Vera Ayres Meloni

List of Publications by Year
in descending order

23	224	1040056 9	1058476 14
papers	citations	h-index	g-index

23	23	23	585
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Duplication 9p and their implication to phenotype. BMC Medical Genetics, 2014, 15, 142.	2.1	32
2	Xâ€linked intellectual disability related genes disrupted by balanced Xâ€autosome translocations. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2015, 168, 669-677.	1.7	32
3	Genetic mechanisms leading to primary amenorrhea in balanced X-autosome translocations. Fertility and Sterility, 2015, 103, 1289-1296.e2.	1.0	25
4	Inactivation of AMMECR1 is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	2.5	15
5	Terminal 18q deletions are stabilized by neotelomeres. Molecular Cytogenetics, 2015, 8, 32.	0.9	12
6	Position effect modifying gene expression in a patient with ring chromosome 14. Journal of Applied Genetics, 2016, 57, 183-187.	1.9	12
7	Breakpoint mapping at nucleotide resolution in X-autosome balanced translocations associated with clinical phenotypes. European Journal of Human Genetics, 2019, 27, 760-771.	2.8	12
8	19q13.33â†'qter trisomy in a girl with intellectual impairment and seizures. Meta Gene, 2014, 2, 799-806.	0.6	11
9	Ring chromosome 10: report on two patients and review of the literature. Journal of Applied Genetics, 2013, 54, 35-41.	1.9	10
10	Deletion of Chromosome 13 due to Different Rearrangements and Impact on Phenotype. Molecular Syndromology, 2019, 10, 139-146.	0.8	10
11	Single-Nucleotide Polymorphism Array-Based Characterization of Ring Chromosome 18. Journal of Pediatrics, 2013, 163, 1174-1178.e3.	1.8	8
12	Downregulation of genes outside the deleted region in individuals with 22q11.2 deletion syndrome. Human Genetics, 2019, 138, 93-103.	3.8	8
13	Malan syndrome in a patient with 19p13.2p13.12 deletion encompassing NFIX and CACNA1A genes: Case report and review of the literature. Molecular Genetics & Genomic Medicine, 2019, 7, e997.	1.2	7
14	Multicentric Carpotarsal Osteolysis Syndrome in a Mother and Daughter with a <i>MAFB</i> Missense Variant and Natural History of the Disease. Molecular Syndromology, 2022, 13, 50-55.	0.8	5
15	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
16	Cytogenomic delineation and clinical followâ€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
17	Unusual Xâ€chromosome inactivation pattern in patients with Xp11.23â€p11.22 duplication: Report and review. American Journal of Medical Genetics, Part A, 2016, 170, 3271-3275.	1.2	4
18	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

#	Article	IF	Citations
19	22q11.2 Deletion Syndrome due to a Translocation t(6;22) in a Patient Conceived via in vitro Fertilization. Molecular Syndromology, 2016, 6, 242-247.	0.8	3
20	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome in an adolescent with complex chromosomal rearrangement and intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2685-2688.	1.2	2
21	Spread of Xâ€chromosome inactivation into autosomal regions in patients with unbalanced Xâ€autosome translocations and its phenotypic effects. American Journal of Medical Genetics, Part A, 2021, 185, 2295-2305.	1.2	2
22	CEDNIK syndrome in a Brazilian patient with compound heterozygous pathogenic variants. European Journal of Medical Genetics, 2022, 65, 104440.	1.3	2
23	Minimal Critical Region and Genes for a Typical Presentation of Langer-Giedion Syndrome. Cytogenetic and Genome Research, 2022, 162, 46-54.	1.1	1