Abstract

The evaluation of the incidence of balanced chromosomal aberrations in the couples with fertility failures

Constitutional aberrant karyotypes can account for infertility or recurrent pregnancy loss. Cytogenetic studies were performed over a period of 10 years (January 1998–Decembre 2007) in The Institute of Biology and Medical Genetics – The University Hospital Motol by 2nd School of Medicine of Charles University on 2121 patients diagnosed as infertile.

Each partner of 815 infertile couples and single each one of 305 women and 186 men was screened for karyotype changes by GTG-banding technique on peripheral lymphocytes. No subject presented with obvious phenotype of chromosomal rearrangement. A total of 126 aberrant karyotypes (5,9 %) was diagnosed, corresponding to an abnormality frequency of 4,6 % (46/1001) for men and 7,1% (80/1120) for women.

These chromosomal abnormalities were found in our study: gonosomal mosaicism in 74 cases, Robertsonian translocations in 8 cases, autosomal reciprocal translocations in 16 cases, inversions in 6 cases, Klinefelter syndrom in 9 cases, Turner syndrom in 1 woman, some other aberrations in 9 cases, a marker chromosome in one case, one case of 46,XY female and one case of 46,XX male. Chromosomal variants (pericentric inversion of the chromosome 9) found in 13 women and 14 men were not included in the above percentages.

Result: Partners of infertile couples appear to be affected by higher frequency of chromosomal rearrangements than the general population.

Key words: *chromosome abnormalities; chromosome analysis; infertility; FISH; genetic counselling; reproductive counselling*

Klíčová slova: *chromozomové aberace; analýza karyotypu; neplodnost; FISH; genetické poradenství; reprodukční poradenství*