

Hanan H Afifi

List of Publications by Year in descending order

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32
papers

572
citations

840119

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642321

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33
all docs

33
docs citations

33
times ranked

1303
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutation in WDR4 impairs tRNA m ⁷ C46 methylation and causes a distinct form of microcephalic primordial dwarfism. <i>Genome Biology</i> , 2015, 16, 210.	3.8	132
2	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.	1.2	91
3	Mutations in ANTXR1 Cause GAPO Syndrome. <i>American Journal of Human Genetics</i> , 2013, 92, 792-799.	2.6	73
4	A homozygous <i>IER3IP1</i> 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, 2788-2796.	0.7	42
5	Distinct Ocular Expression in Infants and Children With Down Syndrome in Cairo, Egypt. <i>JAMA Ophthalmology</i> , 2013, 131, 1057.	1.4	25
6	De Novo Mutation in <i>ABCC9</i> Causes Hypertrichosis Acromegaloid Facial Features Disorder. <i>Pediatric Dermatology</i> , 2016, 33, e109-13.	0.5	19
7	GAPO syndrome: first Egyptian case with ultrastructural changes in the gingiva. <i>Clinical Genetics</i> , 1997, 52, 110-115.	1.0	18
8	Oto-spondylo-megaepiphyseal dysplasia (OSMED): Clinical and radiological findings in sibs homozygous for premature stop codon mutation in the <i>COL11A2</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1189-1195.	0.7	17
9	Growth charts of Down syndrome in Egypt: A study of 434 children 0–36 months of age. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2647-2655.	0.7	17
10	Clinical features of <i>SMARCA2</i> duplication overlap with Coffin–Siris syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2662-2670.	0.7	15
11	Evaluation of Superoxide Dismutase and Glutathione Peroxidase Enzymes and Their Cofactors in Egyptian Children with Down's Syndrome. <i>Biological Trace Element Research</i> , 2001, 81, 21-28.	1.9	14
12	Mutational analysis of the <i>PTPN11</i> gene in Egyptian patients with Noonan syndrome. <i>Journal of the Formosan Medical Association</i> , 2013, 112, 707-712.	0.8	12
13	Expanding the mutation and clinical spectrum of Roberts syndrome. <i>Congenital Anomalies (discontinued)</i> , 2016, 56, 154-162.	0.3	12
14	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.	0.7	11
15	Ectodermal Abnormalities in Patients with Kabuki Syndrome. <i>Pediatric Dermatology</i> , 2011, 28, 507-511.	0.5	9
16	Magnetic resonance imaging of developmental facial paresis: a spectrum of complex anomalies. <i>Neuroradiology</i> , 2018, 60, 1053-1061.	1.1	9
17	Constitutional retinoblastoma gene deletion in Egyptian patients. <i>World Journal of Pediatrics</i> , 2009, 5, 222-225.	0.8	7
18	Chernin syndrome versus rhombencephalosynapsis spectrum: A rare co-occurrence with bipartite parietal bone. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 480-483.	0.7	7

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19	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.	0.7	7
20	Growth curves of Egyptian patients with Turner syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2687-2691.	0.7	6
21	Further delineation of the oculoauricular syndrome phenotype: A new family with a novel truncating HMX1 mutation. Ophthalmic Genetics, 2018, 39, 215-220.	0.5	6
22	Phenotypic and molecular insights into <i>PQBP1</i> -related intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 2446-2450.	0.7	5
23	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, 167, 2418-2424.	0.7	4
24	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
25	Body composition in Egyptian Turner syndrome girls. Indian Journal of Human Genetics, 2013, 19, 150.	0.7	3
26	Lipoid proteinosis: A clinical and molecular study in Egyptian patients. Gene, 2017, 628, 308-314.	1.0	3
27	Lenz-Majewski syndrome in a patient from Egypt. American Journal of Medical Genetics, Part A, 2019, 179, 2039-2042.	0.7	3
28	Congenital isolated leukonychia totalis in three Egyptian sibs. , 2011, 155, 811-814.		1
29	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
30	Clinical Variability of Pallister-Killian Syndrome in Two Egyptian Patients. Journal of Pediatric Genetics, 2020, 09, 207-210.	0.3	0
31	Genetic Implications in High-Risk Pregnancy and Its Outcome: A 2-year Study. American Journal of Perinatology, 2021, , .	0.6	0
32	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.	0.3	0