Hanan H Afifi

List of Publications by Citations

31 papers	399 citations	11 h-index	19 g-index
33 ext. papers	498 ext. citations	3.3 avg, IF	2.85 L-index

#	Paper	IF	Citations
31	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
30	Mutations in ANTXR1 cause GAPO syndrome. *American Journal of Human Genetics*, **2013**, 92, 792-9	11	60
29	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
28	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
27	Distinct ocular expression in infants and children with Down syndrome in Cairo, Egypt: myopia and heart disease. *JAMA Ophthalmology*, **2013**, 131, 1057-66	3.9	18
26	De Novo Mutation in ABCC9 Causes Hypertrichosis Acromegaloid Facial Features Disorder. *Pediatric Dermatology*, **2016**, 33, e109-13	1.9	17
25	GAPO syndrome: first Egyptian case with ultrastructural changes in the gingiva. *Clinical Genetics*, **1997**, 52, 110-5	4	15
24	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
23	Evaluation of superoxide dismutase and glutathione peroxidase enzymes and their cofactors in Egyptian children with Down syndrome. *Biological Trace Element Research*, **2001**, 81, 21-8	4.5	13
22	Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2662-70	2.5	13
21	Growth charts of Down syndrome in Egypt: a study of 434 children 0-36 months of age. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2647-55	2.5	12
20	Expanding the mutation and clinical spectrum of Roberts syndrome. *Congenital Anomalies (discontinued)*, **2016**, 56, 154-62	1.1	10
19	Mutational analysis of the PTPN11 gene in Egyptian patients with Noonan syndrome. *Journal of the Formosan Medical Association*, **2013**, 112, 707-12	3.2	9
18	Ectodermal abnormalities in patients with Kabuki syndrome. *Pediatric Dermatology*, **2011**, 28, 507-11	1.9	7
17	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
16	Magnetic resonance imaging of developmental facial paresis: a spectrum of complex anomalies. *Neuroradiology*, **2018**, 60, 1053-1061	3.2	6
15	Constitutional retinoblastoma gene deletion in Egyptian patients. *World Journal of Pediatrics*, **2009**, 5, 222-5	4.6	6

14	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
13	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
12	Growth curves of Egyptian patients with Turner syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2687-91	2.5	5
11	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
10	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
9	Lipoid proteinosis: A clinical and molecular study in Egyptian patients. *Gene*, **2017**, 628, 308-314	3.8	2
8	Body composition in Egyptian Turner syndrome girls. *Indian Journal of Human Genetics*, **2013**, 19, 150-3		2
7	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
6	Lenz-Majewski syndrome in a patient from Egypt. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2039-2042	2.5	1
5	Phenotypic and molecular insights into PQBP1-related intellectual disability. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2446-2450	2.5	1
4	Clinical Variability of Pallister-Killian Syndrome in Two Egyptian Patients. *Journal of Pediatric Genetics*, **2020**, 9, 207-210	0.7	
3	Congenital isolated leukonychia totalis in three Egyptian sibs. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 811-4	2.5	
2	Familial congenital unilateral cerebral ventriculomegaly: Delineation of a distinct genetic disorder. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1789-94	2.5	
1	Clinical and Cytogenomic Characterization of De Novo 11p14.3-p15.5 Duplication Associated with 18q23 Deletion in an Egyptian Female Infant. *Journal of Pediatric Genetics*, **2021**, 10, 131-138	0.7	