Paul R Mark

List of Publications by Year
in descending order

10 papers	297 citations	1684188 5 h-index	1720034 7 g-index
10 all docs	10 docs citations	10 times ranked	870 citing authors

#	Article	IF	Citations
1	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. New England Journal of Medicine, 2017, 377, 544-552.	27.0	177
2	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
3	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38
4	Narrowing the Critical Region for Congenital Vertical Talus in Patients With Interstitial 18q Deletions. American Journal of Medical Genetics, Part A, 2013, 161, 1117-1121.	1.2	10
5	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.	2.4	6
6	<scp>NAD</scp>+ deficiency in human congenital malformations and miscarriage: A new model of pleiotropy. American Journal of Medical Genetics, Part A, 2022, 188, 2834-2849.	1.2	5
7	Further characterization of <scp>Borjesonâ€Forssmanâ€Lehmann</scp> syndrome in females due to de novo variants in <scp>PHF6</scp>. Clinical Genetics, 2022, 102, 182-190.	2.0	5
8	Comment on critical region for talipes equinovarus in patients with 5q23 deletions. European Journal of Medical Genetics, 2015, 58, 243.	1.3	0
9	Lethal renal anomalies in a fetus with 21q22.11â€q22.12 deletion. American Journal of Medical Genetics, Part A, 2020, 182, 3060-3063.	1.2	0
10	Two cases of different genetic variants of alveolar capillary dysplasia associated with left-sided obstructive CHDs. Cardiology in the Young, 2021, 31, 1368-1370.	0.8	0