

INVERSIONS AND INFERTILITY

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INTRODUCTION

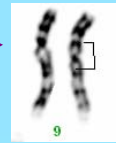
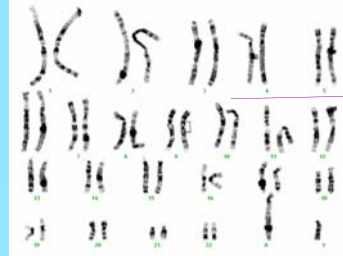
Infertility:

- The inability to achieve conception or to sustain a pregnancy through to livebirth
- 10-15% of couples
- Etiology: genetic (unbalanced – numerical or structural - and balanced rearrangement) malformative, endocrinologic, imunologic and environmental factors

Inversions = intrachromosomal rearrangements in which a segment of the chromosome breaks off and reattaches in the reverse direction

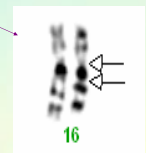
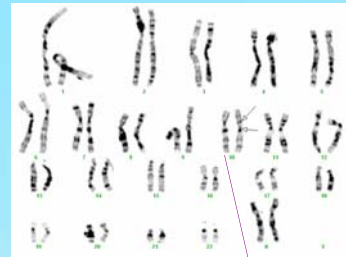
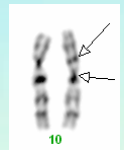
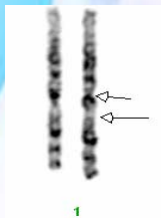
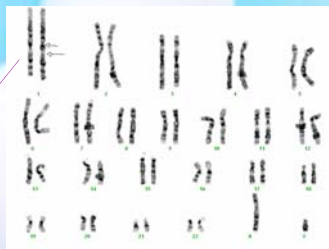
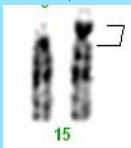
MATERIALS AND METHODS:

- 159 infertile couples analysed in Medical Genetics Center in Iasi, Romania between 2001 and 2008
- Chromosome analysis from peripheral blood lymphocytes cultures using GTG



RESULTS:

- Chromosomal abnormalities were found in 30 couples (18,8%) (just in one person of couple).
- 14 from these are inversions, all pericentric.
- 9 inversions were present in male partner.
- The chromosomes implicated are: chr 9 (9 cases), chr 1 and 15 (2 cases), chr 10 and 16 (1 case).



DISCUSSION:

- The most frequent inversion (as in literature) found was a pericentric one on chromosome 9: $inv(9)(p13q21.2)$. The segment involved is greater than the segment in normal variant of inversion of 9 chromosome – $inv(9)(p11q12)$ and could explain infertility.
- The segment involved in two cases of $inv(15)$ is small, but required prenatal diagnosis because of Prader- Willi syndrome risk.
- One of two inversion chromosome 1 involved centromeric heterochromatin and has no reproductive risk: $46,XY,inv(1)(p11q12)$.
- Another inversions without risk are $inv(16)$ and $inv(10)$.

CONCLUSIONS:

- Chromosome analysis is very important both for etiology and future management
- Inversions are an important cause of infertility