Arjan Bouman

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8871454/publications.pdf

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12	202	1163117	1199594
papers	citations	8 h-index	12 g-index

12	12	12	403
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Three patients with defects in interferon gamma receptor signaling: A challenging diagnosis. Pediatric Allergy and Immunology, 2022, 33, e13768.	2.6	2
2	Whole exome sequencing of known eye genes reveals genetic causes for high myopia. Human Molecular Genetics, 2022, 31, 3290-3298.	2.9	16
3	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
4	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
5	Blepharophimosisâ€“ptosisâ€“epicanthus inversus syndrome caused by a 54-kb microdeletion in a FOXL2 cis-regulatory element. Clinical Dysmorphology, 2018, 27, 58-62.	0.3	8
6	Homozygous DMRT2 variant associates with severe rib malformations in a newborn. American Journal of Medical Genetics, Part A, 2018, 176, 1216-1221.	1.2	16
7	Bloom syndrome does not always present with sun-sensitive facial erythema. European Journal of Medical Genetics, 2018, 61, 94-97.	1.3	16
8	Widespread domain-like perturbations of DNA methylation in whole blood of Down syndrome neonates. PLoS ONE, 2018, 13, e0194938.	2.5	24
9	Oralâ€facialâ€digital syndrome type 1 in males: Congenital heart defects are included in its phenotypic spectrum. American Journal of Medical Genetics, Part A, 2017, 173, 1383-1389.	1.2	16
10	Trisomy 4 mosaicism: Delineation of the phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 1040-1045.	1.2	3
11	Congenital thrombocytopenia in a neonate with an interstitial microdeletion of 3q26.2q26.31. American Journal of Medical Genetics, Part A, 2016, 170, 504-509.	1.2	19
12	ADAMTSL4â€associated isolated ectopia lentis: Further patients, novel mutations and a detailed phenotype description. American Journal of Medical Genetics, Part A, 2015, 167, 2376-2381.	1.2	22