Summary. Turner syndrome was firstly described by N. Shereshevskiy in 1925, and then G. Terner in 1938. In 1959, Ford found that patients with this syndrome miss one X chromosome.

This article summarizes data features of phenotypic manifestations of Turner syndrome, depending on the variant of chromosomal abnormalities.

**Key words**: Turner's syndrome, girls, X-chromosome.

The lack of X-chromosome is only one form of monosomy in humans, which is compatible with both embryonic and postnatal development.

Sex chromosome abnormalities causing Turner's syndrome (TS) are well known and require karyotyping. The frequency of the syndrome is 1 in 50 fertilized. Only 1% of these fetuses are born alive, more than 99% of embryos or fetuses are eliminated at different stages of development. Population TS rate among newborn girls 1: 2000 - 1: 5000. Phenotypic manifestations of chromosomal aberrations inherent clinical variability caused by chromosomal polymorphism and genetic heterogeneity.

Turner syndrome is one of the chromosomal diseases, which have a variety of cytogenetic forms: the complete absence in all cells of one of the X chromosomes - monosomy X chromosome (45, X - 60-70% of cases); mosaicism for X-linked with the existence of normal clone of cells; structural adjustment of one of the X chromosomes, mainly as isochromosome of long arm (Xqi); mosaicism for isochromosome and monosome clone (45H / 46HHqi) and deletion of short (Hr-) and long (Hq-) shoulder; ring chromosome; variety X/X translocations (30-40% of cases) and others.

The predominant karyotypes and their approximate frequencies are: 45, X — 50%; 46, Xi(Xq) — 15%; mosaic 45, X/46, XX — 15%; mosaic 45, X/46, Xi(Xq) — about 5%; 45, X, another anomaly - almost 5%; other mosaic 45, X/? — about 5%.

Common clinical signs of TS: short stature, sexual infantilism (hypogonadism, sufficient genitals), short folded neck, broad (thyroid) chest, well placed nipples, low growth of hair on his head, neck pterygium, high and narrow
palate. Infants with this syndrome often inherit important feature - swelling of the back surface of the feet and hands. In the pathological process are involved other systems, congenital malformations, disorders of the endocrine system (estroheniya, elevated levels of gonadotropins, thyroid dysfunction, diabetes), congenital peripheral lymphedema, cystic hygroma of fetus and others.

For partial or complete X monosomy, indifferent gonads develop in embryonic ovaries with oocytes as in normal condition. This continues for 3 months of development. Next comes the progressive degeneration of herminal cells - their replacement by connective tissue.

Adult patient's gonads are presented by connective strands, undifferentiated rudiments mostly without ovarian cells. This situation is not absolute. Normal ovaries in morphology occur in 18% of patients, but spontaneous menarche recorded only in 2-5% of patients, physiological pregnancy occurs in 3,6-7,6% of patients with Turner's syndrome. Occasionally, there are cases of normal ovarian function in patients with X monosomy.

There are 9 genes localized on the X chromosome that determine the normal development and functioning of the ovaries.

In the pseudoautosomal area of short arm of the X chromosome is mapped a regulatory transcription factor: gene SHOX (short stature homeobox-containing gene) as a major genetic determinant of growth. His expression is found in many tissues: skeletal muscle, kidney, pancreas, heart, brain and tissues of the bone marrow. In featus SHOX gene expression is found in the developing limbs in the first and second pharyngeal somites, which is the evidence of its involvement in the formation of the skeleton.

In 70% of cases of TS, single X chromosome has maternal origin. In passing, we note that the contribution of maternal chromosomes no division is 20-30% of TS by type (45, X). X chromosome of parental origin provides phenotypic signs of disease, such as obesity, abnormalities of the kidneys and eyes, disorder of lipid metabolism. Impacts of imprinting genes, located on the short arm of the X
chromosome is assumed. Thus, PCR allows effective detection of monosomy, hidden mosaicism and parental origin of the X chromosome.

So mosaicism is the result of postzygote violations. In the study of cultures of several tissues, particularly tissues of gonads, is not difficult to detect mosaicism.

Thus, the possibility of phenotypic realization of chromosomal polymorphism in Turner syndrome demonstrates the relevance and feasibility of studies in this area, to understand the place and role of genetic factors in shaping of the variability of pathological processes.