Turner’s syndrome and 46,X,i(Yq) karyotype

Summary. A patient with features of Turner’s syndrome is reported. In her karyotype there were 46 chromosomes including 15 chromosomes in the C group and one metacentric marker the size of a No. 16. This marker was identified through its fluorescence pattern as being an isochromosome for the long arm of the Y. This observation strongly suggests that the male determining factors are situated on the short arm of the Y chromosome.

Case report

The patient (Fig. 1) was referred to us when she was 8 years old with a diagnosis of Turner’s syndrome. She was born after an uneventful pregnancy and normal delivery, when her parents were 25 years old. No data concerning weight and length at birth are available. The parents did not notice that the child was abnormal and were not informed of the presence of abnormalities. The patient has been healthy. Her parents are alive and healthy; she has a younger normal brother. The family history is negative for the existence of congenital anomalies, mental retardation, abortions, or errors in sexual development.

Physical examination. Stature 112 cm, span 110 cm, cephalic circumference 52 cm, weight 17 kg. The patient is normocephalic, the nuchal hairline is very low. Her ears are low-set, and her face mildly asymmetric. Epicanthic folds are present bilaterally. The neck is markedly webbed. The thorax has normal configuration, with widely separated nipples. A systolic murmur is heard on mesocardial area. The hands are short. The external genitalia are normal. Psychoneurological evaluation shows no gross deviation from normalcy. Clinical examination strongly suggests a diagnosis of Turner’s syndrome.

Dermatoglyphics. Dermatoglyphics are summarized in Table I and are in good agreement with a diagnosis of Turner’s syndrome.

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Cytogenetic studies

No Barr bodies were observed in 500 nuclei from Feulgen-stained buccal smears. Chromosomal analysis was performed on peripheral blood leucocytes cultured by a modified Moorhead method. The fluorescence banding studies were performed according to the method described by Caspersson, Lomakka, and Zech (1971). All 32 cells analysed under conventional microscopy showed a 46 chromosome karyotype including 15 apparently normal chromosomes in the C group and one metacentric marker the size of a chromosome 16 (Fig. 2). This chromosome was always distinguishable from the members of the E group for being perfectly metacentric and the ends of both arms showing a fuzzy

![Fig. 1. The proposita at 8 years of age.](http://jmg.bmj.com/)

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appearance. These features strongly suggested that it was an isochromosome for the long arm of the Y, rather than a deleted X. This interpretation would explain why the patient had no Barr bodies, when one should expect a deleted X of this size to be able to form sex chromatin. In all 18 cells studied under UV microscopy a fluorescence spot similar to the typical one on the long arm of the Y chromosome was observed on each arm of the marker chromosome (Fig. 3). This shows that the marker is an isochromosome for the long arm of the Y. The father of the patient has a normal karyotype.

**Discussion**

Only four female patients presenting duplication of the long arm and deletion of the short arm of the Y have been described. Together with the present report these cases allow a discussion on the location on the Y chromosome of (1) the male determining factors and (2) the presumptive loci preventing the manifestation of the somatic features of Turner’s syndrome.

Our case suggests that factors determining male differentiation are located on the short arm of the Y chromosome, since the patient presents female phenotype associated with double doses of the long arm of the Y and absence of its short arm. This hypothesis was advanced by Jacobs and Ross (1966) who detected a presumptive 46,X,i(Yq) karyotype in two females with primary amenorrhoea and sexual infantilism, but no evidence of virilization; in one of these patients a laparotomy showed streak gonads. Recently their cytogenetic interpretation was confirmed by fluorescence studies (Robinson and Buckton, 1971).

Armendares et al (1972) found epididymal structures in a patient with features of Turner’s syndrome and a 46,X,dic(Yq) karyotype. This could be explained by the presence of a segment of the short arm of the Y. Ferguson-Smith et al (1969) reported the presence of rudimentary epididymis in a woman with a 45,X/46,X,i(Yq) karyotype and suggested that factors on both arms of the Y play a part in male determination. However, this interpretation is not the only one possible. Since the patient is a mosaic, it is conceivable that from a
46,XY zygote a transversal fission of the Y centromere produced in a first step a 46,X,i(Yp)/46,X,i(Yq) mosaic and that the cell line with the i(Yp) had persisted during development long enough to slightly influence the gonads. In the present case the family did not give permission for a laparotomy and nothing can be said about gonadal development.

We may conclude that up to now the evidence is compatible with the location of the bulk of virilization factors on the short arm of the Y and the absence from its long arm of any virilization factor of importance.

Since their patients did not show the somatic abnormalities typical of Turner's syndrome, Jacobs and Ross (1966) suggested that genes preventing these features occur on the long arm of the Y, an assumption implying homology between some loci on the X and on the long arm of the Y. However, the phenotypic characteristics of the present case and that described by Armendares et al (1972) do not support this hypothesis, since the patients are of low stature, have shortness and webbing of the neck, and low nuchal hairline. An undetected 45,X cell line could be present both in our patient and in that of Armendares et al (1972). On the other hand, the genes preventing features of Turner's syndrome could be located near the centromere, in the long arm or in the short arm in different patients. It is indeed conceivable that the Y is in a polymorphic state in the normal population with respect to small pericentric inversions, which must not cause meiotic disturbance since X and Y pair end-to-end.

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Chromosomal abnormality associated with congenital macroglossia and other abnormalities

Congenital macroglossia is a rare clinical abnormality, and when found in an infant with a chromosomal abnormality, it could be a useful chromosomal marker. It is for this reason that we would like to present the following clinical and chromosomal findings.

Case report

The patient is a 5-week-old white female who was born to a gravida II, para I mother after an uncomplicated, full-term pregnancy with no history of infection or any medications during pregnancy. Amniocentesis was performed twice during the pregnancy because of a possible Rh blood incompatibility factor. At birth the child had microcephaly, macroglossia, and a heart murmur; she weighed 3500 g at birth. Since birth, she has had breathing and feeding difficulties because of her enlarged tongue.

The 21-year-old father and the 20-year-old mother are in good health. One sib, aged 1 year, is normal. There is no history of congenital anomalies in the family of either parent.

On physical examination, the infant had difficulty breathing but no cyanosis. The infant's tongue was partially protruding (Fig. 1). Her height was 56 cm (75th centile), weight 4190 g (10th centile), head circumference 36 cm (10th centile), heart rate was 140/min, and respiratory rate was 36/min. There was marked microcephaly with 2–3 mm opening of the anterior and posterior fontanelle. The tongue was diffusely enlarged filling the entire mouth cavity and remaining partially protruding at all times. There was no evidence of a hemangioma or cyst of the tongue. There was a cleft uvula. She had a precordial thrill and a grade 4/6 holosystolic murmur at left lower sternal border. The

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FIG. 1. The proposita; note macroglossia and microcephaly.