Chiara Palka Bayard De Volo

List of Publications by Year
in descending order

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	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
2	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
3	Discovering a familial Xp11.4 microduplication: Does the mother matter?. Meta Gene, 2018, 16, 90-95.	0.3	2
4	An 11.4-Mb Interstitial Deletion in a Fetus with No Apparent Phenotypic Alterations. Molecular Syndromology, 2017, 8, 42-44.	0.3	1
5	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
6	16p13.3 microduplication syndrome: A new characteristic case without intellectual disability. Gene Reports, 2016, 4, 218-221.	0.4	0
7	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
8	Spectrum of phenotypic anomalies in four families with deletion of the SHOX enhancer region. BMC Medical Genetics, 2014, 15, 87.	2.1	7
9	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
10	Prenatal diagnosis of a family affected by brachydactyly type A1 with a mutation in IHH. Clinical Dysmorphology, 2012, 21, 137-140.	0.1	3
11	Mosaic 7q31 Deletion Involving FOXP2 Gene Associated With Language Impairment. Pediatrics, 2012, 129, e183-e188.	1.0	45
12	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
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	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
15	Comparison of combined, stepwise sequential, contingent, and integrated screening in 7292 highâ€risk pregnant women. Prenatal Diagnosis, 2011, 31, 1077-1081.	1.1	10
16	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
17	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
18	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

#	Article	IF	Citations
19	Cystic hygroma and mid-trimester maternal serum screening. Journal of Medical Screening, 2007, 14, 109-112.	1.1	6
20	Reticulate Vascular Lesions and a Large Head. Pediatric Dermatology, 2007, 24, 555-556.	0.5	4
21	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
22	Spinal muscular atrophy genotyping by gene dosage using multiple ligation-dependent probe amplification. Neurogenetics, 2006, 7, 269-276.	0.7	59
23	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
24	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
25	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
26	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