Mahmoud Y Issa

List of Publications by Year
in descending order

45 papers	1,431 citations	394286 19 h-index	345118 36 g-index
45 all docs	45 docs citations	45 times ranked	3626 citing authors

#	Article	IF	Citations
1	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	6.0	466
2	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
3	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	2.6	71
4	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
5	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	2.6	46
6	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	3.7	46
7	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.	2.6	45
8	Homozygous Mutations in TBC1D23 Lead to a Non-degenerative Form of Pontocerebellar Hypoplasia. American Journal of Human Genetics, 2017, 101, 441-450.	2.6	43
9	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.	1.8	38
10	A homozygous founder mutation in TRAPPC6B associates with a neurodevelopmental disorder characterised by microcephaly, epilepsy and autistic features. Journal of Medical Genetics, 2018, 55, 48-54.	1.5	37
11	PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. Annals of Neurology, 2016, 80, 59-70.	2.8	35
12	Molybdenum cofactor and isolated sulphite oxidase deficiencies: Clinical and molecular spectrum among Egyptian patients. European Journal of Paediatric Neurology, 2016, 20, 714-722.	0.7	33
13	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	2.6	33
14	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	3.8	31
15	Expanding the phenotypic and mutational spectrum in microcephalic osteodysplastic primordial dwarfism type I. American Journal of Medical Genetics, Part A, 2012, 158A, 1455-1461.	0.7	30
16	Hypermanganesemia with dystonia, polycythemia and cirrhosis in 10 patients: Six novel SLC30A10 mutations and further phenotype delineation. Clinical Genetics, 2018, 93, 905-912.	1.0	30
17	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707.	5.8	28
18	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28

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19	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	5.8	24
20	Further delineation of the clinical spectrum in RNU4ATAC related microcephalic osteodysplastic primordial dwarfism type I. American Journal of Medical Genetics, Part A, 2013, 161, 1875-1881.	0.7	20
21	<scp>PGAP3</scp>â€related hyperphosphatasia with mental retardation syndrome: Report of 10 new patients and a homozygous founder mutation. Clinical Genetics, 2018, 93, 84-91.	1.0	20
22	Loss of Protocadherinâ€12<scp>L</scp>eads to <scp>D</scp>iencephalicâ€<scp>M</scp>esencephalic <scp>J</scp>unction <scp>D</scp>ysplasia <scp>S</scp>yndrome. Annals of Neurology, 2018, 84, 638-647.	2.8	19
23	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.	1.1	18
24	The potential impact of COMT gene variants on dopamine regulation and phenotypic traits of ASD patients. Behavioural Brain Research, 2020, 378, 112272.	1.2	15
25	Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Journal of Medical Genetics, 2021, 58, 495-504.	1.5	14
26	Exome sequencing discloses KALRN homozygous variant as likely cause of intellectual disability and short stature in a consanguineous pedigree. Human Genomics, 2016, 10, 26.	1.4	13
27	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	4.2	13
28	Micro and Martsolf syndromes in 34 new patients: Refining the phenotypic spectrum and further molecular insights. Clinical Genetics, 2020, 98, 445-456.	1.0	12
29	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.	0.7	11
30	Prenatal delineation of a distinct lethal fetal syndrome caused by a homozygous truncating <scp>KIDINS220</scp> variant. American Journal of Medical Genetics, Part A, 2020, 182, 2867-2876.	0.7	10
31	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	1.1	9
32	Phenotypic and mutational spectrum of thirty-five patients with SjÃ¶grenâ€“Larsson syndrome: identification of eleven novel ALDH3A2 mutations and founder effects. Journal of Human Genetics, 2019, 64, 859-865.	1.1	8
33	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.	0.7	8
34	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	1.4	8
35	Profound microcephaly, primordial dwarfism with developmental brain malformations: A new syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1823-1831.	0.7	6
36	New recessive syndrome of microcephaly, cerebellar hypoplasia, and congenital heart conduction defect. American Journal of Medical Genetics, Part A, 2011, 155, 3035-3041.	0.7	4

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37	Blepharophimosisâ€ptosisâ€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.	0.7	4
38	Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. BMC Medical Genomics, 2020, 13, 68.	0.7	4
39	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.	1.4	3
40	<scp>ASAH1</scp>â€related disorders: Description of 15 novel pediatric patients and expansion of the clinical phenotype. Clinical Genetics, 2020, 98, 598-605.	1.0	3
41	Prenatal presentation of Walkerâ€“Warburg syndrome with a POMT2 mutation: an extended fetal phenotype. Egyptian Journal of Medical Human Genetics, 2020, 21, .	0.5	2
42	Unbalanced 14;X Translocation and Pattern of X Inactivation in a Female Patient with Multiple Congenital Anomalies. Cytogenetic and Genome Research, 2018, 156, 71-79.	0.6	1
43	Mutation spectrum in the gene encoding methyl-CpG-binding protein 2 in Egyptian patients with Rett syndrome. Meta Gene, 2020, 24, 100620.	0.3	1
44	A founder mutation in PEX12 among Egyptian patients in peroxisomal biogenesis disorder. Neurological Sciences, 2020, 42, 2737-2745.	0.9	1
45	Variable predicted pathogenic mechanisms for novel MECP2 variants in RTT patients. Journal of Genetic Engineering and Biotechnology, 2022, 20, 44.	1.5	0