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JPRAS Open 37 (2023) 139–144

Contents lists available at ScienceDirect

JPRAS Open

journal homepage: www.elsevier.com/locate/jpra

Case Report

Aesthetic shoulder reconstruction using hypoplastic tissue in a patient with phocomelia – A case report☆

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ARTICLE INFO

Article history:
Received 27 October 2022
Accepted 18 June 2023
Available online 1 July 2023

Keywords:
Phocomelia
Surgical treatment
Case report
Shoulder reconstruction

ABSTRACT

We present a case describing a technique for the surgical management and aesthetico-functional reconstruction of a shoulder in an adult with unilateral upper limb phocomelia (ULP).[1] A 25 year old male patient was presented to our clinic with upper left limb non-syndromic phocomelia. His main concerns was an aesthetically unpleasant limb and shoulder shape.

Few older publications describe available options for pediatric patients, such as clavicle transposition or limb amputation, but nowadays ULP treatment options focus on prosthetic rehabilitation (PR) and targeted muscle reinnervation (TMR). Our patient refused any prosthesis, and TMR is expensive, requires an experienced rehabilitation team and has limited availability. We aim to describe a non-expensive, simple and effective option for selected adult patients with limited shoulder functionality wishes.[2–5]

We used the existent hypoplastic limb for shoulder mound reconstruction, providing the patient with a more anatomical shoulder shape. The palmar skin of the hypoplastic hand and three existing fingers were excised. Also, distal phalanges were amputated. Then, the hand was rotated and positioned under the coracoid process, creating a shoulder-like mound.

☆ 12th Congress of Croatian Society for Plastic Reconstructive and Aesthtetic Surgery (CSPRAS) – Dubrovnik, May 4th-8th 2022.

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https://doi.org/10.1016/j.jpra.2023.06.006

Six months later, there has been a loss of shoulder volume compared to early postoperative weeks, but the patient is satisfied with the results.

Nevertheless, we found some limitations to our approach such as long-lasting postoperative pain, sensory symptomatology, and loss of volume of the reconstructed shoulder. Those could be solved with the complete denervation of the limb and a Latissimus Dorsi (LD) transposition flap in a second stage of reconstructive surgery.

Introduction

Phocomelia (from the greek "phōkē" -seal- and "melos" -limb-) is the term used to describe a rare severe congenital longitudinal malformation defined by the absence or severe hypoplasia of the long tubular bones with more or less intact hands and/or feet, resulting in a flipper-like appendage.[1] It is widely known as the most noticeable finding in thalidomide embryopathy.[6]

"True" phocomelia is defined by a total absence of intercalary structures, while "apparent" phocomelia refers to the most severe form of ulnar ray deficiency: the absence of ulna with radio-humeral synostosis.[7]

It is classified as "incomplete" when either the proximal or distal bones of the extremity are missing, or "complete" when it involves both the proximal and the distal bones of the limb.[8]

Although the pathogenesis of Phocomelia is not fully understood, disturbances to the apical ectodermal ridge during the first four to fourteen weeks of embryonic development seem to be closely involved.[7,9]

Phocomelia prevalence is found in 0.62 per every 100,000 live births, and can be sporadic, thalidomide-induced, or associated with at least 25 syndromes such as Roberts syndrome, Thrombocytopenia with radial aplasia (TAR) syndrome, and more.[7]

Treatment options include limb amputation and clavicle transposition, but mainly focus on PR and, more recently, TMR.[4,5] But the difficult prehension, inefficient task performance, and high user dissatisfaction account for the high abandon rates in PR. On the other hand, TMR is expensive, requires an experienced team and is not widely available.[5,10]

This report is the first to present a case of a shoulder surgical aesthetic reconstruction technique in an adult patient with upper limb phocomelia. The presented technique uses the de-epithelised phocomelic limb to create the bulk of a normal shoulder. It is a non-expensive, simple and effective option suited for patients demanding a reasonable aesthetic outcome without prosthesis or TMR.

Case report

A 25 year old male patient was presented to our clinic with upper left limb phocomelia (Figure 1), normal intelligence and cognitive development. His main concerns were an aesthetically unpleasant limb and an abnormal shoulder shape. **Being of adult age, he rejected any prosthetic treatment or flap reconstruction and** demanded to have a normally shaped shoulder, allowing him to dress normally.

The patient had no family history of any congenital anomaly. His mother denied any intake of teratogenic or vascular compromising medications during pregnancy. The case was classified as a non-syndromic phocomelia with associated spinal anomalies.

The hypoplastic left upper limb consisted of an absent humerus, radius, and ulna with a hypoplasic hand, including three developed fingers and hypoplastic carpal bones, **with a normal cutaneous sensation but lacking motor function**, directly attached to the thorax (Figure 2).

Figure 1. Upper left limb phocomelia.

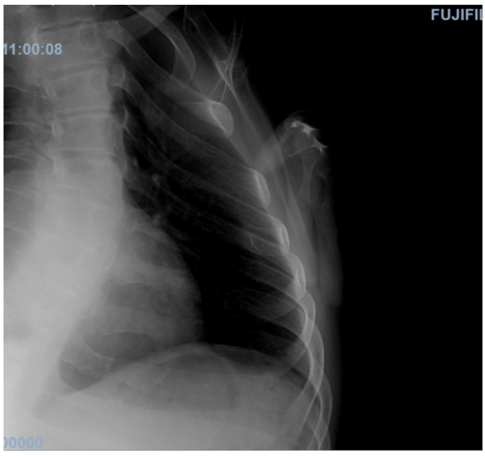

Figure 2. Absent humerus, radius, and ulna with a hypoplastic hand, and thoracolumbar scoliosis.

He presented with associated thoracolumbar scoliosis (Figure 2) that has been treated with multiple surgical corrective procedures until presentation.

Surgical management

The goal of the surgery was to use the existent hypoplastic limb for shoulder mound reconstruction.

The palmar skin of the hypoplastic hand was excised, as well as the skin of the three existing fingers. Subsequently, distal phalanges were amputated to prevent nail growth (Figure 3).

Later, the hand was rotated on its main neurovascular bundle and positioned under the coracoid process. Finally, the dorsal skin of the hand was sutured to the surrounding thoracic skin, consequently creating a shoulder-like mound (Figure 4). No internal fixation was set.

Early postoperative pain, rated by the patient, was between 7/10 and 8/10 on the Mankoski pain scale. The pain was more intense during sleep, associated with a feeling of tightness and stretching in the shoulder, and decreased over a period of two months. At six months, the patient had almost no pain and graded it 1 on the same scale, referring periodical squeezing sensations, tingling, and occasional spasms. Clinically, there has been a loss of shoulder volume compared to early postoperative

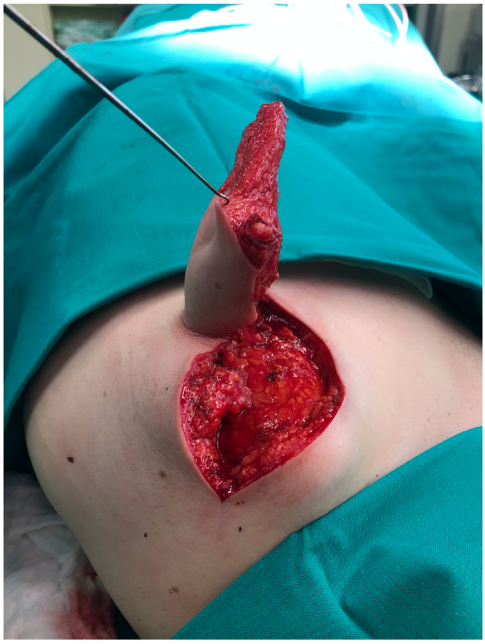

Figure 3. Hypoplastic hand without the excised skin & amputated distal phalanges but remaining dorsal hand skin.

weeks, but we achieved a good rectangular shoulder shape, less bulky than a muscle flap, and overall the patient is satisfied with the result (Figure 4).

Discussion

Clinical management of phocomelia usually includes prosthetics, orthopedic rehabilitation, management of associated anomalies (surgically or not), and psychological support for the patient and the family. There are a few publications available on surgical options for pediatric patients with Phocomelia, dating from the 1960s, but there is a lack of information about surgical management in adult patients.[2–5]

Prosthetic treatment in congenital **limb deformities** should begin before 24 months of age.[5] Central plasticity, familial involvement and cause of deficiency have great impact in prosthesis acceptance. However, because of the inability of the prosthetic device to provide reliable sensory and motor functions, rejection rates are as high as 45–75%.[5]

Other newer proposed treatment is targeted muscle reinnervation in pediatric patients, but until now it has been limited to adults after transhumeral amputation or shoulder disarticulation. However, because of need for an experienced team and high expenses, this is not a commonly offered option.[5]

The presented surgical technique is an adequate option to correct the defect and provide the patient with a more anatomical shoulder shape. A similar technique for augmenting the thenar base in a hypoplastic thumb was described by J.Upton et al. in which they excised skin, nail matrix and bone from the rudimentary thumb forming a filet flap to create an adiopfascial bulk on the thenar region.[10]

Nevertheless, we found some limitations to our technique, such as the possible appearance of long-lasting postoperative pain, sensory symptomatology, and loss of shoulder volume.

One option to prevent post-operative pain and sensory manifestations would be the complete denervation of the hypoplastic limb.

To correct the loss of volume and improve bone coverage, we propose an LD transposition flap in a second stage of reconstructive surgery.

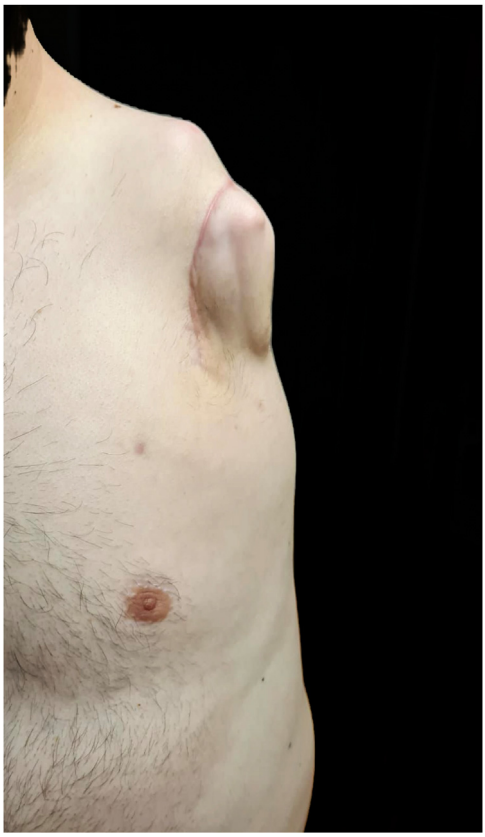

Figure 4. Six months after surgery, anterior view of the left shoulder.

Acknowledgements

None.

Funding

None.

Ethical approval

Not required.

Declaration of Competing Interest

None declared.

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