P Finelli

List of Publications by Year in Descending Order

85 papers	2,057 citations	27 h-index	41 g-index
88 ext. papers	2,294 ext. citations	4.6 avg, IF	4.02 L-index

#	Paper	IF	Citations
85	Histone Deacetylase Inhibitors Ameliorate Morphological Defects and Hypoexcitability of iPSC-Neurons from Rubinstein-Taybi Patients. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
84	Targeted whole exome sequencing and Drosophila modelling to unveil the molecular basis of primary ovarian insufficiency. *Human Reproduction*, **2021**, 36, 2975-2991	5.7	2
83	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. *Molecular Neurobiology*, **2020**, 57, 3685-3701	6.2	6
82	Generation of three iPSC lines (IAIi002, IAIi004, IAIi003) 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	1.6	5
81	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	5.7	23
80	9q34.3 microduplications lead to neurodevelopmental disorders through EHMT1 overexpression. *Neurogenetics*, **2019**, 20, 145-154	3	8
79	Exploring by whole exome sequencing patients with initial diagnosis of Rubinstein-Taybi syndrome: the interconnections of epigenetic machinery disorders. *Human Genetics*, **2019**, 138, 257-269	6.3	16
78	Developmental disorders with intellectual disability driven by chromatin dysregulation: Clinical overlaps and molecular mechanisms. *Clinical Genetics*, **2019**, 95, 231-240	4	29
77	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	5.2	10
76	Rothmund-Thomson Syndrome: Insights from New Patients on the Genetic Variability Underpinning Clinical Presentation and Cancer Outcome. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	16
75	iPSC-derived neurons of CREBBP- and EP300-mutated Rubinstein-Taybi syndrome patients show morphological alterations and hypoexcitability. *Stem Cell Research*, **2018**, 30, 130-140	1.6	10
74	13q mosaic deletion including associated to mild phenotype and no cancer outcome - case report and review of the literature. *Molecular Cytogenetics*, **2018**, 11, 53	2	2
73	Generation of the Rubinstein-Taybi syndrome type 2 patient-derived induced pluripotent stem cell line (IAIi001-A) carrying the EP300 exon 23 stop mutation c.3829A > T, p.(Lys1277*). *Stem Cell Research*, **2018**, 30, 175-179	1.6	3
72	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silver-Russell and Beckwith-Wiedemann syndromes. *Clinical Epigenetics*, **2016**, 8, 23	7.7	42
71	From Whole Gene Deletion to Point Mutations of EP300-Positive Rubinstein-Taybi Patients: New Insights into the Mutational Spectrum and Peculiar Clinical Hallmarks. *Human Mutation*, **2016**, 37, 175-83	4.7	27
70	Clinical and molecular characterization of Rubinstein-Taybi syndrome patients carrying distinct novel mutations of the EP300 gene. *Clinical Genetics*, **2015**, 87, 148-54	4	53
69	Fetal cell microchimerism: a protective role in autoimmune thyroid diseases. *European Journal of Endocrinology*, **2015**, 173, 111-8	6.5	12

68	Familial intragenic duplication of ANKRD11 underlying three patients of KBG syndrome. *Molecular Cytogenetics*, **2015**, 8, 20	2	14
67	Characterization of 14 novel deletions underlying Rubinstein-Taybi syndrome: an update of the CREBBP deletion repertoire. *Human Genetics*, **2015**, 134, 613-26	6.3	31
66	Complex de novo chromosomal rearrangement at 15q11-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**, 167A, 221-30	2.5	10
65	Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. *Scientific Reports*, **2015**, 5, 15454	4.9	10
64	Gene dosage as a relevant mechanism contributing to the determination of ovarian function in Turner syndrome. *Human Reproduction*, **2014**, 29, 368-79	5.7	33
63	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
62	Overall and allele-specific expression of the SMC1A gene in female Cornelia de Lange syndrome patients and healthy controls. *Epigenetics*, **2014**, 9, 973-9	5.7	9
61	Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. *BMC Medical Genetics*, **2013**, 14, 41	2.1	13
60	Oligoclonal bands in the cerebrospinal fluid of amyotrophic lateral sclerosis patients with disease-associated mutations. *Journal of Neurology*, **2013**, 260, 85-92	5.5	19
59	Y chromosome loss in male patients with primary biliary cirrhosis. *Journal of Autoimmunity*, **2013**, 41, 87-91	15.5	73
58	ATRX mutation in two adult brothers with non-specific moderate intellectual disability identified by exome sequencing. *Meta Gene*, **2013**, 1, 102-8	0.7	5
57	Molecular characterization of a mosaic NIPBL deletion in a Cornelia de Lange patient with severe phenotype. *European Journal of Medical Genetics*, **2013**, 56, 138-43	2.6	23
56	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	2	7
55	A novel mosaic NSD1 intragenic deletion in a patient with an atypical phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 611-8	2.5	7
54	Increased loss of the Y chromosome in peripheral blood cells in male patients with autoimmune thyroiditis. *Journal of Autoimmunity*, **2012**, 38, J193-6	15.5	47
53	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	2	17
52	A new structural rearrangement associated to Wolfram syndrome in a child with a partial phenotype. *Gene*, **2012**, 509, 168-72	3.8	17
51	Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. *European Journal of Human Genetics*, **2012**, 20, 734-41	5.3	20

50	Combined characterization of a pituitary adenoma and a subcutaneous lipoma in a MEN1 patient with a whole gene deletion. *Cancer Genetics*, **2011**, 204, 309-15	2.3	13
49	Genotype-phenotype correlations in a new case of 8p23.1 deletion and review of the literature. *European Journal of Medical Genetics*, **2011**, 54, 55-9	2.6	38
48	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	6
47	Characterisation of complex chromosome 18p rearrangements in two syndromic patients with immunological deficits. *European Journal of Medical Genetics*, **2010**, 53, 186-91	2.6	12
46	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. *BMC Medical Genetics*, **2010**, 11, 146	2.1	17
45	Misbehaviour of XIST RNA in breast cancer cells. *PLoS ONE*, **2009**, 4, e5559	3.7	59
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, 42-7		0
43	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, 218-23	2.6	24
42	Fetal cell microchimerism in papillary thyroid cancer: a possible role in tumor damage and tissue repair. *Cancer Research*, **2008**, 68, 8482-8	10.1	51
41	Prenatal diagnosis of a small chromosome 2-derived supernumerary marker, and review of the reported cases. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2200-3	2.5	1
40	Disruption of friend of GATA 2 gene (FOG-2) by a de novo t(8;10) chromosomal translocation is associated with heart defects and gonadal dysgenesis. *Clinical Genetics*, **2007**, 71, 195-204	4	30
39	Evaluation of autism traits in Angelman syndrome: a resource to unfold autism genes. *Neurogenetics*, **2007**, 8, 169-78	3	71
38	13q Deletion and central nervous system anomalies: further insights from karyotype-phenotype analyses of 14 patients. *Journal of Medical Genetics*, **2007**, 44, e60	5.8	80
37	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	5.8	29
36	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, 565-70	3.2	21
35	Trisomy 15q25.2-qter in an autistic child: genotype-phenotype correlations. *American Journal of Medical Genetics, Part A*, **2005**, 133A, 184-8	2.5	16
34	Cytogenetic and molecular evaluation of 241 small supernumerary marker chromosomes: cooperative study of 19 Italian laboratories. *Genetics in Medicine*, **2005**, 7, 620-5	8.1	26
33	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, 867-74	5.7	35

32	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	5.8	29
31	Unbalanced segregation of a complex four-break 5q23-31 insertion in the 5p13 band in a malformed child. *European Journal of Human Genetics*, **2004**, 12, 455-9	5.3	1
30	The evolutionary history of human chromosome 7. *Genomics*, **2004**, 84, 458-67	4.3	37
29	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, 261-5		16
28	Pure 6p22-pter trisomic patient: refined FISH characterization and genotype-phenotype correlation. *American Journal of Medical Genetics Part A*, **2002**, 108, 36-40		15
27	Small familial supernumerary ring chromosome 2: FISH characterization and genotype-phenotype correlation. *American Journal of Medical Genetics Part A*, **2002**, 111, 319-23		12
26	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, 157-64		11
25	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, 308-13		6
24	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, 881-6	5.3	19
23	Characterization of the t(4;14)(p16.3;q32) in the KMS-18 multiple myeloma cell line. *Leukemia*, **2001**, 15, 864-5	10.7	3
22	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, 75-8		1
21	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, 344-50	7.5	25
20	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, 206-11	11.5	73
19	Molecular Analysis of 11q13 Breakpoints in Multiple Myeloma. *Blood*, **1999**, 93, 1330-1337	2.2	68
18	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, 713-8	3.2	55
17	Molecular Analysis of 11q13 Breakpoints in Multiple Myeloma. *Blood*, **1999**, 93, 1330-1337	2.2	6
16	Two mosaic-YY males carrying asymmetric Y chromosomes. *Clinical Genetics*, **1997**, 51, 65-8	4	2
15	Subchromosomal painting libraries (SCPLs) from somatic cell hybrids **1997**, 56-64		

14	The ZNF75 zinc finger gene subfamily: isolation and mapping of the four members in humans and great apes. *Genomics*, **1996**, 35, 312-20	4.3	11
13	The active gene that encodes human high mobility group 1 protein (HMG1) contains introns and maps to chromosome 13. *Genomics*, **1996**, 35, 367-71	4.3	66
12	Structural organization of multiple alphoid subsets coexisting on human chromosomes 1, 4, 5, 7, 9, 15, 18, and 19. *Genomics*, **1996**, 38, 325-30	4.3	42
11	The human skeletal muscle glycogenin gene: cDNA, tissue expression and chromosomal localization. *Biochemical and Biophysical Research Communications*, **1996**, 220, 72-7	3.4	18
10	Progressive deficiencies in blood T cells associated with a 10p12-13 interstitial deletion. *Clinical Immunology and Immunopathology*, **1996**, 80, 9-15		11
9	Comparative fluorescence in situ hybridization mapping of primate chromosomes with Alu polymerase chain reaction generated probes from human/rodent somatic cell hybrids. *Chromosome Research*, **1996**, 4, 38-42	4.4	15
8	A panel of subchromosomal painting libraries representing over 300 regions of the human genome. *Cytogenetic and Genome Research*, **1995**, 68, 25-32	1.9	43
7	Comparative mapping of human alphoid sequences in great apes using fluorescence in situ hybridization. *Genomics*, **1995**, 25, 477-84	4.3	104
6	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, 6067-71	11.5	94
5	Differential electrophysiological features of neuropathies associated with 17p11.2 deletion and duplication. *Muscle and Nerve*, **1995**, 18, 628-35	3.4	56
4	Characterization of chimpanzee-hamster hybrids by chromosome painting. *Somatic Cell and Molecular Genetics*, **1994**, 20, 439-42		2
3	Cloning and comparative mapping of recently evolved human chromosome 22-specific alpha satellite DNA. *Somatic Cell and Molecular Genetics*, **1994**, 20, 443-8		15
2	Mapping of the human NMDAR2B receptor subunit gene (GRIN2B) to chromosome 12p12. *Genomics*, **1994**, 22, 216-8	4.3	20
1	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, 11849-53	11.5	19