P Finelli

List of Publications by Year
in descending order

87	2,582	172457	233421
papers	citations	29 h-index	45 g-index

88	88	88	4070
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Smith-Magenis Syndromeâ€"Clinical Review, Biological Background and Related Disorders. Genes, 2022, 13, 335.	2.4	18
2	Histone Deacetylase Inhibitors Ameliorate Morphological Defects and Hypoexcitability of iPSC-Neurons from Rubinstein-Taybi Patients. International Journal of Molecular Sciences, 2021, 22, 5777.	4.1	7
3	Targeted whole exome sequencing and Drosophila modelling to unveil the molecular basis of primary ovarian insufficiency. Human Reproduction, 2021, 36, 2975-2991.	0.9	9
4	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. Molecular Neurobiology, 2020, 57, 3685-3701.	4.0	15
5	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.	0.7	6
6	High-resolution array-CGH analysis on 46,XX patients affected by early onset primary ovarian insufficiency discloses new genes involved in ovarian function. Human Reproduction, 2019, 34, 574-583.	0.9	32
7	9q34.3 microduplications lead to neurodevelopmental disorders through EHMT1 overexpression. Neurogenetics, 2019, 20, 145-154.	1.4	12
8	Exploring by whole exome sequencing patients with initial diagnosis of Rubinsteinâ€"Taybi syndrome: the interconnections of epigenetic machinery disorders. Human Genetics, 2019, 138, 257-269.	3.8	25
9	Developmental disorders with intellectual disability driven by chromatin dysregulation: Clinical overlaps and molecular mechanisms. Clinical Genetics, 2019, 95, 231-240.	2.0	43
10	A balanced reciprocal translocation t(10;15)(q22.3;q26.1) interrupting ACAN gene in a family with proportionate short stature. Journal of Endocrinological Investigation, 2018, 41, 929-936.	3.3	11
11	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.	0.9	2
12	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.	0.7	4
13	Rothmund-Thomson Syndrome: Insights from New Patients on the Genetic Variability Underpinning Clinical Presentation and Cancer Outcome. International Journal of Molecular Sciences, 2018, 19, 1103.	4.1	20
14	iPSC-derived neurons of CREBBP - and EP300 -mutated Rubinstein-Taybi syndrome patients show morphological alterations and hypoexcitability. Stem Cell Research, 2018, 30, 130-140.	0.7	19
15	From Whole Gene Deletion to Point Mutations ofEP300-Positive Rubinstein-Taybi Patients: New Insights into the Mutational Spectrum and Peculiar Clinical Hallmarks. Human Mutation, 2016, 37, 175-183.	2.5	36
16	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silverâ€"Russell and Beckwithâ€"Wiedemann syndromes. Clinical Epigenetics, 2016, 8, 23.	4.1	54
17	Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. Scientific Reports, 2015, 5, 15454.	3.3	10
18	Clinical and molecular characterization of Rubinsteinâ€Taybi syndrome patients carrying distinct novel mutations of the <scp>EP300</scp> gene. Clinical Genetics, 2015, 87, 148-154.	2.0	75

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19	Fetal cell microchimerism: a protective role in autoimmune thyroid diseases. European Journal of Endocrinology, 2015, 173, 111-118.	3.7	16
20	Familial intragenic duplication of ANKRD11 underlying three patients of KBG syndrome. Molecular Cytogenetics, 2015, 8, 20.	0.9	18
21	Characterization of 14 novel deletions underlying Rubinsteinâ€“Taybi syndrome: an update of the CREBBP deletion repertoire. Human Genetics, 2015, 134, 613-626.	3.8	38
22	Complex de novo chromosomal rearrangement at 15q11â€“q13 involving an intrachromosomal triplication in a patient with a severe neuropsychological phenotype: Clinical report and review of the literature. American Journal of Medical Genetics, Part A, 2015, 167, 221-230.	1.2	14
23	Overall and allele-specific expression of theSMC1Agene in female Cornelia de Lange syndrome patients and healthy controls. Epigenetics, 2014, 9, 973-979.	2.7	10
24	Gene dosage as a relevant mechanism contributing to the determination of ovarian function in Turner syndrome. Human Reproduction, 2014, 29, 368-379.	0.9	39
25	New case of trichorinophalangeal syndrome-like phenotype with a de novo t(2;8)(p16.1;q23.3) translocation which does not disrupt the TRPS1 gene. BMC Medical Genetics, 2014, 15, 52.	2.1	9
26	Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. BMC Medical Genetics, 2013, 14, 41.	2.1	15
27	Oligoclonal bands in the cerebrospinal fluid of amyotrophic lateral sclerosis patients with disease-associated mutations. Journal of Neurology, 2013, 260, 85-92.	3.6	24
28	Y chromosome loss in male patients with primary biliary cirrhosis. Journal of Autoimmunity, 2013, 41, 87-91.	6.5	93
29	ATRX mutation in two adult brothers with non-specific moderate intellectual disability identified by exome sequencing. Meta Gene, 2013, 1, 102-108.	0.6	9
30	Molecular characterization of a mosaic NIPBL deletion in a Cornelia de Lange patient with severe phenotype. European Journal of Medical Genetics, 2013, 56, 138-143.	1.3	24
31	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.	0.9	8
32	A novel mosaic NSD1 intragenic deletion in a patient with an atypical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 611-618.	1.2	8
33	Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. European Journal of Human Genetics, 2012, 20, 734-741.	2.8	23
34	Increased loss of the Y chromosome in peripheral blood cells in male patients with autoimmune thyroiditis. Journal of Autoimmunity, 2012, 38, J193-J196.	6.5	64
35	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.	0.9	22
36	A new structural rearrangement associated to Wolfram syndrome in a child with a partial phenotype. Gene, 2012, 509, 168-172.	2.2	20

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37	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.	0.4	16
38	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.	1.3	47
39	SNPs and real-time quantitative PCR method for constitutional allelic copy number determination, the VPREB1 marker case. BMC Medical Genetics, 2011, 12, 61.	2.1	8
40	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. BMC Medical Genetics, 2010, 11, 146.	2.1	86
41	Characterisation of complex chromosome 18p rearrangements in two syndromic patients with immunological deficits. European Journal of Medical Genetics, 2010, 53, 186-191.	1.3	13
42	Genetic Investigations in a 45,X Turner Patient with Spontaneous Puberty.. , 2010, , P1-305-P1-305.		0
43	Misbehaviour of XIST RNA in Breast Cancer Cells. PLoS ONE, 2009, 4, e5559.	2.5	75
44	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
45	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.	1.3	24
46	Fetal Cell Microchimerism in Papillary Thyroid Cancer: A Possible Role in Tumor Damage and Tissue Repair. Cancer Research, 2008, 68, 8482-8488.	0.9	70
47	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.	1.2	1
48	Disruption of Friend of GATA 2 gene (FOGâ€2) by a de novo t(8;10) chromosomal translocation is associated with heart defects and gonadal dysgenesis. Clinical Genetics, 2007, 71, 195-204.	2.0	33
49	Evaluation of autism traits in Angelman syndrome: a resource to unfold autism genes. Neurogenetics, 2007, 8, 169-178.	1.4	81
50	13q Deletion and central nervous system anomalies: further insights from karyotype-phenotype analyses of 14 patients. Journal of Medical Genetics, 2006, 44, e60-e60.	3.2	97
51	Prenatal diagnosis of a de novo complex chromosome rearrangement (CCR) mediated by six breakpoints, and a review of 20 prenatally ascertained CCRs. Prenatal Diagnosis, 2006, 26, 565-570.	2.3	27
52	Molecular and genomic characterisation of cryptic chromosomal alterations leading to paternal duplication of the 11p15.5 Beckwith-Wiedemann region. Journal of Medical Genetics, 2006, 43, e39-e39.	3.2	32
53	Trisomy 15q25.2-qter in an autistic child: Genotype-phenotype correlations. American Journal of Medical Genetics, Part A, 2005, 133A, 184-188.	1.2	17
54	Cytogenetic and molecular evaluation of 241 small supernumerary marker chromosomes: Cooperative study of 19 Italian laboratories. Genetics in Medicine, 2005, 7, 620-625.	2.4	30

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55	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.	3.1	40
56	FISH characterisation of an identical (16)(p11.2p12.2) tandem duplication in two unrelated patients with autistic behaviour. Journal of Medical Genetics, 2004, 41, e90-e90.	3.2	32
57	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.	2.8	2
58	The evolutionary history of human chromosome 7. Genomics, 2004, 84, 458-467.	2.9	42
59	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
60	Pure 6p22-pter trisomic patient: Refined FISH characterization and genotype-phenotype correlation. American Journal of Medical Genetics Part A, 2002, 108, 36-40.	2.4	17
61	Small familial supernumerary ring chromosome 2: FISH characterization and genotypeâ€phenotype correlation. American Journal of Medical Genetics Part A, 2002, 111, 319-323.	2.4	13
62	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
63	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
64	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.	2.8	21
65	Characterization of the t(4;14)(p16.3;q32) in the KMS-18 multiple myeloma cell line. Leukemia, 2001, 15, 864-865.	7.2	4
66	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
67	Non-random trisomies of chromosomes 5, 8 and 12 in the prolactinoma sub-type of pituitary adenomas: Conventional cytogenetics and interphase fish study. International Journal of Cancer, 2000, 86, 344-350.	5.1	37
68	Molecular cytogenetic dissection of human chromosomes 3 and 21 evolution. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 206-211.	7.1	83
69	Molecular Analysis of 11q13 Breakpoints in Multiple Myeloma. Blood, 1999, 93, 1330-1337.	1.4	80
70	Reciprocal chromosome painting shows that the great difference in diploid number between human and African green monkey is mostly due to non-Robertsonian fissions. Mammalian Genome, 1999, 10, 713-718.	2.2	58
71	Molecular Analysis of 11q13 Breakpoints in Multiple Myeloma. Blood, 1999, 93, 1330-1337.	1.4	6
72	Two mosaicâ€YY males carrying asymmetric Y chromosomes. Clinical Genetics, 1997, 51, 65-68.	2.0	2

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73	Subchromosomal painting libraries (SCPLs) from somatic cell hybrids. , 1997, , 56-64.		0
74	The ZNF75 Zinc Finger Gene Subfamily: Isolation and Mapping of the Four Members in Humans and Great Apes. Genomics, 1996, 35, 312-320.	2.9	12
75	The Active Gene That Encodes Human High Mobility Group 1 Protein (HMG1) Contains Introns and Maps to Chromosome 13. Genomics, 1996, 35, 367-371.	2.9	70
76	Structural Organization of Multiple Alphoid Subsets Coexisting on Human Chromosomes 1, 4, 5, 7, 9, 15, 18, and 19. Genomics, 1996, 38, 325-330.	2.9	45
77	The Human Skeletal Muscle Glycogenin Gene: cDNA, Tissue Expression, and Chromosomal Localization. Biochemical and Biophysical Research Communications, 1996, 220, 72-77.	2.1	20
78	Progressive Deficiencies in Blood T Cells Associated with a 10p12-13 Interstitial Deletion. Clinical Immunology and Immunopathology, 1996, 80, 9-15.	2.0	12
79	Comparative fluorescencein situ hybridization mapping of primate chromosomes withAlu polymerase chain reaction generated probes from human/rodent somatic cell hybrids. Chromosome Research, 1996, 4, 38-42.	2.2	17
80	The Ste locus, a component of the parasitic cry-Ste system of Drosophila melanogaster, encodes a protein that forms crystals in primary spermatocytes and mimics properties of the beta subunit of casein kinase 2.. Proceedings of the National Academy of Sciences of the United States of America, 1995, 92, 6067-6071.	7.1	106
81	Differential electrophysiological features of neuropathies associated with 17p11.2 deletion and duplication. Muscle and Nerve, 1995, 18, 628-635.	2.2	60
82	A panel of subchromosomal painting libraries representing over 300 regions of the human genome. Cytogenetic and Genome Research, 1995, 68, 25-32.	1.1	63
83	Comparative mapping of human alphoid sequences in great apes using fluorescence in situ hybridization. Genomics, 1995, 25, 477-484.	2.9	110
84	Characterization of chimpanzee-hamster hybrids by chromosome painting. Somatic Cell and Molecular Genetics, 1994, 20, 439-442.	0.7	4
85	Cloning and comparative mapping of recently evolved human chromosome 22-specific alpha satellite DNA. Somatic Cell and Molecular Genetics, 1994, 20, 443-448.	0.7	17
86	Mapping of the Human NMDAR2B Receptor Subunit Gene (GRIN2B) to Chromosome 12p12. Genomics, 1994, 22, 216-218.	2.9	29
87	The genes encoding the glutamate receptor subunits KA1 and KA2 (GRIK4 and GRIK5) are located on separate chromosomes in human, mouse, and rat.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 11849-11853.	7.1	24