Larizza L

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3050234/publications.pdf

Version: 2024-02-01

157 papers	4,972 citations	101535 36 h-index	114455 63 g-index
158 all docs	158 docs citations	158 times ranked	6139 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	Rothmund-Thomson syndrome. Orphanet Journal of Rare Diseases, 2010, 5, 2.	2.7	237
3	Germline mutation in the juxtamembrane domain of the kit gene in a family with gastrointestinal stromal tumors and urticaria pigmentosa. Cancer, 2001, 92, 657-662.	4.1	194
4	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2009, 17, 611-619.	2.8	194
5	Constitutional Chromothripsis Rearrangements Involve Clustered Double-Stranded DNA Breaks and Nonhomologous Repair Mechanisms. Cell Reports, 2012, 1, 648-655.	6.4	193
6	Cancer Risk in Beckwith-Wiedemann Syndrome: A Systematic Review and Meta-Analysis Outlining a Novel (Epi)Genotype Specific Histotype Targeted Screening Protocol. Journal of Pediatrics, 2016, 176, 142-149.e1.	1.8	119
7	Mental retardation and cardiovascular malformations in NF1 microdeleted patients point to candidate genes in 17q11.2. Journal of Medical Genetics, 2004, 41, 35-41.	3.2	113
8	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190.	2.8	113
9	NF1 Microdeletion Syndrome: Refined FISH Characterization of Sporadic and Familial Deletions with Locus-Specific Probes. American Journal of Human Genetics, 2000, 66, 100-109.	6.2	105
10	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
11	Prevalence of beckwithâ€“wiedemann syndrome in North West of Italy. American Journal of Medical Genetics, Part A, 2013, 161, 2481-2486.	1.2	93
12	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
13	Recognition of the Cornelia de Lange syndrome phenotype with facial dysmorphology novel analysis. Clinical Genetics, 2016, 89, 557-563.	2.0	77
14	The Kasumi-1 cell line: a t(8;21)-kit mutant model for acute myeloid leukemia. Leukemia and Lymphoma, 2005, 46, 247-255.	1.3	75
15	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
16	Rothmundâ€“Thomson syndrome and RECQL4 defect: Splitting and lumping. Cancer Letters, 2006, 232, 107-120.	7.2	74
17	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
18	A tumor suppressor locus in familial and sporadic chordoma maps to 1p36. International Journal of Cancer, 2000, 87, 68-72.	5.1	69

#	Article	IF	Citations
19	Broadening of cohesinopathies: exome sequencing identifies mutations in <scp>ANKRD11</scp> in two patients with Cornelia de Langeâ€overlapping phenotype. Clinical Genetics, 2016, 89, 74-81.	2.0	69
20	Chromosomal instability in fibroblasts and mesenchymal tumors from 2 sibs with Rothmund-Thomson syndrome. , 1998, 77, 504-510.		61
21	TRIM8 downregulation in glioma affects cell proliferation and it is associated with patients survival. BMC Cancer, 2015, 15, 470.	2.6	61
22	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	3.8	61
23	Rubinstein-Taybi Syndrome: spectrum of CREBBP mutations in Italian patients. BMC Medical Genetics, 2006, 7, 77.	2.1	60
24	De novo balanced chromosome rearrangements in prenatal diagnosis. Prenatal Diagnosis, 2009, 29, 257-265.	2.3	60
25	First cytogenetic study of a recurrent familial chordoma of the clivus. , 1999, 81, 24-30.		58
26	Histone acetylation deficits in lymphoblastoid cell lines from patients with Rubinsteinâ€“Taybi syndrome. Journal of Medical Genetics, 2012, 49, 66-74.	3.2	58
27	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome: a paradigm for genomic medicine. Clinical Genetics, 2016, 89, 403-415.	2.0	57
28	Mapping of candidate region for chordoma development to 1p36.13 by LOH analysis. International Journal of Cancer, 2003, 107, 493-497.	5.1	55
29	Nephrological findings and genotypeâ€“phenotype correlation in Beckwithâ€“Wiedemann syndrome. Pediatric Nephrology, 2012, 27, 397-406.	1.7	55
30	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
31	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
32	Inherited and Sporadic Epimutations at the IGF2-H19 Locus in Beckwith-Wiedemann Syndrome and Wilms’ Tumor. Endocrine Development, 2009, 14, 1-9.	1.3	48
33	Novel C16orf57 mutations in patients with Poikiloderma with Neutropenia: bioinformatic analysis of the protein and predicted effects of all reported mutations. Orphanet Journal of Rare Diseases, 2012, 7, 7.	2.7	48
34	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
35	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	2.8	44
36	Developmental disorders with intellectual disability driven by chromatin dysregulation: Clinical overlaps and molecular mechanisms. Clinical Genetics, 2019, 95, 231-240.	2.0	43

#	Article	IF	Citations
37	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
38	Evidence for non-homologous end joining and non-allelic homologous recombination in atypical NF1 microdeletions. Human Genetics, 2004, 115, 69-80.	3.8	41
39	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
40	Expanding the clinical spectrum of the â€˜<scp>HDAC8</scp>â€phenotypeâ€™ â€“ implications for molecular diagnostics, counseling and risk prediction. Clinical Genetics, 2016, 89, 564-573.	2.0	38
41	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
42	Microtubule-associated protein/microtubule affinity-regulating kinase 4 (MARK4) plays a role in cell cycle progression and cytoskeletal dynamics. European Journal of Cell Biology, 2014, 93, 355-365.	3.6	36
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	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	1.2	36
45	Fetal growth patterns in Beckwithâ€“Wiedemann syndrome. Clinical Genetics, 2016, 90, 21-27.	2.0	34
46	Quantitative DNA methylation analysis improves epigenotype-phenotype correlations in Beckwith-Wiedemann syndrome. Epigenetics, 2013, 8, 1053-1060.	2.7	33
47	Mutations and novel polymorphisms in coding regions and UTRs of CDK5R1 and OMG genes in patients with non-syndromic mental retardation. Neurogenetics, 2006, 7, 59-66.	1.4	32
48	Functional analysis of splicing mutations in exon 7 of NF1gene. BMC Medical Genetics, 2007, 8, 4.	2.1	32
49	Developmental Abnormalities and Cancer Predisposition in Neurofibromatosis Type 1. Current Molecular Medicine, 2009, 9, 634-653.	1.3	32
50	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
51	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
52	RNA processing defects of the helicase geneRECQL4 in a compound heterozygous Rothmund-Thomson patient. American Journal of Medical Genetics Part A, 2003, 120A, 395-399.	2.4	29
53	Pathogenic Variants in STXBP1 and in Genes for GABAa Receptor Subunities Cause Atypical Rett/Rett-like Phenotypes. International Journal of Molecular Sciences, 2019, 20, 3621.	4.1	29
54	Characterization of a cytogenetic 17q11.2 deletion in an NF1 patient with a contiguous gene syndrome. Human Genetics, 1996, 98, 646-650.	3.8	26

#	Article	IF	Citations
55	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
56	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
57	A tumor suppressor locus in familial and sporadic chordoma maps to 1p36. International Journal of Cancer, 2000, 87, 68-72.	5.1	24
58	Differential Signature of the Centrosomal MARK4 Isoforms in Glioma. Analytical Cellular Pathology, 2011, 34, 319-338.	1.4	23
59	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
60	Evidence by Expression Analysis of Candidate Genes for Congenital Heart Defects in the NF1 Microdeletion Interval. Annals of Human Genetics, 2005, 69, 508-516.	0.8	22
61	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
62	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
63	Viable phenotype of ILNEB syndrome without nephrotic impairment in siblings heterozygous for unreported integrin alpha3 mutations. Orphanet Journal of Rare Diseases, 2016, 11, 136.	2.7	21
64	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
65	Update on the Cytogenetics and Molecular Genetics of Chordoma. Hereditary Cancer in Clinical Practice, 2005, 3, 29.	1.5	20
66	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
67	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
68	Maternal chromosome 7 hetero/isodisomy in Silver-Russell syndrome and PEG1 biallelic expression. Clinical Dysmorphology, 2000, 9, 157-162.	0.3	18
69	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
70	Clinical utility gene card for: Rothmundâ€“Thomson syndrome. European Journal of Human Genetics, 2013, 21, 792-792.	2.8	18
71	Familial intragenic duplication of ANKRD11 underlying three patients of KBG syndrome. Molecular Cytogenetics, 2015, 8, 20.	0.9	18
72	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

#	Article	IF	Citations
73	Complex rearrangement involving 9p deletion and duplication in a syndromic patient: Genotype/phenotype correlation and review of the literature. Gene, 2012, 502, 40-45.	2.2	16
74	Familial gastrointestinal stromal tumors, lentigines, and cafÃ©â€‰â€‰lait macules associated with germline câ€‹kit mutation treated with imatinib. International Journal of Dermatology, 2017, 56, 195-201.	1.0	16
75	Genotype-phenotype correlation in two sets of monozygotic twins with Williams syndrome. , 1997, 69, 107-111.		15
76	Atypical Rothmund-Thomson syndrome in a patient with compound Heterozygous Mutations in RECQL4 Gene and phenotypic features in RECQL4 syndromes. European Journal of Pediatrics, 2008, 167, 175-181.	2.7	15
77	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
78	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. Molecular Neurobiology, 2020, 57, 3685-3701.	4.0	15
79	FISH characterization of small supernumerary marker chromosomes in two Prader-Willi patients. , 1997, 68, 99-104.		14
80	Audiological findings, genotype and clinical severity score in Cornelia de Lange syndrome. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 1045-1048.	1.0	14
81	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
82	Ring Chromosome 20 Syndrome: Genetics, Clinical Characteristics, and Overlapping Phenotypes. Frontiers in Neurology, 2020, 11, 613035.	2.4	14
83	A 12-bp deletion (7818del12) in the c-kit protooncogene in a large Italian kindred with piebaldism. Human Mutation, 1995, 6, 343-345.	2.5	13
84	Molecular characterization of FRAXE-positive subjects with mental impairment in two unrelated Italian families. American Journal of Medical Genetics Part A, 1998, 75, 304-308.	2.4	13
85	FISH characterization of two supernumerary r(1) associated with distinct clinical phenotypes. American Journal of Medical Genetics Part A, 1999, 84, 377-380.	2.4	13
86	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
87	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
88	Uncommon Alu-mediated NF1 microdeletion with a breakpoint inside the NF1 gene. Genomics, 2005, 85, 273-279.	2.9	13
89	Novel physiological RECQL4 alternative transcript disclosed by molecular characterisation of Rothmundâ€“Thomson Syndrome sibs with mild phenotype. European Journal of Human Genetics, 2014, 22, 1298-1304.	2.8	13
90	A zebrafish model of Poikiloderma with Neutropenia recapitulates the human syndrome hallmarks and traces back neutropenia to the myeloid progenitor. Scientific Reports, 2015, 5, 15814.	3.3	13

#	Article	IF	Citations
91	9q34.3 microduplications lead to neurodevelopmental disorders through EHMT1 overexpression. Neurogenetics, 2019, 20, 145-154.	1.4	12
92	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
93	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
94	Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	2.3	11
95	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
96	8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. Genes, 2021, 12, 652.	2.4	11
97	Karyotypic characterization of a new human embryonal rhabdomyosarcoma cell line. Cancer Genetics and Cytogenetics, 1991, 54, 83-89.	1.0	10
98	A 12.4ÂMb duplication of 17q11.2q12 in a patient with psychomotor developmental delay and minor anomalies. European Journal of Medical Genetics, 2010, 53, 325-328.	1.3	10
99	Clinical utility gene card for: poikiloderma with neutropenia. European Journal of Human Genetics, 2013, 21, 1185-1185.	2.8	10
100	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
101	Suggestive evidence on the involvement of polypyrimidine-tract binding protein in regulating alternative splicing of MAP/microtubule affinity-regulating kinase 4 in glioma. Cancer Letters, 2015, 359, 87-96.	7.2	10
102	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
103	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
104	7p22.1 microduplication syndrome: Clinical and molecular characterization of an adult case and review of the literature. European Journal of Medical Genetics, 2015, 58, 578-583.	1.3	9
105	SETD5 Gene Haploinsufficiency in Three Patients With Suspected KBG Syndrome. Frontiers in Neurology, 2020, 11, 631.	2.4	9
106	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
107	Interstitial telomeres of an inv(9)(p11.2;q34) involved in a jumping translocation found in a woman through a stable unbalanced translocation in her malformed child. Journal of Medical Genetics, 2002, 39, 42e-42.	3.2	8
108	Deletion of the AP1S2 gene in a child with psychomotor delay and hypotonia. European Journal of Medical Genetics, 2012, 55, 124-127.	1.3	8

#	Article	IF	Citations
109	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
110	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
111	Assessment of intrafamilial clinical variability of poikiloderma with neutropenia by a 10-year follow-up of three affected siblings. European Journal of Medical Genetics, 2019, 62, 73-76.	1.3	8
112	A rare chromosome 5 heterochromatic variant derived from insertion of 9qh satellite 3 sequences. Chromosome Research, 1998, 6, 411-414.	2.2	7
113	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
114	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
115	A familial t(4;8) translocation segregates with epilepsy and migraine with aura. Annals of Clinical and Translational Neurology, 2020, 7, 855-859.	3.7	7
116	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
117	Correspondence. Cancer Genetics and Cytogenetics, 1998, 103, 180-181.	1.0	6
118	Recurrence and Familial Inheritance of Intronic NIPBL Pathogenic Variant Associated With Mild CdLS. Frontiers in Neurology, 2018, 9, 967.	2.4	6
119	Insights into Mutation Effect in Three Poikiloderma with Neutropenia Patients by Transcript Analysis and Disease Evolution of Reported Patients with the Same Pathogenic Variants. Journal of Clinical Immunology, 2018, 38, 494-502.	3.8	6
120	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
121	A novel RAD21 mutation in a boy with mild Cornelia de Lange presentation: Further delineation of the phenotype. European Journal of Medical Genetics, 2020, 63, 103620.	1.3	6
122	Ten new cases of Balanced Reciprocal Translocation Mosaicism (BRTM): Reproductive implications, frequency and mechanism. European Journal of Medical Genetics, 2020, 63, 103639.	1.3	6
123	Mutations in CREBBP and EP300 genes affect DNA repair of oxidative damage in Rubinstein-Taybi syndrome cells. Carcinogenesis, 2020, 41, 257-266.	2.8	6
124	Testing single/combined clinical categories on 5110 Italian patients with developmental phenotypes to improve arrayâ€based detection rate. Molecular Genetics & Genomic Medicine, 2020, 8, e1056.	1.2	6
125	Maternal Uniparental Disomy of Chromosome 20 (UPD(20)mat) as Differential Diagnosis of Silver Russell Syndrome: Identification of Three New Cases. Genes, 2021, 12, 588.	2.4	6
126	Expanding the Molecular Spectrum of ANKRD11 Gene Defects in 33 Patients with a Clinical Presentation of KBG Syndrome. International Journal of Molecular Sciences, 2022, 23, 5912.	4.1	6

#	Article	IF	Citations
127	Molecular cytogenetics characterization of seven small supernumerary marker chromosomes derived from chromosome 19: Genotype-phenotype correlation and review of the literature. European Journal of Medical Genetics, 2018, 61, 173-180.	1.3	5
128	Genes for RNA-binding proteins involved in neural-specific functions and diseases are downregulated in Rubinstein-Taybi iNeurons. Neural Regeneration Research, 2022, 17, 5.	3.0	5
129	Prognostic Impact of C-Kit Mutations in Core Binding Factor-Leukemia.. Blood, 2004, 104, 2013-2013.	1.4	5
130	Loss of Y chromosome with retention of Y heterochromatin in a marker chromosome from a human melanoma. International Journal of Cancer, 1991, 47, 154-157.	5.1	4
131	Activation of c-fos and c-fes in metastatic lympho-macrophage hybrids. International Journal of Cancer, 1992, 52, 478-482.	5.1	4
132	Differential induction of the two early genesc-jun andc-fos in weakly and strongly metastatic murine lymphoma cell lines. International Journal of Cancer, 1994, 57, 86-89.	5.1	4
133	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
134	Segmental Maternal UPD of Chromosome 7q in a Patient With Pendred and Silver Russell Syndromes-Like Features. Frontiers in Genetics, 2018, 9, 600.	2.3	4
135	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
136	Rings and Bricks: Expression of Cohesin Components is Dynamic during Development and Adult Life. International Journal of Molecular Sciences, 2018, 19, 438.	4.1	4
137	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
138	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
139	Survey of medical genetic services in Italy: year 2011. BMC Health Services Research, 2016, 16, 96.	2.2	2
140	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
141	Prenatal diagnosis of an extranumerary i(22p) with normal phenotype. Annales De GÃ©nÃ©tique, 1993, 36, 154-8.	0.4	2
142	Rapid construction of competitive templates for quantitative PCR of reverse-transcribed mRNAs.. Biotechnology Letters, 1995, 9, 879-884.	0.5	1
143	Delineation and physical separation of novel translocation breakpoints on chromosome 1p in two genetically closely associated childhood tumors. Cytogenetic and Genome Research, 2000, 88, 289-295.	1.1	1
144	Recombinant Chromosome 7 Driven by Maternal Chromosome 7 Pericentric Inversion in a Girl with Features of Silver-Russell Syndrome. International Journal of Molecular Sciences, 2020, 21, 8487.	4.1	1

#	Article	IF	Citations
145	Chromosomal instability in fibroblasts and mesenchymal tumors from 2 sibs with Rothmundâ€Thomson syndrome. International Journal of Cancer, 1998, 77, 504-510.	5.1	1
146	RECQL4-Related Recessive Conditions. , 2016, , 1141-1147.		1
147	Somatic Mosaicism as Modulator of the Global and Intellectual Phenotype in Epimutated Angelman Syndrome Patients. Journal of Intellectual Disability - Diagnosis and Treatment, 2015, 3, 126-137.	0.3	1
148	On the Parental Origin of the X Chromosomes in a 49,XXXXX Fetus. Journal of Maternal-Fetal and Neonatal Medicine, 1994, 3, 235-238.	1.5	0
149	Reply to letter by Volpi regarding: ?RNA processing defects of the helicase geneRECQL4 in a compound heterozygous Rothmund-Thomson patient?. American Journal of Medical Genetics Part A, 2004, 129A, 103-103.	2.4	0
150	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
151	Rubinstein Taybi Syndrome in an Indian Child due to EP300 Gene Mutation: Correspondence. Indian Journal of Pediatrics, 2017, 84, 91-92.	0.8	0
152	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
153	Interconnected Gene Networks Underpin the Clinical Overlap of HNRNPH1-Related and Rubinsteinâ€“Taybi Intellectual Disability Syndromes. Frontiers in Neuroscience, 2021, 15, 745684.	2.8	0
154	KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog). Atlas of Genetics and Cytogenetics in Oncology and Haematology, 2011, , .	0.1	0
155	RILEVANZA DEL SESSO NELLA MALATTIA DA COVID-19: QUANDO UN CROMOSOMA IN PIÃ™ FA LA DIFFERENZA!. Istituto Lombardo - Accademia Di Scienze E Lettere - Rendiconti Di Scienze, 0, , .	0.0	0
156	COMPRENDERE LE BASI MOLECOLARI DELLA DISABILITÃƒâ‚¬Â INTELLETTIVA ATTRAVERSO IL MODELLO DEI NEURONI DI CELLULE STAMINALI PLURIPOTENTI INDOTTE. Istituto Lombardo - Accademia Di Scienze E Lettere - Rendiconti Di Scienze, 0, , .	0.0	0
157	12q21 Interstitial Deletions: Seven New Syndromic Cases Detected by Array-CGH and Review of the Literature. Genes, 2022, 13, 780.	2.4	0