Eleven new cases of del(9p) and features from 80 cases

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SUMMARY We report 11 cases of del(9p) and review 69 previously published ones. Of the 80 cases, 39 have a del(9p) as the sole anomaly. The symptoms are typical and diagnosis should be suspected at birth. The sex ratio does not appear to be unbalanced. A cardiac murmur is often present but surgery is rarely necessary. Mean IQ is 48. The number of reported cases with an associated trisomy has previously been underestimated. Death in infancy, owing mainly to gross visceral malformations, occurs more often in cases of del(9p) with another unbalanced chromosome segment (16/41) than in cases of del(9p) as the sole anomaly (1/39).

Over 30 cases of del(9p) have been reviewed recently, the majority being de novo. We report 11 new cases of which seven have del(9p) as the sole anomaly and four have del(9p) with another unbalanced chromosome segment. In each of these two groups of patients, the order in which our cases are presented is a function of the length of the deleted segment, starting with the longest. It appears that dysmorphic features do not differ according to the length of the deletion. We summarise the features from 80 cases (table).

Patients and methods

In the following descriptions, only features which are not included in the appendix will be described. A diagram showing the breakpoints on chromosome 9 is shown in fig 1. Each karyotype was performed with RHG banding except in case 2 where GTG banding was used.

CASE 1*: 46,XX,del(9)(p21)

The proband (fig 2A) was the first child of healthy parents. Delivery was induced at 43 weeks' gestation.

CASE 2: 46,XY,del(9)(p22)

The parents of the proband had previously had 11 children, three of whom died in early infancy without any detectable malformation. There was apparent hypertelorism but a decreased interorbital

distance was ascertained by radiography, small alae nasi, truncal hypotonia, and hypertonia of the extremities. Numerous seizures occurred but repeated electroencephalography did not show any anomaly. Surgery for craniostenosis was performed at the age of nine months. At the age of two years three months, dysmorphic features were unchanged (fig 2B). The patient understood simple requests but did not speak.

FIG 1 Breakpoints on chromosome 9p in present cases (left, cases 1 to 7: del(9p) as the sole anomaly; right, cases 8 to 11: del(9p) with another unbalanced segment).

*Cases cited in reference 56.

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TABLE Main features from 80 cases of del (9p).

9p-	As the sole anomaly (39 cases)		With other unbalanced segment (41 cases)	
	No of cases or mean value	No of informative cases	No of cases or mean value	No of informative cases
Epidemiology				
De novo		34/34		2/40
Inherited (maternal)		0/34		25/40
(paternal)		0/34		13/40
Breakpoint p24	0	n=39*	23	n=38
p23	3		4	
p22	25		9	
p21	5		2	
p13	4		0	
p12	1		0	
Sex ratio	15 M/24 F	n=39	15 M/26 F	n=41
Maternal age (y)	28.7	(n=31)	25.2	(n=26)
Paternal age (y)	31.5	(n=30)	27.9	(n=20)
Family history				
Abnormal pregnancy		1/30		15/31
		5/28		2/26
Birth				
Normal term		26/29		25/27
Weight (kg)	3.2	(n=27)	2.9	(n=29)
Length (cm)	49	(n=11)	48	(n=17)
Head circumference (cm)	34.4	↓ 6/13	33.0	↓ 7/18
Apgar	9	(n=7)	5	(n=6)
Mental development				
Delay		36/36		30/30
Abnormal EEG		2/9		4/8
Hypertonia	7	n=22	4	n=19
Hypotonia	12		14	
Cranium				
Trigonocephaly/prominent forehead		32/32		21/32
Flat occiput		10/11		5/10 (prominent occiput 4/10)
Mid face hypoplasia		6/7		4/6
Eyes				
Upward slanting	26	n=34	12	n=23
Downward slanting	5		7	
Epicanthus		22/27		7/14
Hypertelorism	21	n=25	12	n=22
Apparent hypertelorism	3		1	
Hypotelorism	0		5	
Small palpebral fissures		14/14		6/8
Highly arched eyebrows		9/12		8/15
Nose				
Flat bridge		27/28		20/28 (prominent bridge 6/28)
Anteverted nostrils		30/32		10/17
Mouth				
Long philtrum		32/32		26/32
Small mouth		8/11		9/20 (large mouth 5/12)
High arched and/or cleft palate		25/27		23/25
Micro/retrognathia		26/30		18/21
Ears				
Low set		27/30		23/24
Abnormal lobuli		17/21		9/10
Abnormal auricles		9/11		5/6
Neck				
Short/broad		26/27		25/26
Webbing		15/18		8/11
Low hair line		12/12		8/8
Trunk				
Widely spaced nipples		31/31		14/14
Stiff joints, scoliosis		9/21		8/22
Hernia		9/35		14/24
Cardiac murmur/cardiopathy		16/35		14/24
Abnormal external genitalia		15/36		10/24
Hand/foot				
Abnormal calcaneum		9/28		11/18
Dolichomesophalangy		22/23		16/19
Extra finger flexion crease		10/15		0/3
Thumbs proximally placed		6/7		0/4
Number of whorls	6.3	↑ 18/22	4.5	↑ 2/17
Increased total ridge count		8/10		2/3
Distal axial triradius		9/14		13/16
Simian crease		5/18		11/17
Square, hyperconvex nails		18/19		5/6
Hypoplastic ridges		4/17		2/5

Early reports (refs 1 to 3) have not been included in this review since no banding was available, and it cannot be ascertained if del(9p) was the sole anomaly. Cases of del(9p) as the sole anomaly were reported in refs 4 to 28 and cases of del(9p) associated with another unbalanced segment in refs 5 and 29 to 54. Present cases are included. *In one case, the deletion was interstitial: del(9)(p13.3→p23). ↑ = increased. ↓ = decreased.

Note: Other unbalanced segments were: dup(2)(q31→qter); dup(2)(q33→qter); dup(3)(q25→qter); dup(4)(p14→pter); dup(4)(q23→qter); dup(5)(p13→pter); dup(5)(p7); dup(5)(q31→qter); dup(5)(q32→qter); dup(6)(p21→pter); dup(7)(q21→qter); dup(7)(q31→qter); dup(10)(q25→qter); dup(10)(qter); dup(12)(p11→pter); dup(12)(q21→qter); dup(12)(qter); dup(13)(q21→qter); dup(13)(q22→qter); dup(15)(qter); dup(16)(q13→qter); dup(16)(qter); dup(18)(q12→qter); dup(20)(p?); del(9)(q00→q21); del(15)(pter→q11); del(21)(q11 or 21→qter); t(9;13)(p22;q34) apparently balanced.

FIG 2 A, B, C Patients 1 to 3: cases of *del(9p)* as the sole anomaly. Arrow: dolichomesophalangy with extra finger flexion crease.

CASE 3: 46,XX,*del(9)(p22)*

The proband (fig 2C) was the first child of healthy parents.

CASE 4*: 46,XX,*del(9)(p22)*

A girl with seven healthy sibs was referred to us at the age of one year four months (fig 2D). She presented with microcephaly, turriccephaly, and triangular shaped auditory canals. Her bone age was advanced (two years at one year four months).

CASE 5: 46,XX,*del(9)(p23)*

A girl with four healthy older sibs was referred to us when she was four years old. There were absent lacrimal ducts, deafness, kyphoscoliosis, and hypoplasia of the labia majora (fig 2E).

CASE 6: 46,XX,*del(9)(p23)*

A girl (fig 2F) with one healthy sib was born after a pregnancy complicated by hydramnios. A cardiac murmur was present but no anomalies were detected with ultrasound. She rapidly developed respiratory distress and was transferred to the intensive

care unit. Radiography ascertained choanal atresia. Surgery was performed but the patient died at the age of two months from respiratory failure.

CASE 7: 46,XX,*del(9)(p23)*

A four year old girl with two normal sibs was referred to us to confirm a probable trisomy 21. Clinical examination showed a moon shaped face, flat occiput, epicanthus, low nasal bridge, but also trigonocephaly, long philtrum, widely spaced nipples, dolichomesophalangy, proximally placed thumbs, and five whorls (fig 2G).

CASE 8*: 46,XY,-9,+*der(9),t(9;12)(p21;q24)*mat

The proband was born after an uneventful pregnancy. There had been a previous miscarriage. At delivery, an infarction of the placenta was noted. The proband presented with trigonocephaly and turriccephaly, hypoplastic alae nasi, narrow ear canals, bilateral hexadactyly, hypospadias, and deafness (fig 2H). Numerous episodes of pneumonia occurred. At the age of two months an advanced bone age

FIG 2 D, E, F, G Patients 4 to 7: cases of *del(9p)* as the sole anomaly. Arrow: *dolichomesophalangy* with extra finger flexion crease.

was noted. He died from pneumonia at seven months. Necropsy showed a coarctation of the aorta. The karyotype showed monosomy 9p21→pter and trisomy 12q24→qter.

CASE 9†: 45,XX,-9,-21,+der(9),t(9;21)(p21 or 22;q11 or 21)

The proband was the first child of healthy parents. Her Apgar scores were 8 at one minute and 4 at five minutes. Clinical examination (fig 2I) showed a high, flat forehead, scaphocephaly, microphthalmos, very marked nasolabial furrow, slight prognathism of the upper maxillary bone, vermilion border of the upper lip visible only at the level of the cupid's bow, normal philtrum, crux cimbae and adherent lobuli, narrow auditory canals, small nails, lack of flexion

crease on the third and fourth fingers, camptodactyly of the fifth finger, bilateral talipes valgus, no whorls, seven archs, umbilical and inguinal herniae, and a cardiac murmur. She died at the age of two years. Necropsy showed a ventricular septal defect and malformed kidneys. The karyotype showed monosomy 9p21 or 22→pter and monosomy 21pter→q11 or 21.

CASE 10*: 46,XX,-9,+der(9),t(5;9)(q32;p23)pat

The proband (fig 2J) was born at 32 weeks of pregnancy. Her parents had previously had five miscarriages. Growth retardation was noted one week before birth. There was a severe cardiac anomaly. The anus was anteriorly placed with a cleft towards the vagina. She died suddenly at the age of three months. Necropsy showed hypertrophy of the right ventricle, hypoplastic pulmonary arteries, a

†Case partially reported in reference 55.

FIG 2 H, I, J, K Patients 8 to 11: cases of *del(9p)* with another unbalanced segment.
 H: *del(9)(p21→pter)* and *dup(12)(q24→qter)*;
 I: *del(9)(p21→pter)* and *del(21)(pter→q21)*;
 J: *del(9)(p23→pter)* and *dup(5)(q32→qter)*;
 K: *del(9)(p24→pter)* and *dup(7)(q21→qter)*.

transposed aorta, a large ventricular septal defect (Kirklin type III), and a large ostium secundum (pentology of Fallot). Brain examination revealed diffuse micropolygyria. The karyotype showed monosomy 9p23→pter and trisomy 5q32→qter.

CASE 11: 46,XX,-9,+der(9),t(7;9) (q21;p24)pat

The proband was born after an uneventful pregnancy and appeared to be dead at birth. The first cry occurred at three minutes. Apgar score was 10 at five minutes. The parents had had two healthy children and one miscarriage. Clinical examination showed a flat face, generalised hypertrichosis, high forehead with frontal bossing, scaphocephaly and cranial asymmetry, large fontanelles, exophthalmos, partial syndactyly, simian crease, kyphoscoliosis, and hypertrophy of the labia minora (fig 2K). She died on day 11. The karyotype showed monosomy 9p24→pter and trisomy 7q21→qter.

Discussion

PURE 9p DELETION

A total of 39 cases with 9p- as the sole anomaly is known (main features and references are shown in the table). All these cases are de novo. The breakpoint usually occurs in 9p22. Dysmorphic features, even when carefully analysed, do not seem to differ with the breakpoint or the length of the deleted segment, as has been described in some other deleted or trisomic segments.^{57 58} The sex ratio which had previously been reported to be unbalanced (two or three females to one male)^{10 33} appears to be 1.6/1 (χ^2 test, NS). Mean parental ages at birth are 28.7 (SD 6.5) (mother) and 31.5 (SD 8.2) (father). Family history is negative, pregnancy uneventful, and birth weight is 3.2 kg (SD 0.6). Clinical signs are typical with trigonocephaly, upward slanting palpebral fissures (usually more pronounced than in cases of trisomy 21), long

philtrum, nose with stunted tip, short, broad, and webbed neck with low hair line, excess of whorls on the fingers, dolichomesophalangy with extra finger flexion creases (countertype of trisomy 9p syndrome), and the diagnosis should be suspected at birth. Other signs, although often present, are not specific for this condition (epicanthus, hypertelorism, microretrognathia, low set ears, abnormal tone, etc). A cardiac murmur is often present but cardiac surgery is rarely necessary. Herniae and minor abnormalities of the external genitalia and scoliosis or kyphosis are often encountered. After infancy, pyramidal tract signs, epilepsy, strabismus, nystagmus, and dental anomalies can occur (reported in three or four cases each). Mean IQ is 49 (SD 13) (range 33 to 73, $n=14$). Social adaptation is often good. One case has been diagnosed at the age of 61.

9p DELETION WITH ANOTHER UNBALANCED CHROMOSOME SEGMENT

The number of del(9p) cases associated with a partial trisomy has been underestimated in the past since cases are often reported as 'a case of trisomy...'. We reviewed 41 cases of 9p- with an associated unbalanced chromosome segment, which is approximately half of the total number of cases. Most of them are inherited from a balanced translocation carrier (mother 25 cases, father 13 cases). The breakpoint can occur at 9p22 (nine out of 38 cases), the most common site in pure 9p-, but occurs mainly at 9p24 (23 out of 38 cases). This breakpoint has not been described in cases where 9p- is the sole anomaly, which may in part be because of the difficulty of karyotypic ascertainment of such a terminal deletion when isolated. Sex ratio appears to be 1.7 females/l male (χ^2 test, NS)*. Mean parental ages at birth are 25.2 (SD 3.7) (mother) and 27.9 (SD 4.3) (father). A family history of miscarriages or previous malformed children is present in half of the cases. The pregnancy is usually uneventful: mean birth weight is 2.9 kg (SD 0.6). Mean IQ is 46 (SD 9) (range 33 to 57, $n=7$). The associated unbalanced chromosome segment is variable (table) but four trisomy 13q, three trisomy 12q, three trisomy 10q, two trisomy 2q, two trisomy 5p, two trisomy 5q, two trisomy 7q, and two trisomy 16q were found in unrelated patients. Breakpoints on the trisomic segment are variable apart from 13q21 or 22 (four cases), 10q25 or 26 (three cases), 5q31 or 32, and 12q24 (two cases each). Trigonoccephaly, long philtrum, dolichomesophalangy, and hernia are often found in cases of del(9p)

*When cases of 9p- as the sole anomaly and cases with another unbalanced segment are pooled, a significantly unbalanced sex ratio appears. However, as the chromosomal mechanism is different, it does not seem valid to pool these two groups of patients.

with another unbalanced segment. However, the phenomenon of epistasis is difficult to ascertain in certain cases since it cannot be based on objective investigations and measurements. It should be noted that 16 del(9p) patients with another unbalanced segment and only one patient with del(9p) as the sole anomaly have been reported to die in infancy.

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APPENDIX Present cases of *del(9p)*.

	As the sole anomaly										With other unbalanced segment				
	Case 1	Case 2	Case 3	Case 4	Case 5	Case 6	Case 7	Case 8	Case 9	Case 10	Case 11				
Epidemiology															
De novo	+	+	+	+	+	+	+	+	+	+	+				
Inherited (maternal)															
Inherited (paternal)															
Breakpoint p24															
p23		+	+	+	+	+	+	+	+	+	+				
p22															
p21															
p13															
p12															
Sex															
Maternal age (y)	F 26	M 44	F 23	F 31	F 34	F 22	F 36	M 20	F 32	F 35	F 35				F 28
Paternal age (y)	30	51	27	34	22	23	33	28	34	35	35				27
Family history	-	-	-	-	-	-	-	-	-	+	+				-
Abnormal pregnancy	+	-	-	-	-	-	-	-	-	-	-				-
Birth															
Term (N: normal)	43	40	N	N	N	3-3	3-3	39	40	32	32				39
Weight (kg)	3.8	3.1	3-2	4.5	50	51	3-3	2.5	2.2	1.2	1.2				1.8
Length (cm)	48	48	50	37	35	31	35	50	48	35	35				42
Head circumference (cm)	37	37	10	60	40	10	73	32	32	27	27				31
Apgar		10						5	8						0
Menial development															
Delay (or DO or IQ)	+	+	37	-	40	+	73	50	-	+	+				
Abnormal EEG		-			-	-		-							
Hypertonia					+	+		+							
Hypotonia			-		-	-		+							
Cranium															
Trigonocephaly/prominent forehead	+	+	+	+	+	+	+	+	-	+	+				+
Flat occiput	+	+	+	+	+	+	+	-	+						-
Mid face hypoplasia	+	+	+	+	+	+	+	-	+						-
Eyes															
Upward slanting	+	+	+	-	+	-	-	+	-	+	+				+
Downward slanting	-	+	+	+	+	+	+	-	-	-	-				+
Epicanthus	+	+	+	+	+	+	+	+	+	+	+				+
Hypertelorism	+	-	-	-	+	+	+	+	-	-	-				+
Hypotelorism	-	-	-	-	+	+	+	+	-	-	-				+
Small palpebral fissures	+	-	+	+	+	+	+	+	-	+	+				+
Highly arched eyebrows	+	-	+	+	-	-	-	+	-	+	+				+
Nose															
Flat bridge	+	+	+	+	+	+	+	+	+	+	+				+
Anteverted nostrils		+	+	-	+	+	+	+	+	+	+				-
Mouth															
Long philtrum	+	+	+	+	+	+	+	+	+	+	+				-
Small mouth		+	+	+	+	+	+	+	-	+	+				+
High arched and/or cleft palate		+	+	+	+	+	+	+	-	+	+				+
Micro/retrognathia	+	-	+	+	+	+	+	+	+	+	+				-
Ears															
Low set	+	+	+	+	+	+	+	+	+	+	+				+
Adherent lobuli	+	+	+	+	+	+	+	+	+	+	+				+
Abnormal auricles		-							+						+

continued

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APPENDIX Present cases of *del*(9p)—continued.

	As the sole anomaly					With other unbalanced segment					
	Case 1	Case 2	Case 3	Case 4	Case 5	Case 6	Case 7	Case 8	Case 9	Case 10	Case 11
Neck											
Short/broad	+	-	+	+	+	+	-	+	+	+	+
Webbing	+	-	+	+	+	-	+	+	+	+	+
Low hair line		-									
Trunk											
Widely spaced nipples	+	+		+	+	+	+	+		+	+
Stiff joints, scoliosis		-									+
Hernia	-	-	-	-	-	-	-	+	+	-	-
Cardiac murmur/cardiopathy	-	-	+	-	+	-	-	+	+	+	+
Abnormal external genitalia	-	+	-	-	+	-	-	+	-	+	+
Hand/foot											
Abnormal calcaneum		+	+	-	+	+	-	+	+		
Dolichomesophalangy		+	-								
Extra finger flexion crease	+	+		+	+	+	+	-			
Thumbs proximally placed		+		+	+	+	+				
Square hyperconvex nails		-		+	+	+	+				
Number of whorls	10	8		10	9		5	5	0	10	
Distal axial triradius	+	-		+	-	-	-	-	+	+	+
Simian crease	+	-	-	+	-	-	-	-	+	+	+
Hypoplastic ridges	-	+	-	-				-	-	+	+