Ryznychuk M.O.

PECULIARITIES OF THE PHENOTYPE AND GENETIC VARIABILITY IN CHILDREN WITH TURNER SYNDROME

Department of Pediatrics and Medical Genetics
Bukovinian State Medical University

About 0.5% of all newborns are born with chromosomal abnormalities, which is a total of 1 case per 170 newborns. More than half of these rearrangements are caused by the abnormality of the number or structure of sex chromosomes.

The aim of the work was to study the characteristics of the phenotype and genetic component in children with Turner syndrome (TS) in the Chernivtsi region.

Thirteen patients with Turner syndrome with monosomy of the X chromosome and 1 child with a mosaic variant of the syndrome, registered at the regional children's clinical hospital in Chernivtsi, were examined. Medical and genetic counseling was carried out, which included the assessment of anamnestic data, somatogenetic examination, clinical and genealogical analysis. The somatogenetic examination of the patients was carried out in accordance with the methodological recommendations "Description of the phenotype" (Y. Grechanina et al., 1999). The assessment of physical development was carried out on the basis of the results of anthropometric indicators - height and body weight. To assess the degree of deviation of the patient's height from the average height in the population, the coefficient of standard deviation SDS (Standard Deviation Score) was calculated using the formula: SDS = XX'/ SD, where X is the patient's height, X' is the average final height for a given patient, SD is a standard deviation for a given chronological age and gender. The growth standards developed by J. Tanner and R. Whitehouse (1976) were used. Growth rates were compared with percentile curves of A.J. Lyon for girls with STS.

Children of the observation group were residents of rural areas in 67% of cases, and in 33% residents of the city. All children were stunted (100%). Hypergonadotropic hypogonadism was revealed in 87% of children. Sporadic puberty was observed in only one child (6.7%) with a mosaic variant of the syndrome.

The analysis showed that 33% of girls with STSappealed for medical help at puberty, while other children were diagnosed by puberty, 67%. The main complaints of girls upon treatment were growth retardation and absence of secondary sex characteristics.

The studied subjects had the following frequency of the main clinical and phenotypic disorders: antimongoloid eye incision (80%), ptosis (80%), eye hypertelorism (87%), epicanthic fold (33%), strabismus (20%), micrognathia (20%), high Gothic palate (87%), cleft palate (6.7%), nasal tone of voice (33%), abnormalities in the structure of the ears (87%), low hair growth on the neck (100%), short neck (100%), barrel chest (87%), coarctation of the aorta (6.7%), mitral valve prolapse (40%), hypertelorism and nipple retraction (87%), scoliosis (33%), brachydactyly (100%), short fourths and fifth metacarpal bones (6.7%), nail dysplasia (6.7%), sandal gap (33%), delayed skeletal maturation (87%), multiple pigmented nevi (20%), excess weight (67%).

Average age at the time of examination was 13.78 ± 0.11 years, average height 131.9 cm, stunting -4.2 ± 0.22 SDS, bone age 12.10 ± 0.12 years, difference between chronological and bone the age was 1.68 ± 0.01 years, the average weight was 34.9 kg.

When analyzing the growth rates of girls with TS, it was found that the indicator was on average within the 3rdand 10thpercentiles of the growth curves for girls with TS.

Thus, most of the girls who applied for help with TS were under the age of 11 (67%). In the Chernivtsi region, the following types of karyotype were mainly identified in girls with TS: monosomal (45 X0) , 93.3%, mosaic variant,6.7%.

The phenotypic features of TS remain without due attention from pediatricians and pediatric gynecologists, which indicates the need for a more attentive attitude towards girls with growth retardation and sexual development with mandatory karyotype research.