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List of Publications by Year in descending order

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papers

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docs citations

14
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533
citing authors

#	ARTICLE	IF	CITATIONS
1	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. American Journal of Human Genetics, 2020, 106, 872-884.	6.2	85
2	X-chromosome terminal deletion in a female with premature ovarian failure: Haploinsufficiency of X-linked genes as a possible explanation. Molecular Cytogenetics, 2010, 3, 14.	0.9	24
3	Drug transporters play a key role in the complex process of Imatinib resistance in vitro. Leukemia Research, 2015, 39, 355-360.	0.8	18
4	Copy number variants prioritization after array-CGH analysis â€“ a cohort of 1000 patients. Molecular Cytogenetics, 2015, 8, 103.	0.9	17
5	Insertional translocation leading to a 4q13 duplication including the <i>EPHA5</i> gene in two siblings with attentionâ€deficit hyperactivity disorder. American Journal of Medical Genetics, Part A, 2013, 161, 1923-1928.	1.2	14
6	Three Unusual but Cytogenetically Similar Cases With up to Five Different Cell Lines Involving Structural and Numerical Abnormalities of Chromosome 18. Journal of Histochemistry and Cytochemistry, 2007, 55, 1123-1128.	2.5	13
7	Partial tetrasomy of chromosome 3q and mosaicism in a child with autism. Journal of Autism and Developmental Disorders, 2003, 33, 177-185.	2.7	12
8	Cryptic 7q36.2q36.3 deletion causes multiple congenital eye anomalies and craniofacial dysmorphism. American Journal of Medical Genetics, Part A, 2013, 161, 589-593.	1.2	9
9	Critical region in 2q31.2q32.3 deletion syndrome: Report of two phenotypically distinct patients, one with an additional deletion in Alagille syndrome region. Molecular Cytogenetics, 2012, 5, 25.	0.9	8
10	Molecular cytogenetic characterisation of a mosaic add(12)(p13.3) with an inv dup(3)(q26.31 â†’ qter) detected in an autistic boy. Molecular Cytogenetics, 2009, 2, 16.	0.9	6
11	Molecular Cytogenetic Characterization of Two Cases with de novo Small Mosaic Supernumerary Marker Chromosomes Derived from Chromosome 16: Towards a Genotype/Phenotype Correlation. Cytogenetic and Genome Research, 2009, 125, 109-114.	1.1	5
12	Interstitial 287Åkb deletion of 4p16.3 including FGFR1 gene associated with language impairment and overgrowth. Molecular Cytogenetics, 2014, 7, 87.	0.9	5
13	Prevalence of cytogenetic abnormalities and FMR1 gene premutation in a Portuguese population with premature ovarian insufficiency. Acta Medica Portuguesa, 2021, 34, 580-585.	0.4	2
14	Inv21p12q22del21q22 and intellectual disability. Gene, 2013, 517, 120-124.	2.2	0