Short report

A familial case of chromosome 16p variant

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A variant of chromosome 16, 16p+, was recently described for the first time in six subjects. We present a further case of 16p+ detected prenatally. A 38 year old, gravida 3 woman, who had had one spontaneous abortion and one healthy daughter, was referred for prenatal diagnosis because of advanced maternal age. Cytogenetic investigation of the cultured amniocytes showed a 16p+ in the fetal karyotype. The short arm of chromosome 16 contained additional material proximal to the centromere which stained positively with GTG and QFQ banding and negatively with RBA, CBG, and DA–DAPI banding (fig 1). An identical 16p+ was detected in the karyotype of the mother and of two other healthy relatives (fig 2). The pregnancy continued and a normal male was born at term.

This is the seventh reported case of a chromosome 16 short arm variant. G, C, and DA–DAPI banding patterns of the extra material on 16p appear to be the same as in the cases described previously. In addition, we performed RBA banding which showed a negative stain.

There were two stillbirths without malformations (II-1 and II-2) and one spontaneous abortion in our family. However, the absence of phenotypic effects in the family members who carry the 16p+ supports the current view that this variant has no clinical significance.


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Figure 1 Banding patterns of the 16p+ (left) and normal 16. (a) QFQ, (b) GTG, (c) RBA, (d) CBG, and (e) DA–DAPI banding.

Figure 2 Pedigree of the reported case.