Martijn Breuning

List of Publications by Year
in descending order

11	601	1040056 9	1281871 11
papers	citations	h-index	g-index

11	11	11	1052
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hypermorphic and hypomorphic AARS alleles in patients with CMT2N expand clinical and molecular heterogeneities. Human Molecular Genetics, 2018, 27, 4036-4050.	2.9	22
2	Analysis of mutations within the intron20 splice donor site of CREBBP in patients with and without classical RSTS. European Journal of Human Genetics, 2016, 24, 1639-1643.	2.8	4
3	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. European Journal of Medical Genetics, 2014, 57, 163-168.	1.3	11
4	An unanticipated copy number variant ofÂchromosome 15 disrupting <scp>SMAD3</scp> reveals a threeâ€generation family at serious risk for aortic dissection. Clinical Genetics, 2013, 83, 337-344.	2.0	21
5	Hereditary leiomyomatosis and renal cell cancer in families referred for fumarate hydratase germline mutation analysis. Clinical Genetics, 2011, 79, 49-59.	2.0	154
6	Germline mutations in APC and MUTYH are responsible for the majority of families with attenuated familial adenomatous polyposis. Clinical Genetics, 2007, 71, 427-433.	2.0	134
7	Duplications in theDMD gene. Human Mutation, 2006, 27, 938-945.	2.5	145
8	Bifurcation of the femur with tibial agenesis and additional anomalies. American Journal of Medical Genetics, Part A, 2005, 138A, 45-50.	1.2	15
9	Bannayan-Riley-Ruvalcaba syndrome: further delineation of the phenotype and management of PTEN mutation-positive cases. Familial Cancer, 2003, 2, 79-85.	1.9	69
10	Using a roster and haplotyping is useful in risk assessment for persons with intermediate and reduced penetrance alleles in Huntington disease. American Journal of Medical Genetics Part A, 2001, 105, 737-744.	2.4	17
11	A Rett syndrome patient with a ring X chromosome: further evidence for skewing of X inactivation and heterogeneity in the aetiology of the disease. European Journal of Human Genetics, 2001, 9, 171-177.	2.8	9