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	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17
2	Biallelic FRA10AC1 variants cause a neurodevelopmental disorder with growth retardation. Brain, 2022, 145, 1551-1563.	3.7	9
3	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
4	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	3.7	3
5	<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
6	Variable predicted pathogenic mechanisms for novel MECP2 variants in RTT patients. Journal of Genetic Engineering and Biotechnology, 2022, 20, 44.	1.5	0
7	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
8	Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	3.7	6
9	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	1.0	6
10	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
11	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
12	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
13	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	3.8	31
14	Clinical, Biochemical, and Molecular Characterization of Metachromatic Leukodystrophy Among Egyptian Pediatric Patients: Expansion of the ARSA Mutational Spectrum. Journal of Molecular Neuroscience, 2021, 71, 1112-1130.	1.1	1
15	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
16	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
17	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
18	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

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19	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
20	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	2.6	11
21	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
22	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	1.1	12
23	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
24	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
25	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
26	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
27	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
28	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
29	Samia Temtamy. American Journal of Medical Genetics, Part A, 2021, 185, 3613-3614.	0.7	0
30	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
31	Undiagnosed Phenylketonuria Can Exist Everywhere: Results From an International Survey. Journal of Pediatrics, 2021, 239, 231-234.e2.	0.9	9
32	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	4.2	13
33	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
34	Bi-allelic TTC5 variants cause delayed developmental milestones and intellectual disability. Journal of Medical Genetics, 2021, 58, 237-246.	1.5	4
35	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
36	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

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37	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
38	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29
39	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83.	3.7	8
40	MINPP1 prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. Nature Communications, 2020, 11, 6087.	5.8	28
41	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
42	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
43	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
44	<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
45	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
46	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	4.9	53
47	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
48	A founder mutation in PEX12 among Egyptian patients in peroxisomal biogenesis disorder. Neurological Sciences, 2020, 42, 2737-2745.	0.9	1
49	Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. BMC Medical Genomics, 2020, 13, 68.	0.7	4
50	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
51	Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. Neuron, 2020, 107, 82-94.e6.	3.8	30
52	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
53	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
54	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

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55	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	3.7	6
56	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
57	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	3.7	46
58	Genetic pattern of SMN1, SMN2, and NAIP genes in prognosis of SMA patients. Egyptian Journal of Medical Human Genetics, 2020, 21, .	0.5	10
59	Variants in the NPC1 Gene in Egyptian Patients with Niemann-Pick Type C. Open Access Macedonian Journal of Medical Sciences, 2020, 8, 134-145.	0.1	0
60	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
61	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	2.6	46
62	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
63	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
64	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
65	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
66	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	5.8	24
67	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707.	5.8	28
68	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
69	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	1.1	85
70	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
71	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
72	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	3.9	68

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73	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	1.1	92
74	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
75	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	1.1	46
76	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	1.4	52
77	<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
78	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
79	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
80	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
81	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
82	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
83	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
84	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
85	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
86	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
87	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
88	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
89	Aicardi-GoutiÃ¨res syndrome: unusual neuro-radiological manifestations. Metabolic Brain Disease, 2017, 32, 679-683.	1.4	10
90	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

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91	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
92	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
93	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
94	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
95	Recurrent and Prolonged Infections in a Child with a Homozygous IFIH1 Nonsense Mutation. Frontiers in Genetics, 2017, 8, 130.	1.1	35
96	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
97	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
98	PEX6 is Expressed in Photoreceptor Cilia and Mutated in Deafblindness with Enamel Dysplasia and Microcephaly. Human Mutation, 2016, 37, 170-174.	1.1	36
99	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
100	PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. Annals of Neurology, 2016, 80, 59-70.	2.8	35
101	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
102	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
103	Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly. American Journal of Human Genetics, 2016, 99, 501-510.	2.6	70
104	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
105	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
106	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
107	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	2.6	71
108	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

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109	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
110	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	2.8	64
111	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
112	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
113	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
114	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	9.4	111
115	Overexpression ofKLC2due to a homozygous deletion in the non-coding region causes SPOAN syndrome. Human Molecular Genetics, 2015, 24, ddv388.	1.4	34
116	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
117	Mutations in ADAR1, IFIH1, and RNASEH2B Presenting As Spastic Paraplegia. Neuropediatrics, 2014, 45, 386-391.	0.3	72
118	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	3.8	95
119	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
120	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	2.6	75
121	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	6.0	466
122	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	1.1	75
123	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
124	AMPD2 Regulates GTP Synthesis and Is Mutated in a Potentially Treatable Neurodegenerative Brainstem Disorder. Cell, 2013, 154, 505-517.	13.5	94
125	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
126	Mutations in LAMB1 Cause Cobblestone Brain Malformation without Muscular or Ocular Abnormalities. American Journal of Human Genetics, 2013, 92, 468-474.	2.6	96

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127	Whole-Exome Sequencing Identifies Mutated C12orf57 in Recessive Corpus Callosum Hypoplasia. American Journal of Human Genetics, 2013, 92, 392-400.	2.6	28
128	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
129	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	1.1	114
130	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	2.6	73
131	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
132	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
133	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
134	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84
135	Exome Sequencing Can Improve Diagnosis and Alter Patient Management. Science Translational Medicine, 2012, 4, 138ra78.	5.8	226
136	Diencephalic-mesencephalic junction dysplasia: a novel recessive brain malformation. Brain, 2012, 135, 2416-2427.	3.7	34
137	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157
138	Molecular analysis of MECP2 gene in Egyptian patients with Rett syndrome. Egyptian Journal of Medical Human Genetics, 2012, 13, 19-27.	0.5	1
139	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
140	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
141	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
142	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
143	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
144	Defective Wnt-dependent cerebellar midline fusion in a mouse model of Joubert syndrome. Nature Medicine, 2011, 17, 726-731.	15.2	138

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145	Congenital isolated leukonychia totalis in three Egyptian sibs. , 2011, 155, 811-814.		1
146	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
147	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
148	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
149	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
150	Familial congenital unilateral cerebral ventriculomegaly: Delineation of a distinct genetic disorder. American Journal of Medical Genetics, Part A, 2009, 149A, 1789-1794.	0.7	0
151	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
152	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
153	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
154	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
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