Amarilis Sanchez-Valle

List of Publications by Year
in descending order

9 papers	262 citations	1307594 7 h-index	1474206 9 g-index
10 all docs	10 docs citations	10 times ranked	572 citing authors

#	Article	IF	Citations
1	Biliary Atresia. Advances in Pediatrics, 2017, 64, 285-305.	1.4	59
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	Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. Genetics in Medicine, 2019, 21, 1851-1867.	2.4	56
4	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
5	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
6	Expanding phenotype with severe midline brain anomalies and missense variant supports a causal role for FOXA2 in 20p11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1783-1790.	1.2	10
7	Long-term preservation of intellectual functioning in sapropterin-treated infants and young children with phenylketonuria: A seven-year analysis. Molecular Genetics and Metabolism, 2021, 132, 119-127.	1.1	8
8	CDK19-related disorder results from both loss-of-function and gain-of-function de novo missense variants. Genetics in Medicine, 2021, 23, 1050-1057.	2.4	7
9	PIGA Mutations Can Mimic Neonatal Hemochromatosis. Pediatrics, 2021, 147, .	2.1	4