	4.3	2
16	A novel frameshift mutation in the sterol 27-hydroxylase gene in an Egyptian family with cerebrotendinous xanthomatosis without cataract. <i>Metabolic Brain Disease</i> , 2017 , 32, 311-315	3.9	1
15	Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. <i>BMC Medical Genomics</i> , 2020 , 13, 68	3.7	1
14	Lenz-Majewski syndrome in a patient from Egypt. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 2039-2042	2.5	1

13	A Homozygous Missense Variant in Hedgehog Acyltransferase (HHAT) Gene Associated with 46,XY Gonadal Dysgenesis.. <i>Sexual Development</i> , 2022 , 1-5	1.6	1
12	Expanding the phenotypic and allelic spectrum of SMG8: Clinical observations reveal overlap with SMG9-associated disease trait. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 188, 648	2.5	1
11	Genetic Susceptibility in Family Members of Egyptian Hepatitis C Virus Infected Patients: Role of Interleukin-12 B Gene Polymorphism. <i>Infectious Disorders - Drug Targets</i> , 2019 , 19, 81-87	1.1	1
10	ASAH1-related disorders: Description of 15 novel pediatric patients and expansion of the clinical phenotype. <i>Clinical Genetics</i> , 2020 , 98, 598-605	4	1
9	Blepharophimosis-ptosis-intellectual disability syndrome: A report of nine Egyptian patients with further expansion of phenotypic and mutational spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2857-2866	2.5	1
8	Phenotypic and molecular insights into PQBP1-related intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2446-2450	2.5	1
7	Osteoporosis-pseudoglioma syndrome in four new patients: identification of two novel LRP5 variants and insights on patients management using bisphosphonates therapy.. <i>Osteoporosis International</i> , 2022 , 1	5.3	0
6	Asparagine Synthetase Deficiency with Intracranial Hemorrhage Can Mimic Molybdenum Cofactor Deficiency. <i>Neuropediatrics</i> , 2021 , 52, 201-207	1.6	0
5	Homozygous missense variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. <i>Brain Communications</i> , 2021 , 3, fcab183	4.5	0
4	First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. <i>Molecular Syndromology</i> , 2021 , 12, 279-288	1.5	0
3	Fetal brain arrest broadens the spectrum of WDR81-related developmental brain malformations. <i>Neurogenetics</i> , 2021 , 22, 287-295	3	
2	Clinical and molecular characterization of Unverricht-Lundborg disease among Egyptian patients. <i>Epilepsy Research</i> , 2021 , 176, 106746	3	
1	Spectrum of NPHS1 and NPHS2 variants in Egyptian children with focal segmental glomerular sclerosis: identification of six novel variants and founder effect.. <i>Molecular Genetics and Genomics</i> , 2022 , 1	3.1	