Lois J Starr

List of Publications by Year
in descending order

27 papers	325 citations	1040056 9 h-index	888059 17 g-index
28 all docs	28 docs citations	28 times ranked	758 citing authors

#	Article	IF	Citations
1	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
2	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
3	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
4	Myhre syndrome: Clinical features and restrictive cardiopulmonary complications. American Journal of Medical Genetics, Part A, 2015, 167, 2893-2901.	1.2	31
5	Newly described recessive MYH11 disorder with clinical overlap of Multisystemic smooth muscle dysfunction and Megacystis microcolon hypoperistalsis syndromes. American Journal of Medical Genetics, Part A, 2018, 176, 1011-1014.	1.2	23
6	Clinical and Echocardiographic Prevalence and Detection of Congenital and Acquired Cardiac Abnormalities in Girls and Women with the Turner Syndrome. American Journal of Cardiology, 2018, 122, 327-330.	1.6	15
7	Patient with anomalous skin pigmentation expands the phenotype of ARID2 lossâ€ofâ€function disorder, a SWI/SNFâ€related intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 808-812.	1.2	13
8	Utilization of echocardiography in Ehlersâ€Danlos syndrome. Congenital Heart Disease, 2019, 14, 864-867.	0.2	12
9	Gainâ€ofâ€function pathogenic variants in SMAD4 are associated with neoplasia in Myhre syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 328-337.	1.2	12
10	PIGQ glycosylphosphatidylinositolâ€anchored protein deficiency: Characterizing the phenotype. American Journal of Medical Genetics, Part A, 2019, 179, 1270-1275.	1.2	11
11	Autism spectrum disorder and genetic testing: Parental perceptions and decisionâ€making. Journal for Specialists in Pediatric Nursing, 2018, 23, e12211.	1.1	9
12	Occurrence of nephroblastomatosis with dup(18)(q11.2â€q23) implicates trisomy 18 tumor screening protocol in select patients with 18q duplication. American Journal of Medical Genetics, Part A, 2014, 164, 1079-1082.	1.2	7
13	Assessing the utility of confirmatory studies following identification of large-scale genomic imbalances by microarray. Genetics in Medicine, 2015, 17, 875-879.	2.4	7
14	Vascular dissection in women with Turner syndrome. International Journal of Cardiology, 2021, 325, 127-131.	1.7	7
15	The QT Interval in Patients With the Turner Syndrome. American Journal of Cardiology, 2021, 140, 118-121.	1.6	6
16	Eliciting Narratives to Inform Care for Infants With Trisomy 18. Pediatrics, 2018, 142, .	2.1	5
17	SEARCHING FOR ELECTRICAL PROPERTIES, PHENOMENA AND MECHANISMS IN THE CONSTRUCTION AND FUNCTION OF CHROMOSOMES. Computational and Structural Biotechnology Journal, 2013, 6, e201303007.	4.1	4
18	Valve-Sparing Root and Total Arch Replacement for Cutis Laxa Aortopathy. World Journal for Pediatric & Congenital Heart Surgery, 2019, 10, 376-379.	0.8	4

#	Article	IF	Citations
19	<scp>CHEDDA</scp> syndrome is an underrecognized neurodevelopmental disorder with a highly restricted <scp>ATN1</scp> mutation spectrum. Clinical Genetics, 2021, 100, 468-477.	2.0	4
20	Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. American Journal of Medical Genetics, Part A, 2015, 167, 2168-2175.	1.2	3
21	Duplication of 20qter and deletion of 20pter due to paternal pericentric inversion: Patient report and review of 20qter duplications. American Journal of Medical Genetics, Part A, 2014, 164, 2020-2024.	1.2	2
22	The first patient with tandem duplication of 6q14q16: Molecular and phenotypic characterization. American Journal of Medical Genetics, Part A, 2016, 170, 2416-2420.	1.2	2
23	Second reported individual with a partial STAG2 deletion: middle interhemispheric variant holoprosencephaly in STAG2-related cohesinopathy. Clinical Dysmorphology, 2021, 30, 159-163.	0.3	2
24	Hb Gibbon [Î²124(H2)Proâ†'Thr (HBB: c.373C>A, p.P125T)], an Asymptomatic Novel Hemoglobin Variant Detected by Newborn Screening. Hemoglobin, 2019, 43, 207-209.	0.8	1
25	Kawasaki Disease in a Patient With Williams Syndrome. World Journal for Pediatric & Congenital Heart Surgery, 2020, 11, NP144-NP147.	0.8	0
26	First report of de novo 12q14.2-q23.3 duplication: patient with multiple congenital anomalies, neurodevelopmental delay, and a connective tissue disorder-like phenotype including cutis laxa. Clinical Dysmorphology, 2020, 29, 132-136.	0.3	0
27	Lack of resemblance between Myhre syndrome and other â€œsegmental progeroidâ€•syndromes warrants restraint in applying this classification. GeroScience, 2021, 43, 459-461.	4.6	0