

Ghada M Abdel-Salam

List of Publications by Year in Descending Order

Source: <https://exaly.com/author-pdf/8566817/ghada-m-abdel-salam-publications-by-year.pdf>
Version: 2024-04-09

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.
The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

| | | | |
|-------------------|-------------------------|----------------|-----------------|
| 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 | A founder mutation in PEX12 among Egyptian patients in peroxisomal biogenesis disorder. <i>Neurological Sciences</i> , 2021 , 42, 2737-2745 | 3.5 | |
| 92 | Expanding the phenotypic and allelic spectrum of SMG8: Clinical observations reveal overlap with SMG9-associated disease trait. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 188, 648 | 2.5 | 1 |
| 91 | Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. <i>American Journal of Medical Genetics, Part A</i> , 2021 , | 2.5 | 3 |
| 90 | Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1829 | 2.3 | 2 |
| 89 | Asparagine Synthetase Deficiency with Intracranial Hemorrhage Can Mimic Molybdenum Cofactor Deficiency. <i>Neuropediatrics</i> , 2021 , 52, 201-207 | 1.6 | 0 |
| 88 | Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. <i>Genetics in Medicine</i> , 2021 , 23, 524-533 | 8.1 | 6 |
| 87 | Expanding the KIF4A-associated phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3728-3739 | 2.5 | 0 |
| 86 | OTUD6B-associated intellectual disability: novel variants and genetic exclusion of retinal degeneration as part of a refined phenotype. <i>Journal of Human Genetics</i> , 2021 , | 4.3 | 2 |
| 85 | Fetal brain arrest broadens the spectrum of WDR81-related developmental brain malformations. <i>Neurogenetics</i> , 2021 , 22, 287-295 | 3 | |
| 84 | DTYMK is essential for genome integrity and neuronal survival.. <i>Acta Neuropathologica</i> , 2021 , 143, 245 | 14.3 | 3 |
| 83 | Clinical Variability of Pallister-Killian Syndrome in Two Egyptian Patients. <i>Journal of Pediatric Genetics</i> , 2020 , 9, 207-210 | 0.7 | |
| 82 | KBG syndrome in two patients from Egypt. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 13092-1312 | 3.12 | 3 |
| 81 | Prenatal ultrasound findings of holoprosencephaly spectrum: Unusual associations. <i>Prenatal Diagnosis</i> , 2020 , 40, 565-576 | 3.2 | 5 |
| 80 | Microcephalic osteodysplastic primordial dwarfism type II: Additional nine patients with implications on phenotype and genotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1407-1420 | 2.5 | 6 |
| 79 | Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. <i>Frontiers in Genetics</i> , 2020 , 11, 580484 | 4.5 | 4 |
| 78 | Further Insights into Developmental Brain Malformations and Leukoencephalopathy Associated with 6p25.3 Deletion. <i>Neuropediatrics</i> , 2020 , 51, 76-82 | 1.6 | 0 |
| 77 | Raine syndrome: Prenatal diagnosis based on recognizable fetal facial features and characteristic intracranial calcification. <i>Prenatal Diagnosis</i> , 2020 , 40, 1578-1597 | 3.2 | 2 |

| | | | |
|----|--|-----|----|
| 76 | Response to letter from Okoye JO and Ngokere AA "Are the prevalence of Trisomy 13 and the incidence of severe holoprosencephaly increasing in Africa?". <i>Prenatal Diagnosis</i> , 2020 , 40, 1618-1619 | 3.2 | |
| 75 | Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. <i>Journal of Human Genetics</i> , 2019 , 64, 1173-1186 | 4.3 | 20 |
| 74 | Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. <i>American Journal of Human Genetics</i> , 2019 , 104, 1182-1201 | 11 | 95 |
| 73 | Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellar-vermis hypoplasia. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 1053-1057 | 2.5 | 8 |
| 72 | Lenz-Majewski syndrome in a patient from Egypt. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 2039-2042 | 2.5 | 1 |
| 71 | Phenotypic and mutational spectrum of thirty-five patients with Sjögren-Larsson syndrome: identification of eleven novel ALDH3A2 mutations and founder effects. <i>Journal of Human Genetics</i> , 2019 , 64, 859-865 | 4.3 | 5 |
| 70 | PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. <i>Human Genetics</i> , 2019 , 138, 231-239 | 6.3 | 25 |
| 69 | Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , 2019 , 21, 545-552 | 8.1 | 55 |
| 68 | GAPO syndrome in seven new patients: Identification of five novel ANTXR1 mutations including the first large intragenic deletion. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 237-242 | 2.5 | 5 |
| 67 | Phenotypic spectrum of NDE1-related disorders: from microlissencephaly to microhydranencephaly. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 494-497 | 2.5 | 6 |
| 66 | Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia type-2. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1190-1194 | 2.5 | 5 |
| 65 | Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , 2018 , 20, 1609-1616 | 8.1 | 20 |
| 64 | GWAS signals revisited using human knockouts. <i>Genetics in Medicine</i> , 2018 , 20, 64-68 | 8.1 | 5 |
| 63 | Magnetic resonance imaging of developmental facial paresis: a spectrum of complex anomalies. <i>Neuroradiology</i> , 2018 , 60, 1053-1061 | 3.2 | 6 |
| 62 | Further delineation of the oculoauricular syndrome phenotype: A new family with a novel truncating HMX1 mutation. <i>Ophthalmic Genetics</i> , 2018 , 39, 215-220 | 1.2 | 3 |
| 61 | Phenotypic and molecular insights into PQBP1-related intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2446-2450 | 2.5 | 1 |
| 60 | Warsaw breakage syndrome: Further clinical and genetic delineation. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2404-2418 | 2.5 | 13 |
| 59 | Genetic analysis of SOX2 and VSX2 genes in 27 Egyptian families with anophthalmia and microphthalmia. <i>Ophthalmic Genetics</i> , 2017 , 38, 498-500 | 1.2 | 6 |

| | | | |
|----|--|------|-----|
| 58 | Band-like calcification with simplified gyration and polymicrogyria: report of 10 new families and identification of five novel OCLN mutations. <i>Journal of Human Genetics</i> , 2017 , 62, 553-559 | 4.3 | 13 |
| 57 | Aicardi-Goutières syndrome: unusual neuro-radiological manifestations. <i>Metabolic Brain Disease</i> , 2017 , 32, 679-683 | 3.9 | 7 |
| 56 | Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. <i>Genetics in Medicine</i> , 2016 , 18, 554-62 | 8.1 | 73 |
| 55 | Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. <i>Metabolic Brain Disease</i> , 2016 , 31, 1171-9 | 3.9 | 4 |
| 54 | Molecular and phenotypic spectrum of ASPM-related primary microcephaly: Identification of eight novel mutations. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2133-40 | 2.5 | 22 |
| 53 | Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2662-70 | 2.5 | 13 |
| 52 | De Novo Mutation in ABCC9 Causes Hypertrichosis Acromegaloid Facial Features Disorder. <i>Pediatric Dermatology</i> , 2016 , 33, e109-13 | 1.9 | 17 |
| 51 | Expanding the mutation and clinical spectrum of Roberts syndrome. <i>Congenital Anomalies (discontinued)</i> , 2016 , 56, 154-62 | 1.1 | 10 |
| 50 | Extending the mutation spectrum for Galloway-Mowat syndrome to include homozygous missense mutations in the WDR73 gene. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 992-8 | 2.5 | 17 |
| 49 | PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. <i>Annals of Neurology</i> , 2016 , 80, 59-70 | 9.4 | 25 |
| 48 | Mutations in CIT, encoding citron rho-interacting serine/threonine kinase, cause severe primary microcephaly in humans. <i>Human Genetics</i> , 2016 , 135, 1191-7 | 6.3 | 26 |
| 47 | 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. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 277-82 | 2.5 | 5 |
| 46 | Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. <i>Nature Genetics</i> , 2015 , 47, 528-34 | 36.3 | 89 |
| 45 | Fetal brain disruption sequence versus fetal brain arrest: A distinct autosomal recessive developmental brain malformation phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 1089-99 | 2.5 | 8 |
| 44 | De Novo 17q24.2-q24.3 microdeletion presenting with generalized hypertrichosis terminalis, gingival fibromatous hyperplasia, and distinctive facial features. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2418-24 | 2.5 | 3 |
| 43 | Mutation in WDR4 impairs tRNA m(7)G46 methylation and causes a distinct form of microcephalic primordial dwarfism. <i>Genome Biology</i> , 2015 , 16, 210 | 18.3 | 77 |
| 42 | Accelerating novel candidate gene discovery in neurogenetic disorders via whole-exome sequencing of prescreened multiplex consanguineous families. <i>Cell Reports</i> , 2015 , 10, 148-61 | 10.6 | 262 |
| 41 | Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. <i>Human Molecular Genetics</i> , 2015 , 24, 2594-603 | 5.6 | 23 |

| | | | |
|----|--|------|-----|
| 40 | Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 296-312 | 2.5 | 321 |
| 39 | The supposed tumor suppressor gene WWOX is mutated in an early lethal microcephaly syndrome with epilepsy, growth retardation and retinal degeneration. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 12 | 4.2 | 54 |
| 38 | Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. <i>Science</i> , 2014 , 343, 506-511 | 33.3 | 374 |
| 37 | Gñez-López-hernández syndrome versus rhombencephalosynapsis spectrum: a rare co-occurrence with bipartite parietal bone. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 480-3 | 2.5 | 5 |
| 36 | Mutations in ADAR1, IFIH1, and RNASEH2B presenting as spastic paraplegia. <i>Neuropediatrics</i> , 2014 , 45, 386-93 | 1.6 | 53 |
| 35 | Genomic analysis of primordial dwarfism reveals novel disease genes. <i>Genome Research</i> , 2014 , 24, 291-9 | 9.7 | 112 |
| 34 | Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. <i>American Journal of Human Genetics</i> , 2013 , 92, 598-604 | 11 | 90 |
| 33 | Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. <i>Lancet Neurology</i> , 2013 , 12, 1159-69 | 24.1 | 267 |
| 32 | Mutation spectrum in RAB3GAP1, RAB3GAP2, and RAB18 and genotype-phenotype correlations in warburg micro syndrome and Martsolf syndrome. <i>Human Mutation</i> , 2013 , 34, 686-96 | 4.7 | 86 |
| 31 | Loss-of-function mutations in TBC1D20 cause cataracts and male infertility in blind sterile mice and Warburg micro syndrome in humans. <i>American Journal of Human Genetics</i> , 2013 , 93, 1001-14 | 11 | 88 |
| 30 | Further delineation of the clinical spectrum in RNU4ATAC related microcephalic osteodysplastic primordial dwarfism type I. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 1875-81 | 2.5 | 16 |
| 29 | A homozygous IER3IP1 mutation causes microcephaly with simplified gyral pattern, epilepsy, and permanent neonatal diabetes syndrome (MEDS). <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 2788-96 | 2.5 | 32 |
| 28 | Expanding the phenotypic and mutational spectrum in microcephalic osteodysplastic primordial dwarfism type I. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 1455-61 | 2.5 | 28 |
| 27 | Profound microcephaly, primordial dwarfism with developmental brain malformations: a new syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 1823-31 | 2.5 | 5 |
| 26 | Exome sequencing can improve diagnosis and alter patient management. <i>Science Translational Medicine</i> , 2012 , 4, 138ra78 | 17.5 | 191 |
| 25 | Ectodermal abnormalities in patients with Kabuki syndrome. <i>Pediatric Dermatology</i> , 2011 , 28, 507-11 | 1.9 | 7 |
| 24 | Muenke syndrome with pigmentary disorder and probable hemimegalencephaly: An expansion of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 207-14 | 2.5 | 12 |
| 23 | A homozygous mutation in RNU4ATAC as a cause of microcephalic osteodysplastic primordial dwarfism type I (MOPD I) with associated pigmentary disorder. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 2885-96 | 2.5 | 24 |

| | | | |
|----|--|-----|-----|
| 22 | Distinguishing 3 classes of corpus callosal abnormalities in consanguineous families. <i>Neurology</i> , 2011 , 76, 373-82 | 6.5 | 34 |
| 21 | Recessive mutations in the gene encoding the tight junction protein occludin cause band-like calcification with simplified gyration and polymicrogyria. <i>American Journal of Human Genetics</i> , 2010 , 87, 354-64 | 11 | 98 |
| 20 | Band-like intracranial calcification (BIC), microcephaly and malformation of brain development: a distinctive form of congenital infection like syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 1565-8 | 2.5 | 8 |
| 19 | The molar tooth sign: a new Joubert syndrome and related cerebellar disorders classification system tested in Egyptian families. <i>Neurology</i> , 2008 , 70, 556-65 | 6.5 | 34 |
| 18 | Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 182-90 | 2.5 | 70 |
| 17 | Microcephaly, malformation of brain development and intracranial calcification in sibs: pseudo-TORCH or a new syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 2929-36 | 2.5 | 23 |
| 16 | Alopecia Universalis, Cleft Palate and Lip, Hypohydrosis, Hypodontia, Nail Dysplasia and Syndactyly: New Ectodermal Dysplasia Syndrome?. <i>Congenital Anomalies (discontinued)</i> , 2008 , 39, 37-42 | 1.1 | |
| 15 | Identification of a novel recessive RELN mutation using a homozygous balanced reciprocal translocation. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 939-44 | 2.5 | 56 |
| 14 | Association of epilepsy with different groups of microcephaly. <i>Developmental Medicine and Child Neurology</i> , 2007 , 42, 760-767 | 3.3 | 1 |
| 13 | CEP290 mutations are frequently identified in the oculo-renal form of Joubert syndrome-related disorders. <i>American Journal of Human Genetics</i> , 2007 , 81, 104-13 | 11 | 118 |
| 12 | Isolated Dandy-Walker malformation associated with brain stem dysgenesis in male sibs. <i>Brain and Development</i> , 2006 , 28, 529-33 | 2.2 | 6 |
| 11 | Oto-spondylo-megaepiphyseal dysplasia (OSMED): clinical and radiological findings in sibs homozygous for premature stop codon mutation in the COL11A2 gene. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 1189-95 | 2.5 | 15 |
| 10 | AHL1 gene mutations cause specific forms of Joubert syndrome-related disorders. <i>Annals of Neurology</i> , 2006 , 59, 527-34 | 9.4 | 111 |
| 9 | Aicardi-Goutières syndrome: clinical and neuroradiological findings of 10 new cases. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2004 , 93, 929-36 | 3.1 | 22 |
| 8 | Aicardi-Goutières syndrome: clinical and neuroradiological findings of 10 new cases 2004 , 93, 929 | | 4 |
| 7 | Hallervorden-Spatz syndrome. Variable imaging findings. <i>Neurosciences</i> , 2004 , 9, 214-7 | 0.1 | |
| 6 | The second unrelated case with isolated microcephaly and normal intelligence (microcephalia vera). <i>Clinical Dysmorphology</i> , 2000 , 9, 151-2 | 0.9 | 4 |
| 5 | A case-control etiologic study of microcephaly. <i>Epidemiology</i> , 2000 , 11, 571-5 | 3.1 | 11 |

| | | | |
|---|---|-----|----|
| 4 | Microcephaly with chorioretinal dysplasia: characteristic facial features. <i>American Journal of Medical Genetics Part A</i> , 2000 , 95, 513-5 | | 9 |
| 3 | Association of epilepsy with different groups of microcephaly. <i>Developmental Medicine and Child Neurology</i> , 2000 , 42, 760-7 | 3.3 | 20 |
| 2 | Microcephaly with normal intelligence, and chorioretinopathy. <i>Ophthalmic Genetics</i> , 1999 , 20, 259-64 | 1.2 | 4 |
| 1 | Anthropometric craniofacial pattern profiles in microcephaly. <i>Anthropological Review</i> , 65, 65-74 | 0.6 | |