Kyra E Stuurman

List of Publications by Year
in descending order

10 papers	229 citations	1478505 6 h-index	1372567 10 g-index
10 all docs	10 docs citations	10 times ranked	405 citing authors

#	Article	IF	Citations
1	Clinical and Functional Consequences of C-Terminal Variants in MCT8: A Case Series. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 539-553.	3.6	4
2	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
3	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	2.4	5
4	Isolated Increased Nuchal Translucency in First Trimester Ultrasound Scan: Diagnostic Yield of Prenatal Microarray and Outcome of Pregnancy. Frontiers in Medicine, 2021, 8, 737936.	2.6	8
5	Inflammatory bowel disease in Shwachman-Diamond syndrome; is there an association?. Clinics and Research in Hepatology and Gastroenterology, 2020, 44, e10-e13.	1.5	3
6	TGDS pathogenic variants cause Catelâ€Manzke syndrome without hyperphalangy. American Journal of Medical Genetics, Part A, 2020, 182, 431-436.	1.2	5
7	De novo variants in SUPT16H cause neurodevelopmental disorders associated with corpus callosum abnormalities. Journal of Medical Genetics, 2020, 57, 461-465.	3.2	7
8	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
9	Prenatal ultrasound findings of rasopathies in a cohort of 424 fetuses: update on genetic testing in the NGS era. Journal of Medical Genetics, 2019, 56, 654-661.	3.2	38
10	Prenatal diagnostic testing of the Noonan syndrome genes in fetuses with abnormal ultrasound findings. European Journal of Human Genetics, 2013, 21, 936-942.	2.8	94