were prominent epicanthic folds, bilateral ptosis, and optic atrophy. The ears were large, prominent, and low set. He had flexion deformities and limitation of movement at the proximal interphalangeal joints of all the digits but mainly the second, third, and fourth fingers bilaterally. The fingers were short and spindle-shaped, thickened at the proximal interphalangeal joint and tapering distally. The middle phalanges were shortened and the distal digital creases were absent on the second and fourth fingers bilaterally. Bilateral talipes equinovarus had been corrected surgically at the age of 27 months. The child had a ventricular septal defect. His genitalia were normal.

Dermatoglyphs showed *a*, *b*, *c*, *d*, and *t* triradii and a distal loop in the interdigital area IV of both palms. Total *a-b* ridge count was 88 and *aid* angles were normal.

G, C, and Q banded chromosome preparations from peripheral blood lymphocyte cultures revealed a complex rearrangement between Y, 1, and 3. Breakpoints were identified at Yp11, 1q21, 3q23, and 3q25. The karyotype was interpreted as 46,X,t(Y;1;3)(Yqter→Yp11::1q21→1qter;1pter→1q21::3q25→3qter;3pter→3q23::) (figure). Segment 3q23→q25 could not be accounted for. The chromosomes of the parents were normal.

FIGURE Karyotype of proband with diagram showing breakpoints.

The proband's phenotypic abnormalities may have resulted from the loss of segment 3q23→q25. We are aware of only one reported case of partial monosomy 3q, involving the loss of 3q22→q24,¹ and it is remarkable that the missing segment and the associated phenotypic abnormalities reported there are strikingly similar to the present case: prenatal growth retardation, developmental delay, mental retardation, microcephaly, blepharophimosis, malformed auricles, talipes, and absence of the distal interphalangeal joint creases on the fingers. It is possible that these features constitute a characteristic syndrome of deletion within the segment 3q22→q25, although position effect cannot be completely excluded. The blepharophimosis and digital abnormalities would appear to be the most characteristic features as, unlike the other features, they are not commonly encountered in other malformation syndromes. The involvement of the Y chromosome indicates that the translocation must have occurred either during spermatogenesis or in the zygote during the first cleavage division.

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Prune belly appearance in a Turner subject

The 'prune belly' syndrome was first reported in 1895 by Parker (referred to by Rabinowitz *et al*¹). It is characterised by the triad abdominal muscle deficiency, urinary tract anomalies, and cryptorchidism in the male. The incidence among live born infants is of the order of one in 30 000. So far only 18 female patients have been reported out of a total of some 200 published case reports.² Chromosomal analysis of four of these female patients has been reported and each had a normal 46,XX karyotype. The only report of an abnormal karyotype was that of Harley *et al* (referred to in Rabinowitz *et al*¹) where two brothers with the syndrome had mosaic monosomy 16 (45,XY,-16/46,XY).

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FIGURE The proband. Note the prune belly appearance, lymphoedema of the feet, and pterigium.

Smith² suggested that 'prune belly' syndrome was the result of an early urethral or ureteral obstruction, either mechanical or the result of fibrous replacement of muscle. The increased male to female ratio would result from a higher incidence of the malformation in the male urethra.

We have observed the features of the 'prune belly' syndrome (figure) in a patient with the Turner phenotype and a chromosomal constitution 45,XO (RHG banding). She was the first child of non-consanguineous parents, the father aged 24 years and the mother 21 years.

At necropsy the findings included hypoplastic abdominal musculature, horseshoe kidney, atrophy of the left ureter at the ureteropelvic junction with atrophy and cystic change in the left kidney, and coarctation of the aorta. The bladder was large and the urethra normal.

Although horseshoe kidney and ureteropelvic junction obstruction are frequent anomalies in the Turner syndrome,³ 'prune belly' syndrome has not been reported in association with the Turner phenotype.

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