Cristina Giovanna Gervasini

List of Publications by Year
in descending order

92	2,424	218677	233421
papers	citations	26 h-index	45 g-index

97	97	97	2997
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	2.4	24
2	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
3	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	4.1	8
4	Identical <scp>EP300</scp> variant leading to Rubinsteinâ€“Taybi syndrome with different clinical and immunologic phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 2129-2134.	1.2	2
5	KMT2A: Umbrella Gene for Multiple Diseases. Genes, 2022, 13, 514.	2.4	17
6	LAM Cells as Potential Drivers of Senescence in Lymphangioleiomyomatosis Microenvironment. International Journal of Molecular Sciences, 2022, 23, 7040.	4.1	4
7	Expanding the phenotype associated to KMT2A variants: overlapping clinical signs between Wiedemannâ€“Steiner and Rubinsteinâ€“Taybi syndromes. European Journal of Human Genetics, 2021, 29, 88-98.	2.8	11
8	Spontaneous chromosomal instability in peripheral blood lymphocytes from two molecularly confirmed Italian patients with Hereditary Fibrosis Poikiloderma: insights into cancer predisposition. Genetics and Molecular Biology, 2021, 44, e20200332.	1.3	10
9	Saliva detection of SARS-CoV-2 for mitigating company outbreaks: a surveillance experience, Milan, Italy, March 2021. Epidemiology and Infection, 2021, 149, e171.	2.1	6
10	Lithium as a possible therapeutic strategy for Cornelia de Lange syndrome. Cell Death Discovery, 2021, 7, 34.	4.7	10
11	Insights into the Role of the Microbiota and of Short-Chain Fatty Acids in Rubinsteinâ€“Taybi Syndrome. International Journal of Molecular Sciences, 2021, 22, 3621.	4.1	4
12	SLC35F1 as a candidate gene for neurodevelopmental disorders resembling Rett syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2238-2240.	1.2	5
13	Chromatin Imbalance as the Vertex Between Fetal Valproate Syndrome and Chromatinopathies. Frontiers in Cell and Developmental Biology, 2021, 9, 654467.	3.7	7
14	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
15	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of NIPBL variants in blood. Scientific Reports, 2021, 11, 15459.	3.3	11
16	Editorial: Impact on Embryonic Development of Chromatin Remodeling Alterations. Frontiers in Cell and Developmental Biology, 2021, 9, 744665.	3.7	0
17	Cohesin Mutations Induce Chromatin Conformation Perturbation of the H19/IGF2 Imprinted Region and Gene Expression Dysregulation in Cornelia de Lange Syndrome Cell Lines. Biomolecules, 2021, 11, 1622.	4.0	3
18	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. Neurogenetics, 2021, , 1.	1.4	2

#	Article	IF	Citations
19	Unexpected phenotype in a frameshift mutation of PTCH1. Molecular Genetics & Genomic Medicine, 2020, 8, e987.	1.2	3
20	Chromatinopathies: A focus on Cornelia de Lange syndrome. Clinical Genetics, 2020, 97, 3-11.	2.0	34
21	Olfactory Malformations in Mendelian Disorders of the Epigenetic Machinery. Frontiers in Cell and Developmental Biology, 2020, 8, 710.	3.7	2
22	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. Molecular Neurobiology, 2020, 57, 3685-3701.	4.0	15
23	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
24	Customised next-generation sequencing multigene panel to screen a large cohort of individuals with chromatin-related disorder. Journal of Medical Genetics, 2020, 57, 760-768.	3.2	15
25	Prevalence of Immunological Defects in a Cohort of 97 Rubinsteinâ€“Taybi Syndrome Patients. Journal of Clinical Immunology, 2020, 40, 851-860.	3.8	19
26	Generation of the Becker muscular dystrophy patient derived induced pluripotent stem cell line carrying the DMD splicing mutation c.1705-8 T>C.. Stem Cell Research, 2020, 45, 101819.	0.7	2
27	Establishment of a Duchenne muscular dystrophy patient-derived induced pluripotent stem cell line carrying a deletion of exons 51â€“53 of the dystrophin gene (CCMi003-A). Stem Cell Research, 2019, 40, 101544.	0.7	4
28	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.	0.7	6
29	Phenotypic Overlap of Roberts and Baller-Gerold Syndromes in Two Patients With Craniosynostosis, Limb Reductions, and ESCO2 Mutations. Frontiers in Pediatrics, 2019, 7, 210.	1.9	0
30	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
31	Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	2.3	11
32	Generation of induced pluripotent stem cells from a Becker muscular dystrophy patient carrying a deletion of exons 45-55 of the dystrophin gene (CCMi002BMD-A-9 âˆ†45-55). Stem Cell Research, 2018, 28, 21-24.	0.7	8
33	Rubinsteinâ€“Taybi syndrome: New neuroradiological and neuropsychiatric insights from a multidisciplinary approach. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 406-415.	1.7	24
34	Perthes disease: A new finding in Floatingâ€Harbor syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 703-706.	1.2	6
35	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.3829Aâ€>â€T, p.(Lys1277*). Stem Cell Research, 2018, 30, 175-179.	0.7	4
36	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

#	Article	IF	Citations
37	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
38	Familial gastrointestinal stromal tumors, lentigines, and cafÃ©â€auâ€lait macules associated with germline câ€kit mutation treated with imatinib. International Journal of Dermatology, 2017, 56, 195-201.	1.0	16
39	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	3.8	61
40	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
41	Derivation of the Duchenne muscular dystrophy patient-derived induced pluripotent stem cell line lacking DMD exons 49 and 50 (CCMi001DMD-A-3, âˆ† 49, âˆ† 50). Stem Cell Research, 2017, 25, 128-131.	0.7	9
42	Rubinstein Taybi Syndrome in an Indian Child due to EP300 Gene Mutation: Correspondence. Indian Journal of Pediatrics, 2017, 84, 91-92.	0.8	0
43	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
44	Hepatoblastoma in Rubinstein-Taybi Syndrome: A Case Report. Pediatric Blood and Cancer, 2016, 63, 572-573.	1.5	6
45	A new prognostic index of severity of intellectual disabilities in Cornelia de Lange syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 179-189.	1.6	7
46	Thrombocytopenia and Cornelia de Lange syndrome: Still an enigma?. American Journal of Medical Genetics, Part A, 2016, 170, 130-134.	1.2	4
47	Phenotype and genotype in 52 patients with Rubinsteinâ€“Taybi syndrome caused by EP300 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3069-3082.	1.2	91
48	Adolescents and adults affected by Cornelia de Lange syndrome: A report of 73 Italian patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 206-213.	1.6	22
49	Electroclinical phenotype in Rubinsteinâ€“Taybi syndrome. Brain and Development, 2016, 38, 563-570.	1.1	8
50	Ultra-Rare Syndromes: The Example of Rubinsteinâ€“Taybi Syndrome. Journal of Pediatric Genetics, 2015, 04, 177-186.	0.7	39
51	Dysmorphologic assessment in 115 Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser patients. Clinical Dysmorphology, 2015, 24, 95-101.	0.3	9
52	Expanding the role of the splicing <scp>USB</scp>1 gene from Poikiloderma with Neutropenia to acquired myeloid neoplasms. British Journal of Haematology, 2015, 171, 557-565.	2.5	7
53	Potential impact of fetal genotype on maternal blood pressure during pregnancy. Journal of Hypertension, 2015, 33, 664-665.	0.5	4
54	Rubinstein-Taybi syndrome: clinical features, genetic basis, diagnosis, and management. Italian Journal of Pediatrics, 2015, 41, 4.	2.6	83

#	Article	IF	Citations
55	Insights into genotypeâ€“phenotype correlations from <scp>CREBBP</scp> point mutation screening in a cohort of 46 Rubinsteinâ€“Taybi syndrome patients. Clinical Genetics, 2015, 88, 431-440.	2.0	51
56	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
57	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
58	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
59	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
60	Functional characterisation of a novel mutation affecting the catalytic domain of MMP2 in siblings with multicentric osteolysis, nodulosis and arthropathy. Journal of Human Genetics, 2014, 59, 631-637.	2.3	11
61	Cervical spine malformation in cornelia de lange syndrome: A report of three patients. American Journal of Medical Genetics, Part A, 2014, 164, 1520-1524.	1.2	6
62	Unusual prenatal presentation of Rubinsteinâ€“Taybi syndrome: A case report. American Journal of Medical Genetics, Part A, 2014, 164, 2663-2666.	1.2	6
63	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
64	Cornelia de Lange individuals with new and recurrent SMC1A mutations enhance delineation of mutation repertoire and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2013, 161, 2909-2919.	1.2	31
65	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
66	Germline mosaicism in cornelia de lange syndrome: Dilemmas and risk figures. American Journal of Medical Genetics, Part A, 2013, 161, 1825-1826.	1.2	2
67	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
68	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
69	Histone acetylation deficits in lymphoblastoid cell lines from patients with Rubinsteinâ€“Taybi syndrome. Journal of Medical Genetics, 2012, 49, 66-74.	3.2	58
70	A new report of Cornelia de Lange syndrome associated with split hand and feet. American Journal of Medical Genetics, Part A, 2012, 158A, 2953-2955.	1.2	3
71	High frequency of copy number imbalances in Rubinsteinâ€“Taybi patients negative to CREBBP mutational analysis. European Journal of Human Genetics, 2010, 18, 768-775.	2.8	13
72	Somatic mosaicism in Cornelia de Lange syndrome: a further contributor to the wide clinical expressivity?. Clinical Genetics, 2010, 78, 560-564.	2.0	30

#	Article	IF	Citations
73	SHOX duplications found in some cases with type I Mayer-Rokitansky-Kuster-Hauser syndrome. Genetics in Medicine, 2010, 12, 634-640.	2.4	67
74	Premature chromatid separation is not a useful diagnostic marker for Cornelia de Lange syndrome. Chromosome Research, 2009, 17, 763-771.	2.2	49
75	Recurrent microdeletion at 17q12 as a cause of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: two case reports. Orphanet Journal of Rare Diseases, 2009, 4, 25.	2.7	105
76	Developmental Abnormalities and Cancer Predisposition in Neurofibromatosis Type 1. Current Molecular Medicine, 2009, 9, 634-653.	1.3	32
77	Search for genomic imbalances in a cohort of 24 Cornelia de Lange patients negative for mutations in the NIPBL and SMC1L1 genes. Clinical Genetics, 2008, 74, 531-538.	2.0	11
78	Prenatal/neonatal pathology in two cases of Cornelia de Lange syndrome harboring novel mutations of NIPBL. Genetics in Medicine, 2007, 9, 188-194.	2.4	18
79	High frequency of mosaic CREBBP deletions in Rubinsteinâ€“Taybi syndrome patients and mapping of somatic and germ-line breakpoints. Genomics, 2007, 90, 567-573.	2.9	42
80	Functional analysis of splicing mutations in exon 7 of NF1gene. BMC Medical Genetics, 2007, 8, 4.	2.1	32
81	Clinical score of 62 Italian patients with Cornelia de Lange syndrome and correlations with the presence and type of NIPBL mutation. Clinical Genetics, 2007, 72, 98-108.	2.0	93
82	Rubinstein-Taybi Syndrome: spectrum of CREBBP mutations in Italian patients. BMC Medical Genetics, 2006, 7, 77.	2.1	60
83	X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. Nature Genetics, 2006, 38, 528-530.	21.4	393
84	Uncommon Alu-mediated NF1 microdeletion with a breakpoint inside the NF1 gene. Genomics, 2005, 85, 273-279.	2.9	13
85	Evidence for non-homologous end joining and non-allelic homologous recombination in atypical NF1 microdeletions. Human Genetics, 2004, 115, 69-80.	3.8	41
86	NF1 exonï¿½7 skipping and sequence alterations in exonic splice enhancers (ESEs) in a neurofibromatosisï¿½1 patient. Human Genetics, 2003, 113, 551-554.	3.8	35
87	Tandem duplication of the NF1 gene detected by high-resolution FISH in the 17q11.2 region. Human Genetics, 2002, 110, 314-321.	3.8	17
88	Genomic evidence versus characterisation of a single (17;22) translocation on NF1 gene duplication: lessons from deletions in "balanced" chromosomal rearrangements. Reply. Human Genetics, 2002, 111, 468-469.	3.8	4
89	FISH with locus-specific probes on stretched chromosomes: a useful tool for genome organization studies. Chromosome Research, 2001, 9, 167-170.	2.2	5
90	Title is missing!. Chromosome Research, 2001, 9, 520-520.	2.2	0

#	Article	IF	Citations
91	Identification of duplicated genes in 17q11.2 using FISH on stretched chromosomes and DNA fibers. Human Genetics, 2001, 109, 48-54.	3.8	6
92	Mapping of genes and ESTs assigned to 17q11.2 to a YAC contig centred on the NF1 gene. GeneScreen, 2000, 1, 21-27.	0.6	3