Sarah E Sheppard

List of Publications by Year
in descending order

27 papers	340 citations	1163117 8 h-index	888059 17 g-index
29 all docs	29 docs citations	29 times ranked	548 citing authors

#	Article	IF	Citations
1	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0
2	50 Years Ago in T J P. Journal of Pediatrics, 2022, 241, 195.	1.8	0
3	Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Science Translational Medicine, 2022, 14, eabm4869.	12.4	14
4	Genetics etiologies and genotype phenotype correlations in a cohort of individuals with central conducting lymphatic anomaly. European Journal of Human Genetics, 2022, 30, 1022-1028.	2.8	9
5	Clinical Effectiveness of Telemedicine-Based Pediatric Genetics Care. Pediatrics, 2022, 150, .	2.1	5
6	Heterozygous recurrent <scp>HNF4A</scp> variant p.<scp>Arg85Trp</scp> causes Fanconi renotubular syndrome 4 with maturity onset diabetes of the young, an autosomal dominant phenocopy of Fanconi Bickel syndrome with colobomas. American Journal of Medical Genetics, Part A, 2021, 185, 566-570.	1.2	7
7	Expanded phenotypic spectrum of <scp>JAG1</scp>â€associated diseases: Central conducting lymphatic anomaly with a pathogenic variant in <scp>JAG1</scp>. Clinical Genetics, 2021, 99, 742-743.	2.0	7
8	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
9	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. American Journal of Medical Genetics, Part A, 2021, 185, 1486-1493.	1.2	3
10	Trisomy 9 mosaic syndrome: Sixteen additional patients with new and/or less commonly reported features, literature review, and suggested clinical guidelines. American Journal of Medical Genetics, Part A, 2021, 185, 2374-2383.	1.2	11
11	50 Years Ago in T J P. Journal of Pediatrics, 2021, 233, 211.	1.8	0
12	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
13	Misdiagnosis of capillary malformations in darker skin phototypes. Pediatric Dermatology, 2021, 38 Suppl 2, 137-139.	0.9	1
14	Hyperinsulinism in an individual with an EP300 variant of Rubinsteinâ€Taybi syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1251-1255.	1.2	2
15	Genetic skin disorders: The value of a multidisciplinary clinic. American Journal of Medical Genetics, Part A, 2021, 185, 1159-1167.	1.2	3
16	Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. Clinical Dysmorphology, 2021, 30, 89-92.	0.3	2
17	Proposed criteria for nevoid basal cell carcinoma syndrome in children assessed using statistical optimization. Scientific Reports, 2021, 11, 19791.	3.3	5
18	Cerebrofacial vascular metameric syndrome is caused by somatic pathogenic variants in PIK3CA. Journal of Physical Education and Sports Management, 2021, 7, a006147.	1.2	6

#	Article	IF	Citations
19	Further delineation of the phenotypic spectrum of nevus comedonicus syndrome to include congenital pulmonary airway malformation of the lung and aneurysm. American Journal of Medical Genetics, Part A, 2020, 182, 746-754.	1.2	9
20	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	6.2	18
21	Kaposiform lymphangiomatosis effectively treated with <scp>MEK</scp> inhibition. EMBO Molecular Medicine, 2020, 12, e12324.	6.9	51
22	Generalized, severe epidermolysis bullosa simplex caused by a Keratin 5 p.E477K mutation. Pediatric Dermatology, 2019, 36, 1007-1009.	0.9	1
23	Muenke syndrome: Medical and surgical comorbidities and longâ€term management. American Journal of Medical Genetics, Part A, 2019, 179, 1442-1450.	1.2	1
24	Androgenetic chimerism as an etiology for Beckwithâ€“Wiedemann syndrome: diagnosis and management. Genetics in Medicine, 2019, 21, 2644-2649.	2.4	15
25	Genotypeâ€“phenotype specificity in Menkeâ€“Hennekam syndrome caused by missense variants in exon 30 or 31 of CREBBP. American Journal of Medical Genetics, Part A, 2019, 179, 1058-1062.	1.2	23
26	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0
27	What is new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. American Journal of Medical Genetics, Part A, 2018, 176, 2058-2069.	1.2	106