

# Daniela Giardino

## List of Publications by Year in descending order

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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	Constitutional Chromothripsis Rearrangements Involve Clustered Double-Stranded DNA Breaks and Nonhomologous Repair Mechanisms. <i>Cell Reports</i> , 2012, 1, 648-655.	2.9	193
2	Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. <i>Human Molecular Genetics</i> , 2003, 12, 849-858.	1.4	131
3	European guidelines for constitutional cytogenomic analysis. <i>European Journal of Human Genetics</i> , 2019, 27, 1-16.	1.4	108
4	13q Deletion and central nervous system anomalies: further insights from karyotype-phenotype analyses of 14 patients. <i>Journal of Medical Genetics</i> , 2006, 44, e60-e60.	1.5	97
5	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. <i>BMC Medical Genetics</i> , 2010, 11, 146.	2.1	86
6	The Italian National Survey for Prader-Willi syndrome: An epidemiologic study. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 861-872.	0.7	81
7	The High Mobility Group A2 gene is amplified and overexpressed in human prolactinomas. <i>Cancer Research</i> , 2002, 62, 2398-405.	0.4	69
8	Deletion of the <i>Snord116/SNORD116</i> Alters Sleep in Mice and Patients with Prader-Willi Syndrome. <i>Sleep</i> , 2016, 39, 637-644.	0.6	61
9	<i>De novo</i> balanced chromosome rearrangements in prenatal diagnosis. <i>Prenatal Diagnosis</i> , 2009, 29, 257-265.	1.1	60
10	Genotype-phenotype correlations in a new case of 8p23.1 deletion and review of the literature. <i>European Journal of Medical Genetics</i> , 2011, 54, 55-59.	0.7	47
11	High-mobility group A2 gene expression is frequently induced in non-functioning pituitary adenomas (NFPAs), even in the absence of chromosome 12 polysomy. <i>Endocrine-Related Cancer</i> , 2005, 12, 867-874.	1.6	40
12	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
13	Cytogenetic study of pituitary adenomas. <i>Cancer Genetics and Cytogenetics</i> , 1997, 98, 131-136.	1.0	36
14	Angelman's Syndrome in the First Year of Life. <i>Developmental Medicine and Child Neurology</i> , 1990, 32, 1011-1016.	1.1	34
15	Electroclinical pattern in <i>MECP2</i> duplication syndrome: Eight new reported cases and review of literature. <i>Epilepsia</i> , 2012, 53, 1146-1155.	2.6	34
16	Cytogenetic and molecular evaluation of 241 small supernumerary marker chromosomes: Cooperative study of 19 Italian laboratories. <i>Genetics in Medicine</i> , 2005, 7, 620-625.	1.1	30
17	Biocompatible fluorescent nanoparticles for <i>in vivo</i> stem cell tracking. <i>Nanotechnology</i> , 2013, 24, 245603.	1.3	29
18	Longitudinal Tracking of Human Fetal Cells Labeled with Super Paramagnetic Iron Oxide Nanoparticles in the Brain of Mice with Motor Neuron Disease. <i>PLoS ONE</i> , 2012, 7, e32326.	1.1	28

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19	Cytogenetic, FISH and array-CGH characterization of a complex chromosomal rearrangement carried by a mentally and language impaired patient. <i>European Journal of Medical Genetics</i> , 2009, 52, 218-223.	0.7	24
20	Juxtaposition of heterochromatic and euchromatic regions by chromosomal translocation mediates a heterochromatic long-range position effect associated with a severe neurological phenotype. <i>Molecular Cytogenetics</i> , 2012, 5, 16.	0.4	22
21	Cryptic subtelomeric translocation t(2;16)(q37;q24) segregating in a family with unexplained stillbirths and a dysmorphic, slightly retarded child. <i>European Journal of Human Genetics</i> , 2001, 9, 881-886.	1.4	21
22	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). <i>American Journal of Medical Genetics Part A</i> , 2003, 122A, 261-265.	2.4	20
23	Genetics and mathematics: Evidence from Prader-Willi syndrome. <i>Neuropsychologia</i> , 2008, 46, 206-212.	0.7	20
24	Cytogenetic abnormalities detected by direct analysis in a case of choriocarcinoma. <i>Cancer Genetics and Cytogenetics</i> , 1993, 68, 149-151.	1.0	19
25	iPSC-derived neurons of CREBBP - and EP300 -mutated Rubinstein-Taybi syndrome patients show morphological alterations and hypoexcitability. <i>Stem Cell Research</i> , 2018, 30, 130-140.	0.3	19
26	Trisomy 15q25.2-qter in an autistic child: Genotype-phenotype correlations. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 184-188.	0.7	17
27	Combined characterization of a pituitary adenoma and a subcutaneous lipoma in a MEN1 patient with a whole gene deletion. <i>Cancer Genetics</i> , 2011, 204, 309-315.	0.2	16
28	Complex rearrangement involving 9p deletion and duplication in a syndromic patient: Genotype/phenotype correlation and review of the literature. <i>Gene</i> , 2012, 502, 40-45.	1.0	16
29	FISH characterization of a supernumerary r(1)(:cen?q22::q22?sq21::) chromosome associated with multiple anomalies and bilateral cataracts. <i>American Journal of Medical Genetics Part A</i> , 2001, 104, 157-164.	2.4	13
30	Small familial supernumerary ring chromosome 2: FISH characterization and genotype-phenotype correlation. <i>American Journal of Medical Genetics Part A</i> , 2002, 111, 319-323.	2.4	13
31	Characterisation of complex chromosome 18p rearrangements in two syndromic patients with immunological deficits. <i>European Journal of Medical Genetics</i> , 2010, 53, 186-191.	0.7	13
32	Exploring Patterns of Unwanted Behaviours in Adults with Prader-Willi Syndrome. <i>Journal of Applied Research in Intellectual Disabilities</i> , 2013, 26, 568-577.	1.3	13
33	A 12.4 Mb duplication of 17q11.2q12 in a patient with psychomotor developmental delay and minor anomalies. <i>European Journal of Medical Genetics</i> , 2010, 53, 325-328.	0.7	10
34	Limited value of echography to predict true fetal mosaicism for trisomy 12. <i>Prenatal Diagnosis</i> , 2006, 26, 1186-1189.	1.1	9
35	7p22.1 microduplication syndrome: Clinical and molecular characterization of an adult case and review of the literature. <i>European Journal of Medical Genetics</i> , 2015, 58, 578-583.	0.7	9
36	Deletion of the AP1S2 gene in a child with psychomotor delay and hypotonia. <i>European Journal of Medical Genetics</i> , 2012, 55, 124-127.	0.7	8

#	ARTICLE	IF	CITATIONS
37	Design and validation of a pericentromeric BAC clone set aimed at improving diagnosis and phenotype prediction of supernumerary marker chromosomes. <i>Molecular Cytogenetics</i> , 2013, 6, 45.	0.4	8
38	A novel mosaic <i>NSD1</i> intragenic deletion in a patient with an atypical phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 611-618.	0.7	8
39	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. <i>American Journal of Medical Genetics Part A</i> , 2001, 99, 308-313.	2.4	7
40	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. <i>Stem Cell Research</i> , 2019, 40, 101553.	0.3	6
41	Ten new cases of Balanced Reciprocal Translocation Mosaicism (BRTM): Reproductive implications, frequency and mechanism. <i>European Journal of Medical Genetics</i> , 2020, 63, 103639.	0.7	6
42	Testing single/combined clinical categories on 5110 Italian patients with developmental phenotypes to improve array-based detection rate. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1056.	0.6	6
43	Molecular cytogenetics characterization of seven small supernumerary marker chromosomes derived from chromosome 19: Genotype-phenotype correlation and review of the literature. <i>European Journal of Medical Genetics</i> , 2018, 61, 173-180.	0.7	5
44	Segmental Maternal UPD of Chromosome 7q in a Patient With Pendred and Silver Russell Syndromes-Like Features. <i>Frontiers in Genetics</i> , 2018, 9, 600.	1.1	4
45	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.3829A>G, p.(Lys1277*). <i>Stem Cell Research</i> , 2018, 30, 175-179.	0.3	4
46	Unbalanced segregation of a complex four-break 5q23q31 insertion in the 5p13 band in a malformed child. <i>European Journal of Human Genetics</i> , 2004, 12, 455-459.	1.4	2
47	1q44-qter Trisomy: Clinical Report and Review of the Literature. <i>Genetic Testing and Molecular Biomarkers</i> , 2009, 13, 79-86.	0.3	2
48	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). <i>Cancer Genetics and Cytogenetics</i> , 2009, 188, 42-47.	1.0	2
49	Survey of medical genetic services in Italy: year 2011. <i>BMC Health Services Research</i> , 2016, 16, 96.	0.9	2
50	13q mosaic deletion including RB1 associated to mild phenotype and no cancer outcome – case report and review of the literature. <i>Molecular Cytogenetics</i> , 2018, 11, 53.	0.4	2
51	FISH characterization of t(8;12)(q12;p13) observed as the sole karyotypic anomaly in a myelodysplastic syndrome patient. <i>Cancer Genetics and Cytogenetics</i> , 2001, 130, 75-78.	1.0	1
52	Prenatal diagnosis of a small chromosome 2-derived supernumerary marker, and review of the reported cases. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2200-2203.	0.7	1
53	The Italian National External Quality Assessment Program in Cytogenetics: 4 years of activity (2013-2016) following the introduction of poor performance criteria. <i>Annali Dell'Istituto Superiore Di Sanita</i> , 2018, 54, 109-116.	0.2	0