**Abstract**

**Objective:** To determine chromosome and gene alterations in ectopic endometrial (EM) tissue which may be implicated in the clinical course or the progression of endometriosis and to review the literature concerning the cytogenetic findings of women with endometriosis.

**Study design:** 15 women who underwent laparoscopic endometriosis surgery at the Athens Genesis Clinic were enrolled in the study. Ectopic endometrial tissue was surgically removed and further analyzed by conventional and molecular cytogenetic techniques. Fluoresent in situ hibridization (FISH) with probes for p53, ATM, MYC, MLL1 and IGH genes, the centromeres of chromosomes 7 and 8 and 7q22/7q31 chromosomal regions was carried out.

**Results:** Karyotypic analysis revealed no clonal chromosomal abnormalities. However, an increased frequency of polyploidy (55.6%) and sporadic chromosomal abnormalities (40.0%) concerning chromosomes 9, 11, 17 and X were noticed involving mainly deletions, trisomies or monosomies. FISH analysis showed IGH gene rearrangements in 54% of the EM cases and MLL gene rearrangements in 73% of the examined samples. Normal hybridization patterns were observed for p53, ATM and MYC. The increased frequency of polyploidy shown by conventional karyotyping was also confirmed by FISH.

**Conclusion:** Polyploidy, sporadic chromosomal abnormalities, as well as IGH and MLL gene rearrangements, may provoke genetic instability and play a potential role in the development of endometriosis. IGH and MLL gene rearrangements indicate a genetic relation between endometriosis and carcinogenesis. Confirmation of the above gene rearrangements in a large series of women may allow the determination of their possible involvement in the pathogenesis of this complex disease and their possible contribution in the early identification of women in danger for malignant transformation.