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The current state of diagnosis and treatment of the congenital tibia pseudoarthrosis

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Congenital pseudarthrosis of the tibia (CPT) is a rare disease that is detected with a frequency of 1 in 140–250,000 newborns. The disease is characterized by a wide range of clinical and radiological signs from progressive antecurvature deformation of the tibia to nonunion with a significant bone defect. Changes in the CPT area are caused by the influence of pathologically altered periosteum, which forms a fibrous hamartoma and is responsible for the deformity of the biomechanical properties of bone tissue. CPT can be formed at the moment of birth or developed spontaneously or as a result of minimal trauma in the early years. The main method of treatment of CPT is a surgery. Nowadays a number of surgical techniques, which are actively used and improved by specialists in the world, has been developed, The most used methods are the Ilizarov's method, application of intramedullary fixators, techniques with the use of vascularized tibial autograft, «induced membrane» technique. However, there are a few studies on comparing the effectiveness of different techniques or metal fixatives, most of them are presented in the format of a retrospective analysis of clinical cases series. This is due to the rarity of the disease and the lack of unified approaches on the choice of surgical treatment techniques. The main aim of surgical treatment of CPT is to achieve consolidation in the area of pseudoarthrosis, which may restore the limb resistance. The part of primary consolidation of CPT after using the surgical treatment various techniques varies very much, range from 60 to 100 %. The percentage of children with CPT tibial amputations has decreased significantly over the past 30 years, which generally indicates an improvement of the results of surgical treatment of the mentioned pathology. However, CPT still remains one of the most difficult diseases of pediatric orthopedics due to the large number of unsatisfactory results and complications after surgery. Key words. Congenital pseudarthrosis of the tibia, neurofibromatosis, surgery, treatment, children.

Уроджений псевдоартроз кісток гомілки (УПКГ) — є рідкісним захворюванням, яке виявляють із частотою 1 на 140–250 000 новонароджених. Хвороба характеризується широким спектром клініко-рентгенологічних проявів від прогресованої антекурваційної деформації кісток гомілки до незрошення зі значним дефектом кісткової тканини. Зміни в ділянці УПКГ обумовлені впливом патологічно зміненого окістя, що формує фіброзну гамартому та відповідає за порушення біомеханічних властивостей кісткової тканини. УПКГ може бути сформованим на момент народження чи розвинути у перші роки життя спонтанно або внаслідок мінімальної травми. Основним методом лікування УПКГ є хірургічний. На сьогодні розроблено велику кількість хірургічних методик, які активно використовують і вдосконалюють фахівці у світі. Найуживанішими серед них є: метод Ілізарова, застосування інтрамедулярних фіксаторів, методики з використанням васкуляризованого автотрансплантата малогомілкової кістки, методика «індукованої мембрани». Проте досліджень щодо порівняння ефективності різних методик чи металофіксаторів наразі небагато, більшість подано у форматі ретроспективного аналізу серії клінічних випадків. Це пояснюється рідкістю захворювання та відсутністю уніфікованих підходів до вибору методики хірургічного лікування. Основною метою хірургічного лікування УПКГ є досягнення консолідації в зоні псевдоартрозу, що має дозволити відновити опорність кінцівки. Зосереджені на цьому хірурги, зазвичай, нехтують супутніми ортопедичними деформаціями кінцівки. Частка первинної консолідація УПКГ після застосування різних методик оперативного втручання дуже варіює, становлячи від 60 до 100 %. Відсоток ампутацій кісток гомілки за УПКГ у дітей значно знизився за останні 30 років, що, загалом, свідчить про покращення результатів хірургічного лікування зазначеної патології. Проте УПКГ і на сьогодні залишається одним із найскладніших захворювань дитячої ортопедії через велику кількість незадовільних результатів та ускладнень.

Key words. Congenital pseudarthrosis of the tibia, neurofibromatosis, surgery, treatment, children

Congenital pseudoarthrosis of the tibia (CPT) is a rare disease that occurs with an incidence of 1 in 140–250,000 newborns [1]. CPT is characterized by a wide range of clinical and radiological manifestations ranging from progressive antecurvature deformity of the tibia to nonunion with an extensive bone defect. Today it remains one of the most difficult in pediatric orthopedics, due to the large number of unsatisfactory results and complications of surgical treatment [2]. An anomaly of the development of the tibia with CPT was recorded in 60–90 % of cases [1]. This disease can be formed both at birth and spontaneously in the first years of life, or as a result of minimal trauma. Spontaneous healing of CPT, without treatment does not occur, the disease is accompanied by the formation of deformities and shortening of the leg, violation of the supporting function of the limb [3]. The etiology of the disease remains unclear, but it has been determined that CPT in 40 % of children is associated with neurofibromatosis type I, less often with fibrous dysplasia and osteofibrous Campanacci dysplasia [4]. Numerous theories on the cause of tibia damage are given. According to modern ideas, the main abnormal changes causing the development of CPT are localized in the periosteal membrane of the tibia. A. Codivilla was the first to note abnormal changes in the periosteum [5]. He proved that in the CPT zone periosteum thickens with an increase in the number of neuron-like cells that surround the capillary-type vessels, triggering their narrowing and obliteration. These changes can lead to local hypoxia of bone tissue in the subperiosteal zone with its subsequent resorption [6]. According to another study, pathological changes in the area of CPT are due to the influence of pathologically altered periosteum, this tissue has a special name — fibrous hamartoma, whose cells have a relatively increased osteoclastic activity on the background of suppressed osteoblastic one, compared to the normal periosteum [7].

Many CPT classifications have been proposed: by Crawford [8], El-Rosassy-Paley [9], Boyd [10], Andersen [11], Pozdeev [12]. However, none of them is generally accepted, and although some have proven prognostic value, they do not allow the choice of surgical treatment. The most commonly used is the X-ray classification by Crawford, according to which CPT is divided into four types, which describe the nature of tibial bone damage from the mildest degree — anterolateral deformity (type I) to pseudoarthrosis with bone defect (type IV) [13]. Another popular classification scheme is the clinical-radiological by El-Rosassy-Paley, which takes into account the geometry

of bone fragments, mobility at the level of pseudoarthrosis and the presence of performed surgical interventions [9]. The Boyd classification is considered prognostic, in which CPT is divided into six types, which allows to predict the course of the disease and the outcome of treatment [10]. The Pozdeev classification reflects the relationship between the etiology and features of the course of CPT [12].

Congenital pseudoarthrosis of the tibia is treated with conservative and surgical methods.

Conservative treatment of CPT

Conservative treatment involves the use of plaster casts and individual orthoses. It, as an independent method, has limited indications, because it involves long-term use of orthoses until the completion of skeletal growth [14]. Only isolated cases of the effectiveness of this approach are described. This treatment is often used as part of an integrated approach and starts from the moment of diagnosis of CPT at the stage of fracture. It helps to postpone surgery, which has a positive prognostic effect on the outcome of treatment. Indications for conservative treatment are also the postoperative period, when orthoses are used to prevent refraction.

Another method of such treatment is therapeutic exercise, which aims to increase volume of movements in adjacent joints of the limb [15].

Physiotherapy is used in the case of myelodysplasia to improve neurotrophic function of the limb [16].

Surgical treatment

Surgical method is the main in the treatment of children with CPT. Even in the late 1990s, its results were often unsatisfactory, characterized by low efficiency (adhesions reached less than 20 %), a significant number of surgeries, frequent complications in the form of refractions, and in 30–35 % of cases such treatment ended in amputation of the leg [17]. The main difficulties faced by the surgeon in the treatment of CPT are the reduced potential for fusion in the area of pseudoarthrosis, the tendency to refraction of the tibia and the difficulty of fixing small bone fragments in children with impaired bone quality. Today, due to significant technical progress and a better understanding of pathogenesis and biomechanical features of osteoporotic bone fixation, treatment outcomes have improved significantly. However, there is no generally accepted protocol for surgical treatment, and various techniques show different effectiveness.

McFarland [18], Pozdeev surgery or the method of controlled growth of the distal tibia are performed during the latent phase of CPT, i.e. before the formation of pseudoarthrosis.

Fundamentally different surgical techniques and methods of fixation are used for the treatment of CPT at the stage of fracture: intramedullary stabilization with bone grafting, Ilizarov's method using an external fixation device (EFD), combined methods of intramedullary and external fixation, replacement of vascularized autotransplant of the fibula, «induced membrane» method by Masqulet.

McFarland's surgery involves performing bone autoplasty using a contralateral tibial autotransplant to prevent a tibial fracture (Fig. 1) [18]. O. Ofluoglu et al. [24] showed positive results of the use of this technique in 10 patients, in whom 5–7 years after the performance of prophylactic bone autoplasty, no case of pseudoarthrosis on the operated limb was identified. However, the authors also found moderate shortening and secondary valgus deformity of the ankle joint in 40% of patients [19].

Pozdeev surgery

A. P. Pozdeev et al. [20] proposed an original technique for the use of preventive bone alloplasty with demineralized grafts. According to the analysis of the clinical group of 38 patients, the implementation of such interventions prevented the development of tibial fractures in 84.2 % of cases. The advantage of the technique is the use of an allograft, which prevents the complications associated with obtaining a graft («donor-site morbidity»).

Surgical treatment by Ilizarov

Ilizarov method for surgical treatment of CPT involves the use of the basic principles of compression-distraction osteosynthesis and distraction osteogenesis in EFD [28] (Fig. 2). There are several techniques using circular EFD and the principles of Ilizarov's

method, including: closed compression of the CPT zone; resection with simultaneous shortening and compression; resection with segmental bone transport; resection, autoosseous plasticity and distraction-compression osteosynthesis.

Ilizarov method has advantages, allowing simultaneous performance of compression osteosynthesis of bone fragments and correction of deformity of tibial bones. According to one of the largest multicenter studies conducted by the European Association of Pediatric Orthopedists in 2000 and included an analysis of the results of surgical treatment of 340 children with CPT (1,287 operations), Ilizarov method is recommended for surgical treatment of CPT in children because it has the highest rate of consolidation. in the area of CPT. Treatment of 108 tibiae with CPT required 194 Ilizarov surgeries.

Consolidation of CPT after the first operation was achieved in 65.4 % of cases. Two or more surgeries were performed on 46 legs. The total consolidation rate was 75.5 % [22].

The main disadvantages of the method are infectious complications at the sites of introduction of transosseous elements, as well as refractions, which occur, usually at the level of the previous CPT and reach 34.8 %. they are prevented by introduction of intramedullary fixators after consolidation is achieved [24].

In addition, Ilizarov surgery is the main one used for subsequent surgical correction of residual deformity and shortening of tibia and foot bones in patients with CPT, which significantly affect the cosmetic condition (appearance) and limb fitness and are essential characteristics of the tibia by CPT [25].

Surgical treatment with the use of intramedullary fixators

The original technique was proposed by J. Charnley in 1956 and involved resection of the CPT area,

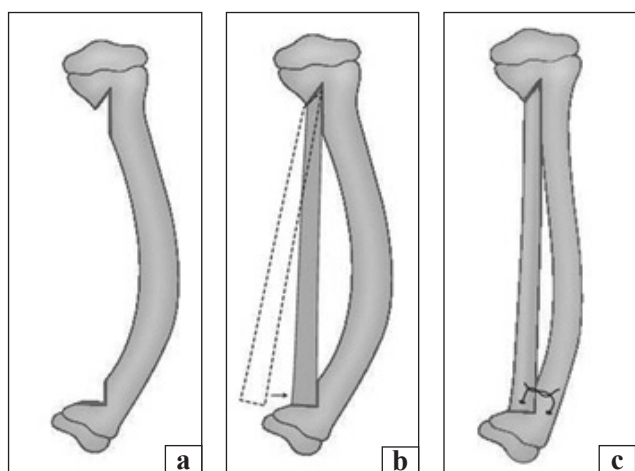


Fig. 1. Scheme of surgical interventions at the stage of latent CPT phase in McFarland surgery: a — the formation of grooves in the tibia; b and c — fixation of the autotransplant of the tibia (by [18])

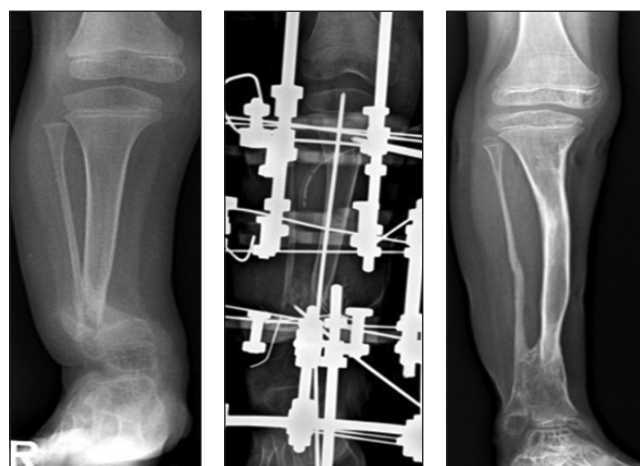


Fig. 2. An example of the use of EFD (according to [23])

followed by intramedullary fixation of fragments and bone grafting [26]. The technique was later modified by P. Williams with the addition of transarticular insertion of the rod through the subtalar and talocrural joints [27]. The technique has become the standard of surgical treatment for decades, demonstrating a fairly high (from 50 to 85%) efficiency of fusion of the CPT area. However, the indicators differed according to different authors due to modifications of intervention techniques and different intramedullary fixators. All researchers confirmed the need for long splinting of the tibia with an intramedullary fixator to prevent fractures (Fig. 3).

The advantages of intramedullary fixation for the treatment of CPT is the possibility of effective correction of angular deformation, which normalizes the distribution of forces during the axial load of the leg, as well as providing a «splinting» effect to prevent the development of refractions.

The choice of intramedullary metal fixator (Kirchner spokes, Steinman rods, telescopic Fassieur-Duval clamps) depends on the surgeon's experience and affordability [28].

Disadvantages of this method of surgical treatment include shortening of the tibia due to resection of the CPT area, as well as the inability to achieve the correction of concomitant deformities, which are usually present in the talocrural joint of the affected tibia.

Surgical treatment by combined methods

The combined use of EFD and intramedullary devices with bone autoplasty allows to apply the biomechanical advantages of both clamps. EFD helps to achieve correction of the position of the bone fragments of the leg, while providing the necessary conditions for consolidation of stability, and intra-

medullary fixator prevents refraction in the area of CPT [30]. The results of surgical treatment of CPT with resection, bone autoplasty and combined use of intramedullary and external devices (EFD) help to obtain consolidation from 40 to 100 % of cases. The frequency of refraction does not exceed 40 %. There is no consensus on the timing of preventive intramedullary fixation of the tibia [31]. In particular, some specialists perform intramedullary fixation during the main stage of surgical treatment, others perform preventive splinting of the tibia after reaching the fusion in the area of operation.

Surgical treatment using a vascularized fibular autograft (VFA) was first described by Taylor et al. in 1975. The technique consists of three stages: resection of abnormal tissues in the area of CPT, obtaining a vascularized autograft from the contralateral fibula and its installation in the area of postresection defect of CPT, vascular anastomosis [32]. Surgical techniques of VFA transfer from contra- and ipsilateral extremities have been described.

The method of CPT treatment with the use of VFA allows to obtain up to 100 % consolidation. It can be performed even for children from the age of one. However, its application requires a multi-team approach involving a vascular surgeon and, consequently, microsurgical instruments. This intervention is technically complex, so it is currently used in few centers of the world [33]. Specific complications of the technique include the formation of valgus deformity of the talocrural joint on the side of the autograft, which is observed in 64 % of cases.

Surgical treatment using the «induced membrane» technique (Masquelet technique) involves two surgeries. The first is the resection of abnormally altered tissues in the area of CPT with the installation



Fig. 3. An example of the use of intramedullary Fassieur-Duval fixator. CPT radiography before (a) in stages (b, c) and after surgical treatment (d). Illustration taken from [29]

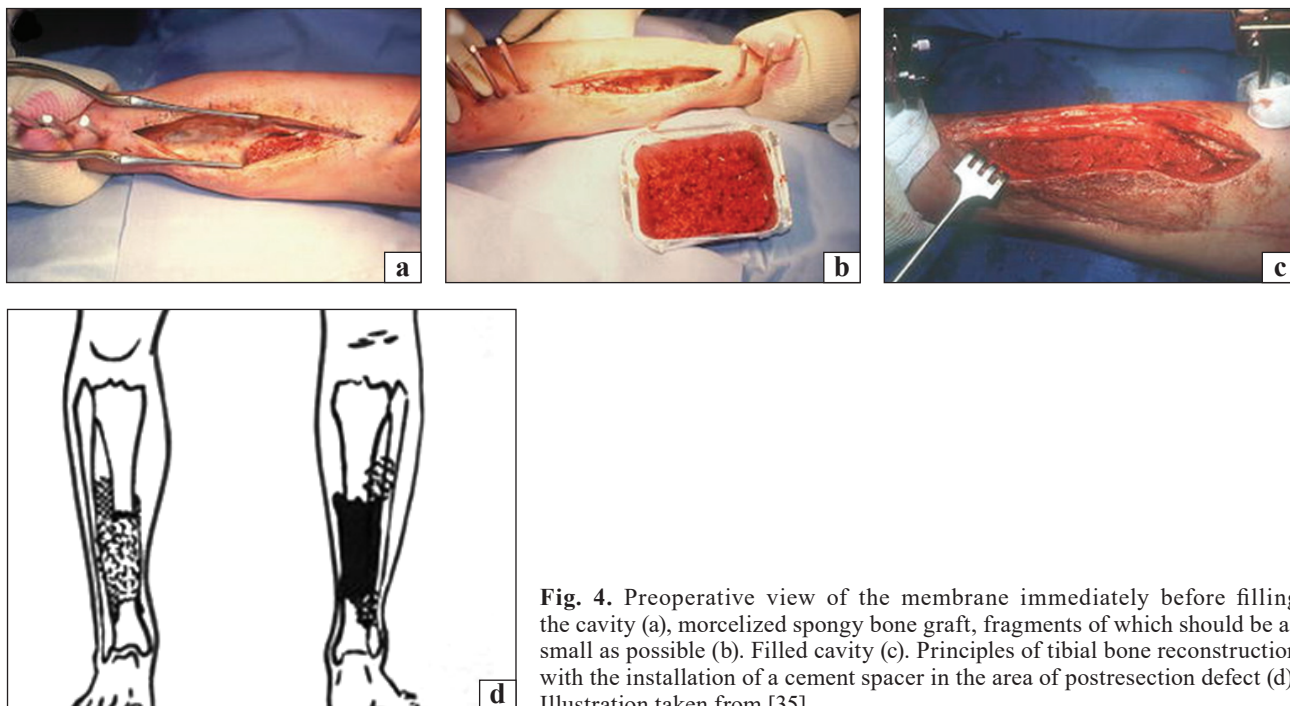


Fig. 4. Preoperative view of the membrane immediately before filling the cavity (a), morcelized spongy bone graft, fragments of which should be as small as possible (b). Filled cavity (c). Principles of tibial bone reconstruction with the installation of a cement spacer in the area of postresection defect (d). Illustration taken from [35]

of a cement spacer in the area of the post-resection defect, the tibia bones are temporarily stabilized by metal osteosynthesis [34]. The second surgery is performed in 6–8 weeks — the time required for the formation of a biologically active membrane, the so-called «induced membrane». The intervention involves the removal of the spacer and plastic repair of the defect with auto- or allobone (Fig. 4). The advantages of this technique are the possibility of its use even in patients who have previously undergone surgical treatment, which proved to be ineffective, and the possibility of replacing sufficiently large defects (up to 8 cm). The disadvantages are the need for double surgery, the duration of treatment and a sufficiently high frequency of refractures after achieving consolidation in the area of CPT.

Amputation

Attitudes toward amputation as a method of surgical treatment of CPT in children vary widely across countries and even medical centers within a country. Some experts consider it as a palliative technique performed in cases of ineffectiveness of reconstructive interventions. Others regard it as one of the effective methods of primary surgical treatment, which avoids multiple surgeries [36]. Several studies have demonstrated good functional results, rapid adaptation and high quality of life of children after amputations due to various abnormalities (injuries, cancer, etc.). L. A. Karol studied the function of walking in children with CPT after surgery and concluded that

the functional results after amputation of the foot are similar to those after standard surgery [37].

Modern views on the indications for amputation for CPT formulated by R. E. McCarthy [38] are as follows: the inability to achieve consolidation after at least three surgeries; significant shortening (more than 5 cm) and significant deformation of the foot and leg with impaired limb support function. Patients are made a prosthesis and allowed early axial loading. Indicators of CPT consolidation after amputation of the foot range from 0 to 90%, while the functionality and resistance of the operated limb does not depend on the radiological picture of consolidation [36].

Conclusions

Congenital pseudoarthrosis of the tibia is a disease that is quite rare and has a wide range of manifestations from progressive antecurvature deformity of the tibia to nonunion with a bone defect. Changes in the CPT area are due to the influence of abnormally altered periosteum, which forms a fibrous hamartoma and is responsible for the violation of the biomechanical properties of bone tissue.

The main method of treatment of CPT is surgical. To date, a large number of operational techniques have been developed that are actively used and improved in the world. The most commonly used are Ilizarov method, treatment with the use of intramedullary fixators, surgical techniques using a vascularized tibial autotransplant and the «induced membrane» technique.

There are currently few studies comparing the effectiveness of different techniques or metal fixatives, most of which are presented in the format of a retrospective analysis of a clinical sample. This is due to the rarity of the disease and the lack of unified approaches to the choice of surgical treatment. The main goal of CPT treatment is to achieve consolidation in the area of pseudoarthrosis, which should restore limb resistance. The disadvantage of the available studies is the concentration on achieving CPT consolidation and neglect of concomitant orthopedic deformities of the limb. The percentage of primary consolidation of CPT in application of various methods of surgical treatment varies greatly, ranging from 60 to 100 %. There is also a significant percentage of complications (nonunion and re-fracture) that require repeated surgery.

Conflict of interest. The authors declare the absence of conflict of interest.

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