Mona S Aglan

List of Publications by Year in Descending Order

61 papers	2,380 citations	24 h-index	48 g-index
65 ext. papers	2,768 ext. citations	6.1 avg, IF	4.02 L-index

#	Paper	IF	Citations
61	Osteoporosis-pseudoglioma syndrome in four new patients: identification of two novel LRP5 variants and insights on patientsUmanagement using bisphosphonates therapy.. *Osteoporosis International*, **2022**, 1	5.3	0
60	Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. *European Journal of Medical Genetics*, **2021**, 65, 104377	2.6	0
59	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
58	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
57	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
56	First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. *Molecular Syndromology*, **2021**, 12, 279-288	1.5	0
55	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
54	Heterozygous pathogenic variants in GLI1 are a common finding in isolated postaxial polydactyly A/B. *Human Mutation*, **2020**, 41, 265-276	4.7	5
53	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
52	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
51	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. *Nature Communications*, **2019**, 10, 797	17.4	10
50	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
49	Genomic and phenotypic delineation of congenital microcephaly. *Genetics in Medicine*, **2019**, 21, 545-552	8.1	55
48	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
47	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
46	Expanding the phenome and variome of skeletal dysplasia. *Genetics in Medicine*, **2018**, 20, 1609-1616	8.1	20
45	mutations are responsible for autosomal recessive osteogenesis imperfecta. *Journal of Medical Genetics*, **2018**, 55, 278-284	5.8	41

44	Phenotypic and molecular insights into PQBP1-related intellectual disability. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2446-2450	2.5	1
43	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
42	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
41	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. *Journal of Proteomics*, **2017**, 167, 46-59	3.9	15
40	Four novel mutations in the N-acetylgalactosamine-6-sulfate sulfatase gene among Egyptian patients with Morquio A disease. *Gene*, **2017**, 600, 48-54	3.8	3
39	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
38	Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. *Metabolic Brain Disease*, **2016**, 31, 1171-9	3.9	4
37	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
36	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
35	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. *Nature Genetics*, **2015**, 47, 528-34	36.3	89
34	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
33	A Novel Loss-of-Sclerostin Function Mutation in a First Egyptian Family with Sclerosteosis. *BioMed Research International*, **2015**, 2015, 517815	3	6
32	Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. *Human Mutation*, **2014**, 35, 1203-10	4.7	67
31	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
30	OSX/SP7 Mutations and Osteogenesis Imperfecta **2014**, 173-179		1
29	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta **2014**, 181-186		1
28	Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1143-50	2.5	23
27	A report of another Egyptian patient with Temtamy preaxial brachydactyly syndrome associated with a novel nonsense CHSY1 mutation. *Middle East Journal of Medical Genetics*, **2014**, 3, 37-41		2

26	Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. *American Journal of Human Genetics*, **2013**, 92, 598-604	11	90
25	Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. *Lancet Neurology, The*, **2013**, 12, 1159-69	24.1	267
24	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotype-phenotype correlations. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1354-69	2.5	39
23	Mutations in WNT1 cause different forms of bone fragility. *American Journal of Human Genetics*, **2013**, 92, 565-74	11	197
22	Further delineation of the clinical spectrum in RNU4ATAC related microcephalic osteodysplastic primordial dwarfism type I. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1875-81	2.5	16
21	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. *Human Mutation*, **2012**, 33, 343-50	4.7	153
20	Growth curves of Egyptian patients with Turner syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2687-91	2.5	5
19	Anthropometric measurements in Egyptian patients with osteogenesis imperfecta. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2714-8	2.5	6
18	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome--osteogenesis imperfecta phenotypic spectrum. *Human Mutation*, **2012**, 33, 1444-9	4.7	67
17	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
16	A scoring system for the assessment of clinical severity in osteogenesis imperfecta. *Journal of Childrens Orthopaedics*, **2012**, 6, 29-35	2.1	15
15	Consanguinity and genetic disorders in Egypt. *Middle East Journal of Medical Genetics*, **2012**, 1, 12-17		30
14	Definition of the phenotypic spectrum of Temtamy preaxial brachydactyly syndrome associated with autosomal recessive CHYS1 mutations. *Middle East Journal of Medical Genetics*, **2012**, 1, 64-70		6
13	Springtime for science in Egypt. *Science*, **2011**, 332, 1032	33.3	
12	Limb malformations with associated congenital constriction rings in two unrelated Egyptian males, one with a disorganization-like spectrum and the other with a probable distinct type of septo-optic dysplasia. *Clinical Dysmorphology*, **2010**, 19, 14-22	0.9	5
11	LRP4 mutations alter Wnt/beta-catenin signaling and cause limb and kidney malformations in Cenani-Lenz syndrome. *American Journal of Human Genetics*, **2010**, 86, 696-706	11	127
10	Identification of a frameshift mutation in Osterix in a patient with recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2010**, 87, 110-4	11	212
9	Temtamy preaxial brachydactyly syndrome is caused by loss-of-function mutations in chondroitin synthase 1, a potential target of BMP signaling. *American Journal of Human Genetics*, **2010**, 87, 757-67	11	77

8	Genetic Disorders in Egypt **2010**, 219-272		3
7	Widening the mutation spectrum of EVC and EVC2: ectopic expression of Weyer variants in NIH 3T3 fibroblasts disrupts Hedgehog signaling. *Human Mutation*, **2009**, 30, 1667-75	4.7	48
6	Dyggve-Melchior-Clausen syndrome: clinical, genetic, and radiological study of 15 Egyptian patients from nine unrelated families. *Journal of Childrens Orthopaedics*, **2009**, 3, 451-8	2.1	17
5	The primordial growth disorder 3-M syndrome connects ubiquitination to the cytoskeletal adaptor OBSL1. *American Journal of Human Genetics*, **2009**, 84, 801-6	11	83
4	Brachydactyly. *Orphanet Journal of Rare Diseases*, **2008**, 3, 15	4.2	98
3	Long interspersed nuclear element-1 (LINE1)-mediated deletion of EVC, EVC2, C4orf6, and STK32B in Ellis-van Creveld syndrome with borderline intelligence. *Human Mutation*, **2008**, 29, 931-8	4.7	43
2	Adams-Oliver syndrome: further evidence of an autosomal recessive variant. *Clinical Dysmorphology* , **2007**, 16, 141-149	0.9	24
1	3-M syndrome: a report of three Egyptian cases with review of the literature. *Clinical Dysmorphology*, **2006**, 15, 55-64	0.9	20