Sahar Mansour

List of Publications by Year in descending order

58 papers	2,663 citations	279798 23 h-index	197818 49 g-index
65 all docs	65 docs citations	65 times ranked	4814 citing authors

#	Article	IF	Citations
1	Mutations in GATA2 cause primary lymphedema associated with a predisposition to acute myeloid leukemia (Emberger syndrome). Nature Genetics, 2011, 43, 929-931.	21.4	440
2	Novel mutations in PIEZO1 cause an autosomal recessive generalized lymphatic dysplasia with non-immune hydrops fetalis. Nature Communications, 2015, 6, 8085.	12.8	247
3	Dominant mutations in ROR2, encoding an orphan receptor tyrosine kinase, cause brachydactyly type B. Nature Genetics, 2000, 24, 275-278.	21.4	210
4	Deep phenotyping of 89 xeroderma pigmentosum patients reveals unexpected heterogeneity dependent on the precise molecular defect. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E1236-45.	7.1	151
5	Mutation in Vascular Endothelial Growth Factor-C, a Ligand for Vascular Endothelial Growth Factor Receptor-3, Is Associated With Autosomal Dominant Milroy-Like Primary Lymphedema. Circulation Research, 2013, 112, 956-960.	4.5	143
6	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
7	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
8	Rapid identification of mutations in GJC2 in primary lymphoedema using whole exome sequencing combined with linkage analysis with delineation of the phenotype. Journal of Medical Genetics, 2011, 48, 251-255.	3.2	96
9	EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088.	8.2	83
10	Emberger syndromeâ€”Primary lymphedema with myelodysplasia: Report of seven new cases. American Journal of Medical Genetics, Part A, 2010, 152A, 2287-2296.	1.2	70
11	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	2.4	66
12	Update and audit of the St Georgeâ€™s classification algorithm of primary lymphatic anomalies: a clinical and molecular approach to diagnosis. Journal of Medical Genetics, 2020, 57, 653-659.	3.2	59
13	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
14	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	2.8	48
15	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
16	p53 Protein Detected By Immunohistochemical Staining is Not Always Mutant. Disease Markers, 1993, 11, 239-250.	1.3	45
17	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	3.2	44
18	Mutations and alternative splicing of theBRCA1 gene in UK breast/ovarian cancer families. , 1997, 18, 102-110.		43

#	Article	IF	Citations
19	Van Maldergem syndrome: further characterisation and evidence for neuronal migration abnormalities and autosomal recessive inheritance. European Journal of Human Genetics, 2012, 20, 1024-1031.	2.8	39
20	Wiedemannâ€“Rautenstrauch syndrome: A phenotype analysis. American Journal of Medical Genetics, Part A, 2017, 173, 1763-1772.	1.2	36
21	Human phenotypes caused by PIEZO1 mutations; one gene, two overlapping phenotypes?. Journal of Physiology, 2018, 596, 985-992.	2.9	36
22	An RYR1 mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. Science Signaling, 2016, 9, ra68.	3.6	34
23	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
24	Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049.	2.5	29
25	Ovarian cancer screeningâ€”ultrasound; impact on ovarian cancer mortality. British Journal of Radiology, 2018, 91, 20170571.	2.2	25
26	Germline CBL mutation associated with a noonanâ€like syndrome with primary lymphedema and teratoma associated with acquired uniparental isodisomy of chromosome 11q23. American Journal of Medical Genetics, Part A, 2014, 164, 1003-1009.	1.2	24
27	Mutations That Alter the Carboxy-Terminal-Propeptide Cleavage Site of the Chains of Type I Procollagen Are Associated With a Unique Osteogenesis Imperfecta Phenotype. Journal of Bone and Mineral Research, 2018, 33, 1260-1271.	2.8	21
28	CCBE1 mutations can cause a mild, atypical form of generalized lymphatic dysplasia but are not a common cause of nonâ€immune hydrops fetalis. Clinical Genetics, 2012, 81, 191-197.	2.0	20
29	An approach to familial lymphoedema. Clinical Medicine, 2017, 17, 552-557.	1.9	20
30	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98.	3.3	20
31	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
32	Biallelic mutations in NRROS cause an early onset lethal microgliopathy. Acta Neuropathologica, 2020, 139, 947-951.	7.7	17
33	Lymphoscintigraphic Abnormalities Associated with Milroy Disease and Lymphedema-Distichiasis Syndrome. Lymphatic Research and Biology, 2019, 17, 610-619.	1.1	15
34	Clinical utility gene card for: Vici Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	2.8	13
35	Detection of submicroscopic subtelomeric chromosome translocations: A new case study. , 2000, 91, 51-55.		12
36	Congenital vascular malformations: A series of five prenatally diagnosed cases. American Journal of Medical Genetics, Part A, 2008, 146A, 2673-2680.	1.2	10

#	Article	IF	Citations
37	A Novel Splice-Site Mutation in VEGFC Is Associated with Congenital Primary Lymphoedema of Gordon. International Journal of Molecular Sciences, 2018, 19, 2259.	4.1	10
38	The St George's Classification Algorithm of Primary Lymphatic Anomalies. Lymphatic Research and Biology, 2021, 19, 25-30.	1.1	10
39	Detection and diagnosis of breast lesions: Performance evaluation of digital breast tomosynthesis and magnetic resonance mammography. Egyptian Journal of Radiology and Nuclear Medicine, 2016, 47, 1159-1172.	0.6	9
40	Fetal hydrops â€“ a review and a clinical approach to identifying the cause. Expert Opinion on Orphan Drugs, 2020, 8, 51-66.	0.8	9
41	SHOX haploinsufficiency presenting with isolated short long bones in the second and third trimester. European Journal of Human Genetics, 2018, 26, 350-358.	2.8	8
42	Redefining WILD syndrome: a primary lymphatic dysplasia with congenital multisegmental lymphoedema, cutaneous lymphovascular malformation, CD4 lymphopaenia and warts. Journal of Medical Genetics, 2023, 60, 84-90.	3.2	8
43	Diffusion-weighted magnetic resonance imaging in the assessment of ovarian masses with suspicious features: Strengths and challenges. Egyptian Journal of Radiology and Nuclear Medicine, 2015, 46, 1279-1289.	0.6	7
44	19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference. European Journal of Human Genetics, 2018, 26, 85-93.	2.8	7
45	Mutations in EPHB4 cause human venous valve aplasia. JCI Insight, 2021, 6, .	5.0	7
46	Autopsy findings in EPG5â€related Vici syndrome with antenatal onset. American Journal of Medical Genetics, Part A, 2017, 173, 2522-2527.	1.2	6
47	Janus-faced EPHB4-associated disorders: novel pathogenic variants and unreported intrafamilial overlapping phenotypes. Genetics in Medicine, 2021, 23, 1315-1324.	2.4	6
48	Ataxia-Pancytopenia Syndrome due to a de Novo SAMD9L Mutation. Neurology: Genetics, 2021, 7, e580.	1.9	5
49	Can artificial intelligence replace ultrasound as a complementary tool to mammogram for the diagnosis of the breast cancer?. British Journal of Radiology, 2021, 94, 20210820.	2.2	5
50	Natural history of NF1 c.2970_2972del p.(Met992del): confirmation of a low risk of complications in a longitudinal study. European Journal of Human Genetics, 2022, 30, 291-297.	2.8	5
51	Short amplicon reverse transcriptionâ€polymerase chain reaction detects aberrant splicing in genes with low expression in blood missed by ribonucleic acid sequencing analysis for clinical diagnosis. Human Mutation, 2022, 43, 963-970.	2.5	4
52	The role of automated breast ultrasound in the assessment of the local extent of breast cancer. Breast Journal, 2021, 27, 113-119.	1.0	3
53	Use of somatostatin analogues to treat chylothorax in a child with Generalised Lymphatic Dysplasia. Respiratory Medicine Case Reports, 2012, 5, 76-77.	0.4	1
54	Overlapping cortical malformations in patients with pathogenic variants inGRIN1andGRIN2B. Journal of Medical Genetics, 2023, 60, 183-192.	3.2	1

#	Article	IF	Citations
55	Diffusion-weighted MR imaging and assessment of ovarian carcinomas with vaginal deposit accidentally detected during pregnancy. BJR|case Reports, 2017, 3, 20150411.	0.2	0
56	O01â€ƒHypophosphatasia in adults at a specialist centre in the UK: the spectrum of musculoskeletal disease. Rheumatology, 2020, 59, .	1.9	0
57	Early prenatal presentation of the cartilage-hair hypoplasia / anauxetic dysplasia spectrum of disorders mimicking recurrent thanatophoric dysplasia. European Journal of Medical Genetics, 2021, 64, 104162.	1.3	0
58	SHORT Syndrome: Systematic Appraisal of the Medical and Dental Phenotype. Cleft Palate-Craniofacial Journal, 2021, , 105566562110268.	0.9	0