Amanda Barone Pritchard

List of Publications by Year
in descending order

7 papers	64 citations	1937685 4 h-index	1872680 6 g-index
7 all docs	7 docs citations	7 times ranked	132 citing authors

#	Article	IF	Citations
1	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
2	Persistent dyslipidemia in treatment of lysosomal acid lipase deficiency. Orphanet Journal of Rare Diseases, 2020, 15, 58.	2.7	8
3	Lossâ€ofâ€function of Endothelin receptor type A results in Oroâ€Otoâ€Cardiac syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1104-1116.	1.2	7
4	Interstitial 4q Deletion Syndrome Including <i>NR3C2</i> Causing Pseudohypoaldosteronism. Molecular Syndromology, 2019, 10, 327-331.	0.8	6
5	Trainee perspectives of COVID-19 impact on medical genetics education. Genetics in Medicine, 2021, 23, 956-962.	2.4	3
6	Inborn error of metabolism patients after liver transplantation: Outcomes of 35 patients over 27â€‰years in one pediatric quaternary hospital. American Journal of Medical Genetics, Part A, 2022, 188, 1443-1447.	1.2	2
7	What not to expect when you're expecting: Unusual cases of placental mosaicism detected on non-invasive prenatal screening. European Journal of Medical Genetics, 2020, 63, 103895.	1.3	0