INVERSIONS AND INFERTILITY

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, immunologic 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.