

Vera Ayres Meloni

List of Publications by Year in descending order

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23
papers

224
citations

1040056

9
h-index

1058476

14
g-index

23
all docs

23
docs citations

23
times ranked

585
citing authors

#	ARTICLE	IF	CITATIONS
1	Multicentric Carpotarsal Osteolysis Syndrome in a Mother and Daughter with a <i>MAFB</i> Missense Variant and Natural History of the Disease. <i>Molecular Syndromology</i> , 2022, 13, 50-55.	0.8	5
2	CEDNIK syndrome in a Brazilian patient with compound heterozygous pathogenic variants. <i>European Journal of Medical Genetics</i> , 2022, 65, 104440.	1.3	2
3	Minimal Critical Region and Genes for a Typical Presentation of Langer-Giedion Syndrome. <i>Cytogenetic and Genome Research</i> , 2022, 162, 46-54.	1.1	1
4	Spread of X-chromosome inactivation into autosomal regions in patients with unbalanced X-autosome translocations and its phenotypic effects. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2295-2305.	1.2	2
5	Malan syndrome in a patient with 19p13.2p13.12 deletion encompassing <i>NFIX</i> and <i>CACNA1A</i> genes: Case report and review of the literature. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e997.	1.2	7
6	Breakpoint mapping at nucleotide resolution in X-autosome balanced translocations associated with clinical phenotypes. <i>European Journal of Human Genetics</i> , 2019, 27, 760-771.	2.8	12
7	Deletion of Chromosome 13 due to Different Rearrangements and Impact on Phenotype. <i>Molecular Syndromology</i> , 2019, 10, 139-146.	0.8	10
8	Downregulation of genes outside the deleted region in individuals with 22q11.2 deletion syndrome. <i>Human Genetics</i> , 2019, 138, 93-103.	3.8	8
9	Inactivation of <i>AMMECR1</i> is associated with growth, bone, and heart alterations. <i>Human Mutation</i> , 2018, 39, 281-291.	2.5	15
10	Unusual X-chromosome inactivation pattern in patients with Xp11.23q11.22 duplication: Report and review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3271-3275.	1.2	4
11	22q11.2 Deletion Syndrome due to a Translocation t(6;22) in a Patient Conceived via in vitro Fertilization. <i>Molecular Syndromology</i> , 2016, 6, 242-247.	0.8	3
12	Position effect modifying gene expression in a patient with ring chromosome 14. <i>Journal of Applied Genetics</i> , 2016, 57, 183-187.	1.9	12
13	X-linked intellectual disability related genes disrupted by balanced X-autosome translocations. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015, 168, 669-677.	1.7	32
14	Terminal 18q deletions are stabilized by neotelomeres. <i>Molecular Cytogenetics</i> , 2015, 8, 32.	0.9	12
15	Genetic mechanisms leading to primary amenorrhea in balanced X-autosome translocations. <i>Fertility and Sterility</i> , 2015, 103, 1289-1296.e2.	1.0	25
16	Duplication 9p and their implication to phenotype. <i>BMC Medical Genetics</i> , 2014, 15, 142.	2.1	32
17	19q13.33qter trisomy in a girl with intellectual impairment and seizures. <i>Meta Gene</i> , 2014, 2, 799-806.	0.6	11
18	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome in an adolescent with complex chromosomal rearrangement and intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2685-2688.	1.2	2

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19	Cytogenomic delineation and clinical follow-up of two siblings with an 8.5â€‰Mb 6q24.2â€‰q25.2 deletion inherited from a paternal insertion. American Journal of Medical Genetics, Part A, 2014, 164, 2378-2384.	1.2	4
20	Trisomy 1q32 and monosomy 11q25 associated with congenital heart defect: cytogenomic delineation and patient fourteen years follow-up. Molecular Cytogenetics, 2014, 7, 57.	0.9	3
21	Single-Nucleotide Polymorphism Array-Based Characterization of Ring Chromosome 18. Journal of Pediatrics, 2013, 163, 1174-1178.e3.	1.8	8
22	Ring chromosome 10: report on two patients and review of the literature. Journal of Applied Genetics, 2013, 54, 35-41.	1.9	10
23	Autistic disorder phenotype associated to a complex 15q intrachromosomal rearrangement. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 823-828.	1.7	4