Samia Ali Li Temtamy

List of Publications by Year in Descending Order

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	Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. *European Journal of Medical Genetics*, **2021**, 65, 104377	2.6	0
91	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
90	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
89	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
88	Using Online Mendelian Inheritance in Man in low- and middle-income countries. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3284-3286	2.5	1
87	First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. *Molecular Syndromology*, **2021**, 12, 279-288	1.5	0
86	Recurrent homozygous damaging mutation in , encoding a protein disulfide isomerase, in four families with microlissencephaly. *Journal of Medical Genetics*, **2020**, 57, 274-282	5.8	5
85	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
84	Heterozygous pathogenic variants in GLI1 are a common finding in isolated postaxial polydactyly A/B. *Human Mutation*, **2020**, 41, 265-276	4.7	5
83	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
82	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
81	The Development of Human Genetics at the National Research Centre, Cairo, Egypt: A Story of 50 Years. *Annual Review of Genomics and Human Genetics*, **2019**, 20, 1-19	9.7	4
80	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. *Nature Communications*, **2019**, 10, 797	17.4	10
79	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
78	Genomic and phenotypic delineation of congenital microcephaly. *Genetics in Medicine*, **2019**, 21, 545-552	8.1	55
77	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
76	Expanding the phenome and variome of skeletal dysplasia. *Genetics in Medicine*, **2018**, 20, 1609-1616	8.1	20

75	mutations are responsible for autosomal recessive osteogenesis imperfecta. *Journal of Medical Genetics*, **2018**, 55, 278-284	5.8	41
74	Phenotypic and molecular insights into PQBP1-related intellectual disability. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2446-2450	2.5	1
73	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
72	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
71	Genetics and Genomic Medicine in Egypt: steady pace. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 8-14	2.3	4
70	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. *Journal of Proteomics*, **2017**, 167, 46-59	3.9	15
69	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
68	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
67	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
66	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
65	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. *Nature Genetics*, **2015**, 47, 528-34	36.3	89
64	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
63	Mother and daughter with a novel phenotype of hand and foot anomalies and severe pectus excavatum. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1867	2.5	
62	Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. *Human Mutation*, **2014**, 35, 1203-10	4.7	67
61	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
60	OSX/SP7 Mutations and Osteogenesis Imperfecta **2014**, 173-179		1
59	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta **2014**, 181-186		1
58	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

57	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
56	Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. *American Journal of Human Genetics*, **2013**, 92, 598-604	11	90
55	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
54	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
53	Mutations in ANTXR1 cause GAPO syndrome. *American Journal of Human Genetics*, **2013**, 92, 792-9	11	60
52	Mutations in WNT1 cause different forms of bone fragility. *American Journal of Human Genetics*, **2013**, 92, 565-74	11	197
51	A boy with hand anomalies similar to those documented with prenatal misoprostol exposure. *Middle East Journal of Medical Genetics*, **2013**, 2, 58-62		1
50	Two different Temtamy syndromes. *Clinical Dysmorphology*, **2013**, 22, 91	0.9	1
49	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. *Human Mutation*, **2012**, 33, 343-50	4.7	153
48	Anthropometric measurements in Egyptian patients with osteogenesis imperfecta. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2714-8	2.5	6
47	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
46	A scoring system for the assessment of clinical severity in osteogenesis imperfecta. *Journal of Childrens Orthopaedics*, **2012**, 6, 29-35	2.1	15
45	Consanguinity and genetic disorders in Egypt. *Middle East Journal of Medical Genetics*, **2012**, 1, 12-17		30
44	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
43	Muenke syndrome with pigmentary disorder and probable hemimegalencephaly: An expansion of the phenotype. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 207-14	2.5	12
42	How many entities exist for the spectrum of disorders associated with brachydactyly, syndactyly, short stature, microcephaly, and intellectual disability?. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 880-4	2.5	6
41	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
40	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

39	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
38	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
37	Genetic Disorders in Egypt **2010**, 219-272		3
36	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
35	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
34	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
33	Brachydactyly. *Orphanet Journal of Rare Diseases*, **2008**, 3, 15	4.2	98
32	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
31	Characteristics of patients with congenital clasped thumb: a prospective study of 40 patients with the results of treatment. *Journal of Childrens Orthopaedics*, **2007**, 1, 313-22	2.1	12
30	Adams-Oliver syndrome: further evidence of an autosomal recessive variant. *Clinical Dysmorphology* , **2007**, 16, 141-149	0.9	24
29	Oto-spondylo-megaepiphyseal dysplasia (OSMED): clinical and radiological findings in sibs homozygous for premature stop codon mutation in the COL11A2 gene. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1189-95	2.5	15
28	3-M syndrome: a report of three Egyptian cases with review of the literature. *Clinical Dysmorphology*, **2006**, 15, 55-64	0.9	20
27	Catel???Manzke digitopalatal syndrome or Temtamy preaxial brachydactyly hyperphalangism syndrome?. *Clinical Dysmorphology*, **2005**, 14, 211	0.9	5
26	Catel-Manzke digitopalatal syndrome or Temtamy preaxial brachydactyly hyperphalangism syndrome?. *Clinical Dysmorphology*, **2005**, 14, 211	0.9	
25	Transposition of external genitalia and associated malformations. *Clinical Dysmorphology*, **2003**, 12, 59-62	0.9	10
24	Mild facial dysmorphism and quasidominant inheritance in Cenani-Lenz syndrome. *Clinical Dysmorphology*, **2003**, 12, 77-83	0.9	20
23	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. *Clinical Genetics*, **2003**, 63, 59-63	4	8
22	Loss-of-function mutations in the cathepsin C gene result in periodontal disease and palmoplantar keratosis. *Nature Genetics*, **1999**, 23, 421-4	36.3	377

21	GAPO syndrome: first Egyptian case with ultrastructural changes in the gingiva. *Clinical Genetics*, **1997**, 52, 110-5	4	15
20	Greig cephalopolysyndactyly syndrome with dysgenesis of the corpus callosum in a Bedouin family. *American Journal of Medical Genetics Part A*, **1996**, 66, 261-4		5
19	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. *Human Genetics*, **1995**, 96, 577-84	6.3	9
18	Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. *Nature Genetics*, **1995**, 11, 338-40	36.3	80
17	Molecular characterization of beta-thalassemia in Egyptians. *Human Mutation*, **1993**, 2, 48-52	4.7	44
16	Karyotype/phenotype correlation in females with short stature. *Clinical Genetics*, **1992**, 41, 147-51	4	16
15	A minority of 46,XX true hermaphrodites are positive for the Y-DNA sequence including SRY. *Human Genetics*, **1992**, 90, 121-5	6.3	85
14	Neu Laxova syndrome in two Egyptian families. *American Journal of Medical Genetics Part A*, **1991**, 41, 30-1		25
13	Cataract, hypertrichosis, and mental retardation (CAHMR): a new autosomal recessive syndrome. *American Journal of Medical Genetics Part A*, **1991**, 41, 432-3		16
12	Hypogenitalism in the acrocallosal syndrome. *American Journal of Medical Genetics Part A*, **1989**, 32, 301-5		24
11	Familial Leydig cell hypoplasia as a cause of male pseudohermaphroditism. *Human Heredity*, **1987**, 37, 36-40	1.1	26
10	The DR syndrome or the Okihiro syndrome?. *American Journal of Medical Genetics Part A*, **1986**, 25, 173-4		7
9	The Genetics of Hand Malformations: Updated*. *Congenital Anomalies (discontinued)*, **1985**, 25, 73-92	1.1	9
8	Cytogenetic effect of two antimonial antibilharzial drugs: tartar emetic and bilharcid. *Environmental Mutagenesis*, **1982**, 4, 83-91		3
7	A postaxial polydactyly-dental-vertebral syndrome. *Journal of Pediatrics*, **1977**, 90, 230-5	3.6	9
6	Macrodactyly, hemihypertrophy, and connective tissue nevi: report of a new syndrome and review of the literature. *Journal of Pediatrics*, **1976**, 89, 924-7	3.6	78
5	The syndrome of aplasia cutis congenita with terminal, transverse defects of limbs. *Journal of Pediatrics*, **1975**, 87, 79-82	3.6	53
4	The Coffin-Lowry syndrome: an inherited faciodigital mental retardation syndrome. *Journal of Pediatrics*, **1975**, 86, 724-31	3.6	61

3	Extending the scope of the VATER association: definition of the VATER syndrome. *Journal of Pediatrics*, **1974**, 85, 345-9	3.6	202
2	Letter: On anomalies associated with radial dysplasia. *Journal of Pediatrics*, **1974**, 85, 585	3.6	1
1	Letter: On the nomenclature of "new" syndrome. *Journal of Pediatrics*, **1974**, 84, 608-9	3.6	1