# 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 | Overlapping cortical malformations in patients with pathogenic variants inGRIN1andGRIN2B. Journal of Medical Genetics, 2023, 60, 183-192. | 3.2 | 1 |
| 2 | 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 |
| 3 | 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 |
| 4 | 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 |
| 5 | The St George's Classification Algorithm of Primary Lymphatic Anomalies. Lymphatic Research and Biology, 2021, 19, 25-30. | 1.1 | 10 |
| 6 | Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356. | 6.2 | 30 |
| 7 | 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 |
| 8 | Ataxia-Pancytopenia Syndrome due to a de Novo SAMD9L Mutation. Neurology: Genetics, 2021, 7, e580. | 1.9 | 5 |
| 9 | Janus-faced EPHB4-associated disorders: novel pathogenic variants and unreported intrafamilial overlapping phenotypes. Genetics in Medicine, 2021, 23, 1315-1324. | 2.4 | 6 |
| 10 | SHORT Syndrome: Systematic Appraisal of the Medical and Dental Phenotype. Cleft Palate-Craniofacial Journal, 2021, , 105566562110268. | 0.9 | 0 |
| 11 | Mutations in EPHB4 cause human venous valve aplasia. JCI Insight, 2021, 6, . | 5.0 | 7 |
| 12 | 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 |
| 13 | 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 |
| 14 | Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464. | 2.5 | 17 |
| 15 | O01â€ƒHypophosphatasia in adults at a specialist centre in the UK: the spectrum of musculoskeletal disease. Rheumatology, 2020, 59, . | 1.9 | 0 |
| 16 | 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 |
| 17 | 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 |
| 18 | Biallelic mutations in NRROS cause an early onset lethal microgliopathy. Acta Neuropathologica, 2020, 139, 947-951. | 7.7 | 17 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98. | 3.3 | 20 |
| 20 | Lymphoscintigraphic Abnormalities Associated with Milroy Disease and Lymphedema-Distichiasis Syndrome. Lymphatic Research and Biology, 2019, 17, 610-619. | 1.1 | 15 |
| 21 | 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 |
| 22 | Human phenotypes caused by PIEZO1 mutations; one gene, two overlapping phenotypes?. Journal of Physiology, 2018, 596, 985-992. | 2.9 | 36 |
| 23 | Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245. | 2.4 | 66 |
| 24 | 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 |
| 25 | 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 |
| 26 | Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846. | 3.2 | 44 |
| 27 | 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 |
| 28 | Ovarian cancer screeningâ€"ultrasound; impact on ovarian cancer mortality. British Journal of Radiology, 2018, 91, 20170571. | 2.2 | 25 |
| 29 | Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908. | 2.4 | 46 |
| 30 | Wiedemannâ€"Rautenstrauch syndrome: A phenotype analysis. American Journal of Medical Genetics, Part A, 2017, 173, 1763-1772. | 1.2 | 36 |
| 31 | Autopsy findings in EPG5â€related Vici syndrome with antenatal onset. American Journal of Medical Genetics, Part A, 2017, 173, 2522-2527. | 1.2 | 6 |
| 32 | An approach to familial lymphoedema. Clinical Medicine, 2017, 17, 552-557. | 1.9 | 20 |
| 33 | 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 |
| 34 | 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 |
| 35 | Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049. | 2.5 | 29 |
| 36 | An RYR1 mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. Science Signaling, 2016, 9, ra68. | 3.6 | 34 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781. | 7.6 | 99 |
| 39 | EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088. | 8.2 | 83 |
| 40 | 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 |
| 41 | Novel mutations in PIEZO1 cause an autosomal recessive generalized lymphatic dysplasia with non-immune hydrops fetalis. Nature Communications, 2015, 6, 8085. | 12.8 | 247 |
| 42 | Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170. | 2.8 | 56 |
| 43 | 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 |
| 44 | 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 |
| 45 | Clinical utility gene card for: Vici Syndrome. European Journal of Human Genetics, 2014, 22, 435-435. | 2.8 | 13 |
| 46 | 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 |
| 47 | 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 |
| 48 | 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 |
| 49 | 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 |
| 50 | 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 |
| 51 | 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 |
| 52 | 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 |
| 53 | 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 |
| 54 | 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 |
|---|---|---|---|
| 55 | Detection of submicroscopic subtelomeric chromosome translocations: A new case study. , 2000, 91, 51-55. | | 12 |
| 56 | Dominant mutations in ROR2, encoding an orphan receptor tyrosine kinase, cause brachydactyly type B. Nature Genetics, 2000, 24, 275-278. | 21.4 | 210 |
| 57 | Mutations and alternative splicing of theBRCA1 gene in UK breast/ovarian cancer families. , 1997, 18, 102-110. | | 43 |
| 58 | p53 Protein Detected By Immunohistochemical Staining is Not Always Mutant. Disease Markers, 1993, 11, 239-250. | 1.3 | 45 |