Kamilla Schlade-Bartusiak

List of Publications by Year
in descending order

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		933447	839539
20	413	10	18
papers	citations	h-index	g-index

20	20	20	863
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	7.6	115
2	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
3	FISH-mapping of telomeric 14q32 deletions: Search for the cause of seizures. American Journal of Medical Genetics, Part A, 2005, 138A, 218-224.	1.2	29
4	Uniparental disomy: can SNP array data be used for diagnosis?. Genetics in Medicine, 2012, 14, 753-756.	2.4	27
5	Polymorphism in nucleotide excision repair gene XPC correlates with bleomycinâ€induced chromosomal aberrations. Environmental and Molecular Mutagenesis, 2007, 48, 666-671.	2.2	21
6	A child with terminal 14q deletion syndrome: Consideration of genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2009, 149A, 1012-1018.	1.2	19
7	A coâ€occurrence of osteogenesis imperfecta type VI and cystinosis. American Journal of Medical Genetics, Part A, 2012, 158A, 1422-1426.	1.2	19
8	Brain MRI abnormalities and spectrum of neurological and clinical findings in three patients with proximal 16p11.2 microduplication. American Journal of Medical Genetics, Part A, 2014, 164, 2003-2012.	1.2	19
9	The influence of GSTM1 and GSTT1 genotypes on the induction of sister chromatid exchanges and chromosome aberrations by 1,2:3,4-diepoxybutane. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2000, 465, 69-75.	1.7	18
10	A child with deletion (14)(q24.3q32.13) and auditory neuropathy. American Journal of Medical Genetics, Part A, 2008, 146A, 117-123.	1.2	13
11	Alternating hypoglycemia and hyperglycemia in a toddler with a homozygous p.R1419H ABCC8 mutation: an unusual clinical picture. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 345-51.	0.9	10
12	A minigene approach for analysis of ATP7B splice variants in patients with Wilson diseaseâ˜†. Biochimie, 2009, 91, 1342-1345.	2.6	9
13	BPES with atypical premature ovarian insufficiency, and evidence of mitotic recombination, in a woman with trisomy X and a translocation t(3;11)(q22.3;q14.1). American Journal of Medical Genetics, Part A, 2012, 158A, 2322-2327.	1.2	8
14	Mosaic embryo transferâ€”first report of a live born with nonmosaic partial aneuploidy and uniparental disomy 15. F&S Reports, 2022, 3, 192-197.	0.7	8
15	Independent post-zygotic breaks of a dicentric chromosome result in mosaicism for an inverted duplication deletion 9p and terminal deletion 9p. European Journal of Medical Genetics, 2013, 56, 229-235.	1.3	7
16	An approach to rapid characterization of DMD copy number variants for prenatal risk assessment. American Journal of Medical Genetics, Part A, 2021, 185, 2541-2545.	1.2	3
17	Clinical and molecular characterization of an almost complete ring chromosome 4 in two sisters, with recurrence due to gonadal mosaicism. Clinical Dysmorphology, 2021, 30, 173-176.	0.3	2
18	Incidental finding of paternal UPD15 in a child with a deletion of 11q21â€“q22.3, presenting with developmental delay, coloboma and characteristic dysmorphic features. Clinical Dysmorphology, 2016, 25, 77-81.	0.3	1

#	Article	IF	Citations
19	MG-117â€...Chromosome microarray and non-coding DNA copy number variants â€“ a case of alveolar capillary dysplasia at FOXF1 locus. Journal of Medical Genetics, 2015, 52, A4.2-A4.	3.2	0
20	MG-127â€...Diagnostic accuracy of chromosome microarray in children with epilepsy and neurological abnormalities of unknown aetiology. Journal of Medical Genetics, 2015, 52, A7.1-A7.	3.2	0