

Case reports

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Ultrasound identification of apparently normal male genitalia in a 46,X,+mar fetus

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SUMMARY A level II ultrasound examination revealed a scrotum and penis in a fetus with a 46,X,+mar chromosome complement. The marker was subsequently considered to be a del(Y)(q11). A phenotypically normal male infant was born. Detailed ultrasound examination of similar cases for visualisation of the genitalia is recommended.

Case report

A 38 year old woman was referred for prenatal chromosome analysis because of advanced maternal age. The patient had had two previous pregnancies resulting in a miscarriage at nine weeks and a stillbirth at 28 weeks' gestation. The stillborn male had a normal karyotype (as determined from amniotic fluid cells), no obvious birth defects, and his death resulted from acute placental abruption. The family history was unremarkable.

Two culture flasks were initiated from the amniotic cells. A total of 36 metaphases was examined from the two flasks. All metaphases had a 46,X,+mar complement by GTG banding (fig 1). QFQ banding and NOR staining failed to help in determining the origin of the marker. Parental karyotypes were normal, and the father's Y chromosome was normal in size and appearance (fig 1).

The marker was reported to the referring physician as being derived from either the missing sex chromosome or an autosome. If derived from an X, a Turner phenotype would be expected. If derived from a Y, the clinical significance would be difficult

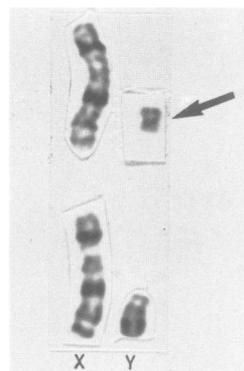


FIG 1 Sex chromosome complement of the proband (top) and father (bottom). Arrow indicates marker chromosome.

to predict because the Y varies in length. If derived from an autosome, Turner's syndrome and possibly other clinical abnormalities would be expected.

A level II sonographic examination was performed on the patient at 20 weeks' gestation because of the unusual cytogenetic findings. All measurements were found to be compatible with normal fetal growth. An attempt to identify the fetal sex at this time was inconclusive, although there was some suggestion that the genitalia were male.

The parents were anxious to have a child and decided to continue the pregnancy since no abnormalities were observed. Another ultrasound scan was to be performed at approximately 23 weeks' gestation. The scan at this time allowed definitive visualisation of a scrotum and penis (fig 2).

The parents were told that we now believed the marker was most likely to have been derived from the Y chromosome. The pregnancy continued to term

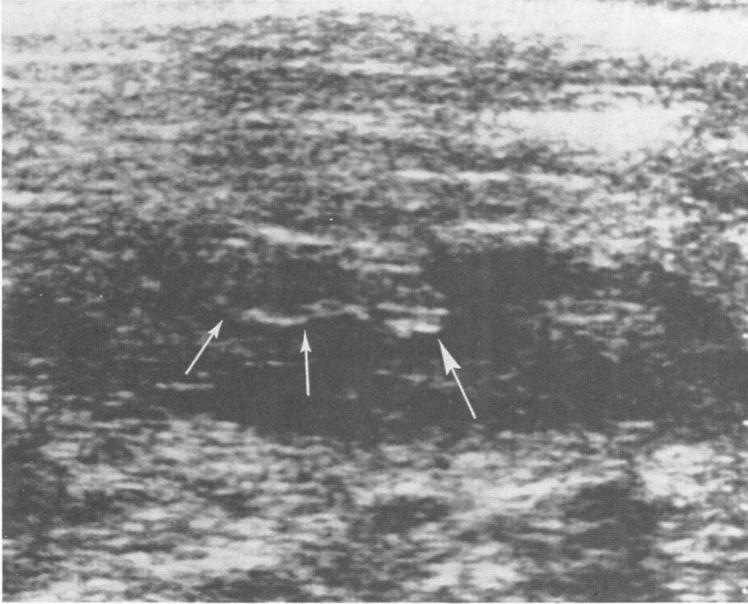


FIG 2 Sonograph showing fetal penis (large arrow) and scrotum (small arrows).

and the patient gave birth to a healthy male infant with no clinical abnormalities. The 46,X,+mar karyotype was confirmed by a peripheral blood culture from the newborn infant. The marker is now considered to be a *de novo* del(Y)(q11).

Discussion

Davis¹ reviewed the structural anomalies of the Y chromosome and determined that a gene, or genes, on the short arm of the Y chromosome near the centromere play an essential role in testicular development. One or more genes near the centromere on the Y long arm may also influence testicular development. Assuming that the deletion in our case resulted from a single break at the end of Yq11, the Yp would be present, as well as the very proximal Yq.

At present, it is not possible to predict whether normal maturation of the testes will occur in this infant at puberty. Fitch *et al*² reviewed 20 reported cases of long arm Y deletions. Fourteen were azoospermic, three were oligospermic, and three were fertile. The latter three patients, however, came from families in which the deletion was also present in other males.

In our case, the marker chromosome was of unknown origin until an ultrasound examination

established the presence of male genitalia. This was the deciding factor in the decision of the parents to continue the pregnancy. DNA probe technology will probably be used to identify such markers in the future, and to determine if the gene(s) for normal testicular development are present once their location is known. This technology, however, is not yet clinically available. In conclusion, a detailed ultrasound examination for visualisation of the genitalia is recommended when similar cases are encountered. The presence of male genitalia may be an important factor in helping the parents decide whether or not to continue the pregnancy, even though the del(Y) carrier may be infertile.

References

- 1 Davis RM. Localisation of male determining factors in man: a thorough review of structural anomalies of the Y chromosome. *J Med Genet* 1981;18:161-95.
- 2 Fitch N, Richer C, Pinsky L, Kahn A. Deletion of the long arm of the Y chromosome and review of Y chromosome abnormalities. *Am J Med Genet* 1985;20:31-42.

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