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TREATMENT OF CONGENITAL CLUBFOOT IN A PATIENT WITH JACOBSEN SYNDROME USING PONSETI METHOD: A CASE REPORT

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Introduction. Jacobsen syndrome, characterized by multiple developmental anomalies, is a rare genetic syndrome caused by a partial deletion of the long arm of the 11th chromosome. The incidence is 1:100,000 live births. Patients of this group have malformations of the heart, kidneys, gastrointestinal tract, central nervous system, and skeleton. The severity of clinical features is diverse. Jacobsen syndrome rarely combines with a congenital clubfoot.
Case report. The clinical case of using the Ponseti method for the treatment of congenital clubfoot in combination with Jacobsen syndrome is presented. As a result, a complete primary correction of the foot was obtained, which did not relapse within 2 years.
Discussion. Only brief references to this pathology were found in the literature. In the case of our patient, a greater number of gypsum dressings were required to complete the primary correction of the foot.
Conclusion. Painless foot has been achieved, which has a full range of motion, which confirms the success of the application of the Ponseti method for the treatment of non-idiopathic congenital clubfoot and the need for using it as a starting method.

Keywords: congenital non-idiopathic clubfoot; Jacobsen syndrome; Ponseti method.

ЛЕЧЕНИЕ ВРОЖДЕННОЙ КОСОЛАПОСТИ В СОЧЕТАНИИ С СИНДРОМОМ ЯКОБСЕНА ПРИ ПОМОЩИ МЕТОДА ПОНСЕТИ

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Введение. Синдром Якобсена — редкий генетический синдром, вызванный частичной делецией длинного плеча 11-й хромосомы. Синдром Якобсена характеризуется множественными аномалиями развития. Частота встречаемости 1 : 100 000 живых новорожденных. Пациенты данной группы имеют мальформации сердца, почек, желудочно-кишечного тракта, ЦНС и скелета. Выраженность клинических особенностей разнообразна. Синдром Якобсена редко сочетается с врожденной косолапостью.
Описание клинического случая. Представлен клинический случай использования метода Понсети для лечения врожденной косолапости в сочетании с синдромом Якобсена. В результате была получена полная первичная коррекция стопы, которая не имеет рецидива в течение двух лет.
Дискуссия. В доступной литературе имеются лишь краткие упоминания об указанной патологии. В случае с нашим пациентом также потребовалось большее число гипсовых повязок для того, чтобы получить полную первичную коррекцию стопы.
Выводы. Достигнута опорная безболезненная стопа, обладающая полным объемом движений, что свидетельствует об успешности метода Понсети в лечении неидиопатической врожденной косолапости и необходимости его применения как стартового метода.

Ключевые слова: врожденная неидиопатическая косолапость; синдром Якобсена; метод Понсети.

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Introduction

Jacobsen syndrome (JS) is a rare group of genetic syndromes caused by a partial deletion of the long arm of chromosome 11. The characteristics of JS include multiple congenital anomalies combined with psychomotor retardation, visceral malformations, and some orthopedic problems [1]. This syndrome was first described by Jacobsen in 1973 in several members of one family who inherited an unbalanced translocation of 11;21 from a parent who carried a balanced translocation [1].

More than 200 JS cases have been described [2, 3] and its incidence is 1 : 100,000 live births. The ratio of males to females is 2 : 1 [2-4].

The most common clinical signs of JS include pre- and postnatal retardation of physical development, psychomotor development, characteristic facial dysmorphism, thrombocytopenia, or pancytopenia. Patients have malformations of the heart, kidneys, gastrointestinal tract, genitalia, central nervous system, and/or skeleton. There may also be impairments of hearing and vision, as well as hormonal and immunological disorders [2, 4].

The clinical severity of symptoms can vary. Cases of JS combined with such orthopedic pathologies as platypodia of the large and long great toe, clinodactyly, brachydactylia, syndactylia of toes 2 and 3, and zygodactyly are described. It should be noted that the combination of JS and congenital clubfoot is extremely rare. The available literature provides only a few references to this pathology, and there are no data on the severity, or the methods and results of treatment.

Currently, the gold standard for treating congenital clubfoot is the Ponseti method [5]. This method has revolutionized the treatment of congenital idiopathic and syndromic clubfoot [6].

In this publication, we present our own clinical observation of a patient of two years with JS combined with congenital clubfoot.

Case description

A full-term male (weight was 2870 g, height was 50 cm, Apgar score was 7/7) was delivered via cesarean section at 38 because of threatening fetal hypoxia (discharge of meconium-colored amniotic fluid and lack of effect of labor induction). During the initial examination on day 2 of life, we noted a moderately severe congenital eqinocavovarus deformity of the right foot, and degree II hypotrophy (Fig. 1). In addition, there were multiple signs of dysembryogenesis, namely low-lying auricles, hypertelorism, high palate, transverse sulci on the palmar surfaces of both hands, cryptorchism, hypoplasia of the preputium, and divergence of the rectus abdominal muscles. Because of the above clinical signs, his blood was sampled and sent to the genetic laboratory for karyotyping.

The severity was 5.5 points on the Pirani system [7] (Table 1) and 17 points on the Dimeglio system [8] (IV degree) (Table 2). Given the patient's generally severe condition, we postponed orthopedic treatment until his condition improvement. Later, frequent abundant regurgitation appeared and the child was transferred to the surgical department, where a duodenal obstruction was detected. He was sent to surgery day 12 where he was diagnosed with atresia of the duodenum. On clinical blood testing, thrombocytopenia was $84 \cdot 10^9$/l. Genetic testing revealed

Table 1

Initial severity on the Pirani scale (points)

Signs	Points
Curvature of the fibular border of the foot	0.5
Medial fold	1
Posterior fold	1
Lateral part of the head of talus	1
Calcaneus position	1
Equinus	1

Table 2

Initial severity on the Dimeglio scale

Signs	Correctability, °	Points	Additional signs	Points
Equinus	37	3	Deep posterior fold above the heel	1
Heel varus	38	3	Deep transverse plantar fold	1
Internal rotation of the foot	60	4	Significant cavus foot	1
Adduction of the forefoot	22	3	Significant muscular atrophy	1

Fig. 1. Patient's appearance before treatment

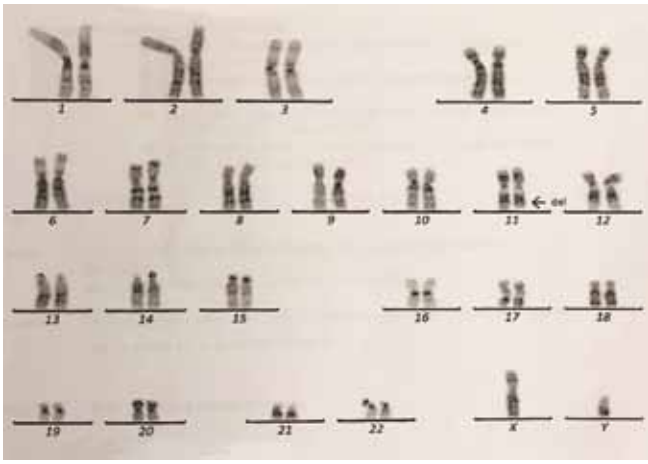
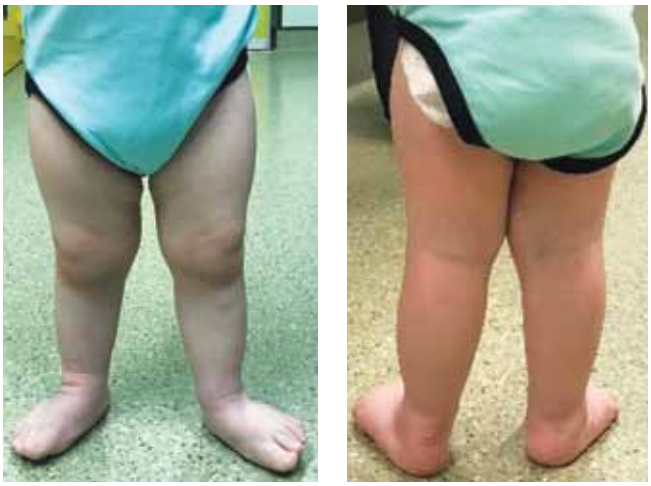


Fig. 2. Male karyotype, deletion of the long arm of chromosome 11 (locus 11q23)



a *b*

Fig. 3. The appearance of the patient after treatment: *a* — front view; *b* — rear view

JS 46,XY, del(11)(q23)[18]/46,XY[2], pathological clone with 11q deletion in 90% of metaphases (Fig. 2).

On day 17 of life, a high plaster cast was applied on the right lower extremity using the Ponseti method [5], from the tips of the toes to the upper third of the thigh in the supination position, for a period of 1 week. Before each subsequent plaster cast application, the foot was manipulated. After 9 staged plaster casts, foot correction was obtained (cavus, adduction, varus were corrected). To obtain complete foot correction, subcutaneous achillotenotomy was necessary. Due to the child's deteriorating condition, and pancytopenia on blood tests, the patient was hospitalized in the hematology department and underwent bone marrow puncture combined with subcutaneous achillotenotomy of the right foot. After 3 weeks, the cast was removed and his foot deformity was completely corrected. Changes in severity after the removal of the last plaster cast, according to Pirani and Dimeglio systems, are shown in Tables 3 and 4. The Denis Brown abduction splint was applied for 23 h per day for a period of 3 months. After the specified period, the time of use was reduced by 3 h for a period of 1 month. Later, the abduction splint use was reduced by 1 h per month upon reaching night sleep use.

Table 3

Severity on the Pirani scale after the removal of the last plaster cast (points)

Signs	Points
Curvature of the fibular border of foot	0
Medial fold	0
Posterior fold	0
Lateral part of the head of talus	0
Calcaneus position	0
Equinus	0

Currently, the child walks independently and without additional supports. The patient's appearance is presented in Figure 3.

Discussion

In everyday practice, cases of congenital idiopathic clubfoot are common; but there are very rare cases associated with connective tissue pathologies, neurological conditions, and chromosomal abnormalities [5, 9, 10]. The available literature provides only single references to this pathology, and there are also no data on its severity, methods, or results of treatment [1]. A small number of

Table 4

Dimeglio scale severity after removing the last plaster cast

Signs	Correctability, °	Points	Additional signs	Points
Equinus	–22	1	Deep posterior fold above the heel	0
Heel varus	–5	1	Deep transverse plantar fold	0
Internal rotation of the foot	–20	1	Significant cavus foot	0
Adduction of the forefoot	–5	1	Significant muscular atrophy	0

reports on the use of the Ponseti method for treating congenital syndromic clubfoot have been published [9, 11–13]. Janicki et al. [11] reported that patients with syndromic clubfoot needed more plaster casts, and often had relapses of the deformities, compared with idiopathic clubfoot. Moroney et al. [13], described the treatment of 29 patients with non-idiopathic clubfoot, and indicated primary correction success in 91% of cases; however, deformity relapse occurred in 44 and 37% cases, which required extensive surgical treatments. Gurnett et al. [10] also believe that more plaster casts were required to treat non-idiopathic clubfoot using the Ponseti method. All authors noted the efficacy of the Ponseti method and recommended it as a first-line treatment for syndromic or non-idiopathic congenital clubfoot.

In the case of our patient, similar conclusions were made. He needed additional plaster casts, and achieved complete primary foot correction, without relapse within two years. Ultimately, we achieved painless foot support with full range of motion.

Conclusions

The Ponseti method should be used as a first-line treatment for syndromic and non-idiopathic clubfoot to obtain a fully functional foot. The treatment of syndromic clubfoot requires multiple plaster casts.

Additional information

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Ethical review. The patient's parents signed a voluntary informed consent to participate in the study, as well as for the processing and publication of personal data.

Contributions of the authors

I.Yu. Kruglov was involved in examination and treatment of the patient, writing all sections of the article, as well as literature collection and processing.

N.Y. Rumyantsev, G.G. Omarov, N.N. Rumyantseva took part in the examination and treatment of the patient.

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