

# Sabrina Rita Giglio

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4289016/publications.pdf>

Version: 2024-02-01

124  
papers

3,986  
citations

117571

34  
h-index

138417

58  
g-index

127  
all docs

127  
docs citations

127  
times ranked

6518  
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic testing in the diagnosis of chronic kidney disease: recommendations for clinical practice. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, 239-254.	0.4	63
2	STOP Pain Projectâ€”Opioid Response in Pediatric Cancer Patients and Gene Polymorphisms of Cytokine Pathways. <i>Pharmaceutics</i> , 2022, 14, 619.	2.0	2
3	A Protective HLA Extended Haplotype Outweighs the Major COVID-19 Risk Factor Inherited From Neanderthals in the Sardinian Population. <i>Frontiers in Immunology</i> , 2022, 13, 891147.	2.2	3
4	Variants Disrupting CD40L Transmembrane Domain and Atypical X-Linked Hyper-IgM Syndrome: A Case Report With Leishmaniasis and Review of the Literature. <i>Frontiers in Immunology</i> , 2022, 13, 840767.	2.2	0
5	Chiari 1 malformation and exome sequencing in 51 trios: the emerging role of rare missense variants in chromatin-remodeling genes. <i>Human Genetics</i> , 2021, 140, 625-647.	1.8	10
6	Leopard-like retinopathy and severe early-onset portal hypertension expand the phenotype of KARS1-related syndrome: a case report. <i>BMC Medical Genomics</i> , 2021, 14, 25.	0.7	2
7	Exclusive Neurogenic Bladder and Fecal Incontinency in an Achondroplastic Child Successfully Treated with Lumbar Foraminal Decompression. <i>Pediatric Neurosurgery</i> , 2021, 56, 471-476.	0.4	0
8	Distal renal tubular acidosis: a systematic approach from diagnosis to treatment. <i>Journal of Nephrology</i> , 2021, 34, 2073-2083.	0.9	20
9	Differential Diagnosis between Marfan Syndrome and Loey'sâ€”Dietz Syndrome Type 4: A Novel Chromosomal Deletion Covering TGFB2. <i>Genes</i> , 2021, 12, 1462.	1.0	2
10	A Novel Splicing Variant of COL2A1 in a Fetus with Achondrogenesis Type II: Interpretation of Pathogenicity of In-Frame Deletions. <i>Genes</i> , 2021, 12, 1395.	1.0	4
11	Prenatal Noninvasive Trio-WES in a Case of Pregnancy-Related Liver Disorder. <i>Diagnostics</i> , 2021, 11, 1904.	1.3	3
12	RB1CC1 duplication and aberrant overexpression in a patient with schizophrenia: further phenotype delineation and proposal of a pathogenetic mechanism. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1561.	0.6	2
13	Expanding the phenotype of Wiedemannâ€”Steiner syndrome: Craniovertebral junction anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2877-2886.	0.7	9
14	Genetic counseling during COVIDâ€”19 pandemic: Tuscany experience. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1433.	0.6	31
15	Variable clinical expression of Stickler Syndrome: A case report of a novel <i>COL11A1</i> mutation. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1353.	0.6	7
16	Clinical correlates in children with autism spectrum disorder and CNVs: Systematic investigation in a clinical setting. <i>International Journal of Developmental Neuroscience</i> , 2020, 80, 276-286.	0.7	6
17	Acute kidney injury promotes development of papillary renal cell adenoma and carcinoma from renal progenitor cells. <i>Science Translational Medicine</i> , 2020, 12, .	5.8	46
18	Novel mutations in <i>MFRP</i> and <i>PRSS56</i> are associated with posterior microphthalmos. <i>Ophthalmic Genetics</i> , 2020, 41, 49-56.	0.5	10

#	ARTICLE	IF	CITATIONS
19	Noninvasive prenatal diagnosis in a family at risk for Fraser syndrome. <i>Prenatal Diagnosis</i> , 2020, 40, 905-908.	1.1	4
20	Reverse Phenotyping after Whole-Exome Sequencing in Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2020, 15, 89-100.	2.2	60
21	Germline mutations and new copy number variants among 40 pediatric cancer patients suspected for genetic predisposition. <i>Clinical Genetics</i> , 2019, 96, 359-365.	1.0	14
22	Clinical and Genetic Profiles of Young Adult Patients with Myelodysplastic Syndromes. <i>Clinical Lymphoma, Myeloma and Leukemia</i> , 2019, 19, S347.	0.2	0
23	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. <i>European Journal of Human Genetics</i> , 2019, 27, 909-918.	1.4	21
24	Opioid response in paediatric cancer patients and the Val158Met polymorphism of the human catechol-O-methyltransferase (COMT) gene: an Italian study on 87 cancer children and a systematic review. <i>BMC Cancer</i> , 2019, 19, 113.	1.1	9
25	A microRNA profile of pediatric glioblastoma: The role of NUCKS1 upregulation. <i>Molecular and Clinical Oncology</i> , 2019, 10, 331-338.	0.4	13
26	Diagnostic implications of genetic copy number variation in epilepsy plus. <i>Epilepsia</i> , 2019, 60, 689-706.	2.6	61
27	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. <i>Human Mutation</i> , 2019, 40, 193-200.	1.1	33
28	Transient Neonatal Diabetes Mellitus in a Very Preterm Infant due to ABCC8 Mutation. <i>AJP Reports</i> , 2018, 08, e39-e42.	0.4	2
29	Diagnostic application of a capture based <sc>NGS</sc> test for the concurrent detection of variants in sequence and copy number as well as <sc>LOH</sc>. <i>Clinical Genetics</i> , 2018, 93, 545-556.	1.0	12
30	Bicuspid Aortic Valve: Role of Multiple Gene Variants in Influencing the Clinical Phenotype. <i>BioMed Research International</i> , 2018, 2018, 1-9.	0.9	6
31	De novo unbalanced translocations have a complex history/aetiology. <i>Human Genetics</i> , 2018, 137, 817-829.	1.8	23
32	A systematic review of the risk factors for clinical response to opioids for all-age patients with cancer-related pain and presentation of the paediatric STOP pain study. <i>BMC Cancer</i> , 2018, 18, 568.	1.1	10
33	Can HbA1c combined with fasting plasma glucose help to assess priority for GCK-MODY vs HNF1A-MODY genetic testing?. <i>Acta Diabetologica</i> , 2018, 55, 981-983.	1.2	14
34	FO057WHOLE-EXOME SEQUENCING FOR PERSONALIZED MANAGEMENT OF IDIOPATHIC NEPHROTIC SYNDROME. <i>Nephrology Dialysis Transplantation</i> , 2018, 33, i43-i43.	0.4	0
35	The genetic and clinical spectrum of a large cohort of patients with distal renal tubular acidosis. <i>Kidney International</i> , 2017, 91, 1243-1255.	2.6	79
36	Chiari I malformation in a child with PTEN hamartoma tumor syndrome: Association or coincidence?. <i>European Journal of Medical Genetics</i> , 2017, 60, 261-264.	0.7	14

#	ARTICLE	IF	CITATIONS
37	Cross-sectional study shows that impaired bone mineral status and metabolism are found in nonmosaic triple X syndrome. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2017, 106, 619-626.	0.7	1
38	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. <i>European Journal of Human Genetics</i> , 2017, 25, 694-701.	1.4	33
39	Metatropic dysplasia in third trimester of pregnancy and a novel causative variant in the TRPV4 gene. <i>European Journal of Medical Genetics</i> , 2017, 60, 365-368.	0.7	2
40	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 671-677.	0.7	25
41	<i>SMARCA4</i> inactivating mutations cause concomitant Coffin-Siris syndrome, microphthalmia and small-cell carcinoma of the ovary hypercalcaemic type. <i>Journal of Pathology</i> , 2017, 243, 9-15.	2.1	47
42	SLMSuite: a suite of algorithms for segmenting genomic profiles. <i>BMC Bioinformatics</i> , 2017, 18, 321.	1.2	3
43	Case report of an atypical early onset X-linked retinoschisis in monozygotic twins. <i>BMC Ophthalmology</i> , 2017, 17, 19.	0.6	12
44	Brain tumors in Li-Fraumeni syndrome: a commentary and a case of a gliosarcoma patient. <i>Future Oncology</i> , 2017, 13, 9-12.	1.1	3
45	GENE-03. MICRORNAS PROFILE IN PAEDIATRIC GBMS. <i>Neuro-Oncology</i> , 2017, 19, iv18-iv18.	0.6	1
46	Monogenic Diabetes Accounts for 6.3% of Cases Referred to 15 Italian Pediatric Diabetes Centers During 2007 to 2012. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 1826-1834.	1.8	88
47	Duplication of FOXP2 binding sites within CNTNAP2 gene in a girl with neurodevelopmental delay. <i>Minerva Pediatrics</i> , 2017, 69, 162-164.	0.2	4
48	Bone Mineral Status in Children and Adolescents with Klinefelter Syndrome. <i>International Journal of Endocrinology</i> , 2016, 2016, 1-9.	0.6	21
49	Bone mineral status and metabolism in patients with Williams-Beuren syndrome. <i>Hormones</i> , 2016, 15, 404-412.	0.9	5
50	Lessons from genetics: is it time to revise the therapeutic approach to children with steroid-resistant nephrotic syndrome?. <i>Journal of Nephrology</i> , 2016, 29, 543-550.	0.9	14
51	Clinical and molecular characterization of a novel INS mutation identified in patients with MODY phenotype. <i>European Journal of Medical Genetics</i> , 2016, 59, 590-595.	0.7	26
52	Next generation sequencing and functional analysis of patient urine renal progenitor-derived podocytes to unravel the diagnosis underlying refractory lupus nephritis. <i>Nephrology Dialysis Transplantation</i> , 2016, 31, 1541-1545.	0.4	11
53	A novel INDEL mutation in the EDA gene resulting in a distinct X-linked hypohidrotic ectodermal dysplasia phenotype in an Italian family. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2016, 30, 341-343.	1.3	5
54	Ruxolitinib is an effective treatment for CALR-positive patients with myelofibrosis. <i>British Journal of Haematology</i> , 2016, 173, 938-940.	1.2	36

#	ARTICLE	IF	CITATIONS
55	Prenatal diagnosis of X-linked adrenoleukodystrophy associated with isolated pericardial effusion. <i>Clinical Case Reports (discontinued)</i> , 2015, 3, 643-645.	0.2	2
56	Determinants of Vitamin D Levels in Children and Adolescents with Down Syndrome. <i>International Journal of Endocrinology</i> , 2015, 2015, 1-11.	0.6	34
57	Human Urine-Derived Renal Progenitors for Personalized Modeling of Genetic Kidney Disorders. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 1961-1974.	3.0	74
58	Heterogeneous Genetic Alterations in Sporadic Nephrotic Syndrome Associate with Resistance to Immunosuppression. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 230-236.	3.0	84
59	Identification of a novel frameshift mutation in the <i>EDAR</i> gene causing autosomal dominant hypohidrotic ectodermal dysplasia. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2015, 29, 1032-1034.	1.3	6
60	Therapeutic implications of novel mutations of the RFX6 gene associated with early-onset diabetes. <i>Pharmacogenomics Journal</i> , 2015, 15, 49-54.	0.9	18
61	Policaptil Gel Retard® significantly reduces body mass index and hyperinsulinism and may decrease the risk of type 2 diabetes mellitus (T2DM) in obese children and adolescents with family history of obesity and T2DM. <i>Italian Journal of Pediatrics</i> , 2015, 41, 10.	1.0	18
62	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. <i>Human Mutation</i> , 2015, 36, 357-368.	1.1	26
63	Comprehensive investigation in patients affected by sperm macrocephaly and globozoospermia. <i>Andrology</i> , 2015, 3, 203-212.	1.9	35
64	Prevalence and prenatal ultrasound detection of clubfoot in a non-selected population: an analysis of 549931 births in Tuscany. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2015, 28, 2066-2069.	0.7	12
65	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. <i>Genetics in Medicine</i> , 2015, 17, 460-466.	1.1	45
66	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. <i>European Journal of Human Genetics</i> , 2015, 23, 1025-1032.	1.4	59
67	Focal dysplasia of the cerebral cortex and infantile spasms associated with somatic 1q21.1q44 duplication including the <i>AKT3</i> gene. <i>Clinical Genetics</i> , 2015, 88, 241-247.	1.0	60
68	Phenotypic heterogeneity and mutational spectrum in a cohort of 45 Italian males subjects with X-linked ectodermal dysplasia. <i>Clinical Genetics</i> , 2015, 87, 338-342.	1.0	16
69	Anti-miR21 oligonucleotide enhances chemosensitivity of T98G cell line to doxorubicin by inducing apoptosis. <i>American Journal of Cancer Research</i> , 2015, 5, 231-42.	1.4	25
70	A SOX3 (Xq26.3-27.3) duplication in a boy with growth hormone deficiency, ocular dyspraxia, and intellectual disability: A long-term follow-up and literature review. <i>Hormones</i> , 2014, 13, 552-60.	0.9	17
71	Characterization of the rs2802292 SNP identifies FOXO3A as a modifier locus predicting cancer risk in patients with PJS and PHTS hamartomatous polyposis syndromes. <i>BMC Cancer</i> , 2014, 14, 661.	1.1	11
72	Reply: Y-chromosome microdeletions are not associated with SHOX haploinsufficiency. <i>Human Reproduction</i> , 2014, 29, 1114-1115.	0.4	0

#	ARTICLE	IF	CITATIONS
73	Long-term auxological and endocrinological evaluation of patients with 9p trisomy: a focus on the growth hormone-insulin-like growth factor-I axis. <i>BMC Endocrine Disorders</i> , 2014, 14, 3.	0.9	12
74	Coeliac disease and risk for other autoimmune diseases in patients with Williams-Beuren syndrome. <i>BMC Medical Genetics</i> , 2014, 15, 61.	2.1	13
75	Genome-wide copy number analysis in pediatric glioblastoma multiforme. <i>American Journal of Cancer Research</i> , 2014, 4, 293-303.	1.4	10
76	Clinical and genetic study of a family with a paternally inherited 15q11-q13 duplication. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1459-1464.	0.7	17
77	Y-chromosome microdeletions are not associated with SHOX haploinsufficiency. <i>Human Reproduction</i> , 2013, 28, 3155-3160.	0.4	13
78	Expression of $\beta$ -adrenergic receptors in pediatric malignant brain tumors. <i>Oncology Letters</i> , 2013, 5, 221-225.	0.8	14
79	Multiorgan Infiltration by CD8+ T Cells and 1p;16p Translocation in a Patient with Hypogammaglobulinemia and a Reduced Number of B Cells. <i>International Archives of Allergy and Immunology</i> , 2012, 158, 206-210.	0.9	2
80	Fabry disease: polymorphic haplotypes and a novel missense mutation in the <i>GLA</i> gene. <i>Clinical Genetics</i> , 2012, 81, 224-233.	1.0	28
81	De Novo Unbalanced Translocations in Prader-Willi and Angelman Syndrome Might Be the Reciprocal Product of inv dup(15)s. <i>PLoS ONE</i> , 2012, 7, e39180.	1.1	5
82	Prenatal manifestation and management of a mother and child affected by spondyloperipheral dysplasia with a C-propeptide mutation in COL2A1: case report. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 7.	1.2	9
83	Constitutional FLCN mutations in patients with suspected Birt-Hogg-Dubé syndrome ascertained for non-cutaneous manifestations. <i>Clinical Genetics</i> , 2011, 79, 345-354.	1.0	36
84	Transient hyperoxaluria in a patient with inherited distal renal tubular acidosis. <i>Pediatric Nephrology</i> , 2011, 26, 323-324.	0.9	2
85	In-frame deletion in <i>FLNA</i> causing familial periventricular heterotopia with skeletal dysplasia in males. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1140-1146.	0.7	12
86	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. <i>PLoS Genetics</i> , 2011, 7, e1002173.	1.5	172
87	Growth hormone therapy-related hyperglycaemia in a boy with renal cystic hypodysplasia and a new mutation of the <i>HNF1A</i> gene. <i>Nephrology Dialysis Transplantation</i> , 2010, 25, 3116-3119.	0.4	4
88	Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H <sup>+</sup> -ATPase genes. <i>Nephrology Dialysis Transplantation</i> , 2009, 24, 2734-2738.	0.4	29
89	Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity. <i>Pediatric Nephrology</i> , 2009, 24, 2147-2153.	0.9	32
90	Type A microsatellite instability in pediatric gliomas as an indicator of Turcot syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 919-927.	1.4	42

#	ARTICLE	IF	CITATIONS
91	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. <i>Neurology</i> , 2009, 72, 784-792.	1.5	110
92	Somatic hypermutability of microsatellite sequences in Turcot syndrome: Implications for forensic genetics. <i>Forensic Science International: Genetics Supplement Series</i> , 2008, 1, 557-558.	0.1	0
93	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. <i>Journal of Medical Genetics</i> , 2007, 44, 750-762.	1.5	244
94	The breakpoint identified in a balanced de novo translocation t(7;9)(p14.1;q31.3) disrupts the A-kinase (PRKA) anchor protein 2 gene (AKAP2) on chromosome 9 in a patient with Kallmann syndrome and bone anomalies. <i>International Journal of Molecular Medicine</i> , 2007, 19, 429.	1.8	4
95	Two classes of low-copy repeats mediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplication at 8p23.2. <i>Human Mutation</i> , 2007, 28, 459-468.	1.1	41
96	13q Deletion and central nervous system anomalies: further insights from karyotype-phenotype analyses of 14 patients. <i>Journal of Medical Genetics</i> , 2006, 44, e60-e60.	1.5	97
97	Inversion Chromosomes. , 2006, , 289-299.		2
98	8.5 Mb deletion at distal 5p in a male ascertained for azoospermia. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 189-192.	0.7	11
99	Reciprocal translocations: a trap for cytogenetists?. <i>Human Genetics</i> , 2005, 117, 571-582.	1.8	54
100	Selective disruption of muscle and brain-specific BPAG1 isoforms in a girl with a 6;15 translocation, cognitive and motor delay, and tracheo-oesophageal atresia. <i>Journal of Medical Genetics</i> , 2004, 41, e71-e71.	1.5	41
101	Inverted duplications: how many of them are mosaic?. <i>European Journal of Human Genetics</i> , 2004, 12, 713-717.	1.4	33
102	Common Long Human Inversion Polymorphism on Chromosome 8p\$. <i>Lecture Notes-monograph Series / Institute of Mathematical Statistics</i> , 2003, , 237-246.	1.0	22
103	Heterozygous Submicroscopic Inversions Involving Olfactory Receptor Gene Clusters Mediate the Recurrent t(4;8)(p16;p23) Translocation. <i>American Journal of Human Genetics</i> , 2002, 71, 276-285.	2.6	185
104	Diabetes mellitus in a girl with thyroid hormone resistance syndrome: a little recognized interaction between the two diseases. <i>Hormones</i> , 2002, 13, 561-7.	0.9	6
105	Gene dosage of the spermidine/spermine N1-acetyltransferase (SSAT) gene with putrescine accumulation in a patient with a Xp21.1p22.12 duplication and keratosis follicularis spinulosa decalvans (KFSD). <i>Human Genetics</i> , 2002, 111, 235-241.	1.8	46
106	Olfactory Receptor Gene Clusters, Genomic-Inversion Polymorphisms, and Common Chromosome Rearrangements. <i>American Journal of Human Genetics</i> , 2001, 68, 874-883.	2.6	338
107	New syndrome of mental retardation, Robin sequence, and brachydactyly. <i>American Journal of Medical Genetics Part A</i> , 2001, 100, 49-51.	2.4	4
108	Inv dup del (1)(pter;q44;q42:) with the classical phenotype of trisomy 1q42-qter. <i>American Journal of Medical Genetics Part A</i> , 2001, 104, 127-130.	2.4	35



#	ARTICLE	IF	CITATIONS
109	Opposite deletions/duplications of the X chromosome: two novel reciprocal rearrangements. <i>European Journal of Human Genetics</i> , 2000, 8, 63-70.	1.4	16
110	Inverted duplications are recurrent rearrangements always associated with a distal deletion: description of a new case involving 2q. <i>European Journal of Human Genetics</i> , 2000, 8, 597-603.	1.4	66
111	CENP-G in neocentromeres and inactive centromeres. <i>Chromosoma</i> , 2000, 109, 328-333.	1.0	26
112	Deletion of a 5-cM Region at Chromosome 8p23 Is Associated With a Spectrum of Congenital Heart Defects. <i>Circulation</i> , 2000, 102, 432-437.	1.6	83
113	Genomic organization and chromosomal localization of the mouse <i>Connexin36</i> (mCx36) gene. <i>Gene</i> , 2000, 251, 123-130.	1.0	30
114	Identification of two paralogous regions mapping to the short and long arms of human chromosome 2 comprising <i>LIS1</i> pseudogenes. <i>Cytogenetic and Genome Research</i> , 1999, 86, 225-232.	0.6	6
115	Transmission of a Fully Functional Human Neocentromere through Three Generations. <i>American Journal of Human Genetics</i> , 1999, 64, 1440-1444.	2.6	113
116	Structure and mutation analysis of the glycogen storage disease type 1b gene. <i>FEBS Letters</i> , 1998, 436, 247-250.	1.3	50
117	Identification and Characterization of a New Human Gene Encoding a Small Protein with High Homology to the Proline-Rich Region of the <i>SH3BGR</i> Gene. <i>Biochemical and Biophysical Research Communications</i> , 1998, 247, 302-306.	1.0	29
118	Characterization of <i>Cxorf5</i> (71-7A), a Novel Human cDNA Mapping to Xp22 and Encoding a Protein Containing Coiled-Coil $\pm$ -Helical Domains. <i>Genomics</i> , 1998, 51, 243-250.	1.3	56
119	A novel pseudoautosomal gene encoding a putative GTP-binding protein resides in the vicinity of the Xp/Yp telomere. <i>Human Molecular Genetics</i> , 1998, 7, 407-414.	1.4	37
120	Agenesis of the corpus callosum with Probst bundles owing to haploinsufficiency for a gene in an 8 cM region of 6q25.. <i>Journal of Medical Genetics</i> , 1998, 35, 1031-1033.	1.5	30
121	Ataxic gait and mental retardation with absence of the paternal chromosome 8 and an <i>idic</i> (8)(p23.3): imprinting effect or nullisomy for distal 8p genes?. <i>Human Genetics</i> , 1997, 99, 766-771.	1.8	25
122	Ring chromosome 13 with loss of the region D13S317-D13S285: Phenotypic overlap with XK syndrome. <i>American Journal of Medical Genetics Part A</i> , 1997, 72, 319-323.	2.4	22
123	Identification and mapping of human cDNAs homologous to <i>Drosophila</i> mutant genes through EST database searching. <i>Nature Genetics</i> , 1996, 13, 167-174.	9.4	177
124	Jumping translocations in acute lymphoblastic leukemia. <i>Cancer Genetics and Cytogenetics</i> , 1995, 80, 80-81.	1.0	20