Chiara Palka Bayard De Volo

List of Publications by Year
in descending order

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26	433	932766 10	713013 21
papers	citations	h-index	g-index

26	26	26	666
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of deletions and duplications of the DMD gene in affected males and carrier females by multiple ligation probe amplification (MLPA). Human Genetics, 2005, 117, 92-98.	1.8	104
2	Spinal muscular atrophy genotyping by gene dosage using multiple ligation-dependent probe amplification. Neurogenetics, 2006, 7, 269-276.	0.7	59
3	Screening of mutations in the CFTR gene in 1195 couples entering assisted reproduction technique programs. European Journal of Human Genetics, 2005, 13, 959-964.	1.4	46
4	Mosaic 7q31 Deletion Involving FOXP2 Gene Associated With Language Impairment. Pediatrics, 2012, 129, e183-e188.	1.0	45
5	Complex rearrangement of chromosomes 7q21.13â€q22.1 confirms the ectrodactylyâ€deafness locus and suggests new candidate genes. American Journal of Medical Genetics, Part A, 2008, 146A, 238-244.	0.7	34
6	Identification and characterization of different SHOX gene deletions in patients with Leriâ€“Weill dyschondrosteosys by MLPA assay. Journal of Human Genetics, 2007, 52, 21-27.	1.1	33
7	A novel mutation of the IRF6 gene in an Italian family with Van der Woude syndrome. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2004, 547, 49-53.	0.4	21
8	Deletion 18p11.32p11.31 in a Child with Global Developmental Delay and Atypical, Drug-Resistant Absence Seizures. Cytogenetic and Genome Research, 2015, 146, 115-119.	0.6	11
9	Sequential combined test, second trimester maternal serum markers, and circulating fetal cells to select women for invasive prenatal diagnosis. PLoS ONE, 2017, 12, e0189235.	1.1	11
10	Comparison of combined, stepwise sequential, contingent, and integrated screening in 7292 highâ€risk pregnant women. Prenatal Diagnosis, 2011, 31, 1077-1081.	1.1	10
11	16q22.1 microdeletion detected by array-CGH in a family with mental retardation and lobular breast cancer. Gene, 2012, 498, 328-331.	1.0	10
12	Novel mutation in the ligand-binding domain of the androgen receptor gene (1790p) associated with complete androgen insensitivity syndrome. Asian Journal of Andrology, 2008, 10, 687-691.	0.8	9
13	Array-CGH characterization of a de novo t(X;Y)(p22;q11) in a female with short stature and mental retardation. Gene, 2012, 504, 107-110.	1.0	7
14	Spectrum of phenotypic anomalies in four families with deletion of the SHOX enhancer region. BMC Medical Genetics, 2014, 15, 87.	2.1	7
15	Cystic hygroma and mid-trimester maternal serum screening. Journal of Medical Screening, 2007, 14, 109-112.	1.1	6
16	Reticulate Vascular Lesions and a Large Head. Pediatric Dermatology, 2007, 24, 555-556.	0.5	4
17	Array-CGH characterization of a prenatally detected de novo 46,X,der(Y)t(X;Y)(p22.3;q11.2) in a male fetus. European Journal of Medical Genetics, 2011, 54, 333-336.	0.7	4
18	Prenatal diagnosis of a family affected by brachydactyly type A1 with a mutation in IHH. Clinical Dysmorphology, 2012, 21, 137-140.	0.1	3

#	Article	IF	Citations
19	Lack of correlation between elevated maternal serum hCG during second-trimester biochemical screening and fetal congenital anomaly. Prenatal Diagnosis, 2005, 25, 220-224.	1.1	2
20	Discovering a familial Xp11.4 microduplication: Does the mother matter?. Meta Gene, 2018, 16, 90-95.	0.3	2
21	A new case of mosaicism for invdup(15) duplicated for Praderâ€“Willi/Angelman syndrome critical region (PWACR) in an adult healthy man. European Journal of Medical Genetics, 2008, 51, 239-244.	0.7	1
22	De novo 9q33 microdeletion identified by array-comparative genomic hybridization in a foetus with sex reversal and congenital heart defects. Clinical Dysmorphology, 2013, 22, 132-134.	0.1	1
23	An 11.4-Mb Interstitial Deletion in a Fetus with No Apparent Phenotypic Alterations. Molecular Syndromology, 2017, 8, 42-44.	0.3	1
24	Case report of newborn with de novo partial trisomy 2q31.2â€“37.3 and monosomy 9p24.3. Journal of Genetics, 2018, 97, 311-317.	0.4	1
25	A 343 Italian cohort of patients analysed with arrayâ€comparative genomic hybridization: unsolved problems and genetic counselling difficulties. Journal of Intellectual Disability Research, 2021, 65, 863-869.	1.2	1
26	16p13.3 microduplication syndrome: A new characteristic case without intellectual disability. Gene Reports, 2016, 4, 218-221.	0.4	0