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THE ROLE OF GENETIC RESEARCH IN THE TREATMENT OF AUTISM. PHELAN-MCDERMID SYNDROME

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The role of genetic factors in the etiology of the majority of psycho-language disorders in children is undeniable. However, the identification of genetic changes and chromosomal syndromes, which are the cause of common diseases associated with autism, are often impossible, due to the number of factors that can cause autism. One of the common autistic syndromes is the Felan-McDermid microdeletion syndrome (SFM) (del 22q13 and r (22)).

The aim of the investigation was to demonstrate the phenotypic manifestations of SFM and the importance of current molecular cytogenetic diagnostic methods for verifying the diagnosis.

The clinical, genealogical, syndromological and cytogenetic methods were used.

SFM includes: 1) craniofacial anomalies: long face, epicanthus, large ears and nose, thin lips, sharp protruding chin, hypertelorism, ptosis, wide eyebrows, long eyelashes, wide cheeks; 2) anomalies of the musculoskeletal system: Syndactyly of the I and III fingers, sandale-shape cleft, clinodactyly, fleshy palms, dysplasia of the toenails; 3) anomalies of internal organs. The diagnosis is genetically determined. Differential diagnosis is performed with Angelman's syndrome, velocardiofacial syndrome, fragile X chromosome, FG syndrome, Prader-Willie syndrome, and Williams syndrome.

Medical-genetic counseling of a girl with signs of autism was conducted. A child was born from the 1st full-term pregnancy on the background of anemia, TORCH infection and the threat of interruption. Fetal ultrasound: intrauterine growth retardation. Childbirth on the 36-37th week, induced. On the 5th day, the child was transferred to the department of premature infants with diagnosis «Antenatal fetal injury, weight insufficiency, prematurity 36-37 weeks». She was observed by a pediatrician, pediatric neurologist and psychiatrist about microcephaly and delay in psycho-linguistic development. Heredity along the line of mother and father is burdened with cardiovascular pathology. Grandfather by father's line abused alcohol. Objectively: dolichocephaly, high forehead, flattening of the middle part of the face, deep-set eyes, full and swollen eyelids, long eyelashes, hypertelorism, full cheeks, enlarged ear lobes. The patient had autistic behavior. Based on the results of karyotyping and modern methods of cytogenetic research (46, XX, r (22) (p11.2q13), SFM was established. Recommendations for the treatment and rehabilitation of the child were given to the family.

This case demonstrates the importance of modern methods of molecular cytogenetic diagnostics for the verification of diagnosis, medical and genetic counseling of families with children with delayed psycho-linguistic development and determination of tactics of medical observation.

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INDICATORS OF BLOOD CELL IMMUNITY IN CHILDREN WITH SEVERE BRONCHIAL ASTHMA

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Although long-term studies of bronchial asthma (BA), conducted by the international community of scientists, the issue of increasing the effectiveness of treatment of this disease in children is important. According to the Global Strategy for the treatment and prevention of asthma, an adequate therapy can control the clinical manifestations of the disease. However, some of the patients developed asthma that difficult to treat. The ineffectiveness of inhaled corticosteroids, which are the basis of the asthma treatment, due, perhaps, the presence of different phenotypes of the disease, in particular phenotype "severe asthma".