Mahmoud Y Issa

List of Publications by Year in Descending Order

43 papers	942 citations	16 h-index	30 g-index
45 ext. papers	1,214 ext. citations	7.1 avg, IF	3.2 L-index

#	Paper	IF	Citations
43	Variable predicted pathogenic mechanisms for novel MECP2 variants in RTT patients.. *Journal of Genetic Engineering and Biotechnology*, **2022**, 20, 44	3.1	
42	A founder mutation in PEX12 among Egyptian patients in peroxisomal biogenesis disorder. *Neurological Sciences*, **2021**, 42, 2737-2745	3.5	
41	Biallelic variants in , encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. *Journal of Medical Genetics*, **2021**, 58, 495-504	5.8	3
40	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. *Nature Communications*, **2021**, 12, 2558	17.4	4
39	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. *Neuron*, **2021**, 109, 241-256.e9	13.9	4
38	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. *European Journal of Human Genetics*, **2021**, 29, 271-279	5.3	3
37	Implication of folate deficiency in CYP2U1 loss of function. *Journal of Experimental Medicine*, **2021**, 218,	16.6	2
36	Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. *BMC Medical Genomics*, **2020**, 13, 68	3.7	1
35	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
34	Mutation spectrum in the gene encoding methyl-CpG-binding protein 2 in Egyptian patients with Rett syndrome. *Meta Gene*, **2020**, 24, 100620	0.7	1
33	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. *American Journal of Human Genetics*, **2020**, 107, 977-988	11	9
32	Micro and Martsolf syndromes in 34 new patients: Refining the phenotypic spectrum and further molecular insights. *Clinical Genetics*, **2020**, 98, 445-456	4	6
31	ASAH1-related disorders: Description of 15 novel pediatric patients and expansion of the clinical phenotype. *Clinical Genetics*, **2020**, 98, 598-605	4	1
30	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
29	Prenatal delineation of a distinct lethal fetal syndrome caused by a homozygous truncating KIDINS220 variant. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2867-2876	2.5	2
28	The potential impact of COMT gene variants on dopamine regulation and phenotypic traits of ASD patients. *Behavioural Brain Research*, **2020**, 378, 112272	3.4	7
27	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. *Brain*, **2020**, 143, 1114-1126	11.2	28

26	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. *Nature Communications*, **2019**, 10, 797	17.4	10
25	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. *American Journal of Human Genetics*, **2019**, 105, 534-548	11	25
24	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
23	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. *Nature Communications*, **2019**, 10, 707	17.4	18
22	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
21	PGAP3-related hyperphosphatasia with mental retardation syndrome: Report of 10 new patients and a homozygous founder mutation. *Clinical Genetics*, **2018**, 93, 84-91	4	14
20	A homozygous founder mutation in associates with a neurodevelopmental disorder characterised by microcephaly, epilepsy and autistic features. *Journal of Medical Genetics*, **2018**, 55, 48-54	5.8	29
19	Hypermanganesemia with dystonia, polycythemia and cirrhosis in 10 patients: Six novel SLC30A10 mutations and further phenotype delineation. *Clinical Genetics*, **2018**, 93, 905-912	4	20
18	Loss of tubulin deglutamylase CCP1 causes infantile-onset neurodegeneration. *EMBO Journal*, **2018**, 37,	13	55
17	Unbalanced 14;X Translocation and Pattern of X Inactivation in a Female Patient with Multiple Congenital Anomalies. *Cytogenetic and Genome Research*, **2018**,	1.9	1
16	Loss of Protocadherin-12 Leads to Diencephalic-Mesencephalic Junction Dysplasia Syndrome. *Annals of Neurology*, **2018**, 84, 638-647	9.4	13
15	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). *Human Genetics*, **2018**, 137, 753-768	6.3	19
14	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
13	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
12	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. *American Journal of Human Genetics*, **2017**, 100, 676-688	11	36
11	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. *American Journal of Human Genetics*, **2017**, 101, 552-563	11	25
10	Homozygous Mutations in TBC1D23 Lead to a Non-degenerative Form of Pontocerebellar Hypoplasia. *American Journal of Human Genetics*, **2017**, 101, 441-450	11	30
9	Exome sequencing discloses KALRN homozygous variant as likely cause of intellectual disability and short stature in a consanguineous pedigree. *Human Genomics*, **2016**, 10, 26	6.8	12

8	Molybdenum cofactor and isolated sulphite oxidase deficiencies: Clinical and molecular spectrum among Egyptian patients. *European Journal of Paediatric Neurology*, **2016**, 20, 714-22	3.8	21
7	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. *American Journal of Human Genetics*, **2016**, 98, 615-26	11	59
6	PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. *Annals of Neurology*, **2016**, 80, 59-70	9.4	25
5	Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. *Science*, **2014**, 343, 506-511	33.3	374
4	Further delineation of the clinical spectrum in RNU4ATAC related microcephalic osteodysplastic primordial dwarfism type I. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1875-81	2.5	16
3	Expanding the phenotypic and mutational spectrum in microcephalic osteodysplastic primordial dwarfism type I. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1455-61	2.5	28
2	Profound microcephaly, primordial dwarfism with developmental brain malformations: a new syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1823-31	2.5	5
1	New recessive syndrome of microcephaly, cerebellar hypoplasia, and congenital heart conduction defect. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 3035-41	2.5	4