Lastivka I.V.

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

Department of Pediatrics and Medical Genetics
Higher State Educational Establishment of Ukraine
«Bukovinian State Medical University»

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.

Marusyk U.I.

INDICATORS OF BLOOD CELL IMMUNITY IN CHILDREN WITH SEVERE BRONCHIAL ASTHMA

Department of Pediatrics and Children Infectious Diseases
Higher State Educational Establishment of Ukraine
«Bukovinian State Medical University»

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".

Considering this fact the objective of our scientific study was to rate indicators of cellular parts of the immune system in school-age children for verification severe phenotype of asthma and improve outcomes.

To achieve this purpose we have conducted our study following the two main tasks: to compare the content of T-helper cells (CD_4) children's in both clinical groups; to compare the content of T- killer/suppressor blood (CD_8) children's in both clinical groups.

60 school-age children with asthma in the remission period were comprehensively examined in the Pulmonology Department of Chernivtsi Regional Children's Clinical Hospital.

Over the course of the disease the patients were divided into two clinical groups. The first (I) clinical group consisted of 30 patients who had been registered severe asthma. The second (II) clinical group formed 30 patients, which was defined moderately severe asthma. For the main clinical features group were not differ.

All children performed immunological blood test II - III levels. The content of T-helper cells (CD_4) and T- killer/suppressor blood (CD_8) were determined by immunofluorescence using a set of monoclonal antibodies.

The results were analyzed by methods of variation statistics using statistical software StatSoft Statistica v5.0 and clinical epidemiology to the definition sensitivity (Se) and specificity (Sp) test, as well as the absolute (AR), relative (RR) and risk odds ratio (OR) indicating the 95% confidence interval (95% CI).

Most children with severe asthma recorded decreased relative content of T-lymphocyte function is associated with helper/inductor. Thus, the content of CD_4 cells less than 26,0% determined in 82,7% of children first clinical group and in 75% of subjects ($P>0,05$) the second. Thus the sensitivity of determination of the above mentioned relative content of CD_4 in the peripheral blood of pupils with severe asthma was 82,7% (95% CI 73,7-89,5) and specificity - only 25,0% (95% CI 16,8-34,6), with odds ratios of 1,5 (95% CI 0,8-3,1).

Elevated levels of CD_8 lymphocytes in peripheral blood (more than 14,0%) was determined in 69,5% of children with severe asthma phenotype, and only 37,5% of II group ($R\varphi<0,05$). Indicators of the diagnostic value of higher concentrations of T-lymphocytes in peripheral blood in detecting severe asthma phenotype relatively medium- severe disease course were as follows: sensitivity – 69,5% (95% CI 59,4-78,3), specificity – 62,5% (95% CI 52,2-71,9). Elevated to the above index content CD_8 lymphocytes in the peripheral blood associated with risk of having severe asthma phenotypes: the absolute risk – 0,3, the relative risk was equal to 1,9 (95% CI 1,4-2,6) with odds ratios – 3,79 (95% CI 2,1-6,7).

Thus, given the low likelihood ratio performance of cellular parts of the immune system in children, they are not appropriate for use on their own verification of the phenotype of severe asthma. Relative content of cytotoxic suppressor which was more then 14,0% was associated with risk of having a severe asthma phenotype with sensitivity of 69,5% and specificity of 62,5%, while the odds ratio – 3,79.

Peryzhniak A.I.

ANALYSIS OF GENERAL CLINICAL BLOOD INDICATORS IN NEWBORNS WITH IMPAIRED FUNCTIONAL STATE OF THE CARDIOVASCULAR SYSTEM

Department of Nursing Care and Higher Medical Nursing Education
Higher state educational establishmment of Ukraine
"Bukovinian State Medical University"

Nonspecific clinical symptomatology of the functional state disorders of the cardiovascular system can occur in a significant part of the pathology of the newborns, stipulating the necessity of its early diagnostics.

The aim of the study was to study the diagnostic information content of a set of indicators for a general blood test for the early diagnostics of posthypoxic myocardial damage in the newborns.