DOI: 10.17816/PTORS3429-36

OCCURRENCE OF RADIAL CLUB HAND IN CHILDREN WITH DIFFERENT SYNDROMES

Avdeichik N.V., Govorov A.V., Golyana S.I., Safonov A.V.

The Turner Scientific and Research Institute for Children's Orthopedics, Saint-Petersburg, Russian Federation

Aim. Clinical analysis of congenital radial club hand as part of some genetic syndromes as well as the optimization of methods of non-surgical and surgical treatment of patients with this pathology.

Material and Methods. From 2007 to 2014, we conducted a survey of 170 children with congenital radial club hand. Among them, 32 patients were diagnosed (18.8%) with different syndromes. We assessed the degree to which the radius was underdeveloped among this group of patients as well as the management features of patients according to various comorbidities.

Results. The assessment identified Holt–Oram syndrome in 17 children (nine boys and eight girls; 53.1%) and TAR-syndrome in nine children (four boys and five girls; 28.1%). VACTERL syndrome was detected in four male patients (12.5%) and Nagera syndrome was observed in two children (one boy and one girl; 6.25%). Surgical treatment of radial club hand in patients with genetic syndromes is the same as that of the patients with isolated congenital radial club hand: a single- or two-stage correction of the hand relative to the ulna with subsequent reconstruction of the rays of the hand. The duration of treatment of such patients did not significantly differ compared to the patients with isolated congenital radial club hand.

Conclusion. Congenital radial club hand, identified as part of genetic syndromes, requires a comprehensive examination to diagnose comorbidities, observation, and treatment by specialists to determine the optimal age for surgical correction of the existing strain of the upper limb.

Keywords: congenital radial club hand, Holt–Oram syndrome, VACTERL syndrome, TAR syndrome Nagera syndrome.

Introduction

Congenital radial club hand is a congenital upper limb malformation, characterized by hypoplasia occurring in a longitudinal direction resulting in the radial deviation of the wrist and shortening of the forearm. The degree of malformation varies from hypoplasia to aplasia of various segments of the forearm and wrist. The typical clinical sign of congenital radial club hand is the shortening of the distal radius.

Radial club hand is a longitudinal deficiency of the radial bone in the forearm. This congenital deficiency is estimated to occur in 1 in 30,000 to 1 in 100,000 live births. It characterized by an absent or incomplete radius, absent or incomplete thumb, malformations of fingers (usually from the second to the fourth), radial deviation of the wrist toward the thumb, and some degree of neuromuscular deficiency. It occurs bilaterally in 38%–58% of cases. The male to female ratio is 3:2 [1, 2, 3].

In 1733, Petit first described radial club hand in an autopsy of a neonate with bilateral club hands and absent radii [4]. Congenital radial club hand may occur as a separate disease or as a symptom of other genetic syndromes. Currently, more than 20 syndromes are known to be associated with radial club hand (radial dysplasia) [5].

Previous studies have failed to show any concrete genetic basis for the etiology of congenital radial syndrome. There are many theories regarding the etiological factors, which can be broadly divided into two groups, namely, exogenous and endogenous. In a study, Wiedemann et al (1962) proposed that 20% of defects were congenital, 20% resulted from exogenous factors and the remaining 60% were of unknown etiology [6, 7].

However, many authors prefer the current theory proposing that deformation results from the destruction or impeded development of the tissue from which the limb is developed [3, 8]. This is because of from various factors, including medication and radiation. In addition, mechanical factors, such as increased intrauterine pressure because of oligohydramnios, small amnion, amniotic constrictions, and tumors of the ovaries and uterus, may also play a part in disease etiology [5–7].

The most frequently associated and clinically significant syndromes are thrombocytopenia with the absence of the radial bone (TAR syndrome), Holt–Oram syndrome (hand–heart syndrome), VACTERL syndrome (VATER association), and Nager syndrome (acrofacial dysostosis) [1–3, 9–12]. Less common syndromes include the Baller–Gerold, Miller, and Roberts syndromes [5, 13]. Each of these syndromes requires an individual approach to formulate an optimal treatment strategy.

TAR syndrome. This syndrome was first described in 1959 by Shaw et al. [14]; in 1969, J. G. Hall with a group of collaborators established its diagnostic criteria [15]. It was concluded to be an autosomal dominant disorder and estimated to occur in 1 in 100,000 live births [14, 15]. It is also known that thrombocytopenia is because of complete absence of megakaryocytes in 66% cases, decreased in the number of megakaryocytes in 12%, and functionally inactive megakaryocytes in 12% and eosinophilia (55%), and anemia [13]. However, more than 50% of children grow up with normal platelet counts [16]. Approximately 40% of children die in infancy because of severe bleeding, while a small percentage of children gradually have a decrease in the number of thrombocytes as they grow [13].

Because of the high risk of bleeding, surgery is recommended in children with TAR syndrome only after the platelet count becomes normal [13, 16, 9, 14–15, 17–18]. In the musculoskeletal system, bilateral congenital anomalies of the shoulder bone and the shoulder joint definitively occur in cases of radial club hand.

The distinguishing feature of this syndrome is that the first ray of the hand remains intact [9]. In 50% of cases, it may affect the lower extremities (congenital dislocation of the hip, femoral neck deformation, and aplasia of the fibula) [16].

Some of the rarely seen symptoms of this disease are cleft palate, congenital heart defects, short stature, strabismus, hyperhidrosis, kidney malformations, agenesis of the uterus and ovaries, and pancreatic cysts [13, 15]. Sixty-two percent of children who suffer from this disease are allergic to dairy products. [17]

Holt–Oram syndrome. Holt and Oram first described this condition in 1960. It is an inherited disorder characterized by abnormalities of the upper limbs and heart. Holt–Oram syndrome is the most common form of heart–hand syndrome, with a prevalence estimated at 1 case per 100,000 total births. Genetic studies have shown mutations in the gene TBX5 which is important in the development of the heart and upper limbs [10]; inheritance patterns may be autosomal dominant (60%) or sporadic (40%) [12].

Clinical manifestations of the syndrome in the musculoskeletal system are variable. Abnormalities may be unilateral or bilateral and asymmetric and may involve the radial, carpal, and thenar bones. Aplasia, hypoplasia, fusion, or anomalous development of these bones produces a spectrum of phenotypes, including triphalangeal or thumb absence [13]. The main clinical manifestation of this syndrome in the cardiovascular system is either an atrial septal defect or a ventricular septal defect. Cardiac anomalies may include cardiac conduction defects, such as progressive atrioventricular block and atrial fibrillation. In the absence of septal defects, anomalies, such as patent ductus arteriosus and peripheral vascular hypoplasia, are frequently present [19].

VACTERL syndrome (VATER association). This syndrome derives its name from the first letters of the non-random co-occurrence of birth defects. It was first described in 1974 based on the study of 34 patients by S. Temtami and D. Miller. Frequency of mutations is 1 in 30,000 live births. The disease occurs sporadically, and its etiology is unknown. Disease incidence is high among children whose mothers are diabetic. Studies have revealed the effect of antibiotics (anthracyclines) in rats that cause defects similar to that of the VACTERL syndrome [11].

Frequent symptoms are a) malformation of the spine (60%–80%) that typically includes segmentation defects, such as hemivertebrae, kyphoscoliosis, and meningocele; b) congenital heart disease (40%–80%), particularly ventricular septal defect; c) imperforate anus/anal atresia (55%–90%); d) esophageal atresia and tracheo-esophageal fistula (50%–80%); e) malformation of the forearm and hand on the side of the radius (40%–50%) that include hypoplasia or aplasia manifested in the first ray, preaxial polydactyly, and syndactyly; and f) renal abnormalities (53%), such as renal agenesis, dysplasia, and hydronephrosis [13, 20, 21].

Nager Syndrome (Acrofacial dysostosis). In 1948, F. Nager and J. deReynier described the characteristics of this syndrome as maxillofacial

dysostosis with hypoplasia of the thumb and radius. Nager syndrome is a rare condition, and although its prevalence is unknown, more than 200 cases have been reported in the medical literature. Inheritance is known to be autosomal dominant with diverse expressivity. The gene maps to chromosome 9q32. The anomalies of the maxillofacial region include palpebral fissures, reduced or absent eyelashes, eyelid coloboma, sharp mandibular hypoplasia, shortening of the hard palate, and hypoplasia of the rudimentary molar. Permanent signs include hearing losses caused by stenosis or atresia of the auditory canal, occasional preauricular growths, and deformation and low positioning of the ear [1,5]. When examining children with multiple malformations, it must be noted that such malformations are not specific to any particular disease and even rare anomalies may have different disease etiologies. Diagnosis should not be based on only one symptom, and it must be noted that minor malformations are not less important than major anomalies.

Objective

The objective of this study was to analyze congenital radial club hand and genetic syndromes associated with it as well as the optimization of methods for conservative and surgical treatments of patients.

Materials and Methods

From 2007 to 2014, a comprehensive study of 170 children with congenital radial club hand was conducted at GI Turner Orthopedic Institute for children. Among these, 32 patients (18.8%) were identified with different syndromes. To assess the stages of underdevelopment of the forearm and hand, we used the classification proposed by VS Prokopovich in 1980. We modified this to include four stages. First stage—underdevelopment of the radius. This was not determined radiographically but clinically, we observed the deviation of the radial side of the wrist. Second stage— hypoplasia of the radius up to 50% of the length of the forearm. Third stage— more than 50% underdevelopment of the radius. The fourth stage— aplasia of the radius. In addition, diagnostic tests were performed on concomitant diseases. All data were retrospectively collected and analyzed, including the different methods used in treating the patients.

Results

Patient distribution was made according to organs that were affected. The most frequent syndrome was Holt–Oram identified in 17 children (9 boys and 8 girls; 53.1%). Nine children were presented with TAR syndrome (4 boys and 5 girls; 28.1%). VACTERL syndrome was observed in 4 male patients (12.5%), whereas Nager syndrome was observed in 2 children (1 boy and 1 girl, 6.25%).

The mean age of outpatients who came for consultation was of 4.2 ± 2.5 (range, 1–12) months. There were more boys (18 patients, 56.2%) than girls (14 patients, 43.8%). Early consultation by patients helped to reveal lesions associated with organs and systems as well as to conservatively treat, reposition, and stretch soft tissues from the radial edge of the forearm and hand.

All 32 patients were admitted for surgical treatment (mean age, 2.9 ± 2.8 years; range, 7 months to 12 years). An assessment of patient history revealed that surgical interventions aimed to address other co-existing pathologies were performed in 12 children (37.5%). Surgeries were performed to correct problems in the cardiovascular (7 children, 58%), digestive (3 children, 9.4%), and urinary (2 children, 6.3%) systems.

The aim of the primary surgery was to eliminate the radial deviation of the hand and to increase the possibly to restore bilateral finger-gripping ability. Therefore, we used one-stage or two-stage methods of correction. Simultaneous middle repositioning of the wrist and its fixation onto the ulna was performed in 28.1% of the patients. This was done to passively reposition the wrist to the middle. This corresponds to the second stage of the two-stage method of correction in the case of a forearm deformity. In situations where it was not possible to reposition the wrist to the middle, another two-stage method was used.

The first stage of surgery was the removal of tensioned soft tissues from the radial edge of the forearm, the excision of the connective cord, which is located along the projection of the radius if necessary, and the mounting of a distraction device on the forearm and the wrist.

In the postoperative period, the single most important procedure was soft tissue distraction,

which began on the third day at a rate of 0.25 mm, three times daily. In addition, conservative treatment was performed to improve blood circulation to the upper extremities (Vitaphone, laser therapy, and treatment with drugs that improve blood circulation in the cervical portion of the spine). Period of distraction of soft tissues ranged from 20 to 30 days with an average of 26.7 ± 4.4 days. We noticed a decrease in the timing of this step and reduction in pains during medical treatment before surgery by an average of 7 ± 2.3 days. The gap between the first stage and the second stage ranged from 0 to 60 days (mean time, 11.4 ± 14.4 days). The second stage of surgery was to fix the wrist onto the head of the ulna. Therefore, an incision was made on the lateral surface of the forearm in transverse direction at the projection of the head of the ulna with an extension to the edge of the elbow.

After gaining access to the extensor tendon of the fifth finger and the hand, it was shifted aside. This allowed for safe incision of the interposed soft-tissue between the epiphysis and the proximal portion of the ulna bone near the wrist, fixing it with Kirschner wires. Using two Kirschner wires passed in cross direction through the second and fifth metacarpal bones, the hand was fixed to the ulna for 6 weeks. After removing the spokes, prostheses of the forearm and hand were applied to save the achieved correction.

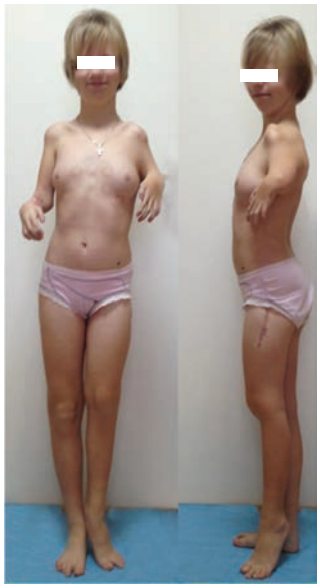
Nine patients with TAR syndrome (Fig. 1) were examined and treated surgically. All cases were patients at stage four (underdeveloped radius).

During examination and hospitalization for surgery blood counts revealed varying platelet levels ranging from 52×10^9/L to 496×10^9/L (mean value, 212.7×10^9/L). X-rays showed a fourth degree underdevelopment of the radius in all cases. The main goal for surgical treatment in this group of patients was removing and fixing the hand to the ulna.

All cases were performed using a two-stage procedure (Fig. 2). Grip function of the hand was accessed bilaterally (between the thumb and the first finger). In 5 children (55.5%), abduction and repositioning of the thumb was done using a combination of skin grafts.

Postoperative use of NSAIDs led to a sharp decrease in platelet count (10% and 32% in 2 patients, respectively) compared with the preoperative platelet count, and this was followed by a prolonged severe edema of the forearm and hand and subcutaneous hematoma.

Holt–Oram Syndrome was seen in 17 patients (53.1%) with congenital club hand (Fig. 3). Significant gender differences in terms of treatment were not observed among children (9 boys, 8 girls). Assessing the cardiovascular system, we found ventricular septal defects in 7 children, patent ductus arteriosus in 6, false chordae tendineae in 2, and atrial septal defect and dextrocardia in 1 patient. Some patients had a combination of ventricular septal defect with other heart defects. Two patients were diagnosed with congenital hydronephrosis, whereas bilateral lesions in the upper extremities were observed in 3 children. Among these, 2 cases were reported with Type IV bilateral radial bone hypoplasia, whereas in the third case, Types II and IV were observed in different limbs. Isolated lesions were



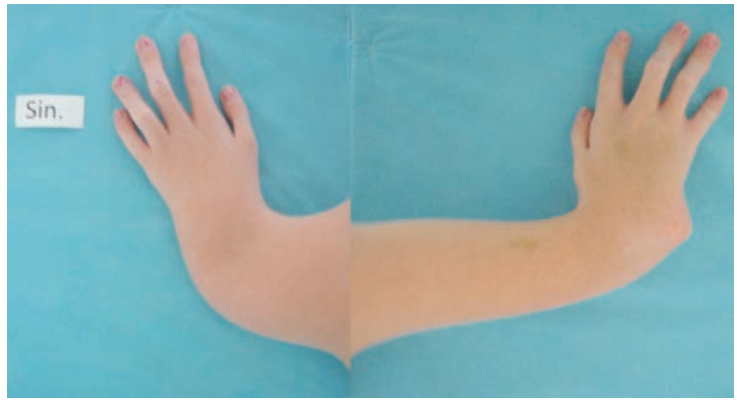


Fig. 1. A 12-year-old Patient D; the appearance of the forearms

observed on the right forearm and hand in 8 patients, whereas on the left in 5 patients. X-ray examination revealed varying degrees of hypoplasia of the radius: Type II was identified in three forearms, Type III in two forearms, and Type IV in 13 forearms. This is an important diagnostic criterion, indicating that there is the possibility of connective tissue deformation located at the projection of the radius in Type III and IV.

Figure 3 shows the correction of the radial deviation of the hand by a two-stage procedure. In patients with third and fourth types of underdevelopment, there was further assessment and dissecting of the spiked connective tissue located at the projection of the radius during the first stage of operation. The first ray of the hand (in all cases of aplasia) was restored by pollicization of the second finger of the hand (Fig. 4).

VACTERL syndrome was diagnosed in 4 patients. Bilateral lesion of the upper limbs was observed in 2 patients. There were unilateral lesions in 2 patients with deformation of the forearm and hand at the right side. The degree of underdevelopment of the radius in all cases was Type IV. In addition, the first ray was absent on the affected side (Fig. 5). Surgical correction of the radial deviation of the wrist was performed in a two-stage procedure. The first ray of the hand was reduced by pollicization of the second finger.

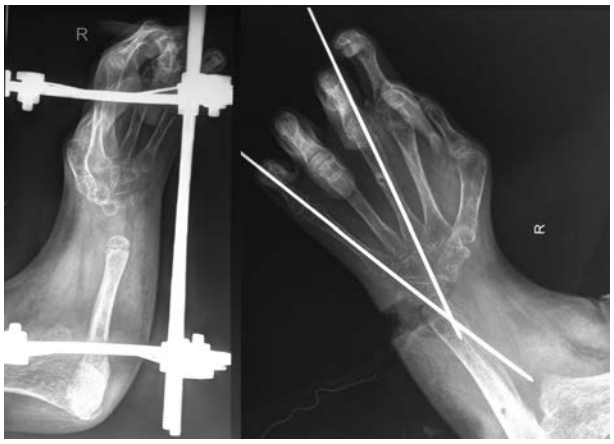


Fig. 2. A 12-year-old patient D; X-ray of the forearm and hand at distraction stage and after centralization of the hand onto the ulna

With respect to existing comorbidities in the patients, esophageal atresia was operated on during the first days of life. Kidney damage was also observed in all patients. The most common cardiovascular lesion, the tetralogy of Fallot, was identified in 2 cases. Anomalies of the spine were found in 3 patients. All symptoms were detected in only 2 children.

A study was conducted and surgical treatments were performed on 2 patients with Nager syndrome (acrofacial dysostosis). Type IV deficiency of the radius, combined with aplasia of the first finger of the hand (Fig. 6), was observed in all patients.

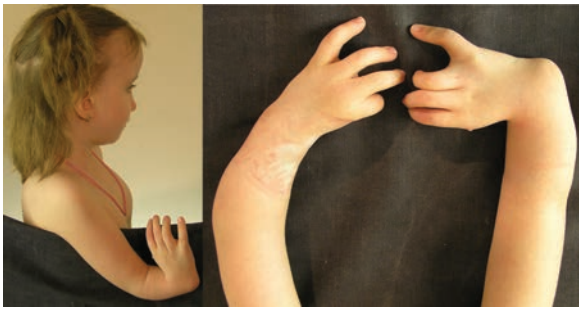


Fig. 3. A 4-year-old Patient M with Holt–Oram syndrome. The appearance of the forearms and hands

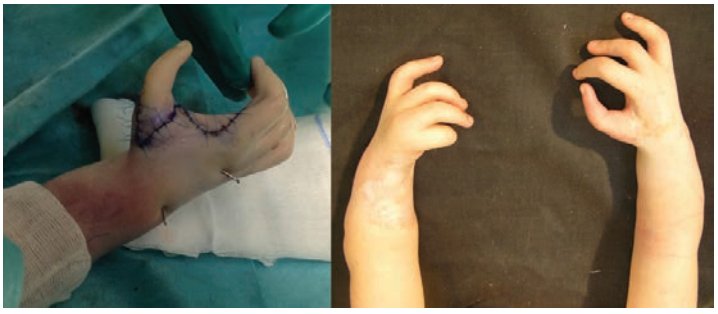


Fig. 4. A 5-year-old Patient M with Holt–Oram syndrome. The appearance of the right hand intraoperatively and at one year after operation

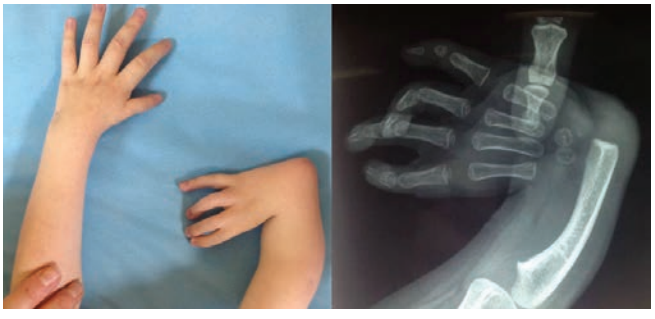


Fig. 5. A 4-year-old Patient B. with VACTERL syndrome. The image and radiograph of both forearms and hands

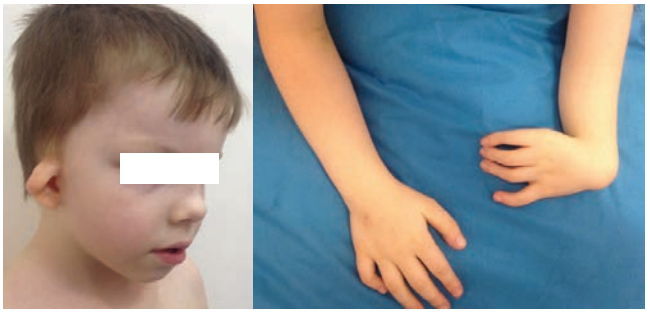


Fig. 6. A 5-year-old Patient A. with Nager syndrome. The appearance of the face and the forearm

We also detected palpebral fissures and various malformations of the facial region of the skull (Fig. 6). In all cases, there was atresia of the ear canal.

Surgical treatment consisted of a two-stage procedure involving the correction of radial deviation, followed by pollicization of the second finger.

Discussion

Congenital radial club hand can manifest as an independent disease and can also be part of a collection of syndromes. Differentiating the pathological causes is important because choosing the best treatment strategy depends on existing comorbidities [4, 6, 7, 16].

The correction of comorbidities requires preoperative preparation and examination as well as the postoperative management of the patient. Thus, the study of blood parameters in patients with TAR syndrome showed no anemia and eosinophilia, despite the description of these features by many authors [14–15]. We observed an increase in the number of platelets towards normal values during the growth of the children that is consistent with other studies [16, 17–18]. Because of the reduction of platelets in patients receiving NSAIDs, we reconsidered the tactics of postoperative pain management to reduce the risk of bleeding. Heart diseases, such as ventricular septal defect and patent ductus arteriosus, described as characteristic of the Holt–Oram syndrome in the literature were also present in our clinical observations [19, 22–23]. The most severe comorbidity was the VACTERL syndrome [21].

Despite the description in the medical literature regarding difficulties in correcting deformities of the upper extremity in this group of patients [4, 6, 16], we found no significant difference in surgical tactics. In some cases, an early conservative treatment allowed us to simultaneously operate on the hand (centralization), achieving positive functional and cosmetic results.

Conclusions

Congenital club hand requires a comprehensive study designed to identify comorbidities that require additional counseling, specialists, and determination of the age for surgical correction of deformities of the upper extremity. Only physical state compensation allows for the correction of existing deformities of the upper extremity. Early conservative treatment to stretch the soft tissues from the radial edge of the forearm allows for reduced surgical treatment time, reduced surgical trauma, and improvement of the cosmetic and functional condition of the entire affected upper limb.

Application of the two-stage method of correcting radial deviation of the wrist with the distraction of soft tissues in combination with physiotherapy is proposed to reduce or eliminate pain, prevent flexion contractures of the fingers, and reduce the time between stages of surgical treatment. TAR syndrome requires monitoring the changes in blood parameters at all stages of treatment. Selective drug therapy in the postoperative period, with the exclusion of NSAIDS, is needful to avoid complications caused by a sharp decline in platelet count. Various syndromes with congenital radial club hand often make the orthopedic traumatologist engage in a sequence of surgical corrections of accompanying deformities. Often, surgical intervention is not the first-line choice for treatment but allows you to restore the function of the upper extremity and improve the social lives of children.

References

1. Flatt AE. The Care of Congenital Hand Anomalies, 2d ed. St Louis: Quality Medical Publishing. 1994:366-410. doi: 10.1016/0266-7681(94)90269-0.
2. Lamb DW. The treatment of radial club hand, absent radius, aplasia of the radius, hypoplasia of the radius, and radial paraxial hemimelia. *Hand*. 1972;4:22-30. doi: 10.1016/0072-968X(72)90004-6
3. Urban MA, Osterman AL. Management of radial dysplasia. *Hand Clin.* 1990;6:589-605.
4. Стариков О.В. Реконструктивно-восстановительное лечение врожденной косорукости по методу Г.А. Илизарова: дис. ... канд. мед. наук. – Курган, 2002. [Starikov OV. Rekonstruktivno-vosstanovitel'noe lechenie vrozhdennoj kosorukosti po metodu G.A.Ilizarova [dissertation]. Kurgan; 2002. (In Russ).]
5. Бочков Н.П. Клиническая генетика. – М.: ГЭОТАРТ-МЕД, 2002. [Bochkov NP. *Klinicheskaya genetika*. Moscow: GEHOTART-MED; 2002. (In Russ).]
6. Барашнев Ю.И., Бахарев В.А., Новиков П.В. Диагностика и лечение врожденных и наследственных заболеваний у детей. – М.: Триада-Х, 2004. [Barashnev YI, Baharev VA, Novikov PV. *Diagnostika i lechenie vrozhdennyh i nasledstvennyh zabolevanij u detej*. Moscow: Triada-X, 2004. (In Russ).]

7. Прокопович В.С., Прокопович Е.С. К вопросу об этиопатогенезе врожденной косорукости у детей / X юбилейный Российский национальный конгресс «Человек и его здоровье» (ортопедия-травматология-протезирование-реабилитация). Ноябрь 21-25, 2005; Санкт-Петербург. [Prokopovich VS, Prokopovich ES. K voprosu ob ehtiopatogeneze vrozhdennoj kosorukosti u detej. (Conference proceedigs) X yubilejnyj Rossijskij Nacional'nyj kongress «Chelovek i ego zdorov'e» (ortopediya-travmatologiya-protezirovanie-reabilitaciya); nov 21-25; Saint-Petersburg. (In Russ).]

8. Фищенко В.А. Врожденная и приобретенная косорукость у детей и подростков: дис. ... канд. мед. наук. – Киев, 1985. [Fishchenko VA. Vrozhdennaya i priobretennaya kosorukost' u detej i podrostkov. [dissertation] Kiev; 1985. (In Russ).]

9. Greenhalg KL, Howell RT, Bottani A, et al. Thrombocytopenia-absent radius syndrome: a clinical genetic study. *J Med Genet.* 2002;39:876-881. doi: 10.1136/jmg.39.12.876

10. Patel C, Silcock L, McMullan D, et al. TBX5 intragenec duplication: a famyly with an atypical Holt-Orama syndrome phenotype. *Eur J Hum Genet.* 2012;20(8):863-9. doi: 10.1038/ejhg.2012.16. Epub 2012 Feb 15.

11. Mc Lauqhlin D, Hajduk P, Murphy P, et al. Adriamycin - Induced models of VACTERL association. *Mol Syndromol.* 2013;4(1-2):46-62. doi: 10.1159/000345579.

12. Holt M., Oram S. Familial heart diasese with skeletal malformations. *Br Heart J.* 1960;22:236-42. doi: 10.1136/hrt.22.2.236.

13. Джонс К. Наследственные синдромы по Дэвиду Смиту. – М.: Практика, 2011. – С. 302-305, 352-355, 376-385, 784-786. [Dzhons K. *Nasledstvennye sindromy po Dehvidu Smitu*. Moscow: Praktika, 2011. (In Russ).]

14. Greenhalgh KL, Howell RT, Bottani A, Ancliff PJ, et al. Thrombocytopenia-absent radius syndrome: a clinical genetic study. *J Med Genet*. 2002;39:876-881. doi: 10.1136/jmg.39.12.876.

15. Hall JG, Levin J, Kuhn JP, Ottenheimer EJ, et al. Thrombocytopenia with absent radius (TAR). *Medicine.* 1969;48:411-39.

16. Прокопович В.С. Лечение врожденной косорукости при TAR-синдроме. Заболевания и повреждения верхних конечностей у детей. – Л., 1988. – С. 97-101. [Prokopovich VS. *Lechenie vrozhdennoj kosorukosti pri TAR-sindrome. Zabolevaniya i povrezhdeniya verhnih konechnostej u detej*. Leningrad; 1988. (In Russ).]

17. Hedberg VA, Lipton JM. Thrombocytopenia absent radii. A review of 100 cases. *Am J Pediatr Hematol/Oncol*. 1988;10:51-64. doi: 10.1097/00043426-198821000-00010.

18. Scott N. Oishi, Peter Carter, et al. Thrombocytopenia-absent radius syndrome: presence of brachiocarpalis muscle and it importance. *J Hand Surg.* 2009;34:1696-1699. doi: http://dx.doi.org/10.1016/j.jhsa.2009.06.025

19. Newbury-Ecob RA, Leanage R, Raeburn JA, Young ID. Holt-Oram syndrome: a clinical genettic study. *J Med Genet.* 1996;33(4):300-307. doi: 10.1136/jmg.33.4.300.

20. Wattanasirichaigoon D, Prasad C, Schneider G, et al. Rib defects in patterns of multiple malformations: A retrospective review and phenotypic analysis of 47 cases. *Am J Medical Genetics.* 2003;122(1):63-9. doi: 10.1002/ajmg.a.20241.

21. Solomon BD, Pineda-Alvarez DE, et al. Analysis of component findings in 79 patients diagnosed with VACTERL association. *Am J Med Genet*. 2010;152:2236-2244. doi: 10.1002/ajmg.a.33572.

22. Sunagawa S, Kikuchi A, et al. Prenatal diagnosis of Holt-Oram syndrome: Role of 3-D ultrasonography *J Congenital Anomalies.* 2009;49:38–41. doi: 10.1111/j.1741-4520.2008.00211.x.

23. Muller LM, De Jong G, Van Heerden KM. The antenatal ultrasonographic detection of the Holt-Oram syndrome. *S Afr Med J.* 1985;68:313.

ВРОЖДЕННАЯ ЛУЧЕВАЯ КОСОРУКОСТЬ У ДЕТЕЙ В СТРУКТУРЕ ГЕНЕТИЧЕСКИХ СИНДРОМОВ

© Авдейчик Н.В., Говоров А.В., Голяна С.И., Сафонов А.В.

ФГБУ «НИДОИ им. Г.И. Турнера» Минздрава России, Санкт-Петербург

Цель исследования. Клинический анализ врожденной лучевой косорукости, входящей в состав генетических синдромов, а также оптимизация методов консервативного и хирургического лечения пациентов с этим видом патологии.

Материалы и методы. С 2007 по 2014 год проведено комплексное обследование 170 детей с врожденной лучевой косорукостью. Среди них было выявлено 32 пациента (18,8 %) с различными синдромами. Оценена степень недоразвития лучевой кости среди данной группы пациентов. Определены особенности ведения пациентов в зависимости от сопутствующих патологий.

Результаты. Проведенная оценка выявила синдром Холта — Орама (Holt-Oram Syndrom) у 17 детей (9 мальчиков и 8 девочек) — 53,1 %. Также выявлен TAR-синдром у 9 детей (4 мальчика и 5 девочек) — 28,1 %. Синдром VACTERL обнаружен у 4 пациентов мужского пола — 12,5 %. Синдром Нагера наблюдался у 2 детей (1 мальчик и 1 девочка) — 6,25 %. Тактика хирургического лечения лучевой косорукости в структуре

генетических синдромов применялась та же, что и у пациентов с изолированной врожденной косорукостью: выполнялась центрация кисти на локтевую кость предплечья одномоментным или двухэтапным способом с последующей реконструкцией лучей. Сроки лечения пациентов с этими патологиями не отличались от сроков лечения пациентов с изолированной врожденной косорукостью.

Заключение. Врожденная лучевая косорукость, выявленная в составе генетических синдромов, требует комплексного обследования для диагностики сопутствующих патологий, наблюдения и лечения у специалистов, определения возрастных показаний для хирургической коррекции имеющихся деформаций верхней конечности.

Ключевые слова: врожденная лучевая косорукость, Holt-Oram-синдром, VACTERL-синдром, TAR-синдром, Нагера синдром.

Information about the authors

Avdeychik Natalia Valerievna — MD, PhD student of the department of reconstructive microsurgery and hand surgery. The Turner Scientific and Research Institute for Children's Orthopedics. E-mail: natali_avdeichik@mail.ru.

Govorov Anton Vladimirovich — MD, PhD, research associate of the department of reconstructive microsurgery and hand surgery. The Turner Scientific and Research Institute for Children's Orthopedics. E-mail: agovorov@yandex.ru.

Golyana Sergei Ivanovich — MD, PhD, head of the department of reconstructive microsurgery and hand surgery. The Turner Scientific and Research Institute for Children's Orthopedics. E-mail: ser.golyana@yandex.ru.

Safonov Andrey Valerievich — MD, PhD, chief of the department of reconstructive microsurgery and hand surgery. The Turner Scientific and Research Institute for Children's Orthopedics. E-mail: safo125@ gmail.com.

Авдейчик Наталья Валерьевна — аспирант отделения реконструктивной микрохирургии и хирургии кисти ФГБУ «НИДОИ им. Г.И. Турнера» Минздрава России. E-mail: natali_avdeichik@mail.ru.

Говоров Антон Владимирович — к. м. н., научный сотрудник отделения реконструктивной микрохирургии и хирургии кисти ФГБУ «НИДОИ им. Г.И. Турнера» Минздрава России. E-mail: agovorov@yandex.ru.

Голяна Сергей Иванович — к. м. н., руководитель отделения реконструктивной микрохирургии и хирургии кисти ФГБУ «НИДОИ им. Г.И. Турнера» Минздрава России. E-mail: ser.golyana@yandex.ru.

Сафонов Андрей Валерьевич — к. м. н., заведующий отделением реконструктивной микрохирургии и хирургии кисти ФГБУ «НИДОИ им. Г.И. Турнера» Минздрава России. E-mail: safo125@gmail.com.