S. GNJIDIĆ[1]
B. TODORIĆ-ŽIVANOVIĆ[1]
K. GARDAŠEVIĆ[2]
M. STRNAD[1]
Z. HAJDUKOVIĆ[3]

[1]INSTITUTE OF PATOLOGY, MILITARY MEDICAL ACADEMY, BELGRADE, YUGOSLAVIA
[2]CLINIC OF REUMATOLOGY, MILITARY MEDICAL ACADEMY, BELGRADE, YUGOSLAVIA
[3]CLINIC OF ENDOCRINOLOGY, MILITARY MEDICAL ACADEMY, BELGRADE, YUGOSLAVIA

Klinefelter's syndrome with a pericentric inversion of chromosome 9, 47,XXY, inv(9)(p11q13) - A case report

KEYWORDS: Klinefelter's syndrome; Mosaicism; Down's Syndrome

Klinefelter Syndrome is the most common disorder among males, which is associated with sex chromosomes. It is characterized by an extra X chromosome, which is the result of nondisjunction in parents' gametes. In 80% of all cases the karyotype is 47,XXY, while in 15% of cases there is a mosaicism, 47, XXY/46,XY. The syndrome is associated with hypogonadism and azoospermia (except in rare cases of mosaicism), and with a tall stature and long extremities. Some data indicate disorders in behaviour and speech and lower IQ. The incidence of this syndrome is 1 in 600-1000 newborn males. From some literature data, it is associated with a higher incidence of some malignant diseases, especially with breast cancer. The use of exogenous androgens during and after puberty can improve psychosexual development. The incidence of inversion of the chromosome 9, inv (9) (p11q13), in normal population is very small and it seems that this inversion doesn't have pathological phenotypic expression. It occurs rare in Klinefelter's Syndrome. We report a patient with a hypogonadism in whom Barr bodies were detected on smears of buccal mucosa. The cytogenetic analysis was done with chromosomes from T lymphocytes after 72h of cultivation in the presence of PHA mitogen and GTG banding technique. 16 mitoses were analysed. The presence of an extra X chromosome and pericentric inversion of chromosome 9, inv(9)(p11q13) were detected. This inversion is confirmed by the C banding technique, which stains specific high repetitive sequences of secondary constriction of chromosomes. Population studies in Japan indicate a higher incidence of inversion of chromosome 9 in normal female group than in normal male group. The incidence is lower in Down's Syndrome patients. In Klinefelter syndrome the incidence is slightly higher in the male than in female group. Why is there a difference in the incidence of this inversion among sexes, and what is the genetic effect of this inversion are still the questions.

Archive of Oncology 2002;10(Suppl 1):88.

Meeting abstract
UDC: 616-006.66:616.988.6:616-037

Biljana HADŽI-NICHEVA[1]
Irina PRODANOVA[2]
Genghis YASHAR[2]
Katerina KUBELKA[2]
Zlata GRNCHAROVSKA[1]
Neli BASHESKA[2]

[1]DEPARTMENT OF OBSTETRICS AND GYNAECOLOGY, MEDICAL FACULTY, SKOPJE, MACEDONIA
[2]DEPARTMENT OF HYSTOPATHOLOGY AND CLINICAL CYTOLOGY, INSTITUTE OF RADIOTHERAPY AND ONCOLOGY, MEDICAL FACULTY, SKOPJE, MACEDONIA

Uterine myxoid leiomyosarcoma arising in a leiomyoma - A case report

KEYWORDS: Uterine Neoplasms; Leiomyosarcoma; Immunohistochemistry

Myxoid leiomyosarcoma of the uterus (MLU) is a very rare neoplasm. We report a case of MLU arising from a pre-existing leiomyoma. Between 1989 and 2001, 45 uterine leiomyosarcomas were diagnosed in our laboratory and MLU was established in one case only (2.1%). Subtotal hysterectomy was performed on a 56-year woman with clinical diagnosis of a uterine myoma. The histopathological processing included hematoxylin-eosin, histochemical and immunohistochemical staining of selected specimens. Macroscopically, the uterus measured 15 x 12 x 11 cm, with an indistinctly circumscribed multi-nodular tumorous mass 10.5 cm in diameter, and infiltrative satellite nodules in the surrounding myometrium. The morphology of the neoplasm showed a leiomyoma with distinct degenerative changes, necrosis and hemorrhage. In some areas the cells were round or oval with a vacuolated cytoplasm, slight atypia and rare mitoses. The mitotic count was 1-3 cells/10 HPF. Due to the presence of an abundant mucoid substance, these areas appeared as hypocellular. The additional processing confirmed the smooth muscular origin of the neoplasm (Masson trichrome, azan, desmine, alfa-smooth muscle actin and vimentine positive). The areas of the mixomatose nodules were characterized with hormone independence (estrogen and progesterone receptors negative), high proliferative activity (Ki-67 - 30%) and the presence of p53 protein product (45%). The areas of the pre-existing leiomyoma showed hormone dependence, low proliferative activity and absence of p53 protein product. Additional immunostaining is useful in supporting a diagnosis of MLU in myxoid uterine smooth-muscle tumors with a low mitotic rate.