

Daniela Rusconi

List of Publications by Year in descending order

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12
papers

277
citations

1040056

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docs citations

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734
citing authors

#	ARTICLE	IF	CITATIONS
1	From Whole Gene Deletion to Point Mutations of <i>EP300</i> -Positive Rubinstein-Taybi Patients: New Insights into the Mutational Spectrum and Peculiar Clinical Hallmarks. <i>Human Mutation</i> , 2016, 37, 175-183.	2.5	36
2	Familial intragenic duplication of ANKRD11 underlying three patients of KBG syndrome. <i>Molecular Cytogenetics</i> , 2015, 8, 20.	0.9	18
3	Characterization of 14 novel deletions underlying Rubinstein-Taybi syndrome: an update of the CREBBP deletion repertoire. <i>Human Genetics</i> , 2015, 134, 613-626.	3.8	38
4	Gene dosage as a relevant mechanism contributing to the determination of ovarian function in Turner syndrome. <i>Human Reproduction</i> , 2014, 29, 368-379.	0.9	39
5	Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. <i>BMC Medical Genetics</i> , 2013, 14, 41.	2.1	15
6	Oligoclonal bands in the cerebrospinal fluid of amyotrophic lateral sclerosis patients with disease-associated mutations. <i>Journal of Neurology</i> , 2013, 260, 85-92.	3.6	24
7	Design and validation of a pericentromeric BAC clone set aimed at improving diagnosis and phenotype prediction of supernumerary marker chromosomes. <i>Molecular Cytogenetics</i> , 2013, 6, 45.	0.9	8
8	A novel mosaic <i>NSD1</i> intragenic deletion in a patient with an atypical phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 611-618.	1.2	8
9	Juxtaposition of heterochromatic and euchromatic regions by chromosomal translocation mediates a heterochromatic long-range position effect associated with a severe neurological phenotype. <i>Molecular Cytogenetics</i> , 2012, 5, 16.	0.9	22
10	Combined characterization of a pituitary adenoma and a subcutaneous lipoma in a <i>MEN1</i> patient with a whole gene deletion. <i>Cancer Genetics</i> , 2011, 204, 309-315.	0.4	16
11	SNPs and real-time quantitative PCR method for constitutional allelic copy number determination, the <i>VPREB1</i> marker case. <i>BMC Medical Genetics</i> , 2011, 12, 61.	2.1	8
12	Erythroid differentiation and maturation from peripheral CD34+ cells in liquid culture: Cellular and molecular characterization. <i>Blood Cells, Molecules, and Diseases</i> , 2008, 40, 148-155.	1.4	45