Chromosome aberrations are found in 2–7% of couples with fertility problems (Daniely et al. 1998). Carriers of balanced structural aberrations appear to have an increased risk of progeny with unbalanced karyotypes resulting in miscarriages. A young woman (32) was examined due to a miscarriage in the 5th week of pregnancy. It was her first detected pregnancy. She admitted that she unsuccessfully was trying to get pregnant within the last 5 years. The subfertility of the couple was previously regarded as idiopathic. Both the patient and her partner were offered a cytogenetic examination. The current case report presents two chromosomal inversions in one of the partners from this subfertile couple.

Lymphocytes of the proband and her partner were cultured, pretreated and fixed according to the standard cytogenetic procedure (Gosden et al. 1994). The karyotypes were analysed by use of the conventional banding technique (GTG). The woman was found to have a normal karyotype with enlargement of a heterochromatic region in the long arm of one chromosome 16 (46,XX,16qh+), but her partner was found to have two inverted chromosomes. His karyotype was 46,XY,inv(2)(p11q13),inv(9)(p11q13). The abnormal karyotype is recognised as a possible reason of fertility problems in the investigated couple. The risk of further miscarriages is considered high, but the risk of progeny with abnormal karyotypes is rather low, as small inversions may lead to lethal recombinants.

Abstract. This case report presents two chromosomal inversions in one of partners from a subfertile couple. The woman was referred due to a spontaneous abortion in the 5th week of pregnancy. Cytogenetic examination showed that the proband’s karyotype was normal: 46,XX,16qh+, as centromeric heterochromatin is thought to be clinically insignificant. However, the proband’s partner occurred to be a carrier of two pericentric inversions. His karyotype was 46,XY,inv(2)(p11q13),inv(9)(p11q13). The abnormal karyotype is recognised as a possible reason of fertility problems in the investigated couple. The risk of further miscarriages is considered high, but the risk of progeny with abnormal karyotypes is rather low, as small inversions may lead to lethal recombinants.

Key words: inv(2), inv(9), miscarriage, pericentric inversion.
only the centromere and centromeric heterochromatin, so it seldom results in aberrant chromosomes after crossing-over (Kaiser 1988). Nevertheless, Gardner and Sutherland (1996) claim that some patients carrying inversions have an increased risk of unbalanced progeny, ranging from 1% to 10%.

Colls et al. (1997) investigated semen of a man with karyotype 46,XY,inv(9)(p11q13). There was inv(9) found in 48.7% of analysed cells and disomic sperm cells for chromosome 9 and 21 were not more frequent than in men with the normal karyotype. Those researchers concluded that inv(9)(p11q13) does not cause defects in spermatogenesis.

Some authors observed an increased risk of progeny with trisomy of chromosome 21 in carriers of inv(9) that contains additionally an enlarged heterochromatin region (Wang and Hamerton 1979; Serra et al. 1980; Neri et al. 1981; Serra et al. 1990). Those findings contradict the hypothesis that heterochromatin does not take part in crossing-over. Ameil et al. (2001) found an increased number of disomic (chromosome 9) sperm in such a carrier.

Pericentric inversion of chromosome 2 is also relatively common, as 14% of all inversion cases carry inv(2). The most common breakpoints are p11q13 (Kaiser 1984). Jacobs et al. (1974) reported that the incidence of this aberration is 1 in 11,680 in newborn infants. This aberration is not associated with a specific syndrome and no abnormal phenotype has been described. The carriers were referred due to various disorders, such as psychiatric problems, mental retardation, congenital abnormalities and spontaneous abortions (Baccichetti et al. 1980; Leonard et al. 1975; Romain et al. 1982; Philips 1978). Four cases of deletion (2)(p11.2p13) have been reported, but only one of them was associated with paternal inv(2)(p11q13) (Lacbawan et al. 1999).

The abnormal karyotype of the proband’s partner 46,XY,inv(2)(p11q13),inv(9)(p11q13) can be acknowledged as a reason of fertility problems in the investigated couple. According to Kaiser (1988) the risk of further miscarriages is high, but the risk of progeny with abnormal karyotypes is rather low. Small inversions may lead to recombinants with lethal deletions or addition of large fragments. It is presumed that the woman could have some undetected miscarriages in early pregnancy. Many of her embryos may have had unbalanced karyotypes, which made them unable to develop.

In contrast, Gardner and Sutherland (2004) described both inv(2) and inv(9) as normal variant chromosomes. Two abnormalities were described in families carrying inv(2): one deletion and one duplication in 2p, resulting probably from unequal crossing-over. For that reason those authors do not
recommend prenatal diagnosis to every couple carrying inv(2). However, researchers frequently do not mention subfertility in discussions about clinical consequences of chromosomal aberrations and there are very few publications concerning the clinical picture of variant chromosome carriers.

In this report we have presented a subfertile couple carrying two inversions recently acknowledged as normal variant chromosomes. The subfertility of the couple was regarded as idiopathic. Unfortunately, the cytogenetic examination of the couple was conducted after the first early miscarriage and it was not possible to obtain the miscarriage material for further examination. The laboratory was also not provided with the semen sample for cytogenetic examination. Thus we were not able to confirm directly the semen abnormality and the direct influence of these two inversions on spermatogenesis.

REFERENCES


