neural tube defect. Morrison *et al*[16] showed that the human *T* gene maps to 6q27 and lies between the two other genes of the T complex, *TCP1* and *TCP10*.

The present report suggests that male sex determination and differentiation is a complex process, involving several autosomal loci, and at least one of them is possibly located at 6q27.

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Distal trisomy 2p and arachnodactyly

EDITOR—We report two sibs with an unbalanced translocation of chromosomes 2 and 10 resulting in distal 2p trisomy. We feel that this sib pair could help to delineate the common features that may result from duplication of this specific region, in particular to show that arachnodactyly is a key feature. We suggest that the combination of arachnodactyly with developmental delay should prompt investigation of this region within the differential diagnosis.

This sib pair was first seen in 1995 aged 8 years (male) and 11 years (female). They were referred with a combination of speech and language delay, poor coordination, and concordant dysmorphic features.

The male sib (fig 1) was born at 38 weeks' gestation weighing 3200 g (25th centile). He was noted to have gross motor delay with hypotonia and walked at 22 months. Speech development was severely delayed necessitating Makaton from the age of 2 years. He attended a school for moderate learning difficulties and required intensive speech therapy. His motor development is still delayed and at the age of 11, he cannot ride a bicycle, catch a ball, or fasten shoe laces. He continues to require considerable supplemental educational input and speech therapy.

His height has followed the 90th centile with weight between the 25th and 50th centile and head circumference on the 50th. There is no limb or body segment disproportion. Joint flexibility is normal. He has arachnodactyly (palm length 10.5 cm (75th centile), middle finger length 8.0 cm (97th centile), middle finger to total hand ratio 43%) with fifth finger clinodactyly and prominent finger pads. His feet (fig 2) are narrow with metatarsus varus, medial deviation of the second toe, and a wide sandal gap. His face is long and triangular with semilunar palpebral fissures (length 2.8 cm). There is a bulbous tip to the nose with a high bridge. The palate is high and arched.

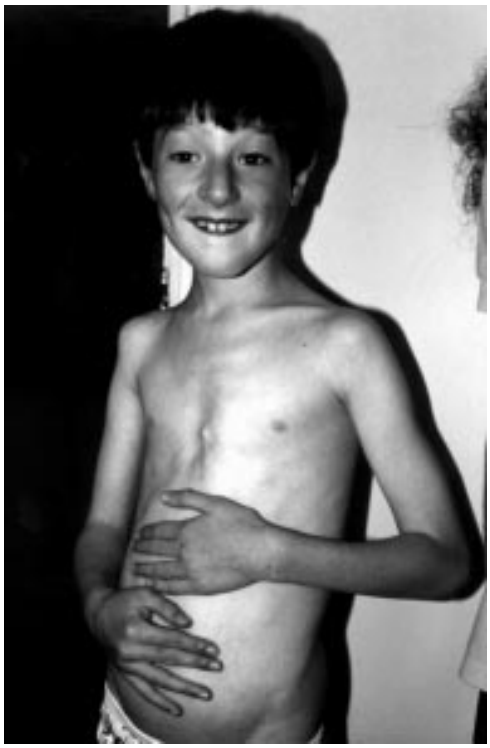

Figure 1 Male sib aged 8 years showing facial dysmorphism and arachnodactyly.

Figure 2 Feet of male child on left aged 8 years, female on right aged 11 years. Their foot abnormalities became more pronounced with increasing age.

Ophthalmological examination showed unilateral myopia only. Hearing is normal. Echocardiography is normal.

His sister (fig 3) was born at 38 weeks' gestation weighing 2940 g (50th centile). She had a splint applied for congenital dislocation of the left hip. Her motor skills were at the extreme of the normal range but she was noted to be clumsy. She was unable to ride a bicycle until 8 years old. Speech and language development was delayed requiring speech therapy. She is educated in a mainstream school with extra help. Her height follows the 95th centile, head circumference on the 50th, and her weight is on the 25th. Upper to lower segment ratio is 0.85, arm span is less than height, and joint flexibility is normal. Her face is elongated, the nose is bulbous with a broad bridge, and the palate is high and arched. Her palpebral fissures are of normal length (3.1/2.9 cm). She has arachnodactyly with finger length on the 97th centile (8.6 cm), palm length 75th centile (10.5 cm), and middle finger to total hand ratio 45%. Her feet (fig 2) are narrow with an exaggerated arch and there is partial 2/3 syndactyly on the right and overriding 1st/2nd and 4th/5th toes on the left. Ophthalmological examination, echocardiography, and audiology are normal.

There is one older male sib who is developmentally normal. His height (aged 17) is 194 cm (>97th centile). In contrast to his sibs, he is of stocky build. Middle finger length is 9.6 cm and palm 11.9 cm. Middle finger to whole hand ratio is 44%. His karyotype is unknown. Three other pregnancies ended in early miscarriage at 12-14 weeks.

Figure 3 Both children displaying concordant dysmorphic features.

Maternal height is 169 cm (75th centile), middle finger length 7.2 cm, and palm length 10.5 cm, and the father's height is 180 cm (75th centile), middle finger length 8.8 cm, and palm length 12.3 cm. They are unrelated.

Cytogenetic studies using G banding showed an unbalanced translocation 46,XY or 46,XX,−10,+der(10)t(2;10)(p25.1;q26.3)mat resulting in distal 2p trisomy (2p+). Fluorescence in situ hybridisation studies were not performed as the same translocation in balanced form was identified in their mother and maternal grandmother. They are phenotypically normal. Previous descriptions of 2p+ have included skeletal abnormalities, facial dysmorphism, severe developmental delay, and cardiac and genital abnormalities. These features have been reported with break-

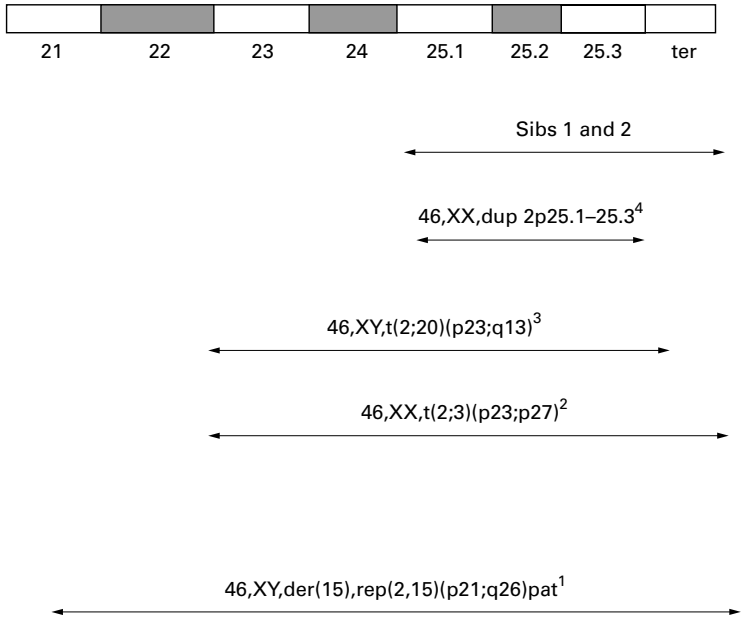

Figure 4 Diagrammatic representation of distal 2p and regions involved in previously described duplications.

Table 1 Comparison of features

Feature	*Female*	*Male*	*Others*
Arachnodactyly	+	+	+
Foot abnormalities	+	+	+
Congenital dislocation of the hip	+	−	+
Vertebral abnormalities	−	−	+
Scoliosis	−	−	+
Pectus excavatum	−	+	+
Developmental delay	Mild	Moderate	Severe
IUGR	−	−	+
Postnatal growth delay	−	−	+
Triangular face	+	+	+
Bulbous nose	+	+	+
High arched palate	+	+	+
Bossed forehead	−	+/−	+
Micrognathia	−	−	+
Hypertelorism	−	−	+
Strabismus	−	−	+
Myopia	−	+	+
Genital abnormalities	−	−	+
Cardiac abnormalities	−	−	+
Fits	−	−	+

points at 2p21,[1] 2p22,[2] and 2p23,[3] usually in combination with partial monosomy involving one of various other chromosomes. However, with the exception of the report from Wakita *et al*,[4] the breakpoint in this case differs from other published cases in that a much shorter duplicated segment is produced (fig 4). The reciprocal chromosome in the translocation also varies. In the case described by Wakita *et al*,[4] trisomy 2p resulted from an isolated duplication 2p and not a translocation. In this case the 10qter deletion is very small (on G banding) and probably does not contribute to the phenotype, although modification of clinical expression by this deletion cannot be excluded.

In these two sibs, the translocation is inherited maternally. In other cases reported, paternal[1] and maternal[2] transmission as well as de novo[4] duplications have been described with features consistent with distal trisomy 2p, making the role of imprinting less likely.

To our knowledge correlation of phenotype to duplicated segment length has not been possible. As this sib pair display some phenotypic differences from other cases described, they may contribute to the understanding of this correlation. The main phenotypic differences are summarised in table 1. In particular, these sibs have neither cardiac nor genital abnormalities and their development is mildly to moderately delayed. They do not have growth failure but have a marfanoid habitus. Arachnodactyly is a feature of Marfan syndrome which is associated with mutations in fibrillin genes mapped to chromosomes 5 and 15.[5] However, the marfanoid habitus with mental retardation has also been described in an X linked disorder.[6] Marfanoid skeletal and cardiovascular phenotypes have also been reported which are not linked to known fibrillin genes.[7] The children presented here do not fulfil the criteria required for a diagnosis of Marfan syndrome.[5]

A combination of retardation (particularly speech and language) with arachnodactyly can also occur in 22q11 microdeletion syndrome, but these children have none of the associated features.[8]

The presence of arachnodactyly coupled with the evident arachnodactyly in the short duplication reported by Wakita *et al*[4] leads us to conclude that this a key feature of duplications in this region. The influence of the reciprocal chromosome involved in the translocation must be considered. In other cases described,[1–3 9] the reciprocal chromosome varies yet arachnodactyly is a constant feature. In this case, the 10qter deletion is unlikely to contribute much to the phenotype (although cannot be excluded) and the case reported by Wakita *et al*[4] was an isolated duplication without a translocation, so the features described can be assumed to result from duplication of this region alone.

The common features evident in these children combined with the notable absence of others described in distal trisomy 2p syndrome aid the phenotype/genotype correlation in this region. We would be interested in other reports of duplications which would help this process.

These sibs also show the importance of investigating this region in children who present with arachnodactyly and developmental delay of unknown origin; very small duplications may produce few additional features.

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