# CLINICAL AND DIAGNOSTIC FEATURES OF ATYPICAL FORMS OF PROGRESSIVE MUSCULAR DUSHEN DYSTROPHY

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Among the hereditary groups of the disease, neuromuscular diseases are considered the most common. The disease is complex, differential diagnosis, especially congenital myopathy. Common signs of congenital structural myopathies are early onset, skeletal anomaly, generalized muscle hypotension, and severe complications from the cardiovascular system and respiratory tract. Statistical data The incidence of progressive Duchenne muscular dystrophy (PMDD) varies from 10 to 33 per 100 thousand boys in the general population. The high growth of the disease is associated with the growth of a mutation in the dystrophy gene, known as the largest human gene (l). The mechanism of the disease is directly related to the gene defect, but the process of pathogenesis of the violation of the immune factor, which triggers the mechanism of impaired regeneration, plays a significant role. The disease goes through a certain stage, a defect in the lipid layer increases permeability (during muscle load), which makes it unprotected from a T-cell attack, creatinenase rushes into the blood, an inflammation cascade is triggered, in turn, the growth factor causes a loss of muscle mass. The continuous stream of degeneration depletes muscle tissue (L). In the literature, you can find a study aimed at the clinical assessment of mitochondrial changes in myopathies, so attention is drawn to the energy supply of organs to the calcium content, thereby ensuring the movement of muscle proteins. During the development of PMDD, the body changes the structure and the system responsible for the transport of calcium ions, the efficiency of calcium filling decreases, and the mitochondria does not retain ions (L). The above stipulates the need for available clinical and instrumental methods for assessing the disease, taking into account the progression. Electroneuromyography is considered the standard instrumental method, but this method has limitations in terms of age. In recent years, the MRI neuroimaging method is considered to be an adequate and potential research method, which makes it possible to give a complete description of the muscle disorder with the determination of the activity of the process, in the stages of treatment and rehabilitation measures.

**Purpose.**
To study the clinical and diagnostic periods of the disease progressive Duchenne muscular dystrophy in children.

**Material and research methods.**

The examination included children aged 5 to 15 years with a diagnosis of PMDD, on the basis of the Department of Pediatric Neurology 1-Clinic SamMI, for the period 2018-2022. The contingent of sick children was admitted to the Clinic from all regions of the Samarkand region, the main district sending for hospitalization is Urgutsky, the distinction of this region is the observance of the tradition of family marriages. The Department of Pediatric Neurology of 1-Clinic SamMI is the only center for the treatment and rehabilitation of such patients, respectively, the number of patients is significant. The first studies of hereditary diseases at the Department of Neurology of SamMI were carried out by Prof. Khananova F.K. (thanks to which a genetics course and a separate screening center were opened at the department in 1990). A total of 68 converts, patients underwent repeated courses of treatment and rehabilitation from 3 to 6 months. Among the examined boys, 58, girls, 10. Children underwent a full clinical and neurological examination, examination by pediatricians, cardiologists. All patients underwent ECG, EEG examination, electroneuromyography in 78%, and MRI studies in 53%. Laboratory research blood biochemistry, CPK analysis, ultrasound examination of internal organs.

**Research result.**

In accordance with the set goal of the work, patients were examined at the age from 5 to 10 years old and from 10 to 15 years old. Having studied in detail the anamnesis, the patients were sent for examination at screening centers in Samarkand and Tashkent at the Institute of Genetics, where they received confirmation of the diagnosis. The hereditary type of definition was found in 25 patients, in other cases it was a gene mutation. The obstetric history of the mothers of the examined patients is favorable. In the history of the pedigree, from the maternal side, cases of the disease were noted, more often in a straight line. The examination of patients, as mentioned above, was carried out in the Department of Pediatric Neurology of the 1-Clinic of SamMI, out of the examined children, 18 patients first applied, the rest have been observed during the last 3 years. The collection of anamnesis and complaints, as a rule, was assessed according to the words of relatives. Table 1 presents the characteristics of complaints (from relatives).

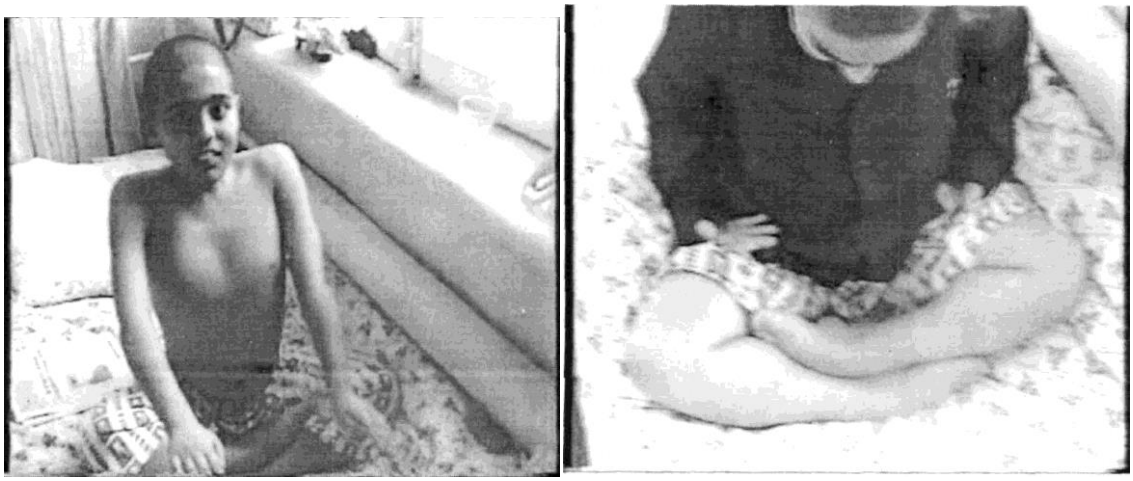
**Card 1**

**Subjective symptoms in patients with PMDD (%)**

| Patient complaints | PMDD n=58 |
|---|---|
| **Movement disorders:** | |
| Weakness in the legs, arms | 100 |
| Difficulty walking, climbing stairs | 100 |
| Restriction of movements in the legs, arms (contractures) | 75 |
| Walking on tiptoes, violation of foot support | 60 |
| Somatic complaints | |
| Dyspnea | Dyspnea |
| Tachycardia | Tachycardia |
| Cognitive impairment | |
| Decreased memory | 78 |
| Carelessness | 45 |
| It's hard to concentrate | 55 |

Attention is drawn to the fact that the decrease in cognitive function, in the form of mental ability in children, worries relatives least of all, in most cases, they explained this with a "calm" balanced character of the child. The severity of muscle weakness depended on the length of the disease, those who first applied had 3-4 points, observed in older age and a longer period of the disease 1-0 points (parents brought children in their arms or in a wheelchair). As well as the condition of the muscles, hyperlardosis increased depending on the duration of the disease. "Duck gait" was noted in all patients, but by its nature, the severity was different, for example, in children with a hereditary type of transmission, the gait had a classic type, children with a gene mutation, a characteristic gait, sometimes "on toes", sometimes "slow" did not differ from the usual slow gait of a healthy person, more often this was observed in girls. Over time, the gait is aggravated by the difficulty of climbing stairs (even through an insignificant threshold of the room); stumbles and falls, all this is due to the difficulty of maintaining the body in an upright position. Rapid progression was noted in 5 patients with adipose variant of the disease. At the time of examination, the atrophic form of PMDD was 80%, the rest of the cases of the adipose variant were boys with a high degree of obesity, of the Itsengo-Kushenga type. Early onset of contractures and retraction, rapid progression, pronounced impairment of the cardiovascular and respiratory systems, immobility occurred within 1 year from the onset of the disease.

In addition to increasing muscle weakness, the examined patients showed symptoms characteristic of some, which were not traced in others. Pseudohypertraphy of the gastrocnemius muscles against the background of general atrophy of all muscle groups, especially the upper girdle, was detected in 13 children, with a vivid manifestation, resembling the legs of a football player or weightlifter (Fig. 1).



The exception was the Achilles reflexes, taking into account atrophy of skeletal muscles, a cut in the legs in 16 children, reflexes were preserved, even at a late stage. Three children had pseudohypertraphy of the tongue - macroglossia, the tongue literally "fell out" from the oral cavity. In the same patients, facial numbness was determined, it was difficult to close the eyes, thickening of the lips (due to the replacement of the muscles of the lips with connective and adipose tissue). One of the qualities of children who are noted by their parents as positive is little emotionalism. Parents do not suspect that lack of initiative, calmness in character, these are the initial manifestations of mental retardation. According to scientific studies, in most cases, an autism spectrum disorder is associated with Duchenne disease, a deficiency of cerebral dystrophin isoforms does not correlate with muscle defect. Another feature of the manifestation of clinical forms of PMDD in our observations is the case of girls. The clinical signs were different from those

in boys. So, if in boys the disease progressed rapidly, then in girls it remained stable. In almost all girls examined, the disease occurred in puberty, which is confirmed by many scientists, hormonal changes are a provocateur. Not expressed atrophy of skeletal muscles, there is no pseudohypertrophy of the gastrocnemius muscles, mental retardation is poorly expressed.

Electroneuromyography showed signs of primary muscular disorders in the legs and arms. There was a decrease in the amplitude of evoked activity, more in the proximal parts of the limbs, the degree of the progressive process varied from mild to pronounced. As the disease lasts, the interference pattern turns into a reduced one due to a decrease in recruitment, and in the end a complete "silence" is determined.

In healthy people, CPK synthesizes creatine and ATP, in patients with PMDD, there is a huge release of CPK, which is the initiator of the inflammation reaction. In the examined patients, in boys, the concentration of CPK exceeded more than 10 times, in girls from 2 to 4 times. A small part of the examined children (mainly children who are able to walk without support) underwent neuroimaging studies. According to MRI data, changes in the femoral muscles were determined in 63%, in the muscles of the lower leg, about 28% of the examined, these were the first time patients. Experienced patients had 100% lesions of the femoral muscles and 76% of the lower leg.

Thus, PMDD is a group of hereditary diseases with the main progression pattern of skeletal muscle atrophy. Mutation of the dystrophin gene is observed in most cases, which leads to staged deformity of the spine, progressive course of cardiopathy, pulmonary insufficiency and, as a result, death.

**OUTPUT**

1. The presented cases of progressive Duchenne muscular dystrophy were distinguished by clinical polymorphism. A special place is occupied by cases of the disease in girls and the adipose variant. Clinical signs are ambiguous, scattered as well as the process of progression itself, this conclusion was made on the basis of early research by Professor of the Department of SamMI FK Hannanova. (1993)

2. Neuroimaging analysis of the skeleton of muscles in patients with PMDD showed a high level of degeneration in the proximal regions, less pronounced in the distal regions, and with the duration of the disease, the nature of degeneration is aggravated. This method is recommended for assessing the preservation of the muscle structure and diagnosing the effectiveness of treatment.

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