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GENOMIC ANOMALIES IN A BOY WITH MENTALLY DISABLED

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The role of genetic factors in the etiology of mental disorders and disorders of psycho-linguistic development is not in doubt. The study of genetic abnormalities in children with various diseases of the central nervous system is one of the urgent tasks in psychiatry and medical genetics [1-4].

Aim. Search for genetic factors in undifferentiated mental retardation in children.

Materials and methods. A clinical case of partial trisomy in a child with undifferentiated mental retardation. Clinical and genealogical, syndromic, cytogenetic, molecular genetic, biochemical, instrumental (ultrasound, MRI) methods were used to establish the diagnosis.

Results and discussion. Proband, a 7-year-old boy, was born from a second physiological birth with asphyxia, at 39-40 weeks of gestation, with facial edema, cyanosis of the skin. During childbirth, the child was delayed in the birth canal, using the Christler method and obstetric forceps with subsequent correction of the airways. Birth weight 3250 g; length – 51 cm; head circumference – 32 cm; chest circumference – 33 cm. On the scale Apgar scored 7-8 points. The child was under the supervision of a pediatric neurologist from birth with a diagnosis of "Left hemiparesis due to organic lesions of the CNS and natal lesions of the cervical spine." The child was under the supervision of a pediatric neurologist from birth with a diagnosis of "Left hemiparesis due to organic lesions of the CNS and natal lesions of the cervical spine." The boy began to hold his head from 9 months of age, sit from 1.5 years of age and walk from 5 years of age. Proband from 6 years is observed at the psychoneurologist concerning: "Organic defeats of the central nervous system with the expressed general delay of cognitive development (F06.8). Severe first-degree speech delay, alalia (F80.0)". Examination of the infant's brain using magnetic resonance imaging revealed a decrease in white matter and parietal lobes of the cerebral hemispheres, cerebellar hypoplasia, deformity of the lateral ventricles.

At the age of 7, the child underwent surgery for bilateral cryptorchidism.

The boy was first admitted to a geneticist at the age of 2 with a delay in psychomotor development of undetected origin and suspicion of amino acid

metabolism disorders. The geneticist recommended examining blood and urine amino acids and screening for microdeletion-related diseases. A molecular genetic study of Angelman and Prader-Willi syndrome was performed, which was negative. Serum and urine amino acid analysis also showed no significant deviations.

Upon repeated examination of the proband by a geneticist at the age of 7, the following phenotypic manifestations were observed: mental retardation, stereotypes, unstable gait, stigma of dimbriogenesis: hyperpigmentation on the toes and hands, protruding forehead; macrotia; wide nose, long filter; clavicle deformity in the form of a “bicycle handle”; Clinodactyly of the little finger; micropenia and microarchism. Family history: heredity of the mother - diabetes mellitus and oncopathology; on the paternal side - cardiovascular pathology. The brother of the mother’s father suffers from cerebral palsy. Parents deny blood marriage.

For further examination and verification of the diagnosis, the boy was sent for cytogenetic examination. Karyotype 47, XYY was detected; diagnosed with "dysomy syndrome Y".

Because the child's phenotype did not match the Y-chromosome dyssomy (high growth, rapid growth, large teeth and ears, stiff hair, slight decrease in intelligence), it was decided to examine the child using the molecular genetic method GTG. An abnormal male karyotype with an additional marker chromosome was identified, which was identified as a derivative of chromosomes 15 and 4 with probable breakpoints at 15q12 and 4p15.2:

47,XY,+ mar. ish der(15)t(4; 15)(p15.2; q12).

Parents were advised to determine the karyotype to exclude carriers of balanced chromosomal rearrangement. The risk of rebirth of a child with unbalanced chromosomal rearrangements in the form of partial trisomy of chromosomes 15 and 4 will depend on the results of parental karyotyping, as mutations may arise de novo or be inherited from one parent, as noted during medical genetic counseling.

Conclusions. The patient's medical history illustrates the difficulty of verifying the diagnosis in a child with neurological symptoms and mental retardation due to chromosomal abnormalities. Proves the need for a comprehensive examination of such patients using methods to detect genome abnormalities. The lack of modern technologies in the arsenal of scientific-practical and medical-diagnostic institutions that study mental and neurological disorders significantly reduces their diagnostic potential.

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