Lucia Castiglia

List of Publications by Year
in descending order

29	1,198	623734	477307
papers	citations	14 h-index	29 g-index

29	29	29	2596
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
2	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
3	Cryptic telomeric rearrangements in subjects with mental retardation associated with dysmorphism and congenital malformations. Journal of Medical Genetics, 2001, 38, 417-420.	3.2	114
4	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 394-400.	6.2	60
5	6q Terminal Deletion Syndrome Associated with a Distinctive EEG and Clinical Pattern: A Report of Five Cases. Epilepsia, 2006, 47, 830-838.	5.1	44
6	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
7	Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. Clinical Genetics, 2007, 71, 599-601.	2.0	38
8	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	32
9	6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. Molecular Cytogenetics, 2013, 6, 4.	0.9	23
10	Narrowing the Candidate Region for Congenital Diaphragmatic Hernia in Chromosome 15q26: Contradictory Results. American Journal of Human Genetics, 2005, 77, 892-894.	6.2	20
11	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. Gene, 2014, 534, 435-439.	2.2	19
12	Genetic variations in human fetal globin gene microsatellites and their functional relevance. Human Genetics, 1999, 104, 307-314.	3.8	18
13	Deletion 2p25.2: A cryptic chromosome abnormality in a patient with autism and mental retardation detected using aCGH. European Journal of Medical Genetics, 2009, 52, 67-70.	1.3	16
14	A t(4;9)(q34;p22) Translocation Associated with Partial Epilepsy, Mental Retardation, and Dysmorphism. Epilepsia, 2005, 46, 1322-1324.	5.1	15
15	Biallelic intragenic duplication in ADGRB3 (BAI3) gene associated with intellectual disability, cerebellar atrophy, and behavioral disorder. European Journal of Human Genetics, 2019, 27, 594-602.	2.8	15
16	Evidence that a dodecamer duplication in the gene HOPA in Xq13 is not associated with mental retardation. Human Genetics, 2000, 106, 36-39.	3.8	12
17	Decreased expression of GRAF1/OPHN-1-L in the X-linked alpha thalassemia mental retardation syndrome. BMC Medical Genomics, 2010, 3, 28.	1.5	12
18	Mutations in ACTL6B, coding for a subunit of the neuron-specific chromatin remodeling complex nBAF, cause early onset severe developmental and epileptic encephalopathy with brain hypomyelination and cerebellar atrophy. Human Genetics, 2019, 138, 187-198.	3.8	12

#	Article	IF	Citations
19	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. Clinical Genetics, 2005, 67, 446-447.	2.0	11
20	Mutational analysis of the ATRX gene by DGGE: A powerful diagnostic approach for the ATRX syndrome. Human Mutation, 2003, 21, 529-534.	2.5	10
21	12q12 deletion: A new patient contributing to genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2008, 146A, 1354-1357.	1.2	10
22	Partial monosomy Xq(Xq23â†'qter) and trisomy 4p(4p15.33â†'pter) in a woman with intractable focal epilepsy, borderline intellectual functioning, and dysmorphic features. Brain and Development, 2008, 30, 425-429.	1.1	10
23	Three new patients with dup(17)(p11.2p11.2) without autism. Clinical Genetics, 2008, 73, 294-296.	2.0	7
24	An unusual presentation ofÂBecker Nevus. European Journal of Dermatology, 2010, 20, 522-523.	0.6	6
25	7q11.23 microduplication syndrome: neurophysiological and neuroradiological insights into a rare chromosomal disorder. Journal of Intellectual Disability Research, 2018, 62, 359-370.	2.0	5
26	Targeted next-generation sequencing identifies the disruption of the SHANK3 and RYR2 genes in a patient carrying a de novo t(1;22)(q43;q13.3) associated with signs of Phelan-McDermid syndrome. Molecular Cytogenetics, 2020, 13, 22.	0.9	4
27	Structural brain anomalies in Cri-du-Chat syndrome: MRI findings in 14 patients and possible genotype-phenotype correlations. European Journal of Paediatric Neurology, 2020, 28, 110-119.	1.6	3
28	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 419.	6.2	2
29	Identification of novel mutations in L1CAM gene by a DHPLC-based assay. Genes and Genomics, 2016, 38, 1159-1164.	1.4	1