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Interstitial deletion of chromosome 10q23: a new case and review

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Abstract

A new case of a deletion of 10q23 is described. Only two other deletions involving this region have been previously noted. A review of clinical features of these three children did not show a distinct pattern of dysmorphic features. Other interstitial deletions of 10q are listed.

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Few interstitial deletions of the long arm of chromosome 10 have been noted. We describe a new case presenting with microcephaly and developmental delay. The deletion of 10q23 of our patient is smaller than any reported previously. The features of others are reviewed but there does not appear to be a specific pattern of dysmorphic features associated with a deletion of this chromosomal region.

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Case report

Developmental delay was observed in the proband, a white male, at 8 months. Chromosome analysis was normal initially but with high resolution banding an interstitial deletion of 10q was detected. His karyotype is 46,XY, del(10)(q22.3q23.2) or (q23.2q24.1) (fig 1). The banding pattern in this region does not allow discrimination between these two bands. Both parents had normal chromosomes. A permanent cell line has not been established.

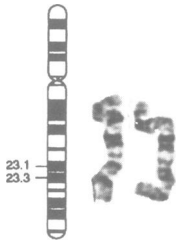

Figure 1 Partial karyotype with del(10) on the left.

His mother consumed a maximum of two drinks of alcohol per week throughout pregnancy. At term, birth weight was 3650 g (75th centile) and OFC was < 3rd centile. The neonatal course was complicated by hypoglycaemia treated briefly by intravenous glucose. A cardiac murmur was audible although an echocardiogram was normal. Early feeding was difficult with frequent gagging and vomiting. By 9 months he was rolling but not sitting. He could reach for toys but was not transferring objects from hand to hand. The OFC was 38 cm (<<3rd centile), height was on the 3rd centile, and weight was >3rd centile. Head shape was dolichocephalic, with a posteriorly sloping forehead and hypoplastic midface, prominent maxilla, long philtrum, and small chin (fig 2). Palpebral fissures were upward slanting while the inner canthal distance was broad (40th centile). The nares were anteverted. He had a wide mouth with downturned corners, thickened alveolar ridges, and a short tongue frenulum. Peg shaped teeth were seen. The nipples were hypoplastic and there was a small umbilical hernia. Penile length was short (<<3rd centile) and both testes were palpable. All toe nails were small. Although distally tone was significantly increased, proximally he was hypotonic. No other dysmorphic features were seen.

Normal investigations included a skull radiograph, head ultrasound, bone age, audiological and ophthalmological assessments, mucopolysaccharide and oligosaccharide screens, and urinary amino acid and organic acid screens. He is one of four sibs born to non-consanguineous parents. His sister and two brothers are well.

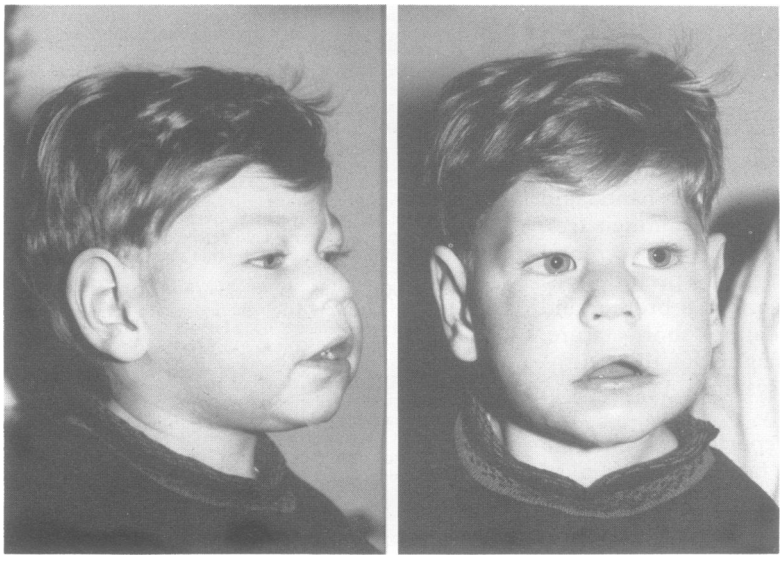

Figure 2 Facial appearance at 28 months.

Discussion

Reports of 10q interstitial deletions are summarised in tables 1 and 2. Three, including this case, involve 10q23.[1,2] The de novo deletion of our patient is the smallest, involving 10q23.1 or 10q23.3. As these two bands are very similar in appearance, it is not possible to determine which is deleted. Non-specific features in common in these three children include postnatal onset of short stature, relative hypertelorism, and developmental delay. Some similarities of the facial area in two of three cases are anteverted nares, long philtrum, wide mouth with downturned corners, ocular anomalies, posi-

Table 1 Interstitial deletions of 10q23.

Characteristic	Mori *et al*[1]	Shapiro *et al*[2]	This report
Karyotype	46,XY,del(10)(q23) (de novo)	46,XX,del(10)(q23) (de novo)	46,XX,del(10)(q23) (de novo)
Height (centile)	< 3rd	3rd	3rd
Head circumference (centile)	98th	< 3rd (> 90th at birth)	< < 3rd
Head			
Prominent occiput		+	–
Broad forehead	+	+	–
Shallow supraorbital ridges		+	Prominent
Low set ears	+	+	–
Ocular region			
Epicanthic folds		+	+
Relative hypertelorism	+	+	+
Other	Glaucoma	Iris coloboma	
Mouth wide, downturned	+		+
Nose small, anteverted		+	+
Palate			
High arch	+		+
Bifid uvula	+		–
Scoliosis or kyphosis	+	+	–
Extremities	Equinovarus 1–2 toe gap	Tapered fingers, proximally placed thumbs, overlapping toes, clinodactyly	–
Nervous system			
Hypotonia	+		Abnormal
Developmental delay	+		+
Abdomen			
Umbilical hernia		+	+
Other	Urinary reflux	Rectal prolapse, sensorineural hearing loss, ptosis, frontal hairline upswept	Peg shaped teeth, small chin, dolichocephaly, small penis

+ = present, – = absent.

Table 2 Other 10q deletions.

Author	Deletion	Height (centile)	Head circumference (centile)	Ocular region	Extremities	Developmental delay	Other
Ray *et al*[3]	q21	45th	50th	Epicanthic folds, hypertelorism	Abnormal palmar creases	+	Short sternum, ichthyosis, small penis
Davis *et al*[4]	q21q22 de novo	N	N	Telecanthus, strabismus		+	Hypotonia
Van de Vooren *et al*[5]	q24.2q25.3(mat)	50th	< 10th	Hypertelorism, stabismus	Pes planovalgus, clinodactyly	+	Sparse eyebrows, cryptorchidism
Holden and Macdonald[6]	q11.2q21 de novo	10th	75th	Ptosis	N	+	Hypotonia, seizures
Glover *et al*[7]	q21.2q22.1 de novo	< 3rd	< 3rd	Telecanthus		+	Plagiocephaly, cutis laxa, hypotonia
Rooney *et al*[8]	q25.2q26.1 de novo			Hypertelorism		+	Microbrachycephaly, hypotonia
Durum and Lawce[9]	q11.2q21.2			Cataract			
Fryns *et al*[10]	q11.23q21.2 de novo	< 3rd	< 3rd				Cockayne syndrome diagnosed as young adult
Curry and Zorn[11]	q21.2q22.1 de novo					+	
Derksen *et al*[12]	q11.2q21.1(mat)						Normal mother and grandmother with same deletion
Lobo *et al*[13]	q11.2q22.1 de novo	N	N	Strabismus, hypertelorism	Digitalised thumbs	+	Hypotonia, prominent philtrum

+ = present, N = normal.

tional ear anomalies, epicanthic folds, broad forehead, and microcephaly. In particular the shape of the mouth of the case of Mori *et al*[1] resembled our patient. An umbilical hernia, sacral dimple, and scoliosis or kyphosis were noted in two of three reports.[1 2] There are no strikingly unusual features present in all three. Phenotypic variability could result from different breakpoints, causing loss or disruption of diverse genes, alternate alleles, position effect, and perhaps imprinting. However, this region has not been noted to have a mouse homologue where imprinting has been suspected.[14] As there are so few cases of deletions of this region, it is difficult to say whether there is a recognisable pattern of features.

Relatively few other deletions of the long arm of chromosome 10 have been reported. These are listed in table 2. These deletions have variable breakpoints, spanning much of the long arm of chromosome 10. Given the differing breakpoints, variable clinical features are expected. These are reviewed further by Lobo *et al*.[13] Shapiro *et al*[2] detected a deletion of 10q21 compounded by a deletion of 10q26.3 and of the terminal portion of 13p. In view of the other chromosomal anomalies, this case is not listed. Terminal deletions with more distal breakpoints have been reviewed by Wulfsberg *et al*.[15] Chieri and Iolster[16] described a large terminal deletion overlapping 10q23. This case was not reported in detail and separation of features found in 10q23 deletions from those associated with more distal deletions is not possible.

Clearly further reports of 10q23 deletions are needed to determine if a recognisable pattern of features is observable. This case reiterates the need to repeat testing when a chromosome anomaly remains a serious clinical consideration, despite an initially normal karyotype.

1 Mori MS, Gomez-Sabrido F, de Bustamante AD, Pinel I, Martinez-Frias ML. De novo 10q23 interstitial deletion. *J Med Genet* 1988;**25**:209–12.
2 Shapiro SD, Hansen KL, Pasztor LM, *et al*. Deletions of the long arm of chromosome 10. *Am J Med Genet* 1985;**20**:181–96.
3 Ray M, Hunter AGW, Josifek K. Interstitial deletion of the long arm of chromosome 10. *Ann Genet (Paris)* 1980;**23**:103–4.
4 Davis JG, Kardon NB, Selman JE. De novo 10q interstitial deletion. *Am J Hum Genet* 1982;**34**:122A.
5 Van de Vooren MJ, Planteydt HT, Hagemeijer A, Peters-Slough MF, Timmerman MJ. Familial balanced insertion (5;10) and monosomy and trisomy (10)(q24.2-q25.3). *Clin Genet* 1984;**24**:52–8.
6 Holden JA, MacDonald EA. Brief clinical report. Interstitial deletion of the long arm of chromosome 10: del(10)(q11.2q21). *Am J Med Genet* 1985;**20**:245–9.
7 Glover G, Gabarrón J, Ballester LJA. De novo 10q(q21q22) interstitial deletion. *Hum Genet* 1987;**76**:205.
8 Rooney DE, Williams K, Coleman DV, Habel A. A case of interstitial deletion of 10q25.2-q26.1. *J Med Genet* 1989;**26**:58–60.
9 Durum C, Lawce H. Brain tickler. *Appl Cytogenet* 1991;**17**:35–6.
10 Fryns JP, Bulcke J, Verdu P, Carton H, Kleczkowska A, Van den Berghe H. Apparent late-onset Cockayne syndrome and interstitial deletion of the long arm of chromosome 10 (del (10)(q11.23q21.2). *Am J Med Genet* 1991;**40**:343–4.
11 Curry C, Zorn E. Cytogenetic findings at variance with referring diagnosis. *Appl Cytogenet* 1991;**17**:62.
12 Derksen C, Ramdhanie R, Wood M, Winsor E. Interstitial deletion in 10q with no apparent abnormalities. *Appl Cytogenet* 1991;**17**:69.
13 Lobo S, Cervenka J, London A, Pierpont MEM. Interstitial deletion of 10q: clinical features and literature review. *Am J Med Genet* 1992;**43**:701–3.
14 Hall JG. Genomic imprinting: review and relevance to human diseases. *Am J Hum Genet* 1990;**46**:857–73.
15 Wulfsberg EA, Weaver RP, Cunniff CM, Jones MC, Jones KL. Chromosome 10qter deletion syndrome: a review and report of three new cases. *Am J Med Genet* 1989; **32**:364–7.
16 Chieri P, Iolster N. Monosomy 10qter due to a balanced maternal translocation: t(10;18)(q23;p23). *Clin Genet* 1983;**24**:147–50.