Ghada A Otaify

List of Publications by Year in Descending Order

30 papers	416 citations	13 h-index	20 g-index
32 ext. papers	569 ext. citations	5 avg, IF	2.89 L-index

#	Paper	IF	Citations
30	Osteoporosis-pseudoglioma syndrome in four new patients: identification of two novel LRP5 variants and insights on patientsVmanagement using bisphosphonates therapy.. *Osteoporosis International*, **2022**, 1	5.3	0
29	Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. *European Journal of Medical Genetics*, **2021**, 65, 104377	2.6	0
28	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
27	Biallelic truncating variants in MAPKAPK5 cause a new developmental disorder involving neurological, cardiac, and facial anomalies combined with synpolydactyly. *Genetics in Medicine*, **2021**, 23, 679-688	8.1	1
26	3D assessment of intervertebral disc degeneration in zebrafish identifies changes in bone density that prime disc disease. *Bone Research*, **2021**, 9, 39	13.3	4
25	First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. *Molecular Syndromology*, **2021**, 12, 279-288	1.5	0
24	Heterozygous pathogenic variants in GLI1 are a common finding in isolated postaxial polydactyly A/B. *Human Mutation*, **2020**, 41, 265-276	4.7	5
23	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. *American Journal of Human Genetics*, **2020**, 107, 977-988	11	9
22	Blepharophimosis-ptosis-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	2.5	1
21	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. *Nature Communications*, **2019**, 10, 797	17.4	10
20	Cellular stress due to impairment of collagen prolyl hydroxylation complex is rescued by the chaperone 4-phenylbutyrate. *DMM Disease Models and Mechanisms*, **2019**, 12,	4.1	16
19	Genomic and phenotypic delineation of congenital microcephaly. *Genetics in Medicine*, **2019**, 21, 545-552	8.1	55
18	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
17	Expanding the phenome and variome of skeletal dysplasia. *Genetics in Medicine*, **2018**, 20, 1609-1616	8.1	20
16	mutations are responsible for autosomal recessive osteogenesis imperfecta. *Journal of Medical Genetics*, **2018**, 55, 278-284	5.8	41
15	PGAP3-related hyperphosphatasia with mental retardation syndrome: Report of 10 new patients and a homozygous founder mutation. *Clinical Genetics*, **2018**, 93, 84-91	4	14
14	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). *Bone*, **2018**, 107, 161-171	4.7	13

13	Genetic study of eight Egyptian patients with pycnodysostosis: identification of novel CTSK mutations and founder effect. *Osteoporosis International*, **2018**, 29, 1833-1841	5.3	8
12	Molecular and clinical analysis of ALPL in a cohort of patients with suspicion of Hypophosphatasia. *American Journal of Medical Genetics, Part A*, **2017**, 173, 601-610	2.5	26
11	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 28-39	2.3	28
10	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	3.9	1
9	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. *Journal of Proteomics*, **2017**, 167, 46-59	3.9	15
8	Raine Syndrome (OMIM #259775), Caused By FAM20C Mutation, Is Congenital Sclerosing Osteomalacia With Cerebral Calcification (OMIM 259660). *Journal of Bone and Mineral Research*, **2017**, 32, 757-769	6.3	26
7	Zoledronic acid in children with osteogenesis imperfecta and Bruck syndrome: a 2-year prospective observational study. *Osteoporosis International*, **2016**, 27, 81-92	5.3	13
6	Specific variants in WDR35 cause a distinctive form of Ellis-van Creveld syndrome by disrupting the recruitment of the EvC complex and SMO into the cilium. *Human Molecular Genetics*, **2015**, 24, 4126-37	5.6	37
5	Mutational spectrum of COL1A1 and COL1A2 in Egyptian patients with autosomal dominant osteogenesis imperfecta with clinical severity score and genotype/phenotype correlation. *Middle East Journal of Medical Genetics*, **2015**, 4, 7-12		1
4	Exome sequencing in the diagnosis of an atypical phenotype of infantile hyalinosis. *Middle East Journal of Medical Genetics*, **2015**, 4, 18-23		1
3	Clinical and molecular characterization of seven Egyptian families with autosomal recessive robinow syndrome: Identification of four novel ROR2 gene mutations. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3054-61	2.5	7
2	A novel homozygous mutation in FGFR3 causes tall stature, severe lateral tibial deviation, scoliosis, hearing impairment, camptodactyly, and arachnodactyly. *Human Mutation*, **2014**, 35, 959-63	4.7	51
1	Anthropometric measurements in Egyptian patients with osteogenesis imperfecta. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2714-8	2.5	6