

# Mona S Aglan

## List of Publications by Citations

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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	Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. <i>Lancet Neurology</i> , <b>2013</b> , 12, 1159-69	24.1	267
60	Identification of a frameshift mutation in Osterix in a patient with recessive osteogenesis imperfecta. <i>American Journal of Human Genetics</i> , <b>2010</b> , 87, 110-4	11	212
59	Mutations in WNT1 cause different forms of bone fragility. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 565-74	11	197
58	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. <i>Human Mutation</i> , <b>2012</b> , 33, 343-50	4.7	153
57	LRP4 mutations alter Wnt/beta-catenin signaling and cause limb and kidney malformations in Cenani-Lenz syndrome. <i>American Journal of Human Genetics</i> , <b>2010</b> , 86, 696-706	11	127
56	Brachydactyly. <i>Orphanet Journal of Rare Diseases</i> , <b>2008</b> , 3, 15	4.2	98
55	Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 598-604	11	90
54	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. <i>Nature Genetics</i> , <b>2015</b> , 47, 528-34	36.3	89
53	The primordial growth disorder 3-M syndrome connects ubiquitination to the cytoskeletal adaptor OBSL1. <i>American Journal of Human Genetics</i> , <b>2009</b> , 84, 801-6	11	83
52	Temtamy preaxial brachydactyly syndrome is caused by loss-of-function mutations in chondroitin synthase 1, a potential target of BMP signaling. <i>American Journal of Human Genetics</i> , <b>2010</b> , 87, 757-67	11	77
51	Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. <i>Human Mutation</i> , <b>2014</b> , 35, 1203-10	4.7	67
50	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome--osteogenesis imperfecta phenotypic spectrum. <i>Human Mutation</i> , <b>2012</b> , 33, 1444-9	4.7	67
49	Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 545-5528.1		55
48	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
47	Widening the mutation spectrum of EVC and EVC2: ectopic expression of Weyer variants in NIH 3T3 fibroblasts disrupts Hedgehog signaling. <i>Human Mutation</i> , <b>2009</b> , 30, 1667-75	4.7	48
46	Long interspersed nuclear element-1 (LINE1)-mediated deletion of EVC, EVC2, C4orf6, and STK32B in Ellis-van Creveld syndrome with borderline intelligence. <i>Human Mutation</i> , <b>2008</b> , 29, 931-8	4.7	43
45	mutations are responsible for autosomal recessive osteogenesis imperfecta. <i>Journal of Medical Genetics</i> , <b>2018</b> , 55, 278-284	5.8	41

44	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotype-phenotype correlations. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 1354-69	2.5	39
43	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
42	Consanguinity and genetic disorders in Egypt. <i>Middle East Journal of Medical Genetics</i> , <b>2012</b> , 1, 12-17		30
41	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
40	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
39	Adams-Oliver syndrome: further evidence of an autosomal recessive variant. <i>Clinical Dysmorphology</i> , <b>2007</b> , 16, 141-149	0.9	24
38	Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. <i>American Journal of Medical Genetics, Part A</i> , <b>2014</b> , 164A, 1143-50	2.5	23
37	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 1609-1616	8.1	20
36	3-M syndrome: a report of three Egyptian cases with review of the literature. <i>Clinical Dysmorphology</i> , <b>2006</b> , 15, 55-64	0.9	20
35	Dyggve-Melchior-Clausen syndrome: clinical, genetic, and radiological study of 15 Egyptian patients from nine unrelated families. <i>Journal of Childrens Orthopaedics</i> , <b>2009</b> , 3, 451-8	2.1	17
34	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
33	Further delineation of the clinical spectrum in RNU4ATAC related microcephalic osteodysplastic primordial dwarfism type I. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 1875-81	2.5	16
32	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
31	A scoring system for the assessment of clinical severity in osteogenesis imperfecta. <i>Journal of Childrens Orthopaedics</i> , <b>2012</b> , 6, 29-35	2.1	15
30	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
29	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> , <b>2012</b> , 158A, 2647-55	2.5	12
28	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
27	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

26	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
25	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> , <b>2020</b> , 182, 1407-1420	2.5	6
24	A Novel Loss-of-Sclerostin Function Mutation in a First Egyptian Family with Sclerosteosis. <i>BioMed Research International</i> , <b>2015</b> , 2015, 517815	3	6
23	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
22	Definition of the phenotypic spectrum of Temtamy preaxial brachydactyly syndrome associated with autosomal recessive CHYS1 mutations. <i>Middle East Journal of Medical Genetics</i> , <b>2012</b> , 1, 64-70		6
21	Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia type-2. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1190-1194	2.5	5
20	Growth curves of Egyptian patients with Turner syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 2687-91	2.5	5
19	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. <i>Clinical Dysmorphology</i> , <b>2010</b> , 19, 14-22	0.9	5
18	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
17	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
16	Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. <i>Metabolic Brain Disease</i> , <b>2016</b> , 31, 1171-9	3.9	4
15	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
14	Four novel mutations in the N-acetylgalactosamine-6-sulfate sulfatase gene among Egyptian patients with Morquio A disease. <i>Gene</i> , <b>2017</b> , 600, 48-54	3.8	3
13	Genetic Disorders in Egypt <b>2010</b> , 219-272		3
12	A report of another Egyptian patient with Temtamy preaxial brachydactyly syndrome associated with a novel nonsense CHSY1 mutation. <i>Middle East Journal of Medical Genetics</i> , <b>2014</b> , 3, 37-41		2
11	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
10	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
9	OSX/SP7 Mutations and Osteogenesis Imperfecta <b>2014</b> , 173-179		1

8	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta <b>2014</b> , 181-186		1
7	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
6	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
5	Phenotypic and molecular insights into PQBP1-related intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 2446-2450	2.5	1
4	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
3	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
2	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
1	Springtime for science in Egypt. <i>Science</i> , <b>2011</b> , 332, 1032	33.3	