risks for females belonging to families with Duchenne muscular dystrophy.

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Identification of a balanced translocation carrier by spouse's low maternal serum α fetoprotein associated with an aneuploid fetus

There have been numerous reports of fetuses with Down's syndrome (trisomy 21), trisomy 18, and trisomy 13 having been identified through a low maternal serum α fetoprotein (MSAFP) level. Recently, Winston *et al*¹ reported a fetus with Turner's syndrome whose mother had a low MSAFP value of 0.34 multiples of the median (MOM) at 16 weeks' gestation. Since that case report, Drugan *et al*² have suggested that genetic counselling for low MSAFP levels should include all chromosome anomalies, not just Down's syndrome. In their experience, women with low MSAFP values have carried fetuses with partial trisomy of chromosome 22, triploidy, duplication of chromosome 5, and sex chromosome abnormalities, in addition to trisomy 21 and 18. We report here the identification of a paternal balanced translocation owing to his wife's low MSAFP value, associated with an aneuploid fetus.

A 29 year old woman (G3P0SAb1 TAB1) presented for genetic counselling owing to a low MSAFP level of 0.19 MOM at 15 weeks' gestation. Correcting for weight, the MSAFP value was 0.22 MOM. The patient was not diabetic. The only significant pregnancy history was that eight months before this pregnancy, the woman had had a spontaneous abortion at six weeks' gestation. Her family history was unremarkable. Her husband's family history was significant for his mother having had a spontaneous abortion at six months' gestation. He had only one sib and his mother was an only child. His father's only sib died in an accident.

The couple elected to have a level II ultrasound scan followed by a genetic amniocentesis. Ultrasound scan was normal with size equal to dates. Fourteen days after amniocentesis, the fetal karyotype showed extra material on chromosome 4. The couple was con-

tacted and their blood karyotypes were performed.

The results of the couple's karyotypes showed the husband to have a balanced reciprocal translocation between chromosomes 4 and 18: 46,XY,t(4;18)(p15.2;q11.2). The woman had a normal female karyotype, 46,XX.

We were thus able to identify the abnormal fetal chromosome 4 as being derived from the father's translocation. The fetus had partial duplication of 18q and partial deletion of 4p: 46,XY,-4,+der(4)t(4;18)(p15.2;q11.2)pat. The couple elected to terminate the pregnancy. However, fetal death occurred one day before the scheduled procedure.

While the use of the MSAFP screening test is still considered investigational for screening for fetal chromosome abnormalities, we feel that it enabled us to identify a balanced reciprocal translocation carrier parent owing to the unbalanced chromosomal complement in the fetus. Based on pregnancy and family history, there was no indication for genetic counselling or amniocentesis in this couple. Without the indication of the low MSAFP, prenatal diagnosis would not have been offered. While the tissue from the fetus could have been studied, this is not always done nor is it always successful. This couple could have gone through another miscarriage, or had a severely affected child without having the option to prepare.

Although MSAFP screening is not able to detect parental translocations directly, data from this case show the occasional ability to detect a balanced translocation in a parent associated with a low MSAFP and an unbalanced fetal chromosome complement.

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