Sabrina Rita Giglio

List of Publications by Year
in descending order

124 papers	3,986 citations	117625 34 h-index	138484 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. Nephrology Dialysis Transplantation, 2022, 37, 239-254.	0.7	63
2	STOP Pain Projectâ€"Opioid Response in Pediatric Cancer Patients and Gene Polymorphisms of Cytokine Pathways. Pharmaceutics, 2022, 14, 619.	4.5	2
3	A Protective HLA Extended Haplotype Outweighs the Major COVID-19 Risk Factor Inherited From Neanderthals in the Sardinian Population. Frontiers in Immunology, 2022, 13, 891147.	4.8	3
4	Variants Disrupting CD40L Transmembrane Domain and Atypical X-Linked Hyper-IgM Syndrome: A Case Report With Leishmaniasis and Review of the Literature. Frontiers in Immunology, 2022, 13, 840767.	4.8	0
5	Chiari 1 malformation and exome sequencing in 51 trios: the emerging role of rare missense variants in chromatin-remodeling genes. Human Genetics, 2021, 140, 625-647.	3.8	10
6	Leopard-like retinopathy and severe early-onset portal hypertension expand the phenotype of KARS1-related syndrome: a case report. BMC Medical Genomics, 2021, 14, 25.	1.5	2
7	Exclusive Neurogenic Bladder and Fecal Incontinency in an Achondroplasic Child Successfully Treated with Lumbar Foraminal Decompression. Pediatric Neurosurgery, 2021, 56, 471-476.	0.7	0
8	Distal renal tubular acidosis: a systematic approach from diagnosis to treatment. Journal of Nephrology, 2021, 34, 2073-2083.	2.0	20
9	Differential Diagnosis between Marfan Syndrome and Loeysâ€"Dietz Syndrome Type 4: A Novel Chromosomal Deletion Covering TGFB2. Genes, 2021, 12, 1462.	2.4	2
10	A Novel Splicing Variant of COL2A1 in a Fetus with Achondrogenesis Type II: Interpretation of Pathogenicity of In-Frame Deletions. Genes, 2021, 12, 1395.	2.4	4
11	Prenatal Noninvasive Trio-WES in a Case of Pregnancy-Related Liver Disorder. Diagnostics, 2021, 11, 1904.	2.6	3
12	RB1CC1 duplication and aberrant overexpression in a patient with schizophrenia: further phenotype delineation and proposal of a pathogenetic mechanism. Molecular Genetics & Genomic Medicine, 2021, 9, e1561.	1.2	2
13	Expanding the phenotype of Wiedemannâ€Steiner syndrome: Craniovertebral junction anomalies. American Journal of Medical Genetics, Part A, 2020, 182, 2877-2886.	1.2	9
14	Genetic counseling during COVIDâ€19 pandemic: Tuscany experience. Molecular Genetics & Genomic Medicine, 2020, 8, e1433.	1.2	31
15	Variable clinical expression of Stickler Syndrome: A case report of a novel COL11A1 mutation. Molecular Genetics & Genomic Medicine, 2020, 8, e1353.	1.2	7
16	Clinical correlates in children with autism spectrum disorder and CNVs: Systematic investigation in a clinical setting. International Journal of Developmental Neuroscience, 2020, 80, 276-286.	1.6	6
17	Acute kidney injury promotes development of papillary renal cell adenoma and carcinoma from renal progenitor cells. Science Translational Medicine, 2020, 12, .	12.4	46
18	Novel mutations in MFRP and PRSS56 are associated with posterior microphthalmos. Ophthalmic Genetics, 2020, 41, 49-56.	1.2	10

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19	Noninvasive prenatal diagnosis in a family at risk for Fraser syndrome. Prenatal Diagnosis, 2020, 40, 905-908.	2.3	4
20	Reverse Phenotyping after Whole-Exome Sequencing in Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 89-100.	4.5	60
21	Germline mutations and new copy number variants among 40 pediatric cancer patients suspected for genetic predisposition. Clinical Genetics, 2019, 96, 359-365.	2.0	14
22	Clinical and Genetic Profiles of Young Adult Patients with Myelodysplastic Syndromes. Clinical Lymphoma, Myeloma and Leukemia, 2019, 19, S347.	0.4	0
23	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. European Journal of Human Genetics, 2019, 27, 909-918.	2.8	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. BMC Cancer, 2019, 19, 113.	2.6	9
25	A microRNA profile of pediatric glioblastoma: The role of NUCKS1 upregulation. Molecular and Clinical Oncology, 2019, 10, 331-338.	1.0	13
26	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
27	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	2.5	33
28	Transient Neonatal Diabetes Mellitus in a Very Preterm Infant due to ABCC8 Mutation. AJP Reports, 2018, 08, e39-e42.	0.7	2
29	Diagnostic application of a capture based <scp>NGS</scp> test for the concurrent detection of variants in sequence and copy number as well as <scp>LOH</scp>. Clinical Genetics, 2018, 93, 545-556.	2.0	12
30	Bicuspid Aortic Valve: Role of Multiple Gene Variants in Influencing the Clinical Phenotype. BioMed Research International, 2018, 2018, 1-9.	1.9	6
31	De novo unbalanced translocations have a complex history/aetiology. Human Genetics, 2018, 137, 817-829.	3.8	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. BMC Cancer, 2018, 18, 568.	2.6	10
33	Can HbA1c combined with fasting plasma glucose help to assess priority for GCK-MODY vs HNF1A-MODY genetic testing?. Acta Diabetologica, 2018, 55, 981-983.	2.5	14
34	FO057WHOLE-EXOME SEQUENCING FOR PERSONALIZED MANAGEMENT OF IDIOPATHIC NEPHROTIC SYNDROME. Nephrology Dialysis Transplantation, 2018, 33, i43-i43.	0.7	0
35	The genetic and clinical spectrum of a large cohort of patients with distal renal tubular acidosis. Kidney International, 2017, 91, 1243-1255.	5.2	79
36	Chiari I malformation in a child with PTEN hamartoma tumor syndrome: Association or coincidence?. European Journal of Medical Genetics, 2017, 60, 261-264.	1.3	14

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37	Crossâ€sectional study shows that impaired bone mineral status andÂmetabolism are found in nonmosaic triple X syndrome. Acta Paediatrica, International Journal of Paediatrics, 2017, 106, 619-626.	1.5	1
38	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. European Journal of Human Genetics, 2017, 25, 694-701.	2.8	33
39	Metatropic dysplasia in third trimester of pregnancy and a novel causative variant in the TRPV4 gene. European Journal of Medical Genetics, 2017, 60, 365-368.	1.3	2
40	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1. European Journal of Paediatric Neurology, 2017, 21, 671-677.	1.6	25
41	SMARCA4 inactivating mutations cause concomitant Coffinâ€“Siris syndrome, microphthalmia and smallâ€cell carcinoma of the ovary hypercalcaemic type. Journal of Pathology, 2017, 243, 9-15.	4.5	47
42	SLMSuite: a suite of algorithms for segmenting genomic profiles. BMC Bioinformatics, 2017, 18, 321.	2.6	3
43	Case report of an atypical early onset X-linked retinoschisis in monozygotic twins. BMC Ophthalmology, 2017, 17, 19.	1.4	12
44	Brain tumors in Li-Fraumeni syndrome: a commentary and a case of a gliosarcoma patient. Future Oncology, 2017, 13, 9-12.	2.4	3
45	GENE-03. MICRORNAS PROFILE IN PAEDIATRIC GBMS. Neuro-Oncology, 2017, 19, iv18-iv18.	1.2	1
46	Monogenic Diabetes Accounts for 6.3% of Cases Referred to 15 Italian Pediatric Diabetes Centers During 2007 to 2012. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1826-1834.	3.6	88
47	Duplication of FOXP2 binding sites within CNTNAP2 gene in a girl with neurodevelopmental delay. Minerva Pediatrics, 2017, 69, 162-164.	0.4	4
48	Bone Mineral Status in Children and Adolescents with Klinefelter Syndrome. International Journal of Endocrinology, 2016, 2016, 1-9.	1.5	21
49	Bone mineral status and metabolism in patients with Williams-Beuren syndrome. Hormones, 2016, 15, 404-412.	1.9	5
50	Lessons from genetics: is it time to revise the therapeutic approach to children with steroid-resistant nephrotic syndrome?. Journal of Nephrology, 2016, 29, 543-550.	2.0	14
51	Clinical and molecular characterization of a novel INS mutation identified in patients with MODY phenotype. European Journal of Medical Genetics, 2016, 59, 590-595.	1.3	26
52	Next generation sequencing and functional analysis of patient urine renal progenitor-derived podocytes to unravel the diagnosis underlying refractory lupus nephritis. Nephrology Dialysis Transplantation, 2016, 31, 1541-1545.	0.7	11
53	A novel <scp>INDEL</scp> mutation in the <scp>EDA</scp> gene resulting in a distinct Xâ€linked hypohidrotic ectodermal dysplasia phenotype in an Italian family. Journal of the European Academy of Dermatology and Venereology, 2016, 30, 341-343.	2.4	5
54	Ruxolitinib is an effective treatment for <scp>CALR</scp>â€positive patients with myelofibrosis. British Journal of Haematology, 2016, 173, 938-940.	2.5	36

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55	Prenatal diagnosis of Xâ€linked adrenoleukodystrophy associated with isolated pericardial effusion. Clinical Case Reports (discontinued), 2015, 3, 643-645.	0.5	2
56	Determinants of Vitamin D Levels in Children and Adolescents with Down Syndrome. International Journal of Endocrinology, 2015, 2015, 1-11.	1.5	34
57	Human Urine-Derived Renal Progenitors for Personalized Modeling of Genetic Kidney Disorders. Journal of the American Society of Nephrology: JASN, 2015, 26, 1961-1974.	6.1	74
58	Heterogeneous Genetic Alterations in Sporadic Nephrotic Syndrome Associate with Resistance to Immunosuppression. Journal of the American Society of Nephrology: JASN, 2015, 26, 230-236.	6.1	84
59	Identification of a novel frameshift mutation in the <scp>EDAR</scp> gene causing autosomal dominant hypohidrotic ectodermal dysplasia. Journal of the European Academy of Dermatology and Venereology, 2015, 29, 1032-1034.	2.4	6
60	Therapeutic implications of novel mutations of the RFX6 gene associated with early-onset diabetes. Pharmacogenomics Journal, 2015, 15, 49-54.	2.0	18
61	Policaptil Gel RetardÂ® 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. Italian Journal of Pediatrics, 2015, 41, 10.	2.6	18
62	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. Human Mutation, 2015, 36, 357-368.	2.5	26
63	Comprehensive investigation in patients affected by sperm macrocephaly and globozoospermia. Andrology, 2015, 3, 203-212.	3.5	35
64	Prevalence and prenatal ultrasound detection of clubfoot in a non-selected population: an analysis of 549â€‰931 births in Tuscany. Journal of Maternal-Fetal and Neonatal Medicine, 2015, 28, 2066-2069.	1.5	12
65	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
66	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	2.8	59
67	Focal dysplasia of the cerebral cortex and infantile spasms associated with somatic 1q21.1â€q44 duplication including the <scp>AKT3</scp> gene. Clinical Genetics, 2015, 88, 241-247.	2.0	60
68	Phenotypic heterogeneity and mutational spectrum in a cohort of 45 Italian males subjects with Xâ€linked ectodermal dysplasia. Clinical Genetics, 2015, 87, 338-342.	2.0	16
69	Anti-miR21 oligonucleotide enhances chemosensitivity of T98G cell line to doxorubicin by inducing apoptosis. American Journal of Cancer Research, 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. Hormones, 2014, 13, 552-60.	1.9	17
71	Characterization of the rs2802292 SNP identifies FOXO3Aas a modifier locus predicting cancer risk in patients with PJS and PHTS hamartomatous polyposis syndromes. BMC Cancer, 2014, 14, 661.	2.6	11
72	Reply: Y-chromosome microdeletions are not associated with SHOX haploinsufficiency. Human Reproduction, 2014, 29, 1114-1115.	0.9	0

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73	Long-term auxological and endocrinological evaluation of patients with 9p trisomy: a focus on the growth hormone-insulin-like growth factor-I axis. BMC Endocrine Disorders, 2014, 14, 3.	2.2	12
74	Coeliac disease and risk for other autoimmune diseases in patients with Williams-Beuren syndrome. BMC Medical Genetics, 2014, 15, 61.	2.1	13
75	Genome-wide copy number analysis in pediatric glioblastoma multiforme. American Journal of Cancer Research, 2014, 4, 293-303.	1.4	10
76	Clinical and genetic study of a family with a paternally inherited 15q11â€“q13 duplication. American Journal of Medical Genetics, Part A, 2013, 161, 1459-1464.	1.2	17
77	Y-chromosome microdeletions are not associated with SHOX haploinsufficiency. Human Reproduction, 2013, 28, 3155-3160.	0.9	13
78	Expression of Î²-adrenergic receptors in pediatric malignant brain tumors. Oncology Letters, 2013, 5, 221-225.	1.8	14
79	Multiorgan Infiltration by CD8+ T Cells and 1p;16p Translocation in a Patient with Hypogammaglobulinemia and a Reduced Number of B Cells. International Archives of Allergy and Immunology, 2012, 158, 206-210.	2.1	2
80	Fabry disease: polymorphic haplotypes and a novel missense mutation in the GLA gene. Clinical Genetics, 2012, 81, 224-233.	2.0	28
81	De Novo Unbalanced Translocations in Prader-Willi and Angelman Syndrome Might Be the Reciprocal Product of inv dup(15)s. PLoS ONE, 2012, 7, e39180.	2.5	5
82	Prenatal manifestation and management of a mother and child affected by spondyloperipheral dysplasia with a C-propeptide mutation in COL2A1: case report. Orphanet Journal of Rare Diseases, 2011, 6, 7.	2.7	9
83	Constitutional FLCN mutations in patients with suspected Birt-Hogg-DubÃ© syndrome ascertained for non-cutaneous manifestations. Clinical Genetics, 2011, 79, 345-354.	2.0	36
84	Transient hyperoxaluria in a patient with inherited distal renal tubular acidosis. Pediatric Nephrology, 2011, 26, 323-324.	1.7	2
85	Inâ€frame deletion in FLNA causing familial periventricular heterotopia with skeletal dysplasia in males. American Journal of Medical Genetics, Part A, 2011, 155, 1140-1146.	1.2	12
86	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	3.5	172
87	Growth hormone therapy-related hyperglycaemia in a boy with renal cystic hypodysplasia and a new mutation of the HNF1Â gene. Nephrology Dialysis Transplantation, 2010, 25, 3116-3119.	0.7	4
88	Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H+-ATPase genes. Nephrology Dialysis Transplantation, 2009, 24, 2734-2738.	0.7	29
89	Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity. Pediatric Nephrology, 2009, 24, 2147-2153.	1.7	32
90	Type A microsatellite instability in pediatric gliomas as an indicator of Turcot syndrome. European Journal of Human Genetics, 2009, 17, 919-927.	2.8	42

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91	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. Neurology, 2009, 72, 784-792.	1.1	110
92	Somatic hypermutability of microsatellite sequences in Turcot syndrome: Implications for forensic genetics. Forensic Science International: Genetics Supplement Series, 2008, 1, 557-558.	0.3	0
93	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. Journal of Medical Genetics, 2007, 44, 750-762.	3.2	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. International Journal of Molecular Medicine, 2007, 19, 429.	4.0	4
95	Two classes of low-copy repeats comediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplication at 8p23.2. Human Mutation, 2007, 28, 459-468.	2.5	41
96	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
97	Inversion Chromosomes. , 2006, , 289-299.		2
98	8.5 Mb deletion at distal 5p in a male ascertained for azoospermia. American Journal of Medical Genetics, Part A, 2005, 133A, 189-192.	1.2	11
99	Reciprocal translocations: a trap for cytogenetists?. Human Genetics, 2005, 117, 571-582.	3.8	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. Journal of Medical Genetics, 2004, 41, e71-e71.	3.2	41
101	Inverted duplications: how many of them are mosaic?. European Journal of Human Genetics, 2004, 12, 713-717.	2.8	33
102	Common Long Human Inversion Polymorphism on Chromosome $8p$. Lecture Notes-monograph Series / Institute of Mathematical Statistics, 2003, , 237-246.	1.0	22
103	Heterozygous Submicroscopic Inversions Involving Olfactory Receptorâ€“Gene Clusters Mediate the Recurrent t(4;8)(p16;p23) Translocation. American Journal of Human Genetics, 2002, 71, 276-285.	6.2	185
104	Diabetes mellitus in a girl with thyroid hormone resistance syndrome: a little recognized interaction between the two diseases. Hormones, 2002, 13, 561-7.	1.9	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). Human Genetics, 2002, 111, 235-241.	3.8	46
106	Olfactory Receptorâ€“Gene Clusters, Genomic-Inversion Polymorphisms, and Common Chromosome Rearrangements. American Journal of Human Genetics, 2001, 68, 874-883.	6.2	338
107	New syndrome of mental retardation, Robin sequence, and brachydactyly. American Journal of Medical Genetics Part A, 2001, 100, 49-51.	2.4	4
108	Inv dup del (1)(pter?q44::q44?q42:) with the classical phenotype of trisomy 1q42-qter. American Journal of Medical Genetics Part A, 2001, 104, 127-130.	2.4	35

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109	Opposite deletions/duplications of the X chromosome: two novel reciprocal rearrangements. European Journal of Human Genetics, 2000, 8, 63-70.	2.8	16
110	Inverted duplications are recurrent rearrangements always associated with a distal deletion: description of a new case involving 2q. European Journal of Human Genetics, 2000, 8, 597-603.	2.8	66
111	CENP-G in neocentromeres and inactive centromeres. Chromosoma, 2000, 109, 328-333.	2.2	26
112	Deletion of a 5-cM Region at Chromosome 8p23 Is Associated With a Spectrum of Congenital Heart Defects. Circulation, 2000, 102, 432-437.	1.6	83
113	Genomic organization and chromosomal localization of the mouse Connexin36 (mCx36) gene. Gene, 2000, 251, 123-130.	2.2	30
114	Identification of two paralogous regions mapping to the short and long arms of human chromosome 2 comprising LIS1 pseudogenes. Cytogenetic and Genome Research, 1999, 86, 225-232.	1.1	6
115	Transmission of a Fully Functional Human Neocentromere through Three Generations. American Journal of Human Genetics, 1999, 64, 1440-1444.	6.2	113
116	Structure and mutation analysis of the glycogen storage disease type 1b gene. FEBS Letters, 1998, 436, 247-250.	2.8	50
117	Identification and Characterization of a New Human Gene Encoding a Small Protein with High Homology to the Proline-Rich Region of the SH3BGR Gene. Biochemical and Biophysical Research Communications, 1998, 247, 302-306.	2.1	29
118	Characterization ofCxorf5(71-7A), a Novel Human cDNA Mapping to Xp22 and Encoding a Protein Containing Coiled-Coil Î±-Helical Domains. Genomics, 1998, 51, 243-250.	2.9	56
119	A novel pseudoautosomal gene encoding a putative GTP-binding protein resides in the vicinity of the Xp/Yp telomere. Human Molecular Genetics, 1998, 7, 407-414.	2.9	37
120	Agenesis of the corpus callosum with Probst bundles owing to haploinsufficiency for a gene in an 8 cM region of 6q25.. Journal of Medical Genetics, 1998, 35, 1031-1033.	3.2	30
121	Ataxic gait and mental retardation with absence of the paternal chromosome 8 and an idic(8)(p23.3): imprinting effect or nullisomy for distal 8p genes?. Human Genetics, 1997, 99, 766-771.	3.8	25
122	Ring chromosome 13 with loss of the region D13S317-D13S285: Phenotypic overlap with XK syndrome. American Journal of Medical Genetics Part A, 1997, 72, 319-323.	2.4	22
123	Identification and mapping of human cDNAs homologous to Drosophila mutant genes through EST database searching. Nature Genetics, 1996, 13, 167-174.	21.4	177
124	Jumping translocations in acute lymphoblastic leukemia. Cancer Genetics and Cytogenetics, 1995, 80, 80-81.	1.0	20