

Lois J Starr

List of Publications by Year in descending order

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Version: 2024-02-01

27
papers

325
citations

1040056

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h-index

888059

17
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28
all docs

28
docs citations

28
times ranked

758
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|-----|-----------|
| 1 | The QT Interval in Patients With the Turner Syndrome. <i>American Journal of Cardiology</i> , 2021, 140, 118-121. | 1.6 | 6 |
| 2 | Vascular dissection in women with Turner syndrome. <i>International Journal of Cardiology</i> , 2021, 325, 127-131. | 1.7 | 7 |
| 3 | Lack of resemblance between Myhre syndrome and other "segmental progeroid" syndromes warrants restraint in applying this classification. <i>GeroScience</i> , 2021, 43, 459-461. | 4.6 | 0 |
| 4 | Second reported individual with a partial STAG2 deletion: middle interhemispheric variant holoprosencephaly in STAG2-related cohesinopathy. <i>Clinical Dysmorphology</i> , 2021, 30, 159-163. | 0.3 | 2 |
| 5 | Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1649-1665. | 1.2 | 34 |
| 6 | CHEDDA syndrome is an underrecognized neurodevelopmental disorder with a highly restricted ATN1 mutation spectrum. <i>Clinical Genetics</i> , 2021, 100, 468-477. | 2.0 | 4 |
| 7 | Kawasaki Disease in a Patient With Williams Syndrome. <i>World Journal for Pediatric & Congenital Heart Surgery</i> , 2020, 11, NP144-NP147. | 0.8 | 0 |
| 8 | Gain-of-function pathogenic variants in SMAD4 are associated with neoplasia in Myhre syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 328-337. | 1.2 | 12 |
| 9 | 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. <i>Clinical Dysmorphology</i> , 2020, 29, 132-136. | 0.3 | 0 |
| 10 | Hb Gibbon [$\beta^{124}(H2)Pro\rightarrow Thr$ (<i>HBB</i> : c.373C>A, p.P125T)], an Asymptomatic Novel Hemoglobin Variant Detected by Newborn Screening. <i>Hemoglobin</i> , 2019, 43, 207-209. | 0.8 | 1 |
| 11 | Utilization of echocardiography in Ehlers-Danlos syndrome. <i>Congenital Heart Disease</i> , 2019, 14, 864-867. | 0.2 | 12 |
| 12 | PIGQ glycosylphosphatidylinositol-anchored protein deficiency: Characterizing the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1270-1275. | 1.2 | 11 |
| 13 | Patient with anomalous skin pigmentation expands the phenotype of ARID2 loss-of-function disorder, a SWI/SNF-related intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 808-812. | 1.2 | 13 |
| 14 | Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotype-phenotype correlation. <i>Genetics in Medicine</i> , 2019, 21, 867-876. | 2.4 | 62 |
| 15 | Valve-Sparing Root and Total Arch Replacement for Cutis Laxa Aortopathy. <i>World Journal for Pediatric & Congenital Heart Surgery</i> , 2019, 10, 376-379. | 0.8 | 4 |
| 16 | Autism spectrum disorder and genetic testing: Parental perceptions and decision-making. <i>Journal for Specialists in Pediatric Nursing</i> , 2018, 23, e12211. | 1.1 | 9 |
| 17 | Newly described recessive MYH11 disorder with clinical overlap of Multisystemic smooth muscle dysfunction and Megacystis microcolon hypoperistalsis syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1011-1014. | 1.2 | 23 |
| 18 | Eliciting Narratives to Inform Care for Infants With Trisomy 18. <i>Pediatrics</i> , 2018, 142, . | 2.1 | 5 |

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|----|---|-----|-----------|
| 19 | Clinical and Echocardiographic Prevalence and Detection of Congenital and Acquired Cardiac Abnormalities in Girls and Women with the Turner Syndrome. <i>American Journal of Cardiology</i> , 2018, 122, 327-330. | 1.6 | 15 |
| 20 | The first patient with tandem duplication of 6q14q16: Molecular and phenotypic characterization. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2416-2420. | 1.2 | 2 |
| 21 | Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2168-2175. | 1.2 | 3 |
| 22 | Myhre syndrome: Clinical features and restrictive cardiopulmonary complications. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2893-2901. | 1.2 | 31 |
| 23 | Assessing the utility of confirmatory studies following identification of large-scale genomic imbalances by microarray. <i>Genetics in Medicine</i> , 2015, 17, 875-879. | 2.4 | 7 |
| 24 | The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 224-228. | 2.8 | 48 |
| 25 | Duplication of 20qter and deletion of 20pter due to paternal pericentric inversion: Patient report and review of 20qter duplications. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2020-2024. | 1.2 | 2 |
| 26 | Occurrence of nephroblastomatosis with dup(18)(q11.2â€“q23) implicates trisomy 18 tumor screening protocol in select patients with 18q duplication. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1079-1082. | 1.2 | 7 |
| 27 | SEARCHING FOR ELECTRICAL PROPERTIES, PHENOMENA AND MECHANISMS IN THE CONSTRUCTION AND FUNCTION OF CHROMOSOMES. <i>Computational and Structural Biotechnology Journal</i> , 2013, 6, e201303007. | 4.1 | 4 |