

P Finelli

List of Publications by Year in descending order

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87
papers

2,582
citations

172457

29
h-index

233421

45
g-index

88
all docs

88
docs citations

88
times ranked

4070
citing authors

#	ARTICLE	IF	CITATIONS
1	Comparative mapping of human alphoid sequences in great apes using fluorescence in situ hybridization. <i>Genomics</i> , 1995, 25, 477-484.	2.9	110
2	The Ste locus, a component of the parasitic cry-Ste system of <i>Drosophila melanogaster</i> , encodes a protein that forms crystals in primary spermatocytes and mimics properties of the beta subunit of casein kinase 2.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1995, 92, 6067-6071.	7.1	106
3	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.	3.2	97
4	Y chromosome loss in male patients with primary biliary cirrhosis. <i>Journal of Autoimmunity</i> , 2013, 41, 87-91.	6.5	93
5	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. <i>BMC Medical Genetics</i> , 2010, 11, 146.	2.1	86
6	Molecular cytogenetic dissection of human chromosomes 3 and 21 evolution. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2000, 97, 206-211.	7.1	83
7	Evaluation of autism traits in Angelman syndrome: a resource to unfold autism genes. <i>Neurogenetics</i> , 2007, 8, 169-178.	1.4	81
8	Molecular Analysis of 11q13 Breakpoints in Multiple Myeloma. <i>Blood</i> , 1999, 93, 1330-1337.	1.4	80
9	Misbehaviour of XIST RNA in Breast Cancer Cells. <i>PLoS ONE</i> , 2009, 4, e5559.	2.5	75
10	Clinical and molecular characterization of Rubinsteinâ€Taybi syndrome patients carrying distinct novel mutations of the <i>EP300</i> gene. <i>Clinical Genetics</i> , 2015, 87, 148-154.	2.0	75
11	The Active Gene That Encodes Human High Mobility Group 1 Protein (HMG1) Contains Introns and Maps to Chromosome 13. <i>Genomics</i> , 1996, 35, 367-371.	2.9	70
12	Fetal Cell Microchimerism in Papillary Thyroid Cancer: A Possible Role in Tumor Damage and Tissue Repair. <i>Cancer Research</i> , 2008, 68, 8482-8488.	0.9	70
13	Increased loss of the Y chromosome in peripheral blood cells in male patients with autoimmune thyroiditis. <i>Journal of Autoimmunity</i> , 2012, 38, J193-J196.	6.5	64
14	A panel of subchromosomal painting libraries representing over 300 regions of the human genome. <i>Cytogenetic and Genome Research</i> , 1995, 68, 25-32.	1.1	63
15	Differential electrophysiological features of neuropathies associated with 17p11.2 deletion and duplication. <i>Muscle and Nerve</i> , 1995, 18, 628-635.	2.2	60
16	Reciprocal chromosome painting shows that the great difference in diploid number between human and African green monkey is mostly due to non-Robertsonian fissions. <i>Mammalian Genome</i> , 1999, 10, 713-718.	2.2	58
17	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silverâ€Russell and Beckwithâ€Wiedemann syndromes. <i>Clinical Epigenetics</i> , 2016, 8, 23.	4.1	54
18	Genotypeâ€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.	1.3	47

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19	Structural Organization of Multiple Alphoid Subsets Coexisting on Human Chromosomes 1, 4, 5, 7, 9, 15, 18, and 19. <i>Genomics</i> , 1996, 38, 325-330.	2.9	45
20	Developmental disorders with intellectual disability driven by chromatin dysregulation: Clinical overlaps and molecular mechanisms. <i>Clinical Genetics</i> , 2019, 95, 231-240.	2.0	43
21	The evolutionary history of human chromosome 7. <i>Genomics</i> , 2004, 84, 458-467.	2.9	42
22	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.	3.1	40
23	Gene dosage as a relevant mechanism contributing to the determination of ovarian function in Turner syndrome. <i>Human Reproduction</i> , 2014, 29, 368-379.	0.9	39
24	Characterization of 14 novel deletions underlying Rubinstein-Taybi syndrome: an update of the CREBBP deletion repertoire. <i>Human Genetics</i> , 2015, 134, 613-626.	3.8	38
25	Non-random trisomies of chromosomes 5, 8 and 12 in the prolactinoma sub-type of pituitary adenomas: Conventional cytogenetics and interphase fish study. <i>International Journal of Cancer</i> , 2000, 86, 344-350.	5.1	37
26	From Whole Gene Deletion to Point Mutations of <i>EP300</i> -Positive Rubinstein-Taybi Patients: New Insights into the Mutational Spectrum and Peculiar Clinical Hallmarks. <i>Human Mutation</i> , 2016, 37, 175-183.	2.5	36
27	Disruption of Friend of GATA 2 gene (<i>FOG2</i>) by a <i>de novo</i> t(8;10) chromosomal translocation is associated with heart defects and gonadal dysgenesis. <i>Clinical Genetics</i> , 2007, 71, 195-204.	2.0	33
28	FISH characterisation of an identical (16)(p11.2p12.2) tandem duplication in two unrelated patients with autistic behaviour. <i>Journal of Medical Genetics</i> , 2004, 41, e90-e90.	3.2	32
29	Molecular and genomic characterisation of cryptic chromosomal alterations leading to paternal duplication of the 11p15.5 Beckwith-Wiedemann region. <i>Journal of Medical Genetics</i> , 2006, 43, e39-e39.	3.2	32
30	High-resolution array-CGH analysis on 46,XX patients affected by early onset primary ovarian insufficiency discloses new genes involved in ovarian function. <i>Human Reproduction</i> , 2019, 34, 574-583.	0.9	32
31	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.	2.4	30
32	Mapping of the Human NMDAR2B Receptor Subunit Gene (<i>GRIN2B</i>) to Chromosome 12p12. <i>Genomics</i> , 1994, 22, 216-218.	2.9	29
33	Prenatal diagnosis of a <i>de novo</i> complex chromosome rearrangement (CCR) mediated by six breakpoints, and a review of 20 prenatally ascertained CCRs. <i>Prenatal Diagnosis</i> , 2006, 26, 565-570.	2.3	27
34	Exploring by whole exome sequencing patients with initial diagnosis of Rubinstein-Taybi syndrome: the interconnections of epigenetic machinery disorders. <i>Human Genetics</i> , 2019, 138, 257-269.	3.8	25
35	The genes encoding the glutamate receptor subunits KA1 and KA2 (<i>GRIK4</i> and <i>GRIK5</i>) are located on separate chromosomes in human, mouse, and rat. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1994, 91, 11849-11853.	7.1	24
36	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.	1.3	24

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37	Oligoclonal bands in the cerebrospinal fluid of amyotrophic lateral sclerosis patients with disease-associated mutations. <i>Journal of Neurology</i> , 2013, 260, 85-92.	3.6	24
38	Molecular characterization of a mosaic NIPBL deletion in a Cornelia de Lange patient with severe phenotype. <i>European Journal of Medical Genetics</i> , 2013, 56, 138-143.	1.3	24
39	Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. <i>European Journal of Human Genetics</i> , 2012, 20, 734-741.	2.8	23
40	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.	0.9	22
41	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.	2.8	21
42	The Human Skeletal Muscle Glycogenin Gene: cDNA, Tissue Expression, and Chromosomal Localization. <i>Biochemical and Biophysical Research Communications</i> , 1996, 220, 72-77.	2.1	20
43	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
44	A new structural rearrangement associated to Wolfram syndrome in a child with a partial phenotype. <i>Gene</i> , 2012, 509, 168-172.	2.2	20
45	Rothmund-Thomson Syndrome: Insights from New Patients on the Genetic Variability Underpinning Clinical Presentation and Cancer Outcome. <i>International Journal of Molecular Sciences</i> , 2018, 19, 1103.	4.1	20
46	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.	0.7	19
47	Familial intragenic duplication of ANKRD11 underlying three patients of KBC syndrome. <i>Molecular Cytogenetics</i> , 2015, 8, 20.	0.9	18
48	Smith-Magenis Syndrome – Clinical Review, Biological Background and Related Disorders. <i>Genes</i> , 2022, 13, 335.	2.4	18
49	Cloning and comparative mapping of recently evolved human chromosome 22-specific alpha satellite DNA. <i>Somatic Cell and Molecular Genetics</i> , 1994, 20, 443-448.	0.7	17
50	Comparative fluorescence in situ hybridization mapping of primate chromosomes with Alu polymerase chain reaction generated probes from human/rodent somatic cell hybrids. <i>Chromosome Research</i> , 1996, 4, 38-42.	2.2	17
51	Pure 6p22-pter trisomic patient: Refined FISH characterization and genotype-phenotype correlation. <i>American Journal of Medical Genetics Part A</i> , 2002, 108, 36-40.	2.4	17
52	Trisomy 15q25.2-qter in an autistic child: Genotype-phenotype correlations. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 184-188.	1.2	17
53	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.	0.4	16
54	Fetal cell microchimerism: a protective role in autoimmune thyroid diseases. <i>European Journal of Endocrinology</i> , 2015, 173, 111-118.	3.7	16

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55	Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. <i>BMC Medical Genetics</i> , 2013, 14, 41.	2.1	15
56	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. <i>Molecular Neurobiology</i> , 2020, 57, 3685-3701.	4.0	15
57	Complex <i>de novo</i> chromosomal rearrangement at 15q11-q13 involving an intrachromosomal triplication in a patient with a severe neuropsychological phenotype: Clinical report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 221-230.	1.2	14
58	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
59	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
60	Characterisation of complex chromosome 18p rearrangements in two syndromic patients with immunological deficits. <i>European Journal of Medical Genetics</i> , 2010, 53, 186-191.	1.3	13
61	The ZNF75 Zinc Finger Gene Subfamily: Isolation and Mapping of the Four Members in Humans and Great Apes. <i>Genomics</i> , 1996, 35, 312-320.	2.9	12
62	Progressive Deficiencies in Blood T Cells Associated with a 10p12-13 Interstitial Deletion. <i>Clinical Immunology and Immunopathology</i> , 1996, 80, 9-15.	2.0	12
63	9q34.3 microduplications lead to neurodevelopmental disorders through EHMT1 overexpression. <i>Neurogenetics</i> , 2019, 20, 145-154.	1.4	12
64	A balanced reciprocal translocation t(10;15)(q22.3;q26.1) interrupting ACAN gene in a family with proportionate short stature. <i>Journal of Endocrinological Investigation</i> , 2018, 41, 929-936.	3.3	11
65	Overall and allele-specific expression of the SMC1A gene in female Cornelia de Lange syndrome patients and healthy controls. <i>Epigenetics</i> , 2014, 9, 973-979.	2.7	10
66	Constitutional <i>de novo</i> deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. <i>Scientific Reports</i> , 2015, 5, 15454.	3.3	10
67	ATRX mutation in two adult brothers with non-specific moderate intellectual disability identified by exome sequencing. <i>Meta Gene</i> , 2013, 1, 102-108.	0.6	9
68	New case of trichorinophalangeal syndrome-like phenotype with a <i>de novo</i> t(2;8)(p16.1;q23.3) translocation which does not disrupt the TRPS1 gene. <i>BMC Medical Genetics</i> , 2014, 15, 52.	2.1	9
69	Targeted whole exome sequencing and Drosophila modelling to unveil the molecular basis of primary ovarian insufficiency. <i>Human Reproduction</i> , 2021, 36, 2975-2991.	0.9	9
70	SNPs and real-time quantitative PCR method for constitutional allelic copy number determination, the VPRED1 marker case. <i>BMC Medical Genetics</i> , 2011, 12, 61.	2.1	8
71	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.	0.9	8
72	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.	1.2	8

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73	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
74	Histone Deacetylase Inhibitors Ameliorate Morphological Defects and Hypoexcitability of iPSC-Neurons from Rubinstein-Taybi Patients. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5777.	4.1	7
75	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.	0.7	6
76	Molecular Analysis of 11q13 Breakpoints in Multiple Myeloma. <i>Blood</i> , 1999, 93, 1330-1337.	1.4	6
77	Characterization of chimpanzee-hamster hybrids by chromosome painting. <i>Somatic Cell and Molecular Genetics</i> , 1994, 20, 439-442.	0.7	4
78	Characterization of the t(4;14)(p16.3;q32) in the KMS-18 multiple myeloma cell line. <i>Leukemia</i> , 2001, 15, 864-865.	7.2	4
79	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.	0.7	4
80	Unbalanced segregation of a complex four-break 5q23³¹ insertion in the 5p13 band in a malformed child. <i>European Journal of Human Genetics</i> , 2004, 12, 455-459.	2.8	2
81	Two mosaic XY males carrying asymmetric Y chromosomes. <i>Clinical Genetics</i> , 1997, 51, 65-68.	2.0	2
82	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
83	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.	0.9	2
84	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
85	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.	1.2	1
86	Genetic Investigations in a 45,X Turner Patient with Spontaneous Puberty.. , 2010, , P1-305-P1-305.		0
87	Subchromosomal painting libraries (SCPLs) from somatic cell hybrids. , 1997, , 56-64.		0