# Mohamed S Abdel-Hamid

## List of Publications by Year in Descending Order



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| 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 | A Homozygous Missense Variant in Hedgehog Acyltransferase (HHAT) Gene Associated with 46,XY Gonadal Dysgenesis.. *Sexual Development*, **2022**, 1-5 | 1.6 | 1 |
| 47 | Osteoporosis-pseudoglioma syndrome in four new patients: identification of two novel LRP5 variants and insights on patientsUmanagement using bisphosphonates therapy.. *Osteoporosis International*, **2022**, 1 | 5.3 | 0 |
| 46 | Spectrum of NPHS1 and NPHS2 variants in egyptian children with focal segmental glomerular sclerosis: identification of six novel variants and founder effect.. *Molecular Genetics and Genomics*, **2022**, 1 | 3.1 | |
| 45 | Expanding the phenotypic and allelic spectrum of SMG8: Clinical observations reveal overlap with SMG9-associated disease trait. *American Journal of Medical Genetics, Part A*, **2021**, 188, 648 | 2.5 | 1 |
| 44 | Asparagine Synthetase Deficiency with Intracranial Hemorrhage Can Mimic Molybdenum Cofactor Deficiency. *Neuropediatrics*, **2021**, 52, 201-207 | 1.6 | 0 |
| 43 | Advances in genomic diagnosis of a large cohort of Egyptian patients with disorders of sex development. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1666-1677 | 2.5 | 4 |
| 42 | Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. *Neuron*, **2021**, 109, 241-256.e9 | 13.9 | 4 |
| 41 | Homozygous missense variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. *Brain Communications*, **2021**, 3, fcab183 | 4.5 | 0 |
| 40 | Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. *Brain*, **2021**, | 11.2 | 2 |
| 39 | OTUD6B-associated intellectual disability: novel variants and genetic exclusion of retinal degeneration as part of a refined phenotype. *Journal of Human Genetics*, **2021**, | 4.3 | 2 |
| 38 | Fetal brain arrest broadens the spectrum of WDR81-related developmental brain malformations. *Neurogenetics*, **2021**, 22, 287-295 | 3 | |
| 37 | Clinical and molecular characterization of Unverricht-Lundborg disease among Egyptian patients. *Epilepsy Research*, **2021**, 176, 106746 | 3 | |
| 36 | First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. *Molecular Syndromology*, **2021**, 12, 279-288 | 1.5 | 0 |
| 35 | Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. *BMC Medical Genomics*, **2020**, 13, 68 | 3.7 | 1 |
| 34 | KBG syndrome in two patients from Egypt. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1309-1312 | 2.5 | 3 |
| 33 | Microcephalic osteodysplastic primordial dwarfism type II: Additional nine patients with implications on phenotype and genotype correlation. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1407-1420 | 2.5 | 6 |
| 32 | Two new families with enamel renal syndrome: A novel FAM20A gene mutation and review of literature. *European Journal of Medical Genetics*, **2020**, 63, 104045 | 2.6 | 2 |

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| 31 | Micro and Martsolf syndromes in 34 new patients: Refining the phenotypic spectrum and further molecular insights. *Clinical Genetics*, **2020**, 98, 445-456 | 4 | 6 |
| 30 | Raine syndrome: Prenatal diagnosis based on recognizable fetal facial features and characteristic intracranial calcification. *Prenatal Diagnosis*, **2020**, 40, 1578-1597 | 3.2 | 2 |
| 29 | ASAH1-related disorders: Description of 15 novel pediatric patients and expansion of the clinical phenotype. *Clinical Genetics*, **2020**, 98, 598-605 | 4 | 1 |
| 28 | Blepharophimosis-ptosis-intellectual disability syndrome: A report of nine Egyptian patients with further expansion of phenotypic and mutational spectrum. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2857-2866 | 2.5 | 1 |
| 27 | Lenz-Majewski syndrome in a patient from Egypt. *American Journal of Medical Genetics, Part A*, **2019** , 179, 2039-2042 | 2.5 | 1 |
| 26 | Phenotypic and mutational spectrum of thirty-five patients with Sjögren-Larsson syndrome: identification of eleven novel ALDH3A2 mutations and founder effects. *Journal of Human Genetics*, **2019**, 64, 859-865 | 4.3 | 5 |
| 25 | Genetic Susceptibility in Family Members of Egyptian Hepatitis C Virus Infected Patients: Role of Interleukin-12 B Gene Polymorphism. *Infectious Disorders - Drug Targets*, **2019**, 19, 81-87 | 1.1 | 1 |
| 24 | GAPO syndrome in seven new patients: Identification of five novel ANTXR1 mutations including the first large intragenic deletion. *American Journal of Medical Genetics, Part A*, **2019**, 179, 237-242 | 2.5 | 5 |
| 23 | Phenotypic spectrum of NDE1-related disorders: from microlissencephaly to microhydranencephaly. *American Journal of Medical Genetics, Part A*, **2019**, 179, 494-497 | 2.5 | 6 |
| 22 | Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia type-2. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1190-1194 | 2.5 | 5 |
| 21 | Genetic mutation in Egyptian children with steroid-resistant nephrotic syndrome. *Journal of the Formosan Medical Association*, **2018**, 117, 48-53 | 3.2 | 5 |
| 20 | Further delineation of the oculoauricular syndrome phenotype: A new family with a novel truncating HMX1 mutation. *Ophthalmic Genetics*, **2018**, 39, 215-220 | 1.2 | 3 |
| 19 | Phenotypic and molecular insights into PQBP1-related intellectual disability. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2446-2450 | 2.5 | 1 |
| 18 | Genetic polymorphisms of human cytochrome P450 CYP1A1 in an Egyptian population and tobacco-induced lung cancer. *Genes and Environment*, **2017**, 39, 7 | 2.8 | 15 |
| 17 | A novel frameshift mutation in the sterol 27-hydroxylase gene in an Egyptian family with cerebrotendinous xanthomatosis without cataract. *Metabolic Brain Disease*, **2017**, 32, 311-315 | 3.9 | 1 |
| 16 | Band-like calcification with simplified gyration and polymicrogyria: report of 10 new families and identification of five novel OCLN mutations. *Journal of Human Genetics*, **2017**, 62, 553-559 | 4.3 | 13 |
| 15 | Aicardi-Goutières syndrome: unusual neuro-radiological manifestations. *Metabolic Brain Disease*, **2017**, 32, 679-683 | 3.9 | 7 |
| 14 | Novel AMH and AMHR2 Mutations in Two Egyptian Families with Persistent Müllerian Duct Syndrome. *Sexual Development*, **2017**, 11, 29-33 | 1.6 | 3 |

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| 13 | Aromatase Deficiency due to a Homozygous CYP19A1 Mutation in a 46,XX Egyptian Patient with Ambiguous Genitalia. *Sexual Development*, **2017**, 11, 275-279 | 1.6 | 12 |
| 12 | Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. *Metabolic Brain Disease*, **2016**, 31, 1171-9 | 3.9 | 4 |
| 11 | Molecular and phenotypic spectrum of ASPM-related primary microcephaly: Identification of eight novel mutations. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2133-40 | 2.5 | 22 |
| 10 | De Novo Mutation in ABCC9 Causes Hypertrichosis Acromegaloid Facial Features Disorder. *Pediatric Dermatology*, **2016**, 33, e109-13 | 1.9 | 17 |
| 9 | PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. *Annals of Neurology*, **2016**, 80, 59-70 | 9.4 | 25 |
| 8 | Identification of NR5A1 Mutations and Possible Digenic Inheritance in 46,XY Gonadal Dysgenesis. *Sexual Development*, **2016**, 10, 147-51 | 1.6 | 27 |
| 7 | 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. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 277-82 | 2.5 | 5 |
| 6 | Clinical and molecular characterization of seven Egyptian families with autosomal recessive robinow syndrome: Identification of four novel ROR2 gene mutations. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3054-61 | 2.5 | 7 |
| 5 | 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 |
| 4 | Evaluating the association of APOA2 polymorphism with insulin resistance in adolescents. *Meta Gene*, **2014**, 2, 366-73 | 0.7 | 8 |
| 3 | A novel mutation (c.2735_2736delTC) in the androgen receptor gene in 46,XY females with complete androgen insensitivity syndrome in an Egyptian family. *Hormone Research in Paediatrics*, **2014**, 82, 411-4 | 3.3 | 3 |
| 2 | Mutations in ADAR1, IFIH1, and RNASEH2B presenting as spastic paraplegia. *Neuropediatrics*, **2014**, 45, 386-93 | 1.6 | 53 |
| 1 | APOA2 Polymorphism in Relation to Obesity and Lipid Metabolism. *Cholesterol*, **2013**, 2013, 289481 | | 14 |