DOI: https://doi.org/10.17816/PTORS492255

Case Report

Polymelia of the upper limb

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ABSTRACT

BACKGROUND: The most rare and extremely rare malformation of the upper limb is polymelia, i.e., doubling (triple) limb. In the literature, no more than 20 cases of an increase in the number of lower limbs and only three cases of doubling (triple) of the upper limb (in humans) have been described.

CLINICAL CASE: A 2-year-old child diagnosed with polymelia of the right upper limb was examined and treated at our clinic. Clinical examination revealed two shoulder blades and one clavicle, with a normal anatomical location and development. Behind, an upper limb ended with the stump of the forearm, represented by an underdeveloped ulna. Moreover, the elbow joint was in a state of flexion contracture at an angle of 170°, and the ulna was turned posteriorly and 15°–20° medially. The "anterior" humerus in the proximal section was underdeveloped, and in the distal section, it articulated in a normally formed elbow joint with the radius and ulna. The hand had five fingers with moderate underdevelopment and deformity of the 5th finger. After examining the bone, muscle, and vascular anatomy of the doubled limb, surgery was performed; as a result, a nearly complete anatomical and functional limb was reconstructed from the two segments of one upper limb.

DISCUSSION: Polymelia is an extremely rare variant of a congenital anomaly of the limbs; thus, only a few cases are reported in the literature, which differs significantly from each other in clinical manifestations. Many questions related to the etiology and pathogenesis of these malformations are unexplored. The doubling of the limbs in animals is caused by both teratogenic environmental factors and genetic mutations. In any case, reconstructive surgery makes it possible to effectively treat children with such anomalies in the development of the limbs and obtain good results.

CONCLUSIONS: The restoration of the upper limb of children with polymelia is quite difficult, but doable. Adequate analysis of all examination data and rational planning of surgical intervention in such cases creates conditions that enable achieving the maximum anatomical and functional results of upper limb reconstruction in children with this developmental anomaly.

Keywords: upper limb; congenital malformation; anomaly; surgical treatment; case report; polymelia; reconstruction.

To cite this article
Golyana SI. Polymelia of the upper limb. *Pediatric Traumatology, Orthopaedics and Reconstructive Surgery.* 2023;11(3):371–380.
DOI: https://doi.org/10.17816/PTORS492255

Received: 10.06.2023 **Accepted:** 28.08.2023 **Published:** 29.09.2023

УДК 617.576-007-053.1
DOI: https://doi.org/10.17816/PTORS492255

Клинический случай

Полимелия верхней конечности

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АННОТАЦИЯ

Обоснование. Крайне редко встречающийся порок развития верхней конечности — полимелия, то есть удвоение (утроение) конечности. На сегодняшний день в литературе описаны не более 20 случаев увеличения количества нижних конечностей и только три случая удвоения (утроения) верхней конечности (у людей).

Клиническое наблюдение. В клинике нашего центра находился на обследовании и лечении ребенок 2 лет с диагнозом «полимелия правой верхней конечности». При клиническом обследовании левая верхняя конечность анатомически сформирована нормально. Справа верхняя конечность удвоена, включая лопатку, плечевую кость и кости предплечья. Конечности на всем протяжении были сращены мягкими тканями и находились в сагиттальной плоскости по отношению друг к другу, поэтому сзади расположенную конечность мы назвали задней (основной), а спереди расположенную — передней (дополнительной). «Задняя» верхняя конечность была нормально развита в проксимальном отделе (лопатка, плечевая кость), но заканчивалась культей недоразвитого предплечья, тогда как «передняя» верхняя конечность была умеренно недоразвита в проксимальном отделе и гораздо более развита в дистальном. В связи с этим проведено хирургическое вмешательство, в ходе которого выполнена реконструкция одной, практически полноценной анатомически и функционально, конечности из проксимального отдела «задней» и дистального отдела «передней» правых верхних конечностей.

Обсуждение. Полимелия — чрезвычайно редкий вариант врожденной аномалии конечностей, поэтому в литературе описаны только единичные случаи, значительно отличающиеся друг от друга клиническими проявлениями. Остается много неизученных вопросов, связанных с этиологией и патогенезом данных пороков развития. Причинами удвоения конечностей у животных называют как тератогенные факторы окружающей среды, так и генетические мутации. В любом случае возможности реконструктивной хирургии позволяют эффективно лечить детей с подобными аномалиями развития и получать хорошие результаты.

Заключение. Задача восстановления верхней конечности у детей с полимелией довольно сложна, но выполнима. Адекватный анализ всех данных обследования и рациональное планирование оперативного вмешательства в подобных случаях создают условия для достижения максимального анатомического и функционального результата реконструкции верхней конечности у детей с данной аномалией развития.

Ключевые слова: верхняя конечность; врожденный порок развития; аномалия; хирургическое лечение; клинический случай; полимелия; реконструкция.

Как цитировать

Голяна С.И. Полимелия верхней конечности // Ортопедия, травматология и восстановительная хирургия детского возраста. 2023. Т. 11. № 3. С. 371–380. DOI: https://doi.org/10.17816/PTORS492255

Рукопись получена: 10.06.2023 **Рукопись одобрена:** 28.08.2023 **Опубликована:** 29.09.2023

BACKGROUND

Among upper limb malformations, which are characterized by increased number of segments, polydactyly (or polyphalangism) of finger I is the most common, occurring in 0.8–1.4 cases per 1000 newborns [1–3].

Ulnar dimelia (duplication of the hand and forearm), known in the literature as "mirror hand" is much less common in this group of congenital upper limb anomalies [4]. In 2007, Irani provided statistics on this developmental anomaly based on an analysis of all literary sources reporting ulnar dimelia [5]. Accordingly, approximately 60 cases of this pathology have been described since 1587. Over 16 years since the publication of this article, the data, certainly, have changed; however, corresponding updated indicators could not be found in the available literature. Some authors presented their experience in the surgical correction of this congenital deformity of the hand and forearm [6–10].

Limb polymelia is an extremely rare variant of congenital pathology. To date, no more than 20 cases of an increase in the number of lower limbs [11–13] and only three cases of doubling (triplication) of the upper limb are known in humans. However, Professor R. O'Rahilly of the Department of Anatomy found a very voluminous article (43 pages of text with tables and diagrams + 189 references) [14], which collected information about all known cases of congenital limb anomalies, associated with "longitudinal insufficiency" (which is more commonly called "radial and ulnar clubhand") and "longitudinal redundancy" of the tissues of the segments [meaning polydactyly, "mirror hand," ulnar or radial dimelia and polymelia, namely, doubling (triplication) of the limb]. In addition, the article highlighted the difficulty in unambiguously determining the degree of doubling of the upper limb in the described clinical cases, as it is mainly referred to an increase in the number of fingers, metacarpals and carpals, and ulnar and radial bones, which indicates the presence of various forms of polydactyly and the "mirror hand" (ulnar or radial dimelia). Recent studies have presented only three cases of complete duplication (polymelia) of the upper limb, which are presented in photographs and radiographs [15–17]. All three pediatric patients underwent surgical treatment using various surgical methods because the manifestations of this pathology differed significantly from each other.

Thus, this study presents the treatment course of a child with the described rare congenital pathology of the upper limb.

CLINICAL CASE

The patient was a 26-month-old boy (Kazakhstan) who presented with a clinical case of complete right upper limb duplication (polymelia).

Clinical examination revealed no pathological changes in the internal organs, lower limbs, chest, spine, and head. The left upper limb was also normally developed, and no pathological changes were detected. On the right limb, a significant increase in the volume of the shoulder girdle and upper limb was noted. Upon palpation, two shoulder blades, two humerus bones, and forearm bones were identified. The limbs along their entire length were fused with soft tissues and were in the sagittal plane in relation to each other; therefore, the posteriorly located limb was called posterior (main) and the anteriorly located one anterior (additional). The "posterior" upper limb was normally developed in the proximal part, and the scapula was of normal shape, with correct anatomical localization and articulated with a normally developed and correctly positioned clavicle. The humerus was articulating with the scapula at the shoulder joint, which was 2 cm shorter than the left humerus. At the elbow joint, the "posterior" upper limb articulated with the shortened ulna, which was the stump of the underdeveloped forearm. Here, the elbow joint was in a state of flexion contracture at an angle of 170°, and the ulna was located posteriorly and 15°–20° inwards. In this elbow joint, rocking passive movements were determined (within 10°–15°), and no hand was found on the "posterior" limb.

The "anterior" (accessory) upper limb was moderately underdeveloped in the proximal region and much more developed in the distal region. The "anterior" humerus in the distal section articulated in a normally formed elbow joint with the radius and ulna. The forearm was moderately shortened (approximately 1 cm compared with the left). The five-fingered hand included normally developed fingers I–IV, with finger I in the opposition. Finger V was moderately shortened, and a moderate flexion contracture in its interphalangeal joints (approximately 30°) and ulnar deviation were detected. Active and passive movements in the shoulder joints were synchronous and limited, with shoulder rise possible up to 70°. Movements in the elbow, wrist, and metacarpophalangeal and interphalangeal joints of fingers I–IV were not limited. In the joints of finger V, extension was limited, soft tissue tension was noted along the palmar surface of the finger, and the fourth interdigital space was deepened and widened (Fig. 1).

Radiography and multislice computed tomography (MSCT) of the upper limbs showed the normal location and correct anatomical shape of the "posterior" scapula, which articulated with the single clavicle at the acromioclavicular joint. The normally developed proximal epiphysis of the humerus of the "posterior" upper limb articulated with the articular surface of the scapula. Toward the distal part, the humerus became moderately thinner and ended with an underdeveloped, small epiphysis, which in turn articulated with the only underdeveloped and shortened ulna of the forearm, ending in a stump and located posteriorly at an angle of 170° to

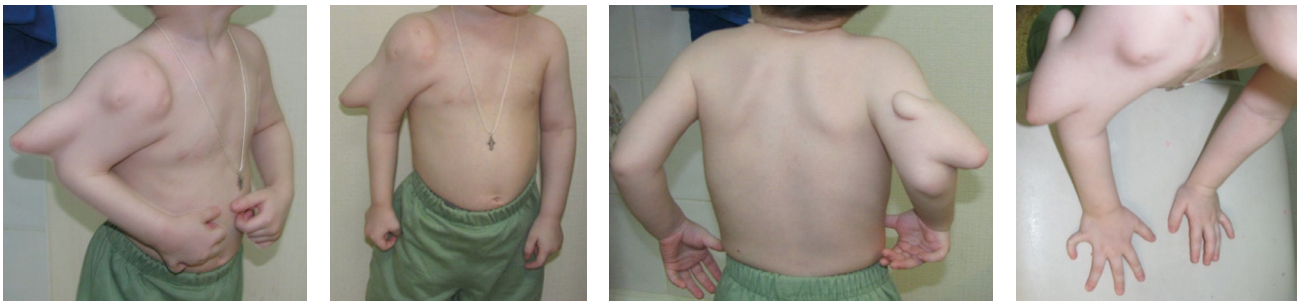

Fig. 1. Pediatric patient P., 2 years old, with doubling of the right upper limb

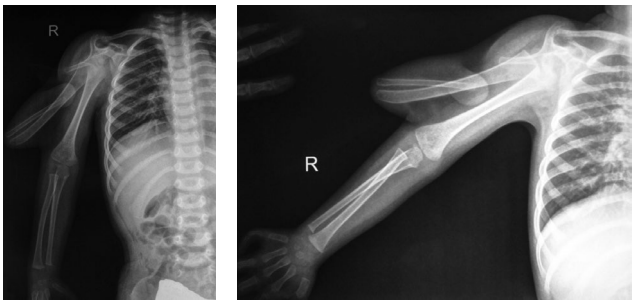

Fig. 2. Radiographs of the right upper limb before surgery

the humerus (Fig. 2). The hand of the "posterior" upper limb was missing.

The other shoulder blade of the "anterior" (accessory) limb was underdeveloped and small, located on the anterior surface of the chest under the right clavicle, and did not articulate with the clavicle based on MSCT data. The proximal part of the "anterior" humerus, which formed the shoulder joint with the described scapula, was small, and toward the distal part, the humerus had correct anatomical dimensions and shape, articulating at the elbow joint with the normally developed radius and ulna (Fig. 3). The hand was also normally formed, except for the fifth ray, as metacarpal bone V was shortened, deviated ulnarly at an angle of 15° from the metacarpal bone IV. The phalanges of the finger V were also moderately reduced in size (Fig. 1, 2).

To determine the anatomical aspects of the vessels of the doubled upper limb, contrast angiography was performed together with MSCT (Fig. 3). Analysis of CT data revealed the following situation. On the right, the subclavian artery was displaced medially, and its continuation, the axillary artery, emerged from under the clavicle in the projection of

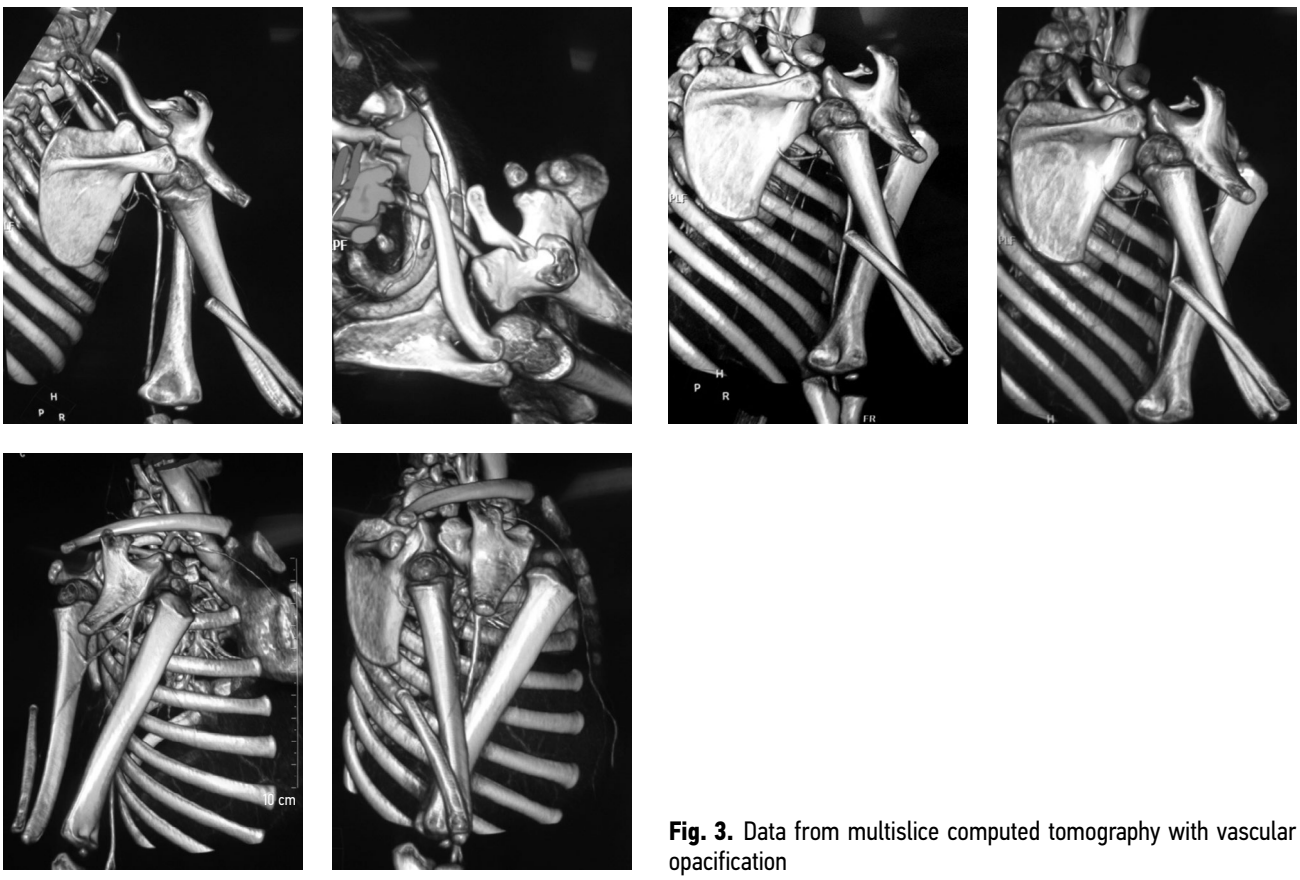

Fig. 3. Data from multislice computed tomography with vascular opacification

DOI: https://doi.org/10.17816/PTORS492255

the border of the medial and middle thirds of the clavicle (normally it is projected to the middle of the clavicle). Then, it passed under the additional "anterior" scapula and continued as a single brachial artery common to both shoulder segments, giving branches to both humerus bones. 3D muscular CT images clearly show that the musculus deltoideus is attached to the proximal part of the "posterior" humerus, and single muscle fibers could be traced over the head of the "anterior" humerus (Fig. 4). Further, the biceps and triceps of the shoulder "covered" the "anterior" humerus, and on the "posterior" one, these muscles were practically absent. On the rudimentary ulna of the "posterior" limb, no muscle formations were seen at all.

After a detailed examination of the anatomical aspects of the doubled right upper limb, the approach of the surgical intervention was decided to create one maximally complete arm from two segments, as shown in the surgery scheme (Fig. 5).

On the operating table, after marking, shaped skin incisions were made from the shoulder girdle to the elbow joint (Fig. 6). The rudimentary "anterior" scapula, which was attached to the clavicle and ribs by fibrous cords, was isolated, and all branches of the axillary artery supplying the scapula and proximal part of the "anterior" humerus were coagulated. The axillary and brachial arteries with accompanying veins were identified along their entire length through available imaging. Then, a transverse osteotomy of the "anterior" humerus was performed at the level of the border of the proximal and middle thirds. The proximal complex of tissues of the "anterior" upper limb, consisting of a rudimentary scapula and proximal humerus, was removed.

Then, the distal section of the "posterior" limb was isolated, the vascular branches supplying it were coagulated, the branches of the peripheral nerves were dissected away, and the distal 2/3 of the length of the "posterior" humerus along with the rudiment of the ulna was then removed. Thus, (a) the proximal part of the "posterior" upper limb, including a full scapula and proximal 1/3 of the length of the "posterior" humerus along with the muscle sheath and

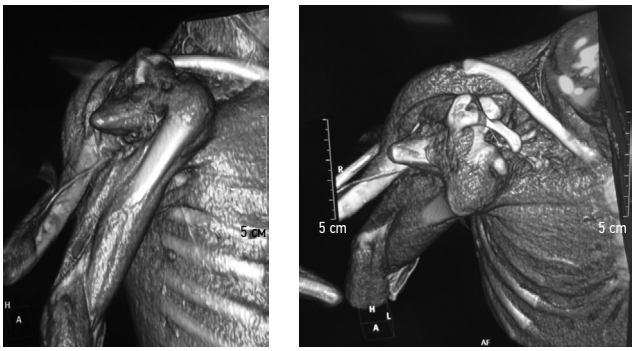

Fig. 4. Multislice computed tomography data (muscle mode). The predominant development of muscles is determined on the proximal part of the "posterior" humerus and the middle and distal thirds of the "anterior" humerus

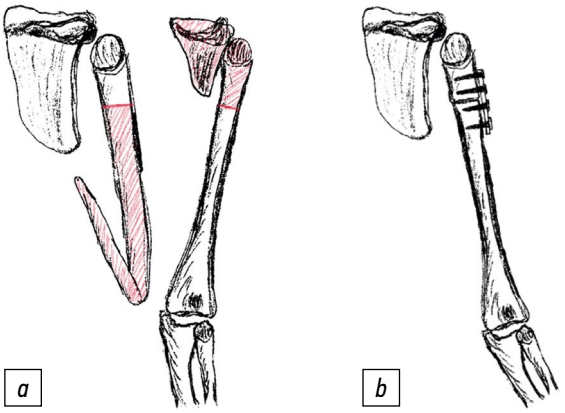

Fig. 5. Surgery scheme: *a,* limb before surgery (the shaded bone fragments are subject to resection); *b,* planned result of surgical intervention (osteosynthesis between the proximal part of the "posterior" humerus and the distal 2/3 of the "anterior" humerus)

(b) distal 2/3 of the length of the "anterior" humerus with a full muscle sheath and, accordingly, with a fully developed forearm and hand, were preserved. These bone fragments of both humeri were juxtaposed end-to-end, after which metal osteosynthesis was performed with a plate. Then, intermuscular sutures were applied, and after excision of the excess soft tissue, the surgical wounds were sutured in layers. The blood supply to the distal parts of the upper

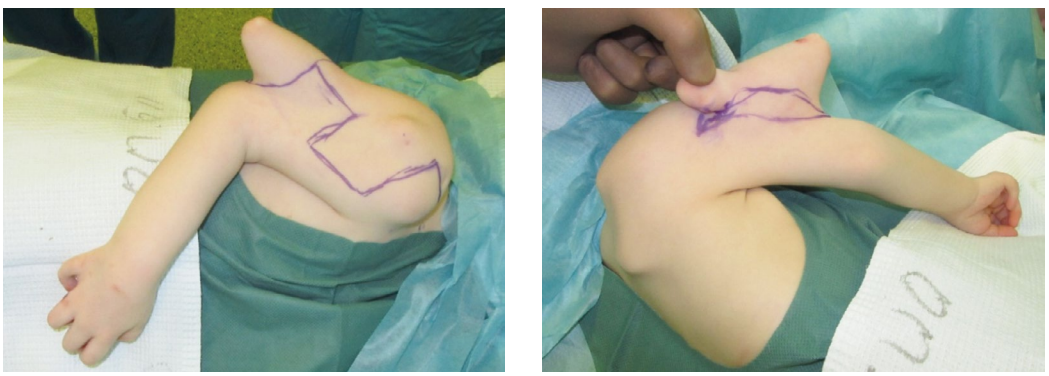

Fig. 6. Marking incisions before surgery

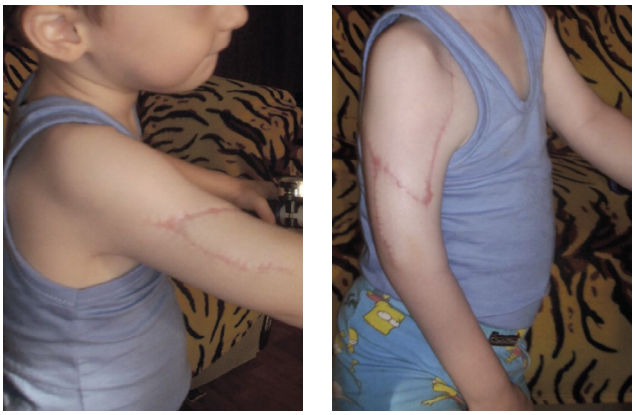

Fig. 7. Start of rehabilitation 3 weeks after surgery and appearance of the right upper limb

limb was monitored. An aseptic dressing was applied, and the limb was placed in a plaster splint. Twenty-one days after the surgery, rehabilitation was started with the recovery of movements in all limb joints (Fig. 7).

Six months after this surgery, the child underwent another hand surgery. When the intermetacarpal ligament between the heads of metacarpal bones IV and V was restored, the interdigital space IV was raised to a normal level by local skin grafting, and the flexion contracture of finger V was partially eliminated.

The child was under absentee follow-up for 10 years postoperatively. It has not yet been possible to conduct an in-person consultation; thus, it was not possible to measure accurately the shortening and strength of the muscles of the reconstructed upper limb. However, the length of the arms varies insignificantly on photographs. The esthetic and functional outcomes of surgical treatment completely satisfy the patient and his mother (Fig. 8). The child plays football, is socializing, and has no problems communicating with peers.

DISCUSSION

The literature describes polymelia as an extremely rare developmental anomaly, mainly in animals and birds [18–23]. This study provides brief information and photographs from these articles (for clarity). A group of authors from Brazil described a case of the birth of twin opossum cubs, one of which had a duplication of the right hind limb [18]. In this case, the femur was single but increased in volume and bifurcated in the area of the knee joint into two articular surfaces, and then complete doubling of the bones of the lower leg and paw (foot) was noted (Fig. 9).

In the case of Swainson's hawk, researchers from California described the duplication of both hind limbs of

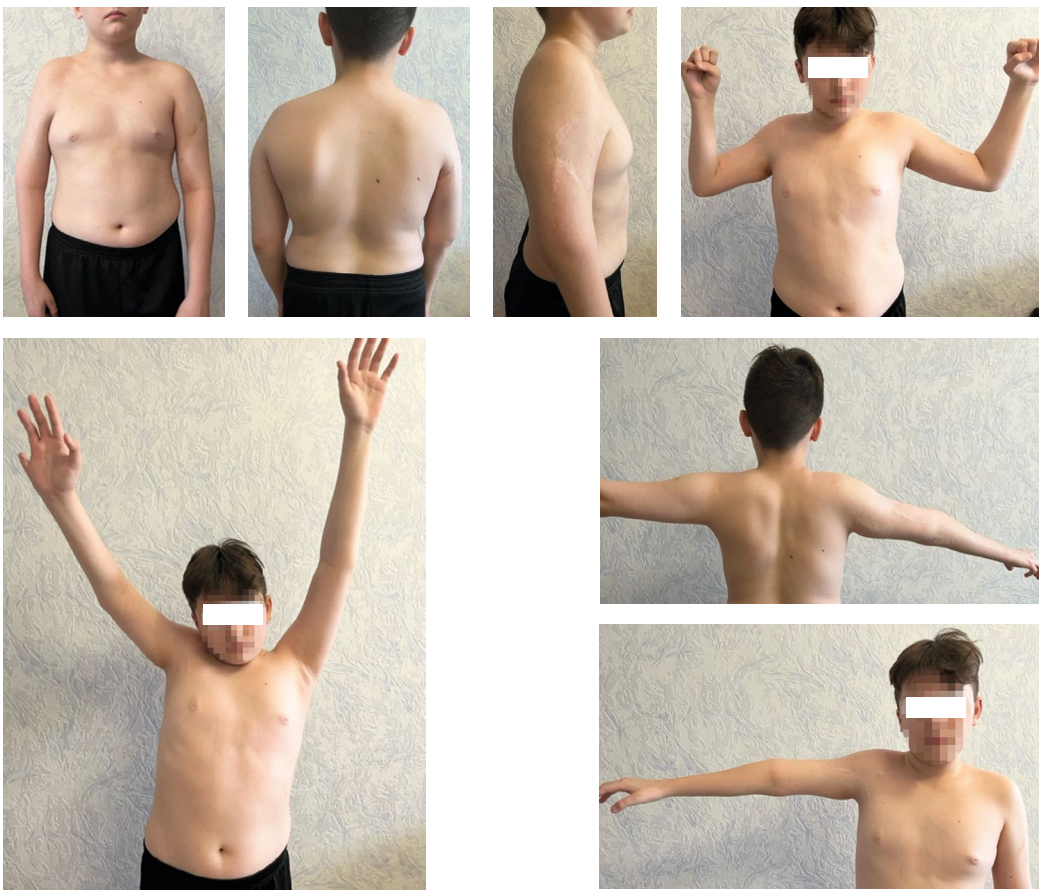

Fig. 8. Treatment results 10 years after surgery

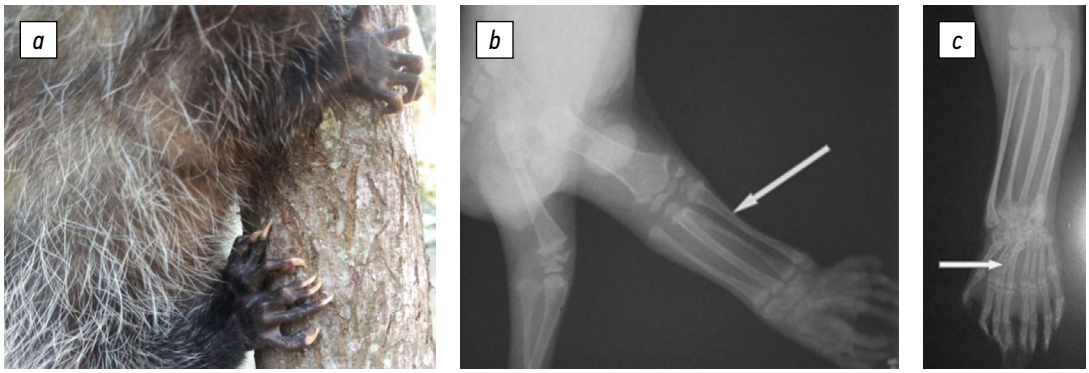

Fig. 9. Duplication of the hind limb in an opossum: *a,* appearance; *b* and *c,* radiographs

the bird [19]. However, this case, like the previous case with the opossum, does not concern complete limb duplication but only the distal sections. The causes of limb duplication in animals include both teratogenic environmental factors (organochlorine insecticides, polychlorinated biphenyls, organophosphates and carbamate insecticides, and heavy metals such as mercury or selenium) and genetic mutations. Authors from the University of Bern [20] presented complete forelimb duplication in a calf, and an article by Egyptian researchers [21] presented a case of complete duplication of the hind legs in a chicken. Swiss scientists call a calf with a double set of forelimbs a "chimera" because, after conducting a genetic analysis of the cells and the additional two limbs of the calf, they discovered two populations of cells with different compositions [22]. The researchers concluded that this is a case of an asymmetrically conjoined dizygotic twin, which distinguishes this from other previously described cases of polymelia cattle that have been identified as an asymmetrically conjoined monozygotic twin (Fig. 10).

One of the theories put forward by scientists is not the doubling of limb segments (or separation) but the "fusion" of twins formed in utero. Ronan O'Rahilly also reported this; in 1951, he carefully analyzed all cases of various doublings and triplication of limbs (or their segments) known at that time. He emphasized that extreme forms of polydactyly are very rare. For example, Voigt reported a patient with 13 fingers on each hand and 12 toes on both feet, and Saviard saw an infant with 40 fingers and toes, 10 on each limb.

Gould and Pyle (1900) and Bradford and Lovett observed a patient with 15 fingers and 13 toes (Steinand Eettmanii, 1940). Extreme cases of this type often involve the duplication of the wrist and forearm (O'Rahilly, 1951). These examples confirm various variants of the duplication of the hand, fingers, and often forearm, which are signs of polydactyly and ulnar dimelia ("mirror hand"), but not complete duplication of the upper limb (polymelia).

Three cases of polymelia of the upper limb were reliably described. Mennen et al. (1997) [15] presented a case of a child born in South Africa, who had two upper limbs on the left, superior and inferior to each other. In this case, the upper limb located above was doubled. Thus, this refers even to the limb triplication! Although the clavicle was doubled, there was only one scapula. The lower limb was hypoplastic and proximally attached to the chest below the blade–humerus joint of the superior limb. The movements of the elbow joint of the accessory upper limb were preserved, whereas flexion contractures were noted in the wrist joint. Movements of the upper limb joints were performed in full. The forearm was represented by two radial bones and one ulnar bone. The hand was completely doubled, all five fingers on each segment were preserved, and the hand duplication was atypical, with an axis of symmetry along the ulnar surface, and fingers V were in total syndactyly. This form of duplication is called radial dimelia and is practically not mentioned in the literature. Surgical treatment consisted of removing the inferiorly located accessory upper limb. Surgical treatment of upper (preserved) limb duplication has not been considered.

Researchers from the University of Washington described the case of a boy from Central America, who had two underdeveloped upper limbs located vertically to the left in relation to each other and was admitted for treatment [16]. Both

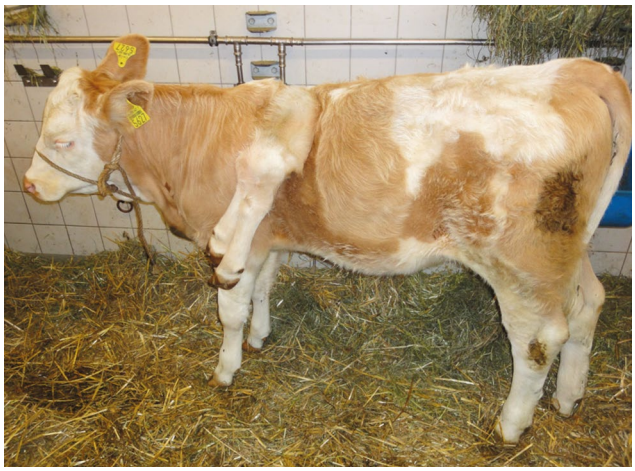

Fig. 10. Extraneous forelimbs in a calf

limbs were significantly underdeveloped, with the "upper" one being normally developed in the proximal part with its scapula and clavicle and a moderately shortened humerus. The significantly shortened forearm was represented by a rudiment of the radius, and only one finger was noted (probably finger I, as a continuation of the radius). The forearm and hand were at 170° (as in our patient). The upper limb located below was attached by soft tissues to the ribs and was represented in the proximal part by a rudiment of the elbow joint (with a small fragment of the humerus), then by the forearm with one ulna and a two-fingered hand. Both fingers had three phalanges, and the child could actively move them and even hold objects. Surgical treatment consisted of isolating and mobilizing the neurovascular pedicle of the "lower" upper limb, which enabled, without imposing microvascular anastomoses, moving of the limb to the distal humerus of the "upper" arm and performing osteosynthesis with the proximal ulna of the graft side-to-side (wire fixation). Thus, the authors, as in our case, managed to "assemble" one, longer, and more functional upper limb from two underdeveloped limbs.

Japanese surgeons from Tokyo described another case of complete upper limb duplication [17]. The patient had two upper limbs on the right, located in the frontal plane, one above the other. The "upper" limb in the proximal section was more developed, and the humerus articulated with the scapula and clavicle. The "lower" arm also had a shoulder joint between the humerus and the underdeveloped scapula. There was no collarbone. The humerus, both forearm bones, and all bones of the hand were completely duplicated, that is, there were anatomically complete two upper limbs. Soft tissue fusion in the limbs was noted in its entire length, and partial bone fusion was found in the elbow joints. The forearms and hands were fused by the ulnar surfaces and, accordingly, by fingers V. Thus, this form of polymelia can be classified as radial dimelia. Interestingly, Japanese surgeons separated the limbs with a conventional linear incision along the line of their fusion, and after removing the additional "lower" upper limb, the postoperative wound was simply closed linearly with interrupted sutures. In our opinion, such sutures are impractical because as the child grows, the linear longitudinal scar on the limbs tightens the soft tissues, which inevitably leads to their deformity and contracture formation in the joints.

All authors offer different theories about the mechanism of occurrence of this pathology; however, they do not agree with their assumptions. Some researchers propose the theory of the "fusion" of twins during embryogenesis, others believe that this anomaly is associated with the "separation" (doubling) of the upper limb, and other researchers explain this phenomenon by the formation of two buds of the upper limb at the initial stages of fetal development due to teratogenic factors. Through animal experiments, geneticists have identified the disorganization gene (Ds), a semidominant mouse model gene of variable penetrance, which has been described to disrupt normal organ development and is associated with significant limb abnormalities, including duplications. However, given the great rarity of this limb malformation, scientists have not come to a common point of view explaining its etiology.

CONCLUSION

The presented case of upper limb polymelia in a child is an extremely rare musculoskeletal pathology. In the available literature, only three cases of upper limb polymelia in humans have been reliably recorded and described in detail with photographs and radiographs. More often, articles have described lower limb duplications, which are also very rare (no more than 20 literary sources). In this era of highly advanced surgical technologies, it is quite possible to manage pediatric patients even with such complex limb malformations. Following a correctly planned surgery performed in our scientific center, a nearly complete, both esthetically and functionally, upper limb was constructed in a child. However, developing any general technology for treating these patients is impossible because these cases are extremely rare and have completely different manifestations of a developmental defect called polymelia (doubling of a limb).

ADDITIONAL INFORMATION

Funding. The study had no external funding.

Conflict of interest. The authors declare no conflict of interest.

Ethical considerations. The consent of the patient (his representatives) was obtained for processing and publication of personal data.

Author contributions. *S.I. Golyana* prepared the article.

The author made significant contributions to the study and preparation of the article and read and approved the final version before its publication.

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