

Samia Ali Li Temtamy

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.

92
papers

3,643
citations

30
h-index

59
g-index

93
ext. papers

4,132
ext. citations

5.9
avg, IF

4.43
L-index

#	Paper	IF	Citations
92	Loss-of-function mutations in the cathepsin C gene result in periodontal disease and palmoplantar keratosis. <i>Nature Genetics</i> , 1999 , 23, 421-4	36.3	377
91	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> , 2013 , 12, 1159-69	24.1	267
90	Identification of a frameshift mutation in Osterix in a patient with recessive osteogenesis imperfecta. <i>American Journal of Human Genetics</i> , 2010 , 87, 110-4	11	212
89	Extending the scope of the VATER association: definition of the VATER syndrome. <i>Journal of Pediatrics</i> , 1974 , 85, 345-9	3.6	202
88	Mutations in WNT1 cause different forms of bone fragility. <i>American Journal of Human Genetics</i> , 2013 , 92, 565-74	11	197
87	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. <i>Human Mutation</i> , 2012 , 33, 343-50	4.7	153
86	LRP4 mutations alter Wnt/beta-catenin signaling and cause limb and kidney malformations in Cenani-Lenz syndrome. <i>American Journal of Human Genetics</i> , 2010 , 86, 696-706	11	127
85	Brachydactyly. <i>Orphanet Journal of Rare Diseases</i> , 2008 , 3, 15	4.2	98
84	Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. <i>American Journal of Human Genetics</i> , 2013 , 92, 598-604	11	90
83	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. <i>Nature Genetics</i> , 2015 , 47, 528-34	36.3	89
82	A minority of 46,XX true hermaphrodites are positive for the Y-DNA sequence including SRY. <i>Human Genetics</i> , 1992 , 90, 121-5	6.3	85
81	The primordial growth disorder 3-M syndrome connects ubiquitination to the cytoskeletal adaptor OBSL1. <i>American Journal of Human Genetics</i> , 2009 , 84, 801-6	11	83
80	Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. <i>Nature Genetics</i> , 1995 , 11, 338-40	36.3	80
79	Macrodactyly, hemihypertrophy, and connective tissue nevi: report of a new syndrome and review of the literature. <i>Journal of Pediatrics</i> , 1976 , 89, 924-7	3.6	78
78	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> , 2010 , 87, 757-67	11	77
77	Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. <i>Human Mutation</i> , 2014 , 35, 1203-10	4.7	67
76	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome--osteogenesis imperfecta phenotypic spectrum. <i>Human Mutation</i> , 2012 , 33, 1444-9	4.7	67

75	The Coffin-Lowry syndrome: an inherited faciodigital mental retardation syndrome. <i>Journal of Pediatrics</i> , 1975 , 86, 724-31	3.6	61
74	Mutations in ANTXR1 cause GAPO syndrome. <i>American Journal of Human Genetics</i> , 2013 , 92, 792-9	11	60
73	Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , 2019 , 21, 545-5528.1		55
72	The syndrome of aplasia cutis congenita with terminal, transverse defects of limbs. <i>Journal of Pediatrics</i> , 1975 , 87, 79-82	3.6	53
71	A novel homozygous mutation in FGFR3 causes tall stature, severe lateral tibial deviation, scoliosis, hearing impairment, camptodactyly, and arachnodactyly. <i>Human Mutation</i> , 2014 , 35, 959-63	4.7	51
70	Widening the mutation spectrum of EVC and EVC2: ectopic expression of Weyer variants in NIH 3T3 fibroblasts disrupts Hedgehog signaling. <i>Human Mutation</i> , 2009 , 30, 1667-75	4.7	48
69	Molecular characterization of beta-thalassemia in Egyptians. <i>Human Mutation</i> , 1993 , 2, 48-52	4.7	44
68	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> , 2008 , 29, 931-8	4.7	43
67	mutations are responsible for autosomal recessive osteogenesis imperfecta. <i>Journal of Medical Genetics</i> , 2018 , 55, 278-284	5.8	41
66	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> , 2013 , 161A, 1354-69	2.5	39
65	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> , 2015 , 24, 4126-37	5.6	37
64	Consanguinity and genetic disorders in Egypt. <i>Middle East Journal of Medical Genetics</i> , 2012 , 1, 12-17		30
63	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. <i>Molecular Genetics & Genomic Medicine</i> , 2017 , 5, 28-39	2.3	28
62	Molecular and clinical analysis of ALPL in a cohort of patients with suspicion of Hypophosphatasia. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 601-610	2.5	26
61	Familial Leydig cell hypoplasia as a cause of male pseudohermaphroditism. <i>Human Heredity</i> , 1987 , 37, 36-40	1.1	26
60	Neu Laxova syndrome in two Egyptian families. <i>American Journal of Medical Genetics Part A</i> , 1991 , 41, 30-1		25
59	Adams-Oliver syndrome: further evidence of an autosomal recessive variant. <i>Clinical Dysmorphology</i> , 2007 , 16, 141-149	0.9	24
58	Hypogenitalism in the acrocallosal syndrome. <i>American Journal of Medical Genetics Part A</i> , 1989 , 32, 301-5		24

57	Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1143-50	2.5	23
56	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , 2018 , 20, 1609-1616	8.1	20
55	3-M syndrome: a report of three Egyptian cases with review of the literature. <i>Clinical Dysmorphology</i> , 2006 , 15, 55-64	0.9	20
54	Mild facial dysmorphism and quasidominant inheritance in Cenani-Lenz syndrome. <i>Clinical Dysmorphology</i> , 2003 , 12, 77-83	0.9	20
53	Dyggve-Melchior-Clausen syndrome: clinical, genetic, and radiological study of 15 Egyptian patients from nine unrelated families. <i>Journal of Children's Orthopaedics</i> , 2009 , 3, 451-8	2.1	17
52	Cellular stress due to impairment of collagen prolyl hydroxylation complex is rescued by the chaperone 4-phenylbutyrate. <i>DMM Disease Models and Mechanisms</i> , 2019 , 12,	4.1	16
51	Karyotype/phenotype correlation in females with short stature. <i>Clinical Genetics</i> , 1992 , 41, 147-51	4	16
50	Cataract, hypertrichosis, and mental retardation (CAHMR): a new autosomal recessive syndrome. <i>American Journal of Medical Genetics Part A</i> , 1991 , 41, 432-3		16
49	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. <i>Journal of Proteomics</i> , 2017 , 167, 46-59	3.9	15
48	A scoring system for the assessment of clinical severity in osteogenesis imperfecta. <i>Journal of Children's Orthopaedics</i> , 2012 , 6, 29-35	2.1	15
47	GAPO syndrome: first Egyptian case with ultrastructural changes in the gingiva. <i>Clinical Genetics</i> , 1997 , 52, 110-5	4	15
46	Oto-spondylo-megaepiphyseal dysplasia (OSMED): clinical and radiological findings in sibs homozygous for premature stop codon mutation in the COL11A2 gene. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 1189-95	2.5	15
45	Zoledronic acid in children with osteogenesis imperfecta and Bruck syndrome: a 2-year prospective observational study. <i>Osteoporosis International</i> , 2016 , 27, 81-92	5.3	13
44	Muenke syndrome with pigmentary disorder and probable hemimegalencephaly: An expansion of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 207-14	2.5	12
43	Characteristics of patients with congenital clasped thumb: a prospective study of 40 patients with the results of treatment. <i>Journal of Children's Orthopaedics</i> , 2007 , 1, 313-22	2.1	12
42	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. <i>Nature Communications</i> , 2019 , 10, 797	17.4	10
41	Transposition of external genitalia and associated malformations. <i>Clinical Dysmorphology</i> , 2003 , 12, 59-62.9	6.9	10
40	Direct molecular analysis of the fragile X syndrome in a sample of Egyptian and German patients using non-radioactive PCR and Southern blot followed by chemiluminescent detection. <i>Human Genetics</i> , 1995 , 96, 577-84	6.3	9

39	The Genetics of Hand Malformations: Updated*. <i>Congenital Anomalies (discontinued)</i> , 1985 , 25, 73-92	1.1	9
38	A postaxial polydactyly-dental-vertebral syndrome. <i>Journal of Pediatrics</i> , 1977 , 90, 230-5	3.6	9
37	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. <i>American Journal of Human Genetics</i> , 2020 , 107, 977-988	11	9
36	A novel point mutation of the androgen receptor (F804L) in an Egyptian newborn with complete androgen insensitivity associated with congenital glaucoma and hypertrophic pyloric stenosis. <i>Clinical Genetics</i> , 2003 , 63, 59-63	4	8
35	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> , 2015 , 167A, 3054-61	2.5	7
34	The DR syndrome or the Okihiro syndrome?. <i>American Journal of Medical Genetics Part A</i> , 1986 , 25, 173-4		7
33	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	2.5	6
32	Anthropometric measurements in Egyptian patients with osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 2714-8	2.5	6
31	How many entities exist for the spectrum of disorders associated with brachydactyly, syndactyly, short stature, microcephaly, and intellectual disability?. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 880-4	2.5	6
30	Definition of the phenotypic spectrum of Temtamy preaxial brachydactyly syndrome associated with autosomal recessive CHYS1 mutations. <i>Middle East Journal of Medical Genetics</i> , 2012 , 1, 64-70		6
29	Recurrent homozygous damaging mutation in , encoding a protein disulfide isomerase, in four families with microlissencephaly. <i>Journal of Medical Genetics</i> , 2020 , 57, 274-282	5.8	5
28	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> , 2010 , 19, 14-22	0.9	5
27	Catel???Manzke digitopalatal syndrome or Temtamy preaxial brachydactyly hyperphalangism syndrome?. <i>Clinical Dysmorphology</i> , 2005 , 14, 211	0.9	5
26	Greig cephalopolysyndactyly syndrome with dysgenesis of the corpus callosum in a Bedouin family. <i>American Journal of Medical Genetics Part A</i> , 1996 , 66, 261-4		5
25	Heterozygous pathogenic variants in GLI1 are a common finding in isolated postaxial polydactyly A/B. <i>Human Mutation</i> , 2020 , 41, 265-276	4.7	5
24	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> , 2019 , 179, 237-242	2.5	5
23	Genetics and Genomic Medicine in Egypt: steady pace. <i>Molecular Genetics & Genomic Medicine</i> , 2017 , 5, 8-14	2.3	4
22	The Development of Human Genetics at the National Research Centre, Cairo, Egypt: A Story of 50 Years. <i>Annual Review of Genomics and Human Genetics</i> , 2019 , 20, 1-19	9.7	4

21	Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. <i>Metabolic Brain Disease</i> , 2016 , 31, 1171-9	3.9	4
20	3D assessment of intervertebral disc degeneration in zebrafish identifies changes in bone density that prime disc disease. <i>Bone Research</i> , 2021 , 9, 39	13.3	4
19	Cytogenetic effect of two antimonial antibilharzial drugs: tartar emetic and bilharcid. <i>Environmental Mutagenesis</i> , 1982 , 4, 83-91		3
18	Genetic Disorders in Egypt 2010 , 219-272		3
17	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> , 2014 , 3, 37-41		2
16	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1829	2.3	2
15	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> , 2015 , 4, 7-12		1
14	OSX/SP7 Mutations and Osteogenesis Imperfecta 2014 , 173-179		1
13	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta 2014 , 181-186		1
12	A boy with hand anomalies similar to those documented with prenatal misoprostol exposure. <i>Middle East Journal of Medical Genetics</i> , 2013 , 2, 58-62		1
11	Two different Temtamy syndromes. <i>Clinical Dysmorphology</i> , 2013 , 22, 91	0.9	1
10	Letter: On anomalies associated with radial dysplasia. <i>Journal of Pediatrics</i> , 1974 , 85, 585	3.6	1
9	Letter: On the nomenclature of "new" syndrome. <i>Journal of Pediatrics</i> , 1974 , 84, 608-9	3.6	1
8	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> , 2020 , 182, 2857-2866	2.5	1
7	Biallelic truncating variants in MAPKAPK5 cause a new developmental disorder involving neurological, cardiac, and facial anomalies combined with synpolydactyly. <i>Genetics in Medicine</i> , 2021 , 23, 679-688	8.1	1
6	Phenotypic and molecular insights into PQBP1-related intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2446-2450	2.5	1
5	Using Online Mendelian Inheritance in Man in low- and middle-income countries. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3284-3286	2.5	1
4	Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. <i>European Journal of Medical Genetics</i> , 2021 , 65, 104377	2.6	0

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| 3 | First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. <i>Molecular Syndromology</i> , 2021 , 12, 279-288 | 1.5 | 0 |
| 2 | Mother and daughter with a novel phenotype of hand and foot anomalies and severe pectus excavatum. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1867 | 2.5 | |
| 1 | Catel-Manzke digitopalatal syndrome or Temtamy preaxial brachydactyly hyperphalangism syndrome?. <i>Clinical Dysmorphology</i> , 2005 , 14, 211 | 0.9 | |