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TURNER SYNDROME IN CHILDREN OF NORTHERN BUKOVINA

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Background. Turner syndrome refers to orphan diseases. From 17 to 25 cases of this syndrome are firstly diagnosing every year in Ukraine. Besides monosomy, pathology of X chromosome (60-70%) in Turner syndrome may be caused by a wide genetic polymorphisms (variants of mosaicism, deletions of short and long arm of the X chromosome, isochromosome, ring chromosomes (30-40%)).

The aim of research was to study the phenotype of children with Turner syndrome depending of genotype

Methods: The study involved 8 patients with Turner syndrome with monosomy of the X chromosome and 1 child with mosaic variant of the syndrome, who were registered in the Regional Children's hospital of Chernivtsi. The average age at the moment of the survey was $13,78 \pm 0,11$ years old. A medical-genetic counseling, somato-genetic testing, clinical and genealogical analysis were performed.

Results. In all children occurred low growth (100%). In 56% of children was found hypergonadotropic hypogonadism. Sporadic puberty was only in one child (11%) with the mosaic variant of syndrome. The main complaints of girls were growth retardation and lack of secondary sexual characteristics. There were such basic phenotypic violation in research: antimongoloid eyes (89%), ptosis (78%), ocular hypertelorism (89%), epikant (33%), strabismus (22%), micrognathia (22%), high gothic palate (89%), cleft soft palate (11%), abnormal structure of ears (89%), low growth of hair on the neck (100%), short neck (100%) barrel chest (89%), coarctation of the aorta (11%), mitral valve prolapse (44%), hypertelorism of nipples (89%), scoliosis (33%), brachydactyly (100%), short fourth and fifth metacarpal bones (11%), nail dysplasia (11%), sandal cleft (33%), delayed skeletal maturation (56%), multiple pigmented nevi (22%) , overweight (67%).

Average growth was - 131,9 cm, growth retardation - $4,2 \pm 0,22$ SDS, bone age $12,10 \pm 0,12$ years, average weight - 34,9 kg.

Conclusion. Phenotypic features of Turner syndrome are overlooking by pediatricians and children gynecologists, suggesting the need for careful attention to girls with delayed growth and sexual development.