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A child with partial monosomy 6q secondary to a maternal direct insertional event

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SUMMARY We report a child, now aged two years, who is monosomic for the region 6q23.1→q24.2. Her mother, older sister, and twin sister have a balanced chromosome complement with this region of 6q being inserted into 11q.

Case report

The proband was one of twins born to healthy non-consanguineous parents; the mother was aged 35 and father 38. The twins were born at term with the proband weighing 2.6 kg and her sister 3.1 kg.

The mother had had two previous pregnancies. The first resulted in a healthy daughter and the second in a son who died aged six weeks from cardiac failure. He was reported as being dysmorphic with micrognathia and a large neck and unfortunately was not karyotyped. Necropsy (Dr R I K Elliott) showed cardiomegaly with a patent ductus arteriosus and a small patent foramen ovale but no explanation for the cardiomegaly. The skin was unusually thick and indurated.

There were no neonatal problems but the proband was slower to gain weight than her healthy sister. At

the age of five months she had the first of several hospital admissions for recurrent chest infections, persistent collapse of the right upper lobe, and failure to thrive. At the age of two years it was noted that length, weight, and head circumference growth rates had all fallen and the liability to infections persisted. She was globally delayed and barely able to sit unsupported. She transferred objects from hand to hand but said no meaningful words.

Her face was slightly dysmorphic (fig 1) with hypertelorism, prominent bridge to the nose, tented nares, and a small mouth with narrow upper lip. Overall her facies was reminiscent of that seen in the Wolf-Hirschhorn syndrome. Her palate was normal, her ears were low set, and her neck short, but no other abnormalities were noted.

Investigations have shown recurrent iron deficiency anaemia but normal immune function and sweat tests.

CYTOGENETIC FINDINGS

Peripheral blood lymphocytes from the proband, her twin sister, and both parents were cultured for chromosome studies with trypsin G banding. The proband showed a small interstitial deletion of the long arm of chromosome 6. Examination of the maternal chromosomes showed a direct insertional event between chromosomes 6 and the long arm of

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FIG 1 The proband aged eight months.

chromosome 11. The maternal karyotype may be described as follows: 46,XX,dir ins(11;6)(11pter→11q21::6q23·1→6q24·2::11q21→11qter;6pter→6q23·1::6q24·2→6qter) (fig 2). The proband's twin sister and her older sister also had the above balanced translocation. The proband's karyotype is 46,XX,der(6),dir ins(11;6)(q21;23·1→q24·2) resulting in partial monosomy of 6q. The paternal chromosomes were normal.

Discussion

Despite the child being monosomic for only a small region of 6q and the dysmorphic features being mild and confined to the facies, there appears to be a marked effect on neurological development, growth, and the ability to withstand infection.

FIG 2 Partial karyotypes of the proband (c) and the mother (a), and an ideogram showing the insertional event (b). The normal chromosomes are in the centre.

The only gene definitely assigned to this region is the 'oncogene' *c-myb* which is localised to 6q22→24,¹ but it would be premature to predict what clinical effect its loss might have.

No other child with this deletion has as yet been reported, but such familial deletions and insertions are becoming more readily recognised as cytogenetic definition improves.

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Interstitial deletion 1p in a 30 year old woman

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SUMMARY High resolution chromosome analysis showed the karyotype 46,XX,del(1)(p22.1 p31.2) in a 30 year old woman with psychomotor retardation and various malformations. Determination of the enzyme phosphoglucomutase 1 (PGM1) showed that she was a heterozygote. Three other cases of interstitial deletion 1p have been reported previously, and one of these cases had several features in common with our case, suggesting a distinct syndrome.

Short arm deletions of chromosome 1 are very rare. A large, presumably terminal, 1p deletion was described by Aarskog.¹ A few other cases of terminal deletion have been reported previously,²⁻⁶ but only three cases of interstitial deletion.⁷⁻⁹ The locus for the enzyme phosphoglucomutase 1 (PGM1) has been localised to band 1p22.1.¹⁰ We present a case of interstitial deletion 1p where the PGM1 phenotype was determined.

Case report

The proband (fig 1) was born at term to a 32 year old mother and a 36 year old father. An older brother was healthy and the parents were not consanguineous. The pregnancy was complicated by a febrile illness during the first and second months. The birth was uncomplicated, birth weight 3100 g, but at

FIG 1 The proband at 15 years of age.

three days of age she became cyanotic due to a throat infection which was successfully treated.

Development was severely retarded. She sat at 22 months of age, stood at three years five months, and walked at four years 11 months. Speech never developed. At 30 years of age her weight was 40 kg, height 128 cm, and head circumference 48 cm.