

# Ghada A Otaify

## List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 545-5528.1		55
29	A novel homozygous mutation in FGFR3 causes tall stature, severe lateral tibial deviation, scoliosis, hearing impairment, camptodactyly, and arachnodactyly. <i>Human Mutation</i> , <b>2014</b> , 35, 959-63	4.7	51
28	mutations are responsible for autosomal recessive osteogenesis imperfecta. <i>Journal of Medical Genetics</i> , <b>2018</b> , 55, 278-284	5.8	41
27	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. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 4126-37	5.6	37
26	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2017</b> , 5, 28-39	2.3	28
25	Molecular and clinical analysis of ALPL in a cohort of patients with suspicion of Hypophosphatasia. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 601-610	2.5	26
24	Raine Syndrome (OMIM #259775), Caused By FAM20C Mutation, Is Congenital Sclerosing Osteomalacia With Cerebral Calcification (OMIM 259660). <i>Journal of Bone and Mineral Research</i> , <b>2017</b> , 32, 757-769	6.3	26
23	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 1609-1616	8.1	20
22	Cellular stress due to impairment of collagen prolyl hydroxylation complex is rescued by the chaperone 4-phenylbutyrate. <i>DMM Disease Models and Mechanisms</i> , <b>2019</b> , 12,	4.1	16
21	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. <i>Journal of Proteomics</i> , <b>2017</b> , 167, 46-59	3.9	15
20	PGAP3-related hyperphosphatasia with mental retardation syndrome: Report of 10 new patients and a homozygous founder mutation. <i>Clinical Genetics</i> , <b>2018</b> , 93, 84-91	4	14
19	Zoledronic acid in children with osteogenesis imperfecta and Bruck syndrome: a 2-year prospective observational study. <i>Osteoporosis International</i> , <b>2016</b> , 27, 81-92	5.3	13
18	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). <i>Bone</i> , <b>2018</b> , 107, 161-171	4.7	13
17	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. <i>Nature Communications</i> , <b>2019</b> , 10, 797	17.4	10
16	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. <i>American Journal of Human Genetics</i> , <b>2020</b> , 107, 977-988	11	9
15	Genetic study of eight Egyptian patients with pycnodysostosis: identification of novel CTSK mutations and founder effect. <i>Osteoporosis International</i> , <b>2018</b> , 29, 1833-1841	5.3	8
14	Clinical and molecular characterization of seven Egyptian families with autosomal recessive robinow syndrome: Identification of four novel ROR2 gene mutations. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167A, 3054-61	2.5	7

13	Anthropometric measurements in Egyptian patients with osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 2714-8	2.5	6
12	Heterozygous pathogenic variants in GLI1 are a common finding in isolated postaxial polydactyly A/B. <i>Human Mutation</i> , <b>2020</b> , 41, 265-276	4.7	5
11	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> , <b>2019</b> , 179, 237-242	2.5	5
10	3D assessment of intervertebral disc degeneration in zebrafish identifies changes in bone density that prime disc disease. <i>Bone Research</i> , <b>2021</b> , 9, 39	13.3	4
9	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2021</b> , 9, e1829	2.3	2
8	A novel frameshift mutation in the sterol 27-hydroxylase gene in an Egyptian family with cerebrotendinous xanthomatosis without cataract. <i>Metabolic Brain Disease</i> , <b>2017</b> , 32, 311-315	3.9	1
7	Mutational spectrum of COL1A1 and COL1A2 in Egyptian patients with autosomal dominant osteogenesis imperfecta with clinical severity score and genotype/phenotype correlation. <i>Middle East Journal of Medical Genetics</i> , <b>2015</b> , 4, 7-12		1
6	Exome sequencing in the diagnosis of an atypical phenotype of infantile hyalinosis. <i>Middle East Journal of Medical Genetics</i> , <b>2015</b> , 4, 18-23		1
5	Blepharophimosis-ptosis-intellectual disability syndrome: A report of nine Egyptian patients with further expansion of phenotypic and mutational spectrum. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 2857-2866	2.5	1
4	Biallelic truncating variants in MAPKAPK5 cause a new developmental disorder involving neurological, cardiac, and facial anomalies combined with synpolydactyly. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 679-688	8.1	1
3	Osteoporosis-pseudoglioma syndrome in four new patients: identification of two novel LRP5 variants and insights on patients management using bisphosphonates therapy. <i>Osteoporosis International</i> , <b>2022</b> , 1	5.3	0
2	Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. <i>European Journal of Medical Genetics</i> , <b>2021</b> , 65, 104377	2.6	0
1	First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. <i>Molecular Syndromology</i> , <b>2021</b> , 12, 279-288	1.5	0