Maha Zaki

List of Publications by Year
in descending order

160 papers	7,417 citations	66315 42 h-index	69214 77 g-index
165 all docs	165 docs citations	165 times ranked	12948 citing authors

#	Article	IF	Citations
1	Assessment of interferon-related biomarkers in Aicardi-GoutiÃ¨res syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. Lancet Neurology, The, 2013, 12, 1159-1169.	4.9	473
2	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	6.0	466
3	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	0.7	447
4	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	9.4	383
5	Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in SLC30A10 , a Manganese Transporter in Man. American Journal of Human Genetics, 2012, 90, 457-466.	2.6	321
6	Exome Sequencing Can Improve Diagnosis and Alter Patient Management. Science Translational Medicine, 2012, 4, 138ra78.	5.8	226
7	Inactivating mutations in MFSD2A, required for omega-3 fatty acid transport in brain, cause a lethal microcephaly syndrome. Nature Genetics, 2015, 47, 809-813.	9.4	180
8	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	2.6	179
9	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157
10	Defective Wnt-dependent cerebellar midline fusion in a mouse model of Joubert syndrome. Nature Medicine, 2011, 17, 726-731.	15.2	138
11	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
12	Recessive Mutations in the Gene Encoding the Tight Junction Protein Occludin Cause Band-like Calcification with Simplified Gyration and Polymicrogyria. American Journal of Human Genetics, 2010, 87, 354-364.	2.6	123
13	Mutations in EOGT Confirm the Genetic Heterogeneity of Autosomal-Recessive Adams-Oliver Syndrome. American Journal of Human Genetics, 2013, 92, 598-604.	2.6	114
14	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	1.1	114
15	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	9.4	111
16	Mutations in LAMB1 Cause Cobblestone Brain Malformation without Muscular or Ocular Abnormalities. American Journal of Human Genetics, 2013, 92, 468-474.	2.6	96
17	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	3.8	95
18	AMPD2 Regulates GTP Synthesis and Is Mutated in a Potentially Treatable Neurodegenerative Brainstem Disorder. Cell, 2013, 154, 505-517.	13.5	94

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19	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	1.1	92
20	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
21	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	1.1	85
22	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84
23	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	2.6	75
24	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	1.1	75
25	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	2.6	73
26	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.3	72
27	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	2.6	71
28	Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly. American Journal of Human Genetics, 2016, 99, 501-510.	2.6	70
29	Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	9.4	70
30	Mutations in MBOAT7 , Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. American Journal of Human Genetics, 2016, 99, 912-916.	2.6	69
31	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	3.9	68
32	Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	9.4	66
33	Identification of a novel recessiveRELN mutation using a homozygous balanced reciprocal translocation. American Journal of Medical Genetics, Part A, 2007, 143A, 939-944.	0.7	65
34	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	1.4	64
35	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	2.8	64
36	Biallelic Truncating Mutations in FMN2, Encoding the Actin-Regulatory Protein Formin 2, Cause Nonsyndromic Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2014, 95, 721-728.	2.6	62

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37	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
38	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	4.9	53
39	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	1.4	52
40	Mutations in GPAA1 , Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. American Journal of Human Genetics, 2017, 101, 856-865.	2.6	49
41	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	2.6	48
42	Fetal MRI in the evaluation of fetuses referred for sonographically suspected neural tube defects (NTDs): impact on diagnosis and management decision. Neuroradiology, 2009, 51, 761-772.	1.1	46
43	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	1.1	46
44	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	2.6	46
45	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	3.7	46
46	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
47	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
48	A homozygous IER3IP1 mutation causes microcephaly with simplified gyral pattern, epilepsy, and permanent neonatal diabetes syndrome (MEDS). American Journal of Medical Genetics, Part A, 2012, 158A, 2788-2796.	0.7	42
49	Coâ€occurrence of distinct ciliopathy diseases in single families suggests genetic modifiers. American Journal of Medical Genetics, Part A, 2011, 155, 3042-3049.	0.7	38
50	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
51	Autosomal dominant SCA5 and autosomal recessive infantile SCA are allelic conditions resulting from SPTBN2 mutations. European Journal of Human Genetics, 2014, 22, 286-288.	1.4	37
52	Mutations in UNC80, Encoding Part of the UNC79-UNC80-NALCN Channel Complex, Cause Autosomal-Recessive Severe Infantile Encephalopathy. American Journal of Human Genetics, 2016, 98, 210-215.	2.6	37
53	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
54	PEX6 is Expressed in Photoreceptor Cilia and Mutated in Deafblindness with Enamel Dysplasia and Microcephaly. Human Mutation, 2016, 37, 170-174.	1.1	36

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55	Extracellular miR-145, miR-223 and miR-326 expression signature allow for differential diagnosis of immune-mediated neuroinflammatory diseases. Journal of the Neurological Sciences, 2017, 383, 188-198.	0.3	36
56	PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. Annals of Neurology, 2016, 80, 59-70.	2.8	35
57	Recurrent and Prolonged Infections in a Child with a Homozygous IFIH1 Nonsense Mutation. Frontiers in Genetics, 2017, 8, 130.	1.1	35
58	Diencephalic-mesencephalic junction dysplasia: a novel recessive brain malformation. Brain, 2012, 135, 2416-2427.	3.7	34
59	Overexpression ofKLC2due to a homozygous deletion in the non-coding region causes SPOAN syndrome. Human Molecular Genetics, 2015, 24, ddv388.	1.4	34
60	Loss of the neural-specific BAF subunit ACTL6B relieves repression of early response genes and causes recessive autism. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 10055-10066.	3.3	34
61	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
62	Adamsâ€“Oliver syndrome: further evidence of an autosomal recessive variant. Clinical Dysmorphology, 2007, 16, 141-149.	0.1	32
63	Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. Human Molecular Genetics, 2015, 24, 2594-2603.	1.4	32
64	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	3.8	31
65	Molecular and phenotypic spectrum of ASPMâ€related primary microcephaly: Identification of eight novel mutations. American Journal of Medical Genetics, Part A, 2016, 170, 2133-2140.	0.7	30
66	Biallelic Mutations in TMTC3, Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly. American Journal of Human Genetics, 2016, 99, 1181-1189.	2.6	30
67	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
68	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genetics in Medicine, 2020, 22, 1851-1862.	1.1	30
69	Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. Neuron, 2020, 107, 82-94.e6.	3.8	30
70	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.	1.1	30
71	Spinocerebellar ataxia type 2 (SCA2) in an Egyptian family presenting with polyphagia and marked CAG expansion in infancy. Journal of Neurology, 2008, 255, 413-419.	1.8	29
72	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29

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73	Whole-Exome Sequencing Identifies Mutated C12orf57 in Recessive Corpus Callosum Hypoplasia. American Journal of Human Genetics, 2013, 92, 392-400.	2.6	28
74	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707.	5.8	28
75	MINPP1 prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. Nature Communications, 2020, 11, 6087.	5.8	28
76	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
77	Extending the mutation spectrum for Gallowayâ€“Mowat syndrome to include homozygous missense mutations in the WDR73 gene. American Journal of Medical Genetics, Part A, 2016, 170, 992-998.	0.7	26
78	Microcephaly, malformation of brain development and intracranial calcification in sibs: Pseudoâ€TORCH or a new syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2929-2936.	0.7	25
79	Mutations in LNPK, Encoding the Endoplasmic Reticulum Junction Stabilizer Lunapark, Cause a Recessive Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 296-304.	2.6	24
80	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	5.8	24
81	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495.	2.6	22
82	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
83	<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
84	Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283.	0.7	20
85	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
86	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
87	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853.	2.6	17
88	Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. Genetics in Medicine, 2021, 23, 524-533.	1.1	17
89	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
90	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17

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91	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
92	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. American Journal of Human Genetics, 2021, 108, 134-147.	2.6	15
93	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
94	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
95	Regulation of human cerebral cortical development by EXOC7 and EXOC8, components of the exocyst complex, and roles in neural progenitor cell proliferation and survival. Genetics in Medicine, 2020, 22, 1040-1050.	1.1	13
96	Neurodevelopmental disorder in an Egyptian family with a biallelic <scp>ALKBH8</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 1288-1293.	0.7	13
97	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	4.2	13
98	Study of C677T variant of methylene tetrahydrofolate reductase gene in autistic spectrum disorder Egyptian children. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 305-309.	1.1	12
99	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
100	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	1.1	12
101	Bandâ€like intracranial calcification (BIC), microcephaly and malformation of brain development: A distinctive form of congenital infection like syndromes. American Journal of Medical Genetics, Part A, 2009, 149A, 1565-1568.	0.7	11
102	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
103	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	2.6	11
104	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	1.1	11
105	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	2.6	11
106	Aicardi-GoutiÃ¨res syndrome: unusual neuro-radiological manifestations. Metabolic Brain Disease, 2017, 32, 679-683.	1.4	10
107	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
108	Homozygous missense WIPI2 variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Brain Communications, 2021, 3, fcab183.	1.5	10

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109	Genetic pattern of SMN1, SMN2, and NAIP genes in prognosis of SMA patients. Egyptian Journal of Medical Human Genetics, 2020, 21, .	0.5	10
110	Expanding the clinical spectrum of SPG11 gene mutations in recessive hereditary spastic paraplegia with thin corpus callosum. European Journal of Medical Genetics, 2011, 54, 82-85.	0.7	9
111	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460.	1.1	9
112	Undiagnosed Phenylketonuria Can Exist Everywhere: Results From an International Survey. Journal of Pediatrics, 2021, 239, 231-234.e2.	0.9	9
113	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. American Journal of Human Genetics, 2021, 108, 2017-2023.	2.6	9
114	Biallelic FRA10AC1 variants cause a neurodevelopmental disorder with growth retardation. Brain, 2022, 145, 1551-1563.	3.7	9
115	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
116	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
117	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
118	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83.	3.7	8
119	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
120	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. European Journal of Human Genetics, 2021, 29, 1226-1234.	1.4	8
121	ASAH1 pathogenic variants associated with acid ceramidase deficiency: Farber disease and spinal muscular atrophy with progressive myoclonic epilepsy. Human Mutation, 2020, 41, 1469-1487.	1.1	8
122	Isolated Dandyâ€“Walker malformation associated with brain stem dysgenesis in male sibs. Brain and Development, 2006, 28, 529-533.	0.6	7
123	Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. Metabolic Brain Disease, 2016, 31, 1171-1179.	1.4	7
124	Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia typeâ€2. American Journal of Medical Genetics, Part A, 2018, 176, 1190-1194.	0.7	7
125	Biallelic hypomorphic mutations in HEATR5B, encoding HEAT repeat-containing protein 5B, in a neurological syndrome with pontocerebellar hypoplasia. European Journal of Human Genetics, 2021, 29, 957-964.	1.4	7
126	<scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540.	1.0	7

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127	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
128	Dandyâ€“Walker malformation, genitourinary abnormalities, and intellectual disability in two families. American Journal of Medical Genetics, Part A, 2015, 167, 2503-2507.	0.7	6
129	Genotype/phenotype correlation in a female patient with 21q22.3 and 12p13.33 duplications. American Journal of Medical Genetics, Part A, 2016, 170, 1050-1058.	0.7	6
130	Recurrent homozygous damaging mutation in TMX2, encoding a protein disulfide isomerase, in four families with microlissencephaly. Journal of Medical Genetics, 2020, 57, 274-282.	1.5	6
131	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	3.7	6
132	Clinical and genetic characterization of ten Egyptian patients with Wolfâ€“Hirschhorn syndrome and review of literature. Molecular Genetics & Genomic Medicine, 2021, 9, e1546.	0.6	6
133	Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	3.7	6
134	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	1.0	6
135	Novel mutation in the fukutin gene in an Egyptian family with Fukuyama congenital muscular dystrophy and microcephaly. Gene, 2014, 539, 279-282.	1.0	5
136	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
137	Clinical, biomarker and genetic spectrum of Niemannâ€Pick type C in Egypt: The detection of nine novel NPC1 mutations. Clinical Genetics, 2019, 95, 537-539.	1.0	4
138	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
139	Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. BMC Medical Genomics, 2020, 13, 68.	0.7	4
140	Bi-allelic TTC5 variants cause delayed developmental milestones and intellectual disability. Journal of Medical Genetics, 2021, 58, 237-246.	1.5	4
141	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. Molecular Genetics & Genomic Medicine, 2021, 9, e1829.	0.6	4
142	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
143	Bilateral Calcification of Basal Ganglia in a Patient with Duplication of Both 11q13.1q22.1 and 4q35.2 with New Phenotypic Features. Cytogenetic and Genome Research, 2019, 159, 130-136.	0.6	3
144	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. Journal of Medical Genetics, 2021, 58, 815-831.	1.5	3

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145	<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
146	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	3.7	3
147	Unusual association of simplified gyral pattern and sparse hair in an Egyptian patient with microcephalyâ€“lymphoedema. Clinical Dysmorphology, 2006, 15, 245-247.	0.1	2
148	Clinical and molecular findings in eight Egyptian patients with suspected mitochondrial disorders and optic atrophy. Egyptian Journal of Medical Human Genetics, 2013, 14, 37-47.	0.5	2
149	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	2.8	2
150	Congenital isolated leukonychia totalis in three Egyptian sibs. , 2011, 155, 811-814.		1
151	Molecular analysis of MECP2 gene in Egyptian patients with Rett syndrome. Egyptian Journal of Medical Human Genetics, 2012, 13, 19-27.	0.5	1
152	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
153	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
154	A founder mutation in PEX12 among Egyptian patients in peroxisomal biogenesis disorder. Neurological Sciences, 2020, 42, 2737-2745.	0.9	1
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