Ghada M Abdel-Salam

List of Publications by Citations

93 papers	3,423 citations	26 h-index	57 g-index
97 ext. papers	4,117 ext. citations	5.1 avg, IF	4.23 L-index

#	Paper	IF	Citations
93	Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. *Science*, **2014**, 343, 506-511	33.3	374
92	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**, 167A, 296-312	2.5	321
91	Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. *Lancet Neurology, The*, **2013**, 12, 1159-69	24.1	267
90	Accelerating novel candidate gene discovery in neurogenetic disorders via whole-exome sequencing of prescreened multiplex consanguineous families. *Cell Reports*, **2015**, 10, 148-61	10.6	262
89	Exome sequencing can improve diagnosis and alter patient management. *Science Translational Medicine*, **2012**, 4, 138ra78	17.5	191
88	CEP290 mutations are frequently identified in the oculo-renal form of Joubert syndrome-related disorders. *American Journal of Human Genetics*, **2007**, 81, 104-13	11	118
87	Genomic analysis of primordial dwarfism reveals novel disease genes. *Genome Research*, **2014**, 24, 291-9	9.7	112
86	AHI1 gene mutations cause specific forms of Joubert syndrome-related disorders. *Annals of Neurology*, **2006**, 59, 527-34	9.4	111
85	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, 354-64	11	98
84	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. *American Journal of Human Genetics*, **2019**, 104, 1182-1201	11	95
83	Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. *American Journal of Human Genetics*, **2013**, 92, 598-604	11	90
82	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. *Nature Genetics*, **2015**, 47, 528-34	36.3	89
81	Loss-of-function mutations in TBC1D20 cause cataracts and male infertility in blind sterile mice and Warburg micro syndrome in humans. *American Journal of Human Genetics*, **2013**, 93, 1001-14	11	88
80	Mutation spectrum in RAB3GAP1, RAB3GAP2, and RAB18 and genotype-phenotype correlations in warburg micro syndrome and Martsolf syndrome. *Human Mutation*, **2013**, 34, 686-96	4.7	86
79	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
78	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. *Genetics in Medicine*, **2016**, 18, 554-62	8.1	73
77	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). *American Journal of Medical Genetics, Part A*, **2008**, 146A, 182-90	2.5	70

76	Identification of a novel recessive RELN mutation using a homozygous balanced reciprocal translocation. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 939-44	2.5	56
75	Genomic and phenotypic delineation of congenital microcephaly. *Genetics in Medicine*, **2019**, 21, 545-552	8.1	55
74	The supposed tumor suppressor gene WWOX is mutated in an early lethal microcephaly syndrome with epilepsy, growth retardation and retinal degeneration. *Orphanet Journal of Rare Diseases*, **2014**, 9, 12	4.2	54
73	Mutations in ADAR1, IFIH1, and RNASEH2B presenting as spastic paraplegia. *Neuropediatrics*, **2014**, 45, 386-93	1.6	53
72	Distinguishing 3 classes of corpus callosal abnormalities in consanguineous families. *Neurology*, **2011**, 76, 373-82	6.5	34
71	The molar tooth sign: a new Joubert syndrome and related cerebellar disorders classification system tested in Egyptian families. *Neurology*, **2008**, 70, 556-65	6.5	34
70	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, 2788-96	2.5	32
69	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
68	Mutations in CIT, encoding citron rho-interacting serine/threonine kinase, cause severe primary microcephaly in humans. *Human Genetics*, **2016**, 135, 1191-7	6.3	26
67	PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. *Annals of Neurology*, **2016**, 80, 59-70	9.4	25
66	PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. *Human Genetics*, **2019**, 138, 231-239	6.3	25
65	A homozygous mutation in RNU4ATAC as a cause of microcephalic osteodysplastic primordial dwarfism type I (MOPD I) with associated pigmentary disorder. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2885-96	2.5	24
64	Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. *Human Molecular Genetics*, **2015**, 24, 2594-603	5.6	23
63	Microcephaly, malformation of brain development and intracranial calcification in sibs: pseudo-TORCH or a new syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2929-36	2.5	23
62	Aicardi-Goutières syndrome: clinical and neuroradiological findings of 10 new cases. *Acta Paediatrica, International Journal of Paediatrics*, **2004**, 93, 929-36	3.1	22
61	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
60	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. *Journal of Human Genetics* , **2019**, 64, 1173-1186	4.3	20
59	Expanding the phenome and variome of skeletal dysplasia. *Genetics in Medicine*, **2018**, 20, 1609-1616	8.1	20

58	Association of epilepsy with different groups of microcephaly. *Developmental Medicine and Child Neurology*, **2000**, 42, 760-7	3.3	20
57	De Novo Mutation in ABCC9 Causes Hypertrichosis Acromegaloid Facial Features Disorder. *Pediatric Dermatology*, **2016**, 33, e109-13	1.9	17
56	Extending the mutation spectrum for Galloway-Mowat syndrome to include homozygous missense mutations in the WDR73 gene. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 992-8	2.5	17
55	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
54	Oto-spondylo-megaepiphyseal dysplasia (OSMED): clinical and radiological findings in sibs homozygous for premature stop codon mutation in the COL11A2 gene. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1189-95	2.5	15
53	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
52	Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2662-70	2.5	13
51	Warsaw breakage syndrome: Further clinical and genetic delineation. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2404-2418	2.5	13
50	Muenke syndrome with pigmentary disorder and probable hemimegalencephaly: An expansion of the phenotype. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 207-14	2.5	12
49	A case-control etiologic study of microcephaly. *Epidemiology*, **2000**, 11, 571-5	3.1	11
48	Expanding the mutation and clinical spectrum of Roberts syndrome. *Congenital Anomalies (discontinued)*, **2016**, 56, 154-62	1.1	10
47	Microcephaly with chorioretinal dysplasia: characteristic facial features. *American Journal of Medical Genetics Part A*, **2000**, 95, 513-5		9
46	Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellar-vermis hypoplasia. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1053-1057	2.5	8
45	Fetal brain disruption sequence versus fetal brain arrest: A distinct autosomal recessive developmental brain malformation phenotype. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1089-99	2.5	8
44	Band-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, 1565-8	2.5	8
43	Aicardi-Goutières syndrome: unusual neuro-radiological manifestations. *Metabolic Brain Disease*, **2017**, 32, 679-683	3.9	7
42	Ectodermal abnormalities in patients with Kabuki syndrome. *Pediatric Dermatology*, **2011**, 28, 507-11	1.9	7
41	Genetic analysis of SOX2 and VSX2 genes in 27 Egyptian families with anophthalmia and microphthalmia. *Ophthalmic Genetics*, **2017**, 38, 498-500	1.2	6

40	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
39	Magnetic resonance imaging of developmental facial paresis: a spectrum of complex anomalies. *Neuroradiology*, **2018**, 60, 1053-1061	3.2	6
38	Isolated Dandy-Walker malformation associated with brain stem dysgenesis in male sibs. *Brain and Development*, **2006**, 28, 529-33	2.2	6
37	Phenotypic spectrum of NDE1-related disorders: from microlissencephaly to microhydranencephaly. *American Journal of Medical Genetics, Part A*, **2019**, 179, 494-497	2.5	6
36	Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. *Genetics in Medicine*, **2021**, 23, 524-533	8.1	6
35	Prenatal ultrasound findings of holoprosencephaly spectrum: Unusual associations. *Prenatal Diagnosis*, **2020**, 40, 565-576	3.2	5
34	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
33	GWAS signals revisited using human knockouts. *Genetics in Medicine*, **2018**, 20, 64-68	8.1	5
32	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
31	Gómez-López-hernández syndrome versus rhombencephalosynapsis spectrum: a rare co-occurrence with bipartite parietal bone. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 480-3	2.5	5
30	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
29	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
28	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
27	The second unrelated case with isolated microcephaly and normal intelligence (microcephalia vera). *Clinical Dysmorphology*, **2000**, 9, 151-2	0.9	4
26	Microcephaly with normal intelligence, and chorioretinopathy. *Ophthalmic Genetics*, **1999**, 20, 259-64	1.2	4
25	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. *Frontiers in Genetics*, **2020**, 11, 580484	4.5	4
24	Aicardi-Goutières syndrome: clinical and neuroradiological findings of 10 new cases **2004**, 93, 929		4
23	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

22	KBG syndrome in two patients from Egypt. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1309-1312	2.5	3
21	De Novo 17q24.2-q24.3 microdeletion presenting with generalized hypertrichosis terminalis, gingival fibromatous hyperplasia, and distinctive facial features. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2418-24	2.5	3
20	Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	3
19	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
18	DTYMK is essential for genome integrity and neuronal survival.. *Acta Neuropathologica*, **2021**, 143, 245	14.3	3
17	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1829	2.3	2
16	Raine syndrome: Prenatal diagnosis based on recognizable fetal facial features and characteristic intracranial calcification. *Prenatal Diagnosis*, **2020**, 40, 1578-1597	3.2	2
15	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
14	Lenz-Majewski syndrome in a patient from Egypt. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2039-2042	2.5	1
13	Association of epilepsy with different groups of microcephaly. *Developmental Medicine and Child Neurology*, **2007**, 42, 760-767	3.3	1
12	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
11	Phenotypic and molecular insights into PQBP1-related intellectual disability. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2446-2450	2.5	1
10	Asparagine Synthetase Deficiency with Intracranial Hemorrhage Can Mimic Molybdenum Cofactor Deficiency. *Neuropediatrics*, **2021**, 52, 201-207	1.6	0
9	Further Insights into Developmental Brain Malformations and Leukoencephalopathy Associated with 6p25.3 Deletion. *Neuropediatrics*, **2020**, 51, 76-82	1.6	0
8	Expanding the KIF4A-associated phenotype. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3728-3739	2.5	0
7	Clinical Variability of Pallister-Killian Syndrome in Two Egyptian Patients. *Journal of Pediatric Genetics*, **2020**, 9, 207-210	0.7	
6	A founder mutation in PEX12 among Egyptian patients in peroxisomal biogenesis disorder. *Neurological Sciences*, **2021**, 42, 2737-2745	3.5	
5	Alopecia Universalis, Cleft Palate and Lip, Hypohydrosis, Hypodontia, Nail Dysplasia and Syndactyly: New Ectodermal Dysplasia Syndrome?. *Congenital Anomalies (discontinued)*, **2008**, 39, 37-42	1.1	

List of Publications

4	Response to letter from Okoye JO and Ngokere AA "Are the prevalence of Trisomy 13 and the incidence of severe holoprosencephaly increasing in Africa?". *Prenatal Diagnosis*, **2020**, 40, 1618-1619	3.2
3	Fetal brain arrest broadens the spectrum of WDR81-related developmental brain malformations. *Neurogenetics*, **2021**, 22, 287-295	3
2	Hallervorden-Spatz syndrome. Variable imaging findings. *Neurosciences*, **2004**, 9, 214-7	0.1
1	Anthropometric craniofacial pattern profiles in microcephaly. *Anthropological Review*,65, 65-74	0.6