was observed. Condition deteriorated in the first weeks of life, when anxiety, flacidity, repeated vomiting, refusal to eat, loss of body weight appeared. Basing on complaints, electrolyte and hormonal disorders congenital adrenal hyperplasia, salt-wasting form was diagnosed. In a clinical blood test there was high thrombocytosis (up to 800×109), granulocyte shift, anemia. Child received substitution corticosteroid therapy, antibiotics, correction of fluid and electrolyte disorders. To eliminate heamoblastosis with primary thrombocytosis myelogram was conducted, pathology was not revealed. After empirical antibiotic therapy by meronem general improvement and normalization of all parameters of blood test were observed. In this case there was a secondary thrombocytosis associated with the underlying disease in combination with microbial infection processes on the background of immunodeficiency.

Conclusions: in the case of a transient thrombocytosis, which disappear after treatment of background cause, heamotological examination is not necessary. In cases of combined reasons of thrombocytosis (congenital adrenal hyperplasia and infections) thrice bacterial inoculation of all biological fluids should be included to plan of examination to provide target antibiotic therapy. Conversely, in the case of persist thrombocytosis in absence of an obvious cause, total hematological examination must be conducted.

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PEDIATRIC SPINAL MUSCULAR ATROPHY AS A SYNDROME OF FLACCID CHILD

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Proximal spinal muscular atrophy (SMA) of childhood is a severe, often fatal monogenic disease, inherited in an autosomal recessive manner. Pathogenetic basis of the disease is an affection of motor alpha neurons of the anterior horns of the spinal cord. This disease is connected with mutation of gene SMN1 (survival motor neuron-1) in 5q13.

Case report. The child N. of 1 year 4 months was admitted to the department of anesthesiology and intensive therapy with complaints of the absence of physical activity in the upper and lower limbs, cramps, refusal to eat, weight loss, dyspnoe. The boy was born from the second normal pregnancy in gestational age of 40 weeks. Delivery was physiological. The older child in

family had trisomy 21. At the first days of life the mother noted decrease in general motor activity, weak cry, skin jaundice. At the age of two weeks, there was a decrease in muscle tone of the upper and lower extremities, absence of sight fixation. After the 2nd week of life there was a general muscular hypotonia, movements of legs disappear, distinct dyspnoe. Because of this, child was examined by a neurologist and a geneticist. Basing on the results of molecular genetic studies the deletion of the 7th and 8th exons telomeric SMN1 gene in the homozygous state (spinal muscular atrophy) has been detected. Results of electromyography detected signs of a lesion of motor neurons of the spinal cord. At the age of 4 months a progressive deterioration with increasing bulbar syndrome, distinct reduced muscle tone and limited active movements was observed. Tendon reflexes from upper and lower extremities were absent. While admission condition was severe with respiratory insufficiency of II-III degree; infant was flaccid, adynamic, malnutrition of III degree, funnel chest deformity was observed. While examination, there were no active movements, areflexia presented, tendon reflexes were absent. Breathing was superficial, arrhythmic, dyspnoe. While percussion in the lower parts of right lung pulmonary sound was dull; while auscultation breathing was weakened. Cardiac tones were rhythmic, muffled with tachycardia. On the chest X-ray there was a right focal confluent pneumonia. Child was supported by ALV, due to a distinct dicrease of oxygen saturation. The child spent 365 days in the department, a tracheostomy was made because efforts to recover spontaneous breathing were not effective, the feeding was carried out through a tube. During his stay in the hospital pneumonia had undulating character; child received symptomatic, pathogenetic therapy and drugs that improve trophism of the nervous tissue. Despite conducted complex therapeutic measures, the child's condition progressively worsened and he died of a cerebral, cardio-respiratory, renal failure.

Suchwise, after the detection of the symptoms of weakness and diffuse hypotonia in children, physicians should suspect high probability of the diagnosis of SMA.