Samia Ali Li Temtamy

List of Publications by Year
in descending order

92 papers	4,696 citations	126708 33 h-index	106150 65 g-index
93 all docs	93 docs citations	93 times ranked	7064 citing authors

#	Article	IF	Citations
1	Assessment of interferon-related biomarkers in Aicardi-GoutiÃ¨res syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. Lancet Neurology, The, 2013, 12, 1159-1169.	4.9	473
2	Loss-of-function mutations in the cathepsin C gene result in periodontal disease and palmoplantar keratosis. Nature Genetics, 1999, 23, 421-424.	9.4	442
3	Identification of a Frameshift Mutation in Osterix in a Patient with Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 87, 110-114.	2.6	246
4	Extending the scope of the VATER association: Definition of the VATER syndrome. Journal of Pediatrics, 1974, 85, 345-349.	0.9	242
5	Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574.	2.6	240
6	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. Human Mutation, 2012, 33, 343-350.	1.1	178
7	LRP4 Mutations Alter Wnt/Î²-Catenin Signaling and Cause Limb and Kidney Malformations in Cenani-Lenz Syndrome. American Journal of Human Genetics, 2010, 86, 696-706.	2.6	151
8	Brachydactyly. Orphanet Journal of Rare Diseases, 2008, 3, 15.	1.2	127
9	Mutations in EOGT Confirm the Genetic Heterogeneity of Autosomal-Recessive Adams-Oliver Syndrome. American Journal of Human Genetics, 2013, 92, 598-604.	2.6	114
10	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	9.4	111
11	A minority of 46,XX true hermaphrodites are positive for the Y-DNA sequence including SRY. Human Genetics, 1992, 90, 121-5.	1.8	100
12	The Primordial Growth Disorder 3-M Syndrome Connects Ubiquitination to the Cytoskeletal Adaptor OBSL1. American Journal of Human Genetics, 2009, 84, 801-806.	2.6	93
13	Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. Nature Genetics, 1995, 11, 338-340.	9.4	89
14	Macrodactyly, hemihypertrophy, and connective tissue nevi: Report of a new syndrome and review of the literature. Journal of Pediatrics, 1976, 89, 924-927.	0.9	87
15	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, 757-767.	2.6	86
16	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	1.1	85
17	The Coffin-Lowry syndrome: An inherited faciodigital mental retardation syndrome. Journal of Pediatrics, 1975, 86, 724-731.	0.9	77
18	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome-Osteogenesis imperfecta phenotypic spectrum. Human Mutation, 2012, 33, 1444-1449.	1.1	77

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19	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	1.1	75
20	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	2.6	73
21	A Novel Homozygous Mutation in FGFR3 Causes Tall Stature, Severe Lateral Tibial Deviation, Scoliosis, Hearing Impairment, Camptodactyly, and Arachnodactyly. Human Mutation, 2014, 35, 959-963.	1.1	64
22	The syndrome of aplasia cutis congenita with terminal, transverse defects of limbs. Journal of Pediatrics, 1975, 87, 79-82.	0.9	57
23	Long interspersed nuclear element-1 (LINE1)-mediated deletion ofEVC,EVC2,C4orf6, andSTK32B in Ellisâ€“van Creveld syndrome with borderline intelligence. Human Mutation, 2008, 29, 931-938.	1.1	55
24	FAM46A mutations are responsible for autosomal recessive osteogenesis imperfecta. Journal of Medical Genetics, 2018, 55, 278-284.	1.5	55
25	Widening the mutation spectrum ofEVCandEVC2: ectopic expression of Weyer variants in NIH 3T3 fibroblasts disrupts hedgehog signaling. Human Mutation, 2009, 30, 1667-1675.	1.1	54
26	Consanguinity and genetic disorders in Egypt. Middle East Journal of Medical Genetics, 2012, 1, 12-17.	0.0	48
27	Molecular characterization of Î²-thalassemia in Egyptians. Human Mutation, 1993, 2, 48-52.	1.1	47
28	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	1.1	46
29	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2013, 161, 1354-1369.	0.7	42
30	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, 4126-4137.	1.4	42
31	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. Molecular Genetics & Genomic Medicine, 2017, 5, 28-39.	0.6	37
32	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.	0.7	36
33	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	2.6	33
34	Adamsâ€“Oliver syndrome: further evidence of an autosomal recessive variant. Clinical Dysmorphology, 2007, 16, 141-149.	0.1	32
35	Cellular stress due to impairment of collagen prolyl hydroxylation complex is rescued by the chaperone 4-phenylbutyrate. DMM Disease Models and Mechanisms, 2019, 12, .	1.2	32
36	Neu Laxova syndrome in two Egyptian families. American Journal of Medical Genetics Part A, 1991, 41, 30-31.	2.4	31

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37	3D assessment of intervertebral disc degeneration in zebrafish identifies changes in bone density that prime disc disease. Bone Research, 2021, 9, 39.	5.4	31
38	Familial Leydig Cell Hypoplasia as a Cause of Male Pseudohermaphroditism. Human Heredity, 1987, 37, 36-40.	0.4	30
39	Hypogenitalism in the acrocallosal syndrome. American Journal of Medical Genetics Part A, 1989, 32, 301-305.	2.4	27
40	Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. American Journal of Medical Genetics, Part A, 2014, 164, 1143-1150.	0.7	27
41	Zoledronic acid in children with osteogenesis imperfecta and Bruck syndrome: a 2-year prospective observational study. Osteoporosis International, 2016, 27, 81-92.	1.3	24
42	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	5.8	24
43	Mild facial dysmorphism and quasidominant inheritance in Cenani-Lenz syndrome. Clinical Dysmorphology, 2003, 12, 77-83.	0.1	22
44	Cytoskeleton and nuclear lamina affection in recessive osteogenesis imperfecta: A functional proteomics perspective. Journal of Proteomics, 2017, 167, 46-59.	1.2	22
45	3-M syndrome: a report of three Egyptian cases with review of the literature. Clinical Dysmorphology, 2006, 15, 55-64.	0.1	21
46	A scoring system for the assessment of clinical severity in osteogenesis imperfecta. Journal of Children's Orthopaedics, 2012, 6, 29-35.	0.4	21
47	Karyotype/phenotype correlation in females with short stature. Clinical Genetics, 1992, 41, 147-151.	1.0	20
48	Cataract, hypertrichosis, and mental retardation (CAHMR): A new autosomal recessive syndrome. American Journal of Medical Genetics Part A, 1991, 41, 432-433.	2.4	19
49	Dyggveâ€“Melchiorâ€“Clausen syndrome: clinical, genetic, and radiological study of 15 Egyptian patients from nine unrelated families. Journal of Children's Orthopaedics, 2009, 3, 451-458.	0.4	19
50	GAPO syndrome: first Egyptian case with ultrastructural changes in the gingiva. Clinical Genetics, 1997, 52, 110-115.	1.0	18
51	Oto-spondylo-megaepiphyseal dysplasia (OSMED): Clinical and radiological findings in sibs homozygous for premature stop codon mutation in theCOL11A2 gene. American Journal of Medical Genetics, Part A, 2006, 140A, 1189-1195.	0.7	17
52	Characteristics of patients with congenital clasped thumb: a prospective study of 40 patients with the results of treatment. Journal of Children's Orthopaedics, 2007, 1, 313-322.	0.4	16
53	Muenke syndrome with pigmentary disorder and probable hemimegalencephaly: An expansion of the phenotype. American Journal of Medical Genetics, Part A, 2011, 155, 207-214.	0.7	15
54	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.	1.8	13

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55	Transposition of external genitalia and associated malformations. Clinical Dysmorphology, 2003, 12, 59-62.	0.1	13
56	The Genetics of Hand Malformations: Updated. Congenital Anomalies (discontinued), 1985, 25, 73-92.	0.3	12
57	A postaxial polydactyly-dental-vertebral syndrome. Journal of Pediatrics, 1977, 90, 230-235.	0.9	11
58	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.	0.7	11
59	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, 2002, 63, 59-63.	1.0	10
60	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, 167, 3054-3061.	0.7	10
61	Anthropometric measurements in Egyptian patients with osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2012, 158A, 2714-2718.	0.7	9
62	The DR syndrome or the Okihiro syndrome?. American Journal of Medical Genetics Part A, 1986, 25, 173-174.	2.4	8
63	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.	0.7	8
64	Genetic Disorders in Egypt. , 2010, , 219-272.		8
65	Greig cephalopolysyndactyly syndrome with dysgenesis of the corpus callosum in a Bedouin family. , 1996, 66, 261-264.		7
66	Megalencephalic leukoencephalopathy with cysts in twelve Egyptian patients: novel mutations in MLC1 and HEPACAM and a founder effect. Metabolic Brain Disease, 2016, 31, 1171-1179.	1.4	7
67	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.	1.1	7
68	Cytogenetic effects of two antimonial antibilharzial drugs: Tartar emetic and bilharcid. Environmental Mutagenesis, 1982, 4, 83-91.	1.4	6
69	Catel???Manzke digitopalatal syndrome or Temtamy preaxial brachydactyly hyperphalangism syndrome?. Clinical Dysmorphology, 2005, 14, 211.	0.1	6
70	How many entities exist for the spectrum of disorders associated with brachydactyly, syndactyly, short stature, microcephaly, and intellectual disability?. , 2011, 155, 880-884.		6
71	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.	0.0	6
72	Genetics and Genomic Medicine in Egypt: steady pace. Molecular Genetics & Genomic Medicine, 2017, 5, 8-14.	0.6	6

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73	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.	2.5	6
74	Heterozygous pathogenic variants inGLI1are a common finding in isolated postaxial polydactyly A/B. Human Mutation, 2020, 41, 265-276.	1.1	6
75	Recurrent homozygous damaging mutation in TMX2, encoding a protein disulfide isomerase, in four families with microlissencephaly. Journal of Medical Genetics, 2020, 57, 274-282.	1.5	6
76	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.	0.1	5
77	Phenotypic and molecular insights into PQBP1â€related intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 2446-2450.	0.7	5
78	Blepharophimosisâ€ptosisâ€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.	0.7	4
79	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. Molecular Genetics & Genomic Medicine, 2021, 9, e1829.	0.6	4
80	First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous <i>CANT1</i> Mutations. Molecular Syndromology, 2021, 12, 279-288.	0.3	3
81	Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. European Journal of Medical Genetics, 2022, 65, 104377.	0.7	3
82	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.	0.0	2
83	On anomalies associated with radial dysplasia. Journal of Pediatrics, 1974, 85, 585.	0.9	1
84	On the nomenclature of a â€œnewâ€•syndrome. Journal of Pediatrics, 1974, 84, 608.	0.9	1
85	A boy with hand anomalies similar to those documented with prenatal misoprostol exposure. Middle East Journal of Medical Genetics, 2013, 2, 58-62.	0.0	1
86	Two different Temtamy syndromes. Clinical Dysmorphology, 2013, 22, 91.	0.1	1
87	OSX/SP7 Mutations and Osteogenesis Imperfecta. , 2014, , 173-179.		1
88	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta. , 2014, , 181-186.		1
89	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.	0.0	1
90	Using Online Mendelian Inheritance in Man in lowâ€and middleâ€income countries. American Journal of Medical Genetics, Part A, 2021, 185, 3284-3286.	0.7	1

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91	Mother and daughter with a novel phenotype of hand and foot anomalies and severe pectus excavatum. American Journal of Medical Genetics, Part A, 2014, 164, 1867-1867.	0.7	0
92	Catel-Manzke digitopalatal syndrome or Temtamy preaxial brachydactyly hyperphalangism syndrome?. Clinical Dysmorphology, 2005, 14, 211.	0.1	0