Larizza L

List of Publications by Citations

151 papers	3,924 citations	35 h-index	57 g-index
158 ext. papers	4,506 ext. citations	4.5 avg, IF	4.79 L-index

#	Paper	IF	Citations
151	X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. *Nature Genetics*, **2006**, 38, 528-30	36.3	351
150	Rothmund-Thomson syndrome. *Orphanet Journal of Rare Diseases*, **2010**, 5, 2	4.2	182
149	Germline mutation in the juxtamembrane domain of the kit gene in a family with gastrointestinal stromal tumors and urticaria pigmentosa. *Cancer*, **2001**, 92, 657-62	6.4	180
148	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2009**, 17, 611-9	5.3	176
147	Constitutional chromothripsis rearrangements involve clustered double-stranded DNA breaks and nonhomologous repair mechanisms. *Cell Reports*, **2012**, 1, 648-55	10.6	159
146	NF1 microdeletion syndrome: refined FISH characterization of sporadic and familial deletions with locus-specific probes. *American Journal of Human Genetics*, **2000**, 66, 100-9	11	93
145	Mental retardation and cardiovascular malformations in NF1 microdeleted patients point to candidate genes in 17q11.2. *Journal of Medical Genetics*, **2004**, 41, 35-41	5.8	90
144	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	3.6	90
143	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
142	(Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2016**, 24, 183-90	5.3	75
141	The Kasumi-1 cell line: a t(8;21)-kit mutant model for acute myeloid leukemia. *Leukemia and Lymphoma*, **2005**, 46, 247-55	1.9	65
140	Rothmund-Thomson syndrome and RECQL4 defect: splitting and lumping. *Cancer Letters*, **2006**, 232, 107-20	9.9	65
139	Phenotype and genotype in 52 patients with Rubinstein-Taybi syndrome caused by EP300 mutations. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3069-3082	2.5	60
138	A tumor suppressor locus in familial and sporadic chordoma maps to 1p36. *International Journal of Cancer*, **2000**, 87, 68-72	7.5	60
137	Recognition of the Cornelia de Lange syndrome phenotype with facial dysmorphology novel analysis. *Clinical Genetics*, **2016**, 89, 557-63	4	56
136	First cytogenetic study of a recurrent familial chordoma of the clivus. *International Journal of Cancer*, **1999**, 81, 24-30	7.5	54
135	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

134	Histone acetylation deficits in lymphoblastoid cell lines from patients with Rubinstein-Taybi syndrome. *Journal of Medical Genetics*, **2012**, 49, 66-74	5.8	53
133	Broadening of cohesinopathies: exome sequencing identifies mutations in ANKRD11 in two patients with Cornelia de Lange-overlapping phenotype. *Clinical Genetics*, **2016**, 89, 74-81	4	51
132	De novo heterozygous mutations in SMC3 cause a range of Cornelia de Lange syndrome-overlapping phenotypes. *Human Mutation*, **2015**, 36, 454-62	4.7	51
131	Chromosomal instability in fibroblasts and mesenchymal tumors from 2 sibs with Rothmund-Thomson syndrome. *International Journal of Cancer*, **1998**, 77, 504-10	7.5	51
130	Mapping of candidate region for chordoma development to 1p36.13 by LOH analysis. *International Journal of Cancer*, **2003**, 107, 493-7	7.5	51
129	Rubinstein-Taybi Syndrome: spectrum of CREBBP mutations in Italian patients. *BMC Medical Genetics*, **2006**, 7, 77	2.1	50
128	Nephrological findings and genotype-phenotype correlation in Beckwith-Wiedemann syndrome. *Pediatric Nephrology*, **2012**, 27, 397-406	3.2	48
127	De novo balanced chromosome rearrangements in prenatal diagnosis. *Prenatal Diagnosis*, **2009**, 29, 257-65	3.1	48
126	Prevalence of Beckwith-Wiedemann syndrome in North West of Italy. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2481-6	2.5	45
125	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
124	Inherited and Sporadic Epimutations at the IGF2-H19 locus in Beckwith-Wiedemann syndrome and Wilms' tumor. *Endocrine Development*, **2009**, 14, 1-9		41
123	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. *Human Genetics*, **2017**, 136, 307-320	6.3	40
122	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	4.2	38
121	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
120	High frequency of mosaic CREBBP deletions in Rubinstein-Taybi syndrome patients and mapping of somatic and germ-line breakpoints. *Genomics*, **2007**, 90, 567-73	4.3	37
119	(Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome: a paradigm for genomic medicine. *Clinical Genetics*, **2016**, 89, 403-415	4	37
118	Evidence for non-homologous end joining and non-allelic homologous recombination in atypical NF1 microdeletions. *Human Genetics*, **2004**, 115, 69-80	6.3	36
117	TRIM8 downregulation in glioma affects cell proliferation and it is associated with patients survival. *BMC Cancer*, **2015**, 15, 470	4.8	35

116	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. *European Journal of Human Genetics*, **2016**, 24, 784-93	5.3	34
115	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
114	Insights into genotype-phenotype correlations from CREBBP point mutation screening in a cohort of 46 Rubinstein-Taybi syndrome patients. *Clinical Genetics*, **2015**, 88, 431-40	4	31
113	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	3	30
112	Expanding the clinical spectrum of the 'HDAC8-phenotype' - implications for molecular diagnostics, counseling and risk prediction. *Clinical Genetics*, **2016**, 89, 564-73	4	29
111	Developmental disorders with intellectual disability driven by chromatin dysregulation: Clinical overlaps and molecular mechanisms. *Clinical Genetics*, **2019**, 95, 231-240	4	29
110	Quantitative DNA methylation analysis improves epigenotype-phenotype correlations in Beckwith-Wiedemann syndrome. *Epigenetics*, **2013**, 8, 1053-60	5.7	28
109	Fetal growth patterns in Beckwith-Wiedemann syndrome. *Clinical Genetics*, **2016**, 90, 21-7	4	28
108	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, 355-65	6.1	27
107	Developmental abnormalities and cancer predisposition in neurofibromatosis type 1. *Current Molecular Medicine*, **2009**, 9, 634-53	2.5	27
106	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
105	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**, 161A, 2909-19	2.5	26
104	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
103	Functional analysis of splicing mutations in exon 7 of NF1 gene. *BMC Medical Genetics*, **2007**, 8, 4	2.1	25
102	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
101	Nomenclature and definition in asymmetric regional body overgrowth. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1735-1738	2.5	24
100	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
99	RNA processing defects of the helicase gene RECQL4 in a compound heterozygous Rothmund-Thomson patient. *American Journal of Medical Genetics Part A*, **2003**, 120A, 395-9		23

98	Characterization of a cytogenetic 17q11.2 deletion in an NF1 patient with a contiguous gene syndrome. *Human Genetics*, **1996**, 98, 646-50	6.3	23
97	A tumor suppressor locus in familial and sporadic chordoma maps to 1p36. *International Journal of Cancer*, **2000**, 87, 68-72	7.5	22
96	Pathogenic Variants in and in Genes for GABAa Receptor Subunities Cause Atypical Rett/Rett-like Phenotypes. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	21
95	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
94	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
93	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
92	Prenatal/neonatal pathology in two cases of Cornelia de Lange syndrome harboring novel mutations of NIPBL. *Genetics in Medicine*, **2007**, 9, 188-94	8.1	17
91	Evidence by expression analysis of candidate genes for congenital heart defects in the NF1 microdeletion interval. *Annals of Human Genetics*, **2005**, 69, 508-16	2.2	17
90	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
89	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
88	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
87	Maternal chromosome 7 hetero/isodisomy in Silver-Russell syndrome and PEG1 biallelic expression. *Clinical Dysmorphology*, **2000**, 9, 157-62	0.9	16
86	Complex rearrangement involving 9p deletion and duplication in a syndromic patient: genotype/phenotype correlation and review of the literature. *Gene*, **2012**, 502, 40-5	3.8	15
85	Differential Signature of the Centrosomal MARK4 Isoforms in Glioma. *Analytical Cellular Pathology*, **2011**, 34, 319-338	3.4	15
84	Update on the cytogenetics and molecular genetics of chordoma. *Hereditary Cancer in Clinical Practice*, **2005**, 3, 29-41	2.3	15
83	Familial intragenic duplication of ANKRD11 underlying three patients of KBG syndrome. *Molecular Cytogenetics*, **2015**, 8, 20	2	14
82	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, 175-81	4.1	14
81	Familial gastrointestinal stromal tumors, lentigines, and café-au-lait macules associated with germline c-kit mutation treated with imatinib. *International Journal of Dermatology*, **2017**, 56, 195-201	1.7	13

80	Audiological findings, genotype and clinical severity score in Cornelia de Lange syndrome. *International Journal of Pediatric Otorhinolaryngology*, **2014**, 78, 1045-8	1.7	13
79	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
78	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
77	FISH characterization of two supernumerary r(1) associated with distinct clinical phenotypes. *American Journal of Medical Genetics Part A*, **1999**, 84, 377-380		13
76	Viable phenotype of ILNEB syndrome without nephrotic impairment in siblings heterozygous for unreported integrin alpha3 mutations. *Orphanet Journal of Rare Diseases*, **2016**, 11, 136	4.2	12
75	Novel physiological RECQL4 alternative transcript disclosed by molecular characterisation of Rothmund-Thomson Syndrome sibs with mild phenotype. *European Journal of Human Genetics*, **2014**, 22, 1298-304	5.3	12
74	Clinical utility gene card for: Rothmund-Thomson syndrome. *European Journal of Human Genetics*, **2013**, 21,	5.3	12
73	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		12
72	Uncommon Alu-mediated NF1 microdeletion with a breakpoint inside the NF1 gene. *Genomics*, **2005**, 85, 273-9	4.3	12
71	Small familial supernumerary ring chromosome 2: FISH characterization and genotype-phenotype correlation. *American Journal of Medical Genetics Part A*, **2002**, 111, 319-23		12
70	Genotype-phenotype correlation in two sets of monozygotic twins with Williams syndrome. *American Journal of Medical Genetics Part A*, **1997**, 69, 107-11		11
69	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
68	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
67	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
66	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
65	FISH characterization of small supernumerary marker chromosomes in two Prader-Willi patients. *American Journal of Medical Genetics Part A*, **1997**, 68, 99-104		10
64	Karyotypic characterization of a new human embryonal rhabdomyosarcoma cell line. *Cancer Genetics and Cytogenetics*, **1991**, 54, 83-9		10
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	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	9.9	9
61	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
60	9q34.3 microduplications lead to neurodevelopmental disorders through EHMT1 overexpression. *Neurogenetics*, **2019**, 20, 145-154	3	8
59	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, 631-7	4.3	8
58	Clinical utility gene card for: poikiloderma with neutropenia. *European Journal of Human Genetics*, **2013**, 21,	5.3	8
57	A 12.4 Mb duplication of 17q11.2q12 in a patient with psychomotor developmental delay and minor anomalies. *European Journal of Medical Genetics*, **2010**, 53, 325-8	2.6	8
56	A 12-bp deletion (7818del12) in the c-kit protooncogene in a large Italian kindred with piebaldism. *Human Mutation*, **1995**, 6, 343-5	4.7	8
55	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	4.9	7
54	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
53	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
52	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, e42	5.8	7
51	Molecular Etiology Disclosed by Array CGH in Patients With Silver-Russell Syndrome or Similar Phenotypes. *Frontiers in Genetics*, **2019**, 10, 955	4.5	7
50	7p22.1 microduplication syndrome: Clinical and molecular characterization of an adult case and review of the literature. *European Journal of Medical Genetics*, **2015**, 58, 578-83	2.6	6
49	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. *Molecular Neurobiology*, **2020**, 57, 3685-3701	6.2	6
48	Expanding the role of the splicing USB1 gene from Poikiloderma with Neutropenia to acquired myeloid neoplasms. *British Journal of Haematology*, **2015**, 171, 557-65	4.5	6
47	Deletion of the AP1S2 gene in a child with psychomotor delay and hypotonia. *European Journal of Medical Genetics*, **2012**, 55, 124-7	2.6	6
46	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
45	Testing single/combined clinical categories on 5110 Italian patients with developmental phenotypes to improve array-based detection rate. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1056	2.3	6

44	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
43	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	2.6	5
42	A rare chromosome 5 heterochromatic variant derived from insertion of 9qh satellite 3 sequences. *Chromosome Research*, **1998**, 6, 411-4	4.4	5
41	Translocation (7;17)(q22;p13) as a sole karyotypic change in an adrenal adenoma. *Cancer Genetics and Cytogenetics*, **1998**, 103, 180-1		5
40	Ring Chromosome 20 Syndrome: Genetics, Clinical Characteristics, and Overlapping Phenotypes. *Frontiers in Neurology*, **2020**, 11, 613035	4.1	5
39	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	6.3	4
38	Differential induction of the two early genes c-jun and c-fos in weakly and strongly metastatic murine lymphoma cell lines. *International Journal of Cancer*, **1994**, 57, 86-9	7.5	4
37	Loss of Y chromosome with retention of Y heterochromatin in a marker chromosome from a human melanoma. *International Journal of Cancer*, **1991**, 47, 154-7	7.5	4
36	Gene Haploinsufficiency in Three Patients With Suspected KBG Syndrome. *Frontiers in Neurology*, **2020**, 11, 631	4.1	4
35	Ten new cases of Balanced Reciprocal Translocation Mosaicism (BRTM): Reproductive implications, frequency and mechanism. *European Journal of Medical Genetics*, **2020**, 63, 103639	2.6	4
34	Mutations in CREBBP and EP300 genes affect DNA repair of oxidative damage in Rubinstein-Taybi syndrome cells. *Carcinogenesis*, **2020**, 41, 257-266	4.6	4
33	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	5.7	4
32	A familial t(4;8) translocation segregates with epilepsy and migraine with aura. *Annals of Clinical and Translational Neurology*, **2020**, 7, 855-859	5.3	3
31	Mapping of genes and ESTs assigned to 17q11.2 to a YAC contig centred on the NF1 gene. *GeneScreen*, **2000**, 1, 21-27		3
30	Activation of c-fos and c-fes in metastatic lympho-macrophage hybrids. *International Journal of Cancer*, **1992**, 52, 478-82	7.5	3
29	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	2.6	3
28	Expanding the phenotype associated to KMT2A variants: overlapping clinical signs between Wiedemann-Steiner and Rubinstein-Taybi syndromes. *European Journal of Human Genetics*, **2021**, 29, 88-98	5.3	3
27	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	2	3

26	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	2.6	3
25	Segmental Maternal UPD of Chromosome 7q in a Patient With Pendred and Silver Russell Syndromes-Like Features. *Frontiers in Genetics*, **2018**, 9, 600	4.5	3
24	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
23	Survey of medical genetic services in Italy: year 2011. *BMC Health Services Research*, **2016**, 16, 96	2.9	2
22	Rings and Bricks: Expression of Cohesin Components is Dynamic during Development and Adult Life. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	2
21	Germline mosaicism in Cornelia de Lange syndrome: dilemmas and risk figures. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1825-6	2.5	2
20	Prenatal diagnosis of an extranumerary i(22p) with normal phenotype. *Annales De G□□ique*, **1993**, 36, 154-8		2
19	Prognostic Impact of C-Kit Mutations in Core Binding Factor-Leukemia.. *Blood*, **2004**, 104, 2013-2013	2.2	2
18	8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. *Genes*, **2021**, 12,	4.2	2
17	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
16	Recurrence and Familial Inheritance of Intronic Pathogenic Variant Associated With Mild CdLS. *Frontiers in Neurology*, **2018**, 9, 967	4.1	2
15	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
14	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
13	Genes for RNA-binding proteins involved in neural-specific functions and diseases are downregulated in Rubinstein-Taybi iNeurons. *Neural Regeneration Research*, **2022**, 17, 5-14	4.5	2
12	Delineation and physical separation of novel translocation breakpoints on chromosome 1p in two genetically closely associated childhood tumors. *Cytogenetic and Genome Research*, **2000**, 88, 289-95	1.9	1
11	Rapid construction of competitive templates for quantitative PCR of reverse-transcribed mRNAs.. *Biotechnology Letters*, **1995**, 9, 879-884		1
10	Maternal Uniparental Disomy of Chromosome 20 (UPD(20)mat) as Differential Diagnosis of Silver Russell Syndrome: Identification of Three New Cases. *Genes*, **2021**, 12,	4.2	1
9	Chromosomal instability in fibroblasts and mesenchymal tumors from 2 sibs with Rothmund-Thomson syndrome **1998**, 77, 504		1

8	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	6.3	0
7	Phenotypic Overlap of Roberts and Baller-Gerold Syndromes in Two Patients With Craniosynostosis, Limb Reductions, and Mutations. *Frontiers in Pediatrics*, **2019**, 7, 210	3.4	
6	Rubinstein Taybi Syndrome in an Indian Child due to EP300 Gene Mutation: Correspondence. *Indian Journal of Pediatrics*, **2017**, 84, 91-92	3	
5	Reply to letter by Volpi regarding: RNA processing defects of the helicase gene RECQL4 in a compound heterozygous Rothmund Thomson patient *American Journal of Medical Genetics Part A* , **2004**, 129A, 103-103		
4	On the Parental Origin of the X Chromosomes in a 49,XXXXX Fetus. *Journal of Maternal-Fetal and Neonatal Medicine*, **1994**, 3, 235-238	2	
3	Interconnected Gene Networks Underpin the Clinical Overlap of -Related and Rubinstein-Taybi Intellectual Disability Syndromes. *Frontiers in Neuroscience*, **2021**, 15, 745684	5.1	
2	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,	6.3	
1	12q21 Interstitial Deletions: Seven New Syndromic Cases Detected by Array-CGH and Review of the Literature. *Genes*, **2022**, 13, 780	4.2	