Daniela Giardino

List of Publications by Year
in descending order

53 papers	1,549 citations	361045 20 h-index	329751 37 g-index
53 all docs	53 docs citations	53 times ranked	2417 citing authors

#	Article	IF	Citations
1	Ten new cases of Balanced Reciprocal Translocation Mosaicism (BRTM): Reproductive implications, frequency and mechanism. European Journal of Medical Genetics, 2020, 63, 103639.	0.7	6
2	Testing single/combined clinical categories on 5110 Italian patients with developmental phenotypes to improve arrayâ€based detection rate. Molecular Genetics & Genomic Medicine, 2020, 8, e1056.	0.6	6
3	Generation of three iPSC lines (IALi002, IALi004, IALi003) from Rubinstein-Taybi syndrome 1 patients carrying CREBBP non sense c.4435G>T, p.(Gly1479*) and c.3474G>A, p.(Trp1158*) and missense c.4627G>T, p.(Asp1543Tyr) mutations. Stem Cell Research, 2019, 40, 101553.	0.3	6
4	European guidelines for constitutional cytogenomic analysis. European Journal of Human Genetics, 2019, 27, 1-16.	1.4	108
5	Molecular cytogenetics characterization of seven small supernumerary marker chromosomes derived from chromosome 19: Genotype-phenotype correlation and review of the literature. European Journal of Medical Genetics, 2018, 61, 173-180.	0.7	5
6	Segmental Maternal UPD of Chromosome 7q in a Patient With Pendred and Silver Russell Syndromes-Like Features. Frontiers in Genetics, 2018, 9, 600.	1.1	4
7	13q mosaic deletion including RB1 associated to mild phenotype and no cancer outcome â€“ case report and review of the literature. Molecular Cytogenetics, 2018, 11, 53.	0.4	2
8	Generation of the Rubinstein-Taybi syndrome type 2 patient-derived induced pluripotent stem cell line (IALi001-A) carrying the EP300 exon 23 stop mutation c.3829Aâ€>â€T, p.(Lys1277*). Stem Cell Research, 2018, 30, 175-179.	0.3	4
9	iPSC-derived neurons of CREBBP - and EP300 -mutated Rubinstein-Taybi syndrome patients show morphological alterations and hypoexcitability. Stem Cell Research, 2018, 30, 130-140.	0.3	19
10	The Italian National External Quality Assessment Program in Cytogenetics: 4 years of activity (2013-2016) following the introduction of poor performance criteria. Annali Dell'Istituto Superiore Di Sanita, 2018, 54, 109-116.	0.2	0
11	Deletion of theSnord116/SNORD116Alters Sleep in Mice and Patients with Prader-Willi Syndrome. Sleep, 2016, 39, 637-644.	0.6	61
12	Survey of medical genetic services in Italy: year 2011. BMC Health Services Research, 2016, 16, 96.	0.9	2
13	7p22.1 microduplication syndrome: Clinical and molecular characterization of an adult case and review of the literature. European Journal of Medical Genetics, 2015, 58, 578-583.	0.7	9
14	Biocompatible fluorescent nanoparticles forin vivostem cell tracking. Nanotechnology, 2013, 24, 245603.	1.3	29
15	Exploring Patterns of Unwanted Behaviours in Adults with <scp>P</scp>raderâ€“<scp>W</scp>illi Syndrome. Journal of Applied Research in Intellectual Disabilities, 2013, 26, 568-577.	1.3	13
16	Design and validation of a pericentromeric BAC clone set aimed at improving diagnosis and phenotype prediction of supernumerary marker chromosomes. Molecular Cytogenetics, 2013, 6, 45.	0.4	8
17	A novel mosaic NSD1 intragenic deletion in a patient with an atypical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 611-618.	0.7	8
18	Deletion of the AP1S2 gene in a child with psychomotor delay and hypotonia. European Journal of Medical Genetics, 2012, 55, 124-127.	0.7	8

#	Article	IF	Citations
19	Complex rearrangement involving 9p deletion and duplication in a syndromic patient: Genotype/phenotype correlation and review of the literature. Gene, 2012, 502, 40-45.	1.0	16
20	Juxtaposition of heterochromatic and euchromatic regions by chromosomal translocation mediates a heterochromatic long-range position effect associated with a severe neurological phenotype. Molecular Cytogenetics, 2012, 5, 16.	0.4	22
21	Constitutional Chromothripsis Rearrangements Involve Clustered Double-Stranded DNA Breaks and Nonhomologous Repair Mechanisms. Cell Reports, 2012, 1, 648-655.	2.9	193
22	Electroclinical pattern in MECP2 duplication syndrome: Eight new reported cases and review of literature. Epilepsia, 2012, 53, 1146-1155.	2.6	34
23	Longitudinal Tracking of Human Fetal Cells Labeled with Super Paramagnetic Iron Oxide Nanoparticles in the Brain of Mice with Motor Neuron Disease. PLoS ONE, 2012, 7, e32326.	1.1	28
24	Combined characterization of a pituitary adenoma and a subcutaneous lipoma in a MEN1 patient with a whole gene deletion. Cancer Genetics, 2011, 204, 309-315.	0.2	16
25	Genotypeâ€“phenotype correlations in a new case of 8p23.1 deletion and review of the literature. European Journal of Medical Genetics, 2011, 54, 55-59.	0.7	47
26	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. BMC Medical Genetics, 2010, 11, 146.	2.1	86
27	Characterisation of complex chromosome 18p rearrangements in two syndromic patients with immunological deficits. European Journal of Medical Genetics, 2010, 53, 186-191.	0.7	13
28	A 12.4ÂMb duplication of 17q11.2q12 in a patient with psychomotor developmental delay and minor anomalies. European Journal of Medical Genetics, 2010, 53, 325-328.	0.7	10
29	1q44-qter Trisomy:Clinical Report and Review of the Literature. Genetic Testing and Molecular Biomarkers, 2009, 13, 79-86.	0.3	2
30	Fluorescence in situ hybridization dissection of a chronic myeloid leukemia case bearing the apparently balanced translocations (9;22)(q34;q11.2) and (11;11)(p15;q13). Cancer Genetics and Cytogenetics, 2009, 188, 42-47.	1.0	2
31	De novo balanced chromosome rearrangements in prenatal diagnosis. Prenatal Diagnosis, 2009, 29, 257-265.	1.1	60
32	Cytogenetic, FISH and array-CGH characterization of a complex chromosomal rearrangement carried by a mentally and language impaired patient. European Journal of Medical Genetics, 2009, 52, 218-223.	0.7	24
33	The Italian National Survey for Praderâ€“Willi syndrome: An epidemiologic study. American Journal of Medical Genetics, Part A, 2008, 146A, 861-872.	0.7	81
34	Genetics and mathematics: Evidence from Prader-Willi syndrome. Neuropsychologia, 2008, 46, 206-212.	0.7	20
35	Prenatal diagnosis of a small chromosome 2â€derived supernumerary marker, and review of the reported cases. American Journal of Medical Genetics, Part A, 2007, 143A, 2200-2203.	0.7	1
36	13q Deletion and central nervous system anomalies: further insights from karyotype-phenotype analyses of 14 patients. Journal of Medical Genetics, 2006, 44, e60-e60.	1.5	97

#	Article	IF	Citations
37	Limited value of echography to predict true fetal mosaicism for trisomy 12. Prenatal Diagnosis, 2006, 26, 1186-1189.	1.1	9
38	Trisomy 15q25.2-qter in an autistic child: Genotype-phenotype correlations. American Journal of Medical Genetics, Part A, 2005, 133A, 184-188.	0.7	17
39	Cytogenetic and molecular evaluation of 241 small supernumerary marker chromosomes: Cooperative study of 19 Italian laboratories. Genetics in Medicine, 2005, 7, 620-625.	1.1	30
40	High-mobility group A2 gene expression is frequently induced in non-functioning pituitary adenomas (NFPAs), even in the absence of chromosome 12 polysomy. Endocrine-Related Cancer, 2005, 12, 867-874.	1.6	40
41	Unbalanced segregation of a complex four-break 5q23â€“31 insertion in the 5p13 band in a malformed child. European Journal of Human Genetics, 2004, 12, 455-459.	1.4	2
42	Narrowing the candidate region of Albright hereditary osteodystrophy-like syndrome by deletion mapping in a patient with an unbalanced cryptic translocation t(2;6)(q37.3;q26). American Journal of Medical Genetics Part A, 2003, 122A, 261-265.	2.4	20
43	Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. Human Molecular Genetics, 2003, 12, 849-858.	1.4	131
44	Small familial supernumerary ring chromosome 2: FISH characterization and genotype-phenotype correlation. American Journal of Medical Genetics Part A, 2002, 111, 319-323.	2.4	13
45	The High Mobility Group A2 gene is amplified and overexpressed in human prolactinomas. Cancer Research, 2002, 62, 2398-405.	0.4	69
46	FISH characterization of a supernumerary r(1)(::cen?q22::q22?sq21::) chromosome associated with multiple anomalies and bilateral cataracts. American Journal of Medical Genetics Part A, 2001, 104, 157-164.	2.4	13
47	Refined FISH characterization of a de novo 1p22-p36.2 paracentric inversion and associated 1p21-22 deletion in a patient with signs of 1p36 microdeletion syndrome. American Journal of Medical Genetics Part A, 2001, 99, 308-313.	2.4	7
48	Cryptic subtelomeric translocation t(2;16)(q37;q24) segregating in a family with unexplained stillbirths and a dysmorphic, slightly retarded child. European Journal of Human Genetics, 2001, 9, 881-886.	1.4	21
49	FISH characterization of t(8;12)(q12;p13) observed as the sole karyotypic anomaly in a myelodysplastic syndrome patient. Cancer Genetics and Cytogenetics, 2001, 130, 75-78.	1.0	1
50	Non-random trisomies of chromosomes 5, 8 and 12 in the prolactinoma sub-type of pituitary adenomas: Conventional cytogenetics and interphase fish study. , 2000, 86, 344-350.		37
51	Cytogenetic study of pituitary adenomas. Cancer Genetics and Cytogenetics, 1997, 98, 131-136.	1.0	36
52	Cytogenetic abnormalities detected by direct analysis in a case of choriocarcinoma. Cancer Genetics and Cytogenetics, 1993, 68, 149-151.	1.0	19
53	Angelman's Syndrome in the First Year of Life. Developmental Medicine and Child Neurology, 1990, 32, 1011-1016.	1.1	34