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DYSFUNCTION IN THE HIP JOINTS IN CHILDREN WITH CHARCOT-MARIE-TOOTH SYNDROME (LITERATURE REVIEW)

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A review of the literature on the treatment of children with dysfunction in the hip joints in motor-sensory neuropathy Charcot-Marie-Tooth is presented. Peculiarities of disease diagnosis and the approach used in the treatment of patients are described.

The Charcot-Marie-Tooth syndrome is a hereditary neuromuscular disease characterized by progressive atrophy of the distal muscle group of the lower limbs. According to international authors, the incidence of hip joint dysfunction in this condition is at least 10%, ranking second only to foot deformities. In the Russian literature, the problem has not been adequately interpreted.

Early diagnosis of dysfunction in the hip joints during Charcot-Marie-Tooth syndrome is complicated by the child's age and is characterized by progression. Conflicting clinical signs and trivial symptoms of the disease also confuse diagnosis, until it becomes clearer in adolescence or the second or third decade of life. Surgical reconstructive operations on the hip joint often occur too late, and they are accompanied by a greater frequency of neurological complications.

Practitioner awareness coupled with an early diagnosis of hip subluxation and decentration and complex orthopedic and neurological examinations of children with the disease of Charcot-Marie-Tooth should result in more favorable outcomes.

Keywords: Charcot-Marie-Tooth disease, hip instability subluxation, hereditary motor and sensory neuropathy, CMTHD, hip dysplasia.

The main cause of discontiguity in the hip joints of children is hip dysplasia and its consequences. In an analysis of patients treated at a department of pathology specializing in hip joints at Research Pediatric Orthopedic Institute of G. I. Turner, some patients, particularly school-age children and adolescents with newly diagnosed subluxation of the hip, also had neural amyotrophy of Charcot–Marie–Tooth disease (CMTD).

CMTD is a hereditary neuromuscular disease first described in 1886 by Jean-Martin Charcot and Pierre Marie from France and Howard Henry Tooth from Great Britain. The disease is based on the primary defect in the structure of the peripheral nerves, the myelin of damaged motor or sensory fibers, which is manifested by weakness and atrophy of the distal muscle groups of the lower extremities [1-11].

It is one of the most common inherited neuromuscular diseases occurring in 36 of 100,000 people and is characterized by clinical and genetic heterogeneity. The disease can be inherited in three different ways: X-linked, autosomal dominant, or autosomal recessive. Currently, mutations of more than 60 genes are known to cause the disease [12-14]. The most frequent form of the disease is type 1A (80% of cases) with an autosomal dominant mode of inheritance, which is based on duplication of the 17r11.2 locus. While point mutations associated with the disease have been recorded in many genes, as a rule, their frequency is low, and they are often unique [15, 16]. Approximately 20% of cases of CTMD type 1 are due to autosomal dominant inheritance caused by mutations de novo, while 89% have a paternal and 11% have a maternal origin [17, 18].

Early diagnosis is difficult because of conflicting clinical signs and a low symptom load. In 10% of patients, clinical manifestations of the disease are minimal, and diagnosis is possible only upon a careful clinical and electromyographic (EMG) study [19]. A characteristic electroneuromyographic (ENMG) sign of the disease is a decrease in the conduction velocity (CV) of the peripheral nerves to below 38 m/s. For the median nerve, this number is 25 to 35 m/s. Given the expressed genetic

heterogeneity of hereditary demyelinating polyneuropathy, DNA analysis plays the most important role in the diagnosis of certain genetic variants. The disease is currently incurable, but in most cases, it is slowly progressive and does not lead to severe disability of the patient. Less than 5% of patients with CTMD type 1 require a wheelchair, and for them, life expectancy is not reduced [1, 2, 5, 20].

Clinical Presentation

Hypotrophy and decreased strength of the peroneal muscles and feet are the main complaints of patients. Other complaints include fatigue during prolonged walking, instability, frequent stumbling, and difficulty going up and down stairs or jumping. Initial clinical symptoms are difficulty in dorsiflexion of the foot and impaired sensitivity in the distal extremity of the "socks" type as well as reduced vibration sensitivity, especially in the lower extremities, as noted in 70%–80% of cases. Impaired sensitivity, however, is rarely observed by patients themselves. The involvement of the upper limbs occurs several years after the onset of the disease [1, 2, 3, 6, 7, 19, 21]. In 60%–70% of patients, tendon reflexes in the distal portions of the extremities are decreased, which fade as the disease progresses [4, 5, 19]. Future progressive muscle hypotrophy leads to foot deformities (high arch or claw toes), and gait takes on the character of steppage [1, 2, 5].

Foot deformities are regarded by most reseachers to be the obligatory indication of CTMD type 1 and have been described in detail [22, 23]. On the contrary, in Russian literature, to our knowledge, there have been no reports of defects of the hip joints in children with CMTD.

The combination of discontiguity of the hip joints and hereditary polyneuropathy was originally described by S. J. Kumar et al. in 1985 [24]. They reported on five 8- to 15-year-old children with decentration and subluxation of the hip and who required surgery. In all 5 children, the disease was asymptomatic, or there were minimal clinical manifestations.

More recent studies report at least 10% incidence of such disorders in the hip in these patients, which is ranked second after foot deformities [25, 26, 27, 28, 29]. Some researchers suggest an even greater prevalence of pathology due to a significant number of erased forms and rare identification of the disease before adolescence [20, 28, 30, 31, 32].

Ushiyama T. et al. (2003) reported on 3 patients from the same family with hereditary motor and sensory neuropathy type 1 and severe bilateral hip subluxation and concluded that focused examination of the hip joints in such families is essential for the early diagnosis of the disease [31].

The definition of the root cause of discontiguity in the hip joints during CMTD, whether it results from congenital dysplasia or the patient's neurological status, is debatable, and there is no single point of view regarding this issue [24, 25, 31, 33, 34]. Naturally, it is possible that there is an element of true congenital hip dysplasia in some patients with CMTD. However, violations of the stability of a dysplastic hip (subluxation of the hip) are usually diagnosed in the first year of life. By the time of the onset of clinical manifestations, the main features of dysplasia must be characteristic changes of the acetabulum, a taper arch, reduction of its length and volume, and the thickening of its bottom [35]. According to literature, in cases of CMTD, more pronounced changes are noted in the femur, and disturbed relationships in the hip joints are caused to a greater extent by subluxation of the hip due to valgus and torsional deformation of the neck and valgus position of the epiphysis [36, 37].

The pathogenesis of hip joint damage in CMTD is not fully understood. There are suggestions that progressive neuromuscular abnormalities lead to weakness of the muscles in the hip region, which, in turn, leads to weakness of the hip extensors. C. J. Newman et al. (2007) studied gait characteristics of patients observed walking with legs wide apart and external rotation of the hip [38]. Van Erve R.H. and Driessen A.P. (1999), G. Chan et al. (2006), and T.D. Bird (2008) concluded that after birth, such patients have a stable hip and that subluxation of the hip and deformation of the acetabulum develop secondarily with the growth of the child [20, 26, 32]. Disturbances in the hip joints on the diseased child's growth are characterized by progression and are clinically expressed by abnormal gait and appearance of pain [25, 26, 38].

An important feature of the disease is that until adolescence or the second to third decade of life, it often is asymptomatic, and clinical signs or decentered hip subluxation in children with CMTD may be the first manifestation of distress [24, 29, 39].

M.J. Hadianfard and A. Ashraf (2012) considered that contrary to prevailing notions, neuropathy is not always to be treated as a process that affects only the distal parts of the extremities [31, 40].

The principles of the surgical correction of the hip in children with CMTD are generally similar to those for the treatment of congenital dysplasia. Depending on the age of the patient, worldwide, corrective osteotomy of the femur and acetabulum, transposition surgery, a Salter procedure, a triple pelvic osteotomy, and the Bernese periacetabular osteotomy are common [26, 41]. The implementation of Salter pelvis osteotomy for the correction of spatial position of the acetabulum and femoral head coverage deficiency is possible in patients up to 7 to 8 years old. For older children and adolescents, more extensive surgical procedures are necessary [42].

According to literature, an essential feature in the surgical treatment of patients with unstable hip joints and CMTD is the high probability of the occurrence of neurological complications [43]. Given the relatively high morbidity of interventions conducted in patients with adverse neurological backgrounds, neuropathy of the peroneal nerve has been repeatedly reported, particularly after triple pelvic osteotomy and periacetabular procedures when surgery is performed in the vicinity of the sciatic nerve (*nervus ischiadicus*) [32, 41]. It is natural to assume that when performing hip replacement in adults with CMTD and violations of the stability of the hip joint, the risk of neuropathy of the sciatic nerve tract significantly increases because of the need of thigh pull-through.

Conclusion

The analysis of publications in literature suggests the reality of this issue. When identifying disturbed relationships in the hip joints of children older than 7 to 8 years, we believe that it is necessary to conduct a comprehensive orthopedic and neurological examination (including EMG and ENMG) to prevent neuromuscular diseases such as peripheral neuropathy. The early diagnosis of decentration and subluxation of the hip in children with CMTD will allow reconstructive surgery to be performed less traumatically and before the development of secondary deformations of joint components, thereby postponing arthroplasty. Careful examination, an individual approach, and complex therapy in these patients will help reduce the number of complications and achieve better treatment results.

Pre- and postoperative medications in these patients, rehabilitation treatment, and evaluation of features of formation of the hip joints during the long-term follow-up need further studies.

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НАРУШЕНИЕ СООТНОШЕНИЙ В ТАЗОБЕДРЕННЫХ СУСТАВАХ У ДЕТЕЙ С БОЛЕЗНЬЮ ШАРКО–МАРИ–ТУТА (ОБЗОР ЛИТЕРАТУРЫ)

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Представлен обзор литературы по проблеме лечения детей с нарушением соотношений в тазобедренных суставах при моторно-сенсорной невропатии Шарко–Мари–Тута. Рассмотрены особенности диагностики заболевания и подхода в лечении больных.

Болезнь Шарко–Мари–Тута (БШМТ) — наследственное нейромышечное заболевание, характеризующееся прогрессирующей атрофией дистальной группы мышц нижних конечностей. По данным зарубежных авторов, частота поражения тазобедренных суставов при БШМТ составляет не менее 10 %, занимая второе место после деформации стоп. В отечественной же литературе проблема не получила должного освещения.

Нарушения соотношений в тазобедренных суставах при БШМТ с возрастом ребенка характеризуются прогрессированием, однако ранняя диагностика часто затруднена из-за противоречивых клинических признаков и незначительной выраженности симптомов заболевания до подросткового возраста или второго-третьего десятилетия жизни. Хирургические реконструктивные вмешательства на тазобедренном суставе часто выполняются запоздало и сопровождаются большей частотой неврологических осложнений.

Осведомленность практических врачей, ранняя диагностика децентрации и подвывиха бедра, комплексное ортопедоневрологическое обследование детей с болезнью Шарко–Мари–Тута позволят рассчитывать на получение лучших результатов лечения.

Ключевые слова: болезнь Шарко–Мари–Тута, БШМТ, наследственная моторно-сенсорная невропатия, тазобедренный сустав, подвывих бедра.

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