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	X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. Nature Genetics, 2006, 38, 528-530.	21.4	393
2	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
3	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
4	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
5	Rubinstein-Taybi syndrome: clinical features, genetic basis, diagnosis, and management. Italian Journal of Pediatrics, 2015, 41, 4.	2.6	83
6	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
7	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
8	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
9	SHOX duplications found in some cases with type I Mayer-Rokitansky-Kuster-Hauser syndrome. Genetics in Medicine, 2010, 12, 634-640.	2.4	67
10	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	3.8	61
11	Rubinstein-Taybi Syndrome: spectrum of CREBBP mutations in Italian patients. BMC Medical Genetics, 2006, 7, 77.	2.1	60
12	Histone acetylation deficits in lymphoblastoid cell lines from patients with Rubinsteinâ€“Taybi syndrome. Journal of Medical Genetics, 2012, 49, 66-74.	3.2	58
13	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
14	Premature chromatid separation is not a useful diagnostic marker for Cornelia de Lange syndrome. Chromosome Research, 2009, 17, 763-771.	2.2	49
15	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
16	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
17	Evidence for non-homologous end joining and non-allelic homologous recombination in atypical NF1 microdeletions. Human Genetics, 2004, 115, 69-80.	3.8	41
18	Ultra-Rare Syndromes: The Example of Rubinsteinâ€“Taybi Syndrome. Journal of Pediatric Genetics, 2015, 04, 177-186.	0.7	39

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19	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
20	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
21	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
22	Chromatinopathies: A focus on Cornelia de Lange syndrome. Clinical Genetics, 2020, 97, 3-11.	2.0	34
23	Functional analysis of splicing mutations in exon 7 of NF1gene. BMC Medical Genetics, 2007, 8, 4.	2.1	32
24	Developmental Abnormalities and Cancer Predisposition in Neurofibromatosis Type 1. Current Molecular Medicine, 2009, 9, 634-653.	1.3	32
25	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
26	Somatic mosaicism in Cornelia de Lange syndrome: a further contributor to the wide clinical expressivity?. Clinical Genetics, 2010, 78, 560-564.	2.0	30
27	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
28	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
29	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
30	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
31	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	2.4	24
32	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
33	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
34	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
35	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
36	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

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37	Prevalence of Immunological Defects in a Cohort of 97 Rubinsteinâ€“Taybi Syndrome Patients. Journal of Clinical Immunology, 2020, 40, 851-860.	3.8	19
38	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
39	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
40	KMT2A: Umbrella Gene for Multiple Diseases. Genes, 2022, 13, 514.	2.4	17
41	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
42	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
43	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. Molecular Neurobiology, 2020, 57, 3685-3701.	4.0	15
44	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
45	Uncommon Alu-mediated NF1 microdeletion with a breakpoint inside the NF1 gene. Genomics, 2005, 85, 273-279.	2.9	13
46	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
47	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
48	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
49	Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	2.3	11
50	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
51	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
52	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
53	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
54	Lithium as a possible therapeutic strategy for Cornelia de Lange syndrome. Cell Death Discovery, 2021, 7, 34.	4.7	10

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55	Dysmorphologic assessment in 115 Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser patients. Clinical Dysmorphology, 2015, 24, 95-101.	0.3	9
56	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
57	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
58	Electroclinical phenotype in Rubinsteinâ€“Taybi syndrome. Brain and Development, 2016, 38, 563-570.	1.1	8
59	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
60	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
61	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
62	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
63	Chromatin Imbalance as the Vertex Between Fetal Valproate Syndrome and Chromatinopathies. Frontiers in Cell and Developmental Biology, 2021, 9, 654467.	3.7	7
64	Identification of duplicated genes in 17q11.2 using FISH on stretched chromosomes and DNA fibers. Human Genetics, 2001, 109, 48-54.	3.8	6
65	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
66	Unusual prenatal presentation of Rubinsteinâ€“Taybi syndrome: A case report. American Journal of Medical Genetics, Part A, 2014, 164, 2663-2666.	1.2	6
67	Hepatoblastoma in Rubinstein-Taybi Syndrome: A Case Report. Pediatric Blood and Cancer, 2016, 63, 572-573.	1.5	6
68	Perthes disease: A new finding in Floatingâ€Harbor syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 703-706.	1.2	6
69	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
70	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
71	FISH with locus-specific probes on stretched chromosomes: a useful tool for genome organization studies. Chromosome Research, 2001, 9, 167-170.	2.2	5
72	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

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73	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
74	Potential impact of fetal genotype on maternal blood pressure during pregnancy. Journal of Hypertension, 2015, 33, 664-665.	0.5	4
75	Thrombocytopenia and Cornelia de Lange syndrome: Still an enigma?. American Journal of Medical Genetics, Part A, 2016, 170, 130-134.	1.2	4
76	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
77	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
78	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
79	LAM Cells as Potential Drivers of Senescence in Lymphangioleiomyomatosis Microenvironment. International Journal of Molecular Sciences, 2022, 23, 7040.	4.1	4
80	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
81	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
82	Unexpected phenotype in a frameshift mutation of PTCH1. Molecular Genetics & Genomic Medicine, 2020, 8, e987.	1.2	3
83	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
84	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
85	Olfactory Malformations in Mendelian Disorders of the Epigenetic Machinery. Frontiers in Cell and Developmental Biology, 2020, 8, 710.	3.7	2
86	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
87	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. Neurogenetics, 2021, , 1.	1.4	2
88	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
89	Title is missing!. Chromosome Research, 2001, 9, 520-520.	2.2	0
90	Rubinstein Taybi Syndrome in an Indian Child due to EP300 Gene Mutation: Correspondence. Indian Journal of Pediatrics, 2017, 84, 91-92.	0.8	0

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91	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
92	Editorial: Impact on Embryonic Development of Chromatin Remodeling Alterations. Frontiers in Cell and Developmental Biology, 2021, 9, 744665.	3.7	0