Described by French physician D. Louis-Bar, and later by American doctors E. Boder, RP Sedgwick, the syndrome (synonym - ataxia telangiectasia) is hereditary, and usually its onset can be marked in infancy. Most of the children suffer from psychomotor retardation. Manifestation of the disease begins with neurological symptoms: cerebellar ataxia with gait impairment, tremor of head and trunk, choreoathetosis, eye-bulbs movement disorders. When a child is 2-6 year old, asymmetric telangiectasia appears on the skin and mucosa, dermatodyschromia is marked.

In addition to the lesions of skin and nervous system, Louis-Bar syndrome is characterized by combined immunodeficiency that causes a tendency to frequent infectious diseases and oncology, the formation of chronic broncho-pulmonary processes. Immunogram demonstrates that immune serum globulin A, sometimes G and E, reaches bottom or is absent. Cytogenetics of lymphocytes shows a chromosomal instability. Pathomorphological study discovers hypoplasia or aplasia of thymus gland, lymph nodes, spleen [1,2].

Morphologically ataxia telangiectasia is characterized by degenerative changes of cerebellum tissue, in particular by progressive death of Purkinje cells. Degenerative changes may affect other brain structures [3].

Louis-Bar syndrome is inherited by autosomal recessive mode, associated with mutations in the ATM (ataxia telangiectasia mutated), whose main function is to repair DNA breaks and maintain the integrity of the genome in the cell. Genomic instability at chromosomal level is a genetic cause not only of Louis-Bar syndrome, but of other central nervous system disorders. It is known that regular and mosaic forms of chromosomal diseases always negatively affect brain function and are manifested in various forms of mental retardation, autism and epilepsy [4,5]. The combination of epilepsy with rare hereditary or congenital diseases can cause significant difficulties in the diagnosis of these diseases, and make their prognosis more complicated.

**Own observation:** There was a case of combination of Louis-Bar syndrome with epilepsy in a 15-year old girl in our clinical practice. Clinical diagnosis: "Louis-Bar syndrome. Focal symptomatic epilepsy with frequent attacks in the form of complex absence seizures and complex partial seizures, mental retardation in the stage imbecility. Chronic suppurative endobronchitis. Chronic pyelonephritis. Delay of physical growth".

It’s known from anamnesis that the delay of child's physical and psychomotor development was observed during the first year of its life. The late development of crawling and walking art, delay in speech development were marked. Since birth the girl repeatedly suffered
from ARVI, bronchitis, streptoderma, enterocolitis. When she was 2-year old the girl suffered from sepsis, bilateral bronchopneumonia, hepatitis. The delay of neuro-mental and physical development, as well as a frequent infectious disease physicians associated with defects of the care and feeding of the patient.

Initial neurological diagnosis "ICP, ataxic form" was made in 3-years-age of patient. Later, the symptoms such as staggering the head, torso, convergent strabismus, dysarthria, hypotension limb muscles. Pediatricians and neurologists noted that disease occurs with negative dynamics: in the girl grew manifestations cerebellar ataxia with progressive loss of movement and self-service skills, there was a delay of the psycho-speech development, dysarthria with a decrease in vocabulary.

At 10-years age the clinical picture of the disease was aggravated by the appearance of seizures. Epilepsy with frequent attacks in the form of complex partial seizures was diagnosed. A combination with epilepsy complicated an early diagnosis of the underlying disease. The diagnosis of "Louis-Bar syndrome" was made when the girl was 11-year old on the basis of clinical symptoms of cerebellar ataxia, dysarthria, telangiectasia, and characteristic changes in immunogram - lymphopenia, a significant reduction of IgA and IgE.

Thus, the above case demonstrates the difficulty of early diagnosis of Louis-Bar syndrome in children, because its possible combination with pathology of the nervous system makes the diagnosing more difficult. Despite the specificity of the clinical picture of this form of primary immunodeficiency, different variants for the development and course of the disease, with a gradual manifestation pathognomonic symptoms can hide syndrome Louis-Bar under the mask of various forms of cerebellar ataxia. An early verification of the diagnosis requires a careful analysis of clinical and anamnestic data in conjunction with a differential finding the causes of recurrence of infectious diseases in children with neurological disorders.