Laurie Demmer

List of Publications by Year
in descending order

10 papers	299 citations	1684188 5 h-index	1474206 9 g-index
10 all docs	10 docs citations	10 times ranked	774 citing authors

#	Article	IF	Citations
1	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	3.2	4
2	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	2.4	4
3	Systematic evidence-based review: outcomes from exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability. Genetics in Medicine, 2020, 22, 986-1004.	2.4	53
4	Spectrum of K_V2.1 Dysfunction in KCNB1â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2019, 86, 899-912.	5.3	52
5	An unusual case of nephrotic syndrome in a microcephalic infant: Questions. Pediatric Nephrology, 2019, 34, 2325-2326.	1.7	0
6	An unusual case of nephrotic syndrome in a microcephalic infant: Answers. Pediatric Nephrology, 2019, 34, 2327-2329.	1.7	1
7	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
8	Development, Implementation, and Assessment of a Genetics Curriculum Across Institutions. AJP Reports, 2016, 06, e372-e377.	0.7	3
9	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
10	Autosomal recessive MFN2â€related Charcotâ€Marieâ€Tooth disease with diaphragmatic weakness: Case report and literature review. American Journal of Medical Genetics, Part A, 2016, 170, 1580-1584.	1.2	17