

describe what is, to our knowledge, the first familial case of pericentric inversion of chromosome 1 involving the whole of the short arm and associated with infertility.

The proband was a male aged 35 who came to our laboratory because of sterility. He reported a 33 year old brother who had been married for two years without children. Both patients had normal phenotypes and no history of testicular pathology. Semen analysis showed severe oligozoospermia in both and the few spermatozoa present were immotile. The patients refused testicular biopsy.

Chromosome preparations in the proband and his brother, obtained from peripheral blood lymphocytes, were examined after G, C, and high resolution R banding. G and R banding revealed in both an inverted short arm of chromosome 1 with the centromere located near the terminal part of the chromosome. C banding showed two blocks of C heterochromatin, the smaller one located in a subterminal position. This suggested that the breakpoints were at 1p36.3 and 1q12 (fig 1).

This unusual pericentric inversion was also found in the mother of the proband, *inv(1)(p36.3q12)*, showing that the inverted chromosome was inherited without meiotic recombination in the inverted segment. The karyotype of II.5 was normal (fig 2).

In this case, as in the three others previously reported, it is reasonable to assume that there may be an association between the inversion and failure of spermatogenesis.

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ALBERTO BARROS*†, M CARMO TAVARES*, M PAULA GOMES*, AND M PURIFICAÇÃO TAVARES*
 *Department of Medical Genetics, Faculty of Medicine, University of Oporto; and
 †Department of Obstetrics, S João Hospital, Oporto, Portugal.

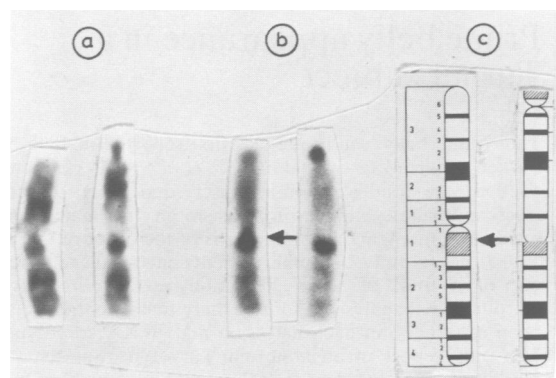


FIG 1 (a) G banding (*inv* on right). (b) C banding (*inv* on right). (c) Diagram of chromosome 1 with the proposed breakpoints (arrow) and the resulting inverted chromosome (right).

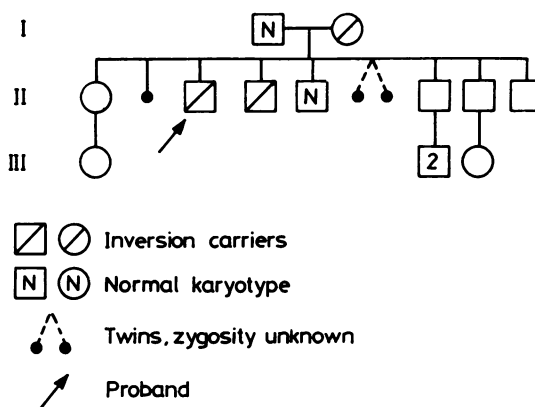


FIG 2 Family pedigree.

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Correspondence and requests for reprints to Dr Alberto Barros, Department of Medical Genetics, Faculty of Medicine, University of Porto, 4200 Porto, Portugal.

Complex translocation involving chromosomes Y, 1, and 3 resulting in deletion of segment 3q23→q25

The proband was the first child of non-consanguineous parents. He has a younger, phenotypically normal sister. Birth weight was 1.9 kg and he had respiratory distress at birth. Psychomotor development was delayed. His height and weight were on the 3rd centile and his head circumference was below the 3rd centile. He started to walk at 2½ years and could say a few words at 3½ years. IQ was assessed to be around 75 to 80.

The proband had microbrachycephaly, a flat occiput, depressed supraorbital ridges, and flat nasal bridge. Palpebral fissures were narrow and upward slanting. There

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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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S A AL-AWADI, K K NAGUIB, T I FARAG, A S TEEBI,
A CUSCHIERI, S A AL-OTHMAN, AND
T S SUNDARESHAN
Medical Genetics Centre, Maternity Hospital, Kuwait.

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Correspondence and requests for reprints to Dr Sadika A Al-Awadi, Kuwait Medical Genetics Centre, PO Box 4080, Safat, Kuwait.

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. 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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