Caroline Schluth-Bolard

List of Publications by Year
in descending order

46	1,642	393982 19	315357 38
papers	citations	h-index	g-index

49	49	49	3471
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Complete characterisation of two new large Xq28 duplications involving F8 using whole genome sequencing in patients without haemophilia A. Haemophilia, 2022, 28, 117-124.	1.0	4
2	Disruption and deletion of the proximal part of TCF4 are associated with mild intellectual disability: About three new patients. European Journal of Medical Genetics, 2022, 65, 104458.	0.7	3
3	Deciphering balanced translocations in infertile males by next-generation sequencing to identify candidate genes for spermatogenesis disorders. Molecular Human Reproduction, 2021, 27, .	1.3	7
4	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	2.6	108
5	Fryns type mesomelic dysplasia of the upper limbs caused by inverted duplications of the HOXD gene cluster. European Journal of Human Genetics, 2020, 28, 324-332.	1.4	12
6	A 14q distal chromoanagenesis elucidated by whole genome sequencing. European Journal of Medical Genetics, 2020, 63, 103776.	0.7	4
7	A Case of Trisomy 13 Mosaicism Presenting with a Severe Aortic Root Dilatation and Marfanoid Habitus due to an Unpredictable Cytogenetic Mechanism. Cytogenetic and Genome Research, 2020, 160, 72-79.	0.6	1
8	Genome sequencing in cytogenetics: Comparison of shortâ€read and linkedâ€read approaches for germline structural variant detection and characterization. Molecular Genetics & Genomic Medicine, 2020, 8, e1114.	0.6	10
9	Supravalvular Aortic Stenosis Caused by a Familial Chromosome 7 Inversion Disrupting the ELN Gene Uncovered by Whole-Genome Sequencing. Molecular Syndromology, 2019, 10, 209-213.	0.3	4
10	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	1.1	4
11	Molecular investigation, using chromosomal microarray and whole exome sequencing, of six patients affected by Williams Beuren syndrome and Autism Spectrum Disorder. Orphanet Journal of Rare Diseases, 2019, 14, 121.	1.2	5
12	Molecular Characterization of a Familial 13.6-Mb 20p11.1p12.1 Duplication without Clinical Consequence. Cytogenetic and Genome Research, 2019, 157, 141-147.	0.6	0
13	Comment on â€œTrisomy 21 noninvasive prenatal testing for twin pregnanciesâ€• Prenatal Diagnosis, 2019, 39, 571-572.	1.1	1
14	Severe hemophilia A caused by an unbalanced chromosomal rearrangement identified using nanopore sequencing. Journal of Thrombosis and Haemostasis, 2019, 17, 1097-1103.	1.9	10
15	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. Journal of Medical Genetics, 2019, 56, 526-535.	1.5	46
16	Asparagine synthetase deficiency: A novel case with an unusual molecular mechanism. Molecular Genetics and Metabolism Reports, 2019, 21, 100509.	0.4	4
17	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
18	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	2.6	37

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19	Autism spectrum disorder associated with 49,XYYYY: case report and review of the literature. BMC Medical Genetics, 2017, 18, 9.	2.1	14
20	Mosaic variegated aneuploidy syndrome: Case report of two brothers. European Journal of Paediatric Neurology, 2017, 21, e54.	0.7	0
21	Prenatal Diagnosis of Trisomy 2p due to Terminal 2p Duplication including Interstitial Telomeric Sequences. Cytogenetic and Genome Research, 2017, 153, 117-124.	0.6	2
22	Genetic Counselling Pitfall: Co-Occurrence of an 11.8-Mb Xp22 Duplication and an Xp21.2 Duplication Disrupting IL1RAPL1. Molecular Syndromology, 2017, 8, 325-330.	0.3	4
23	A Distinct Class of Chromoanagenesis Events Characterized by Focal Copy Number Gains. Human Mutation, 2016, 37, 661-668.	1.1	30
24	Complete Human and Rat Ex Vivo Spermatogenesis from Fresh or Frozen Testicular Tissue. Biology of Reproduction, 2016, 95, 89-89.	1.2	71
25	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	1.4	36
26	Characterization of a de novo Supernumerary Neocentric Ring Chromosome Derived from Chromosome 7. Cytogenetic and Genome Research, 2015, 147, 111-117.	0.6	3
27	A new syndrome of intellectual disability with dysmorphism due to TBL1XR1 deletion. American Journal of Medical Genetics, Part A, 2015, 167, 164-168.	0.7	37
28	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	1.5	501
29	Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations. Journal of Medical Genetics, 2013, 50, 144-150.	1.5	99
30	Jacobsen and Beckwithâ€“Wiedemann syndromes in a child with mosaicism for partial 11pter trisomy and partial 11qter monosomy. American Journal of Medical Genetics, Part A, 2013, 161, 331-337.	0.7	2
31	Neuropathological features in a female fetus with OPHN1 deletion and cerebellar hypoplasia. European Journal of Medical Genetics, 2013, 56, 270-273.	0.7	10
32	Telomere protection and TRF2 expression are enhanced by the canonical Wnt signalling pathway. EMBO Reports, 2013, 14, 356-363.	2.0	72
33	Interstitial 12p13.1 deletion involving GRIN2B in three patients with intellectual disability. American Journal of Medical Genetics, Part A, 2013, 161, 2564-2569.	0.7	23
34	HBB loss of heterozygosity in the hemopoietic lineage gives rise to an unusual sickle-cell trait phenotype. Haematologica, 2013, 98, e7-e8.	1.7	6
35	Direct Visualization of the Highly Polymorphic RNU2 Locus in Proximity to the BRCA1 Gene. PLoS ONE, 2013, 8, e76054.	1.1	16
36	An 800â€‰kb deletion at 17q23.2 including the MED13 (THRAP1) gene, revealed by aCGH in a patient with a SMC 17p. American Journal of Medical Genetics, Part A, 2012, 158A, 400-405.	0.7	15

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37	Chromosomal Position Effects and Gene Variegation. , 2011, , 77-105.		1
38	17p13.1 microdeletion involving the TP53 gene in a boy presenting with mental retardation but no tumor. American Journal of Medical Genetics, Part A, 2010, 152A, 1278-1282.	0.7	20
39	Replication Timing of Human Telomeres Is Chromosome Armâ€"Specific, Influenced by Subtelomeric Structures and Connected to Nuclear Localization. PLoS Genetics, 2010, 6, e1000920.	1.5	94
40	D4Z4as a prototype of CTCF and lamins-dependent insulator in human cells. Nucleus, 2010, 1, 30-36.	0.6	31
41	Unexpected diagnosis of 45,X/47,XX,+18 mosaicism in a girl with mild phenotype. American Journal of Medical Genetics, Part A, 2009, 149A, 2584-2587.	0.7	2
42	Identification of a perinuclear positioning element in human subtelomeres that requires A-type lamins and CTCF. EMBO Journal, 2009, 28, 2428-2436.	3.5	76
43	Cryptic genomic imbalances in de novo and inherited apparently balanced chromosomal rearrangements: Array CGH study of 47 unrelated cases. European Journal of Medical Genetics, 2009, 52, 291-296.	0.7	89
44	Distal Xq duplication and functional Xq disomy. Orphanet Journal of Rare Diseases, 2009, 4, 4.	1.2	58
45	TWIST microdeletion identified by array CGH in a patient presenting Saethreâ€"Chotzen phenotype and a complex rearrangement involving chromosomes 2 and 7. European Journal of Medical Genetics, 2008, 51, 156-164.	0.7	21
46	Monosomy 19pter and trisomy 19q13-qter in two siblings arising from a maternal pericentric inversion: Clinical data and molecular characterization. European Journal of Medical Genetics, 2008, 51, 622-630.	0.7	2