

Patients with disorders of sex development with an increased risk for development of gonadal germ cell tumors mostly undergo prophylactic gonadectomy, that always leads to a need for hormonal substitution, and moreover, in some cases prevents fertility. The aim of the thesis was to better determine the risk for tumor development in relation to the functional and phenotypical characteristics in patients with complete form of androgen insensitivity syndrome (CAIS) and patients with 45,X/46,XY gonadal dysgenesis (GD).

Hematoxiline and eosine staining and immunohistochemical detection of OCT3/4, TSPY and KITLG were used to assess 37 and 36 gonadal tissue samples of 19 CAIS patients, respectively, and 84 samples from 47 patients with 45,X/46,XY GD. The results were correlated with gonadal position and expected level of androgen receptor activity in the first group. The gained data were compared with level of virilization of external genitalia in the second group.

Due to an unequal distribution in relation to the age in patients with CAIS, it was not possible to independently assess the influence of gonadal position (inguinal versus abdominal) on the histological gonadal changes. Expected residual androgen receptor activity has a positive effect on survival of general germ cell population but not on the development of the atypical (neoplastic) changes of the germ cells.

A significant relation between tumor risk and level of virilization of external genitalia was demonstrated in patients with 45,X/46,XY GD. The risk is the highest in patients with ambiguous genitalia; in patients with mild undervirilization or patients with phenotype of Turner syndrome it is much lower.