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CLINICAL POLYMORPHISIS OF GONOSOMAL ANEUPLOIDIES IN HUMAN ON THE EXAMPLE OF SYNDROME XXX

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Introduction. Trisomy X chromosome (TXCh) is one of the most common aneuploidy in humans. In most cases, karyotype 47,XXX is found in women, but isolated cases of TXCh in men have been described. Cases of mosaicism 47,XXX/46,XX are also described in the literature; 45,X/47,XXX; 47,XXX/48,XXXX; 47,XXX/94,XXXXXXX. Etiopathogenesis of TXCh in almost 80% of cases is associated with the mismatch of the maternal sex chromosomes in the first meiotic diverge, less often TXCh can occur during the first diverge of the zygote. In approximately 90% of cases, the syndrome is of maternal origin and only in 10% of cases – paternal. Associations of the appearance of TXCh with an increase in the age of the mother were revealed [1-3].

TXCh syndrome is characterized by clinical polymorphism. The most characteristic phenotypic manifestations: male physique, relative microcephaly, epicenter, hypertelorism, micrognathia, flattened nose bridge, Mongoloid eye incision, auricle deformities, nipple hypertelorism, craniofacial anomalies (cleft hard and soft palate), clinodactyly, hammer-like toes. Flattening of the feet. Growth is above average. Congenital heart and urogenital tract defects, delayed speech development, mental disorders (depression, psychosis in the context of schizophrenia), behavioral disorders; difficulty communicating with peers; retardation of physical development. Intellectual development may remain within normal limits or there may be a slight delay in mental development [4].

In most cases, sexual development is normal, secondary sexual characteristics are pronounced, the menstrual cycle is regular. Less often, the delay in the formation of secondary sexual characteristics, the late formation of the menstrual cycle, are recorded. Reproductive function in women with trisomy X, especially with the mosaic variant, is often preserved, but an increased frequency of miscarriage can be

observed. For patients with CTCh, heterosexual orientation is characteristic. Patients, compared with other women, often experience behavioral disorders and problems associated with low intelligence. 90% of women are not aware that their karyotype has an extra X chromosome. The diagnosis of CTCh is verified by examination of the karyotype. Possible prenatal diagnosis of the syndrome. Differential diagnosis of CTCh including other types of polysomy on the X chromosome, Shereshevsky-Turner syndrome [5, 6].

Materials and methods. A case of trisomy of the X chromosome in a girl with suspected Shereshevsky-Turner syndrome. To establish the diagnosis, a psychoneurological examination of the patient was performed, clinical-genealogical, syndromic, cytogenetic, instrumental (ultrasound, EEG, ECG, MRI) methods were used.

Results and discussion. A ten-year-old girl was referred for genetic consultation with a diagnosis of Cerebral Palsy, Spastic Diplegia. Dislalia. Short stature. Inspection results: hypoplasia of the middle part of the face, Mongoloid eye incision, hypertelorism, micrognathia, clinodactyly of the fifth finger, narrow shoulders. The skin is clean, dry, pink. Height 148 cm, weight 41 kg. Physical development is below the 3rd percentile, proportional.

According to the survey, the age of parents at the time of conception is mother is 37 years old, father is 40 years old. The girl was born from 2 desired pregnancy, which proceeded against the background of the threat of interruption. Born at 39 weeks of gestation, by surgery. Anthropometric data on proband at birth: weight – 3540 g, length – 54 cm, head circumference – 33 cm. From the first year of life, a delay in physical and mental development was observed. Started walking on toes at 1 year 4 months; often fell; from 3.5 years observed by a neurologist about lower spastic paraparesis. Lagged in growth from peers. The family has a senior healthy sibling. Parents are healthy, however they smoke and drink weekly alcohol. Heredity on the mother's side is not burdened on the father's side burdened by oncopathology and cardiovascular pathology.

From the consultative conclusions of pediatric endocrinologist, neurologist, orthopedist it is known: the thyroid gland is not palpated, clinically – euthyroidism. Sexual development by Tanner – I degree. Radiograph of the hands: bone age – 8 years; styloid process of the radial bone is not pronounced, but there is a pea-shaped bone. EEG: cortical rhythm is formed according to age with the phenomena of dysrhythmia, interhemispheric asymmetry with a decrease in amplitude in the left occipital region. Epiactivity was not detected. ECG: pronounced sinus arrhythmia. Heart rate – 93 b/min. Ultrasound of the abdominal cavity: signs of solitary cyst of the spleen, signs of sagittal rotation of the right kidney. Hip radiography: Hip dysplasia. MRI of the brain: a picture of a variant of Dandy-Walker malformation, lateroventriculoasymmetry, expansion of the subarachnoid spaces of the left cerebellar hemisphere. Scoliotic posture. Valgus installation of the lower extremities.

Given the specific phenotype of the child, cytogenetic examination was recommended. According to the results of karyotype 47,XXX, the syndrome of trisomy X of chromosome was established.

Conclusions. When conducting genetic counseling, attention should be paid to the polymorphism of the clinical manifestations of gonosomal aneuploidy. The development of CTCh is not typical for proband siblings. Offspring of patients may have children with chromosomal abnormalities, but the degree of risk is unknown. In most cases, the karyotype of the offspring is normal. Patients are advised to consult a psychologist, psychiatrist, classes with a speech therapist and inclusive training.

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