Chromosome breakage hotspots and delineation of the critical region for the 9p-deletion syndrome

DOI: 10.1086/302606
American Journal of Human Genetics, 1999, 65, 1387-95.

#	Paper	IF	Citations
84	Putting chromosome aberrations on the map. *Trends in Genetics*, **2000**, 16, 420-2	8.5	2
83	Sex-determining gene(s) on distal 9p: clinical and molecular studies in six cases. *Journal of Clinical Endocrinology and Metabolism*, **2000**, 85, 3094-100	5.6	67
82	A new association of congenital hydrocephalus, albinism, megalocornea, and retinal coloboma in a syndromic child: A clinical and genetic study. *Ophthalmic Genetics*, **2000**, 21, 211-216	1.2	6
81	Molecular mechanisms for constitutional chromosomal rearrangements in humans. *Annual Review of Genetics*, **2000**, 34, 297-329	14.5	280
80	Current World Literature. *Current Opinion in Obstetrics and Gynecology*, **2001**, 13, 223-251	2.4	
79	Female gonadal development in XX patients with distal 9p monosomy. *European Journal of Endocrinology*, **2001**, 145, 613-7	6.5	17
78	Two cases with partial trisomy 9p: molecular cytogenetic characterization and clinical follow-up. *American Journal of Medical Genetics Part A*, **2002**, 109, 125-32		22
77	Delimitation of duplicated segments and identification of their parental origin in two partial chromosome 3p duplications. *American Journal of Medical Genetics Part A*, **2002**, 113, 144-50		3
76	Clinical and genetic aspects of trigonocephaly: a study of 25 cases. **2003**, 117A, 127-35		32
75	Gonadoblastoma in a patient with del(9)(p22) and sex reversal: report of a case and review of the literature. *Cancer Genetics and Cytogenetics*, **2003**, 143, 174-7		32
74	Monosomy 1p36 breakpoints indicate repetitive DNA sequence elements may be involved in generating and/or stabilizing some terminal deletions. *Chromosome Research*, **2004**, 12, 133-41	4.4	21
73	Complex chromosome re-arrangement 45,X,t(Y;9) in a girl with sex reversal and mental retardation. *American Journal of Medical Genetics Part A*, **2004**, 124A, 259-62		16
72	Three patients with 9p deletions including DMRT1 and DMRT2: a girl with XY complement, bilateral ovotestes, and extreme growth retardation, and two XX females with normal pubertal development. **2004**, 130A, 415-23		53
71	[Tuberculosis in an area with high incidence and a large percentage of immigrants]. *Anales De Pediatr□*, **2004**, 61, 185-6; author reply 186	0.2	0
70	[Diagnosis of 9p- syndrome at birth. A new case]. *Anales De Pediatr□*, **2004**, 61, 194-6	0.2	
69	Genitourinary phenotype in XX patients with distal 9p monosomy. *Molecular Genetics and Metabolism*, **2004**, 82, 173-9	3.7	13
68	Molecular screening for microdeletions at 9p22-p24 and 11q23-q24 in a large cohort of patients with trigonocephaly. *Clinical Genetics*, **2005**, 67, 503-10	4	46

67	Ionizing radiation and genetic risks XIV. Potential research directions in the post-genome era based on knowledge of repair of radiation-induced DNA double-strand breaks in mammalian somatic cells and the origin of deletions associated with human genomic disorders. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2005**, 578, 333-70	3.3	38
66	Detection of cryptic chromosome aberrations in a patient with a balanced t(1;9)(p34.2;p24) by array-based comparative genomic hybridization. *American Journal of Medical Genetics, Part A*, **2005**, 139, 32-6	2.5	29
65	FISH screening for subtelomeric rearrangements in 219 patients with idiopathic mental retardation and normal karyotype. *European Journal of Medical Genetics*, **2005**, 48, 388-96	2.6	17
64	Genomic Disorders. **2006**,		21
63	Mutational screening of FGFR1, CER1, and CDON in a large cohort of trigonocephalic patients. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 148-51	1.9	14
62	De novo monosomy 9p24.3-pter and trisomy 17q24.3-qter characterised by microarray comparative genomic hybridisation in a fetus with an increased nuchal translucency. *Prenatal Diagnosis*, **2006**, 26, 206-13	3.2	19
61	Breakpoint mapping in a case of mosaicism with partial monosomy 9p23 --> pter and partial trisomy 1q41 --> qter suggests neo-telomere formation in stabilizing the deleted chromosome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 82-7	2.5	13
60	Narrowing candidate region for monosomy 9p syndrome to a 4.7-Mb segment at 9p22.2-p23. *American Journal of Medical Genetics, Part A*, **2006**, 140, 373-7	2.5	41
59	Association of deletion 9p, 46,XY gonadal dysgenesis and autistic spectrum disorder. *Molecular Human Reproduction*, **2007**, 13, 685-9	4.4	54
58	Nasal abnormalities in the 9p deletion syndrome. *JAMA Otolaryngology*, **2007**, 133, 1054-6		1
57	Cz□ciowa monosomia ramienia kr□tkiego chromosomu 9 u siedmiomiesi□cznej dziewczynki □opis przypadku. *Pediatria Polska*, **2007**, 82, 727-730	0.1	
56	Molecular characterisation of a mosaicism with a complex chromosome rearrangement: evidence for coincident chromosome healing by telomere capture and neo-telomere formation. *Journal of Medical Genetics*, **2007**, 44, 250-6	5.8	30
55	Further refinement of the candidate region for monosomy 9p syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2353-6	2.5	30
54	Novel aphidicolin-inducible common fragile site FRA9G maps to 9p22.2, within the C9orf39 gene. *Genes Chromosomes and Cancer*, **2007**, 46, 991-9	5	20
53	Genetic analysis of non-syndromic craniosynostosis. *Orthodontics and Craniofacial Research*, **2007**, 10, 129-37	3	101
52	Partial trisomy and partial monosomy resulting from a reciprocal segregating in a large family. *Indian Journal of Pediatrics*, **2008**, 75, 956-60	3	2
51	Clinical and cytogenetic characterization of 13 Dutch patients with deletion 9p syndrome: Delineation of the critical region for a consensus phenotype. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1430-8	2.5	74
50	Genetics of craniosynostosis: genes, syndromes, mutations and genotype-phenotype correlations. *Frontiers of Oral Biology*, **2008**, 12, 107-143		114

49	Detailed characterization of, and clinical correlations in, 10 patients with distal deletions of chromosome 9p. *Genetics in Medicine*, **2008**, 10, 599-611	8.1	50
48	Molecular mechanisms for subtelomeric rearrangements associated with the 9q34.3 microdeletion syndrome. *Human Molecular Genetics*, **2009**, 18, 1924-36	5.6	55
47	Cytogenetic and array-CGH characterization of a complex de novo rearrangement involving duplication and deletion of 9p and clinical findings in a 4-month-old female. *Cytogenetic and Genome Research*, **2009**, 126, 305-12	1.9	21
46	Investigation of the candidate region for trigonocephaly in a patient with monosomy 9p syndrome using array-CGH. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1076-80	2.5	14
45	Characterization of deletions at 9p affecting the candidate regions for sex reversal and deletion 9p syndrome by MLPA. *European Journal of Human Genetics*, **2009**, 17, 1439-47	5.3	68
44	A de novo unbalanced translocation leading to partial monosomy 9p23-pter and partial trisomy 15q25.3-qter associated with 46,XY complete gonadal dysgenesis, tall stature and mental retardation. *Clinical Dysmorphology*, **2010**, 19, 190-194	0.9	2
43	Chromosomes and the Skin. **2011**, 116.1-116.19		
42	Craniosynostosis and Chromosomal Alterations. *Monographs in Human Genetics*, **2011**, 152-164		4
41	Molecular mechanisms generating and stabilizing terminal 22q13 deletions in 44 subjects with Phelan/McDermid syndrome. *PLoS Genetics*, **2011**, 7, e1002173	6	132
40	Partial 9P Monosomy and Partial 8Q Trisomy by Adjacent 1 Segregation of Balanced Paternal Translocation Revealed by Molecular Karyotyping. *Biotechnology and Biotechnological Equipment*, **2012**, 26, 2773-2776	1.6	
39	Genetic basis of single-suture synostoses: genes, chromosomes and clinical implications. *Childjs Nervous System*, **2012**, 28, 1301-10	1.7	45
38	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. *Nature Genetics*, **2012**, 44, 445-9, S1	36.3	170
37	Cytogenomic characterization of an unexpected 17.6 Mb 9p deletion associated to a 14.8 Mb 20p duplication in a dysmorphic patient with multiple congenital anomalies presenting a normal G-banding karyotype. *Gene*, **2012**, 496, 59-62	3.8	3
36	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
35	Characterization of a complex rearrangement involving duplication and deletion of 9p in an infant with craniofacial dysmorphism and cardiac anomalies. *Molecular Cytogenetics*, **2012**, 5, 31	2	14
34	Chromosome 9p deletion syndrome and sex reversal: novel findings and redefinition of the critically deleted regions. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2266-71	2.5	29
33	7q36 deletion and 9p22 duplication: effects of a double imbalance. *Molecular Cytogenetics*, **2013**, 6, 2	2	7
32	Ring chromosome 9 in a girl with developmental delay and dysmorphic features: case report and review of the literature. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1447-52	2.5	7

31	Independent post-zygotic breaks of a dicentric chromosome result in mosaicism for an inverted duplication deletion 9p and terminal deletion 9p. *European Journal of Medical Genetics*, **2013**, 56, 229-35	2.6	4
30	Normal intelligence and premature ovarian failure in an adult female with a 7.6 Mb de novo terminal deletion of chromosome 9p. *European Journal of Medical Genetics*, **2013**, 56, 458-62	2.6	1
29	Child with deletion 9p syndrome presenting with craniofacial dysmorphism, developmental delay, and multiple congenital malformations. *Case Reports in Genetics*, **2013**, 2013, 785830	0.7	7
28	A Wt1-Dmrt1 transgene restores DMRT1 to sertoli cells of Dmrt1(-/-) testes: a novel model of DMRT1-deficient germ cells. *Biology of Reproduction*, **2013**, 88, 51	3.9	29
27	Patient with terminal 9 Mb deletion of chromosome 9p: refining the critical region for 9p monosomy syndrome with trigonocephaly. *Congenital Anomalies (discontinued)*, **2013**, 53, 49-53	1.1	13
26	Case Report: Opitz C Syndrome with a Rare Chromosomal Abnormality. *International Journal of Human Genetics*, **2014**, 14, 67-71	1	0
25	Case report. Familial balanced translocation leading to an offspring with phenotypic manifestations of 9p syndrome. *Genetics and Molecular Research*, **2014**, 13, 4302-10	1.2	6
24	Visible deletions, duplications and insertions. **2014**, 75-84		
23	Duplication 9p and their implication to phenotype. *BMC Medical Genetics*, **2014**, 15, 142	2.1	23
22	Post-zygotic breakage of a dicentric chromosome results in mosaicism for a telocentric 9p marker chromosome in a boy with developmental delay. *Gene*, **2014**, 533, 403-10	3.8	6
21	Phenotype and genotype in Nicolaides-Baraitser syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 302-14	3.1	51
20	DMRT1 and the road to masculinity. **2015**, 123-174		3
19	Partial trisomy 4q and partial monosomy 9p in a girl with choanal atresia and various dysmorphic findings. *Gene*, **2015**, 568, 211-4	3.8	1
18	Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2662-70	2.5	13
17	Prenatal molecular cytogenetic analysis of a mild dysmorphic fetus with a huge unbalance karyotype involving partial 9p deletion and partial 18q duplication. *Genes and Genomics*, **2016**, 38, 53-57	2.1	
16	Clinical and neuroradiological features of the 9p deletion syndrome. *Childjs Nervous System*, **2016**, 32, 327-35	1.7	13
15	Interstitial 9p24.3 deletion involving only DOCK8 and KANK1 genes in two patients with non-overlapping phenotypic traits. *European Journal of Medical Genetics*, **2016**, 59, 20-5	2.6	10
14	Associations between the clinical findings of cases having submicroscopic chromosomal imbalances at chromosomal breakpoints of apparently balanced structural rearrangements. *Gene Reports*, **2017**, 7, 50-58	1.4	1

13	Chromatin Remodeling BAF (SWI/SNF) Complexes in Neural Development and Disorders. *Frontiers in Molecular Neuroscience*, **2017**, 10, 243	6.1	88
12	Neonatal hyperinsulinemic hypoglycemia in a patient with 9p deletion syndrome. *European Journal of Medical Genetics*, **2018**, 61, 473-477	2.6	3
11	A 46,XY Female with a 9p24.3p24.1 Deletion and a 8q24.11q24.3 Duplication: A Case Report and Review of the Literature. *Cytogenetic and Genome Research*, **2019**, 158, 74-82	1.9	3
10	Precise breakpoint detection in a patient with 9p- syndrome. *Journal of Physical Education and Sports Management*, **2020**, 6,	2.8	2
9	Anaesthetic concerns in an infant with a rare genetic condition; chromosome 9p22 deletion syndrome. *Indian Journal of Anaesthesia*, **2015**, 59, 516-7	1.5	1
8	A rare familial rearrangement of chromosomes 9 and 15 associated with intellectual disability: a clinical and molecular study. *Molecular Cytogenetics*, **2021**, 14, 47	2	
7	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1829	2.3	2
6	Diversity of human chromosome structural rearrangements identified at the Center for Medical Genetics in 2002-2007. *Biologija (Vilnius, Lithuania)*, **2008**, 54, 27-32	1.8	
5	A Case of Partial Short Arm Deletion in Chromosome 9 with Inguinal Hernia, Testicular Cystic Lesion, and Arthrogryposis Multiplex Congenita. *Neonatal Medicine*, **2017**, 24, 88	0.2	1
4	A case of 9p deletion syndrome with congenital infantile glaucoma, effective method of diagnosis, and treatment. *International Journal of Ophthalmology*, **2017**, 10, 318-320	1.4	
3	Monosomy 1p36 As a Model for the Molecular Basis of Terminal Deletions. **2006**, 301-314		1
2	From karyotypes to precision genomics in 9p deletion and duplication syndromes.. *Human Genetics and Genomics Advances*, **2022**, 3, 100081	0.8	
1	Insights into the Cardiac Phenotype in 9p Deletion Syndrome: A Multicenter Italian Experience and Literature Review. **2023**, 14, 146		0