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Understanding Poland syndrome: a collaborative approach to patient care

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ABSTRACT

Background and objectives. Poland syndrome is a rare congenital condition characterized by the underdevelopment of the chest muscles and upper limb abnormalities, commonly presenting with brachy syndactyly. This case report details the successful staging of plastic surgery in a 3-year-old boy with Poland syndrome, who had a significant deformity in his left hand. The aim is to describe the importance of early diagnostic imaging, the surgical strategy, outcomes, and the challenges faced during the staged correction process.

Materials and methods. A 3-year-old boy with Poland syndrome presented with underdevelopment of the left pectoral muscles and significant brachysyndactyly of the left hand. A multidisciplinary team developed a comprehensive, staged surgical plan to address these deformities by corrective surgery on the left hand, including digit lengthening and syndactyly release. Postoperative management included monitoring for complications and a structured rehabilitation program to enhance hand function.

Results. Over 12 months, the staged surgical procedures were completed successfully. Hand surgery significantly improved digit length and separation, resulting in enhanced grip strength and fine motor skills. Postoperative imaging confirmed the correction of bony and soft tissue abnormalities. The patient showed marked improvements in both physical function and psychosocial well-being.

Conclusions. Early diagnosis through appropriate imaging and Staged plastic surgery for the correction of Poland syndrome-related deformities in a young child, particularly involving the left hand, can result in excellent functional and cosmetic outcomes. Early intervention, precise surgical planning, and a multidisciplinary approach are crucial for successful treatment.

Keywords: hand function, reconstructive surgery, pectoralis major, congenital syndactyly, Poland syndrome

INTRODUCTION

After examining the corpse of a prisoner named George Elt at Guy's Hospital in 1841, Sir Alfred Poland initially reported the existence of Poland syndrome [1]. It is a congenital condition where the pectoralis major muscle is unilaterally absent or hypoplasia. This condition often includes associated upper limb anomalies, such as syndactyly [2,3]. The embryological mechanisms underlying Poland syndrome remain largely unknown, but it is speculated that vascular disruptions during fetal development play a significant role. The syndrome's prevalence varies from 1 in 10,000 to 1 in 100,000 live births, with no discernible bias toward either side of the body.

Early recognition of Poland syndrome is crucial for managing associated complications and providing appropriate medical and psychological support. The syndrome's variable presentation and potential for associated anomalies underscore the need for a comprehensive, multidisciplinary approach to patient care. A few other names for the syndrome are “Unilateral defect of pectoralis major with syndac-

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tyly of the hand, Poland's anomaly, Poland's sequence, Poland syndactyly, and Poland's anomaly." Poland syndrome can be classified into several types based on the severity and extent of anomalies. Type 1: the isolated pectoral muscle defect in its minimal form. Type 2: the incomplete form, or pectoral muscle defect, connected to abnormalities of the upper limb or ribs. Type 2 is divided into two subtypes: Type 2a is the upper limb variant; whereas Type 2b, the thoracic variant, shows rib anomalies without upper limb anomalies. Type 3: the full form, characterized by a pectoral muscle defect linked to rib and upper limb abnormalities.

CASE PRESENTATION

A 3-year-old boy delivered vaginally and with an otherwise normal antenatal and perinatal history, was referred to the plastic surgery department due to a congenital hand anomaly. Clinical examination revealed unilateral hypoplasia of the left pectoralis major muscle along with syndactyly involving the second, middle, and ring fingers of the left hand as represented in (Figure 1). The posters anterior chest radiograph showed unilateral hyperlucency on the left side, correlating with the missing of the pectoralis major muscle as given in (Figure 2). The X-ray of the left hand exhibited a relatively short and tubular second metacarpal with irregular cortical surfaces and the absence of the ossification center of the metacarpal head. The remaining metacarpals

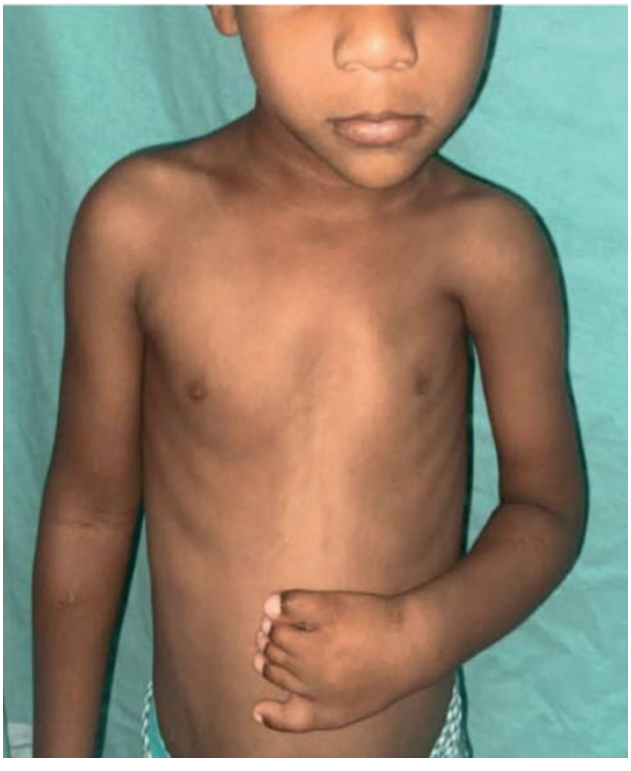


FIGURE 1. Hypoplasia of left pectoralis major muscle with syndactyly of left hand

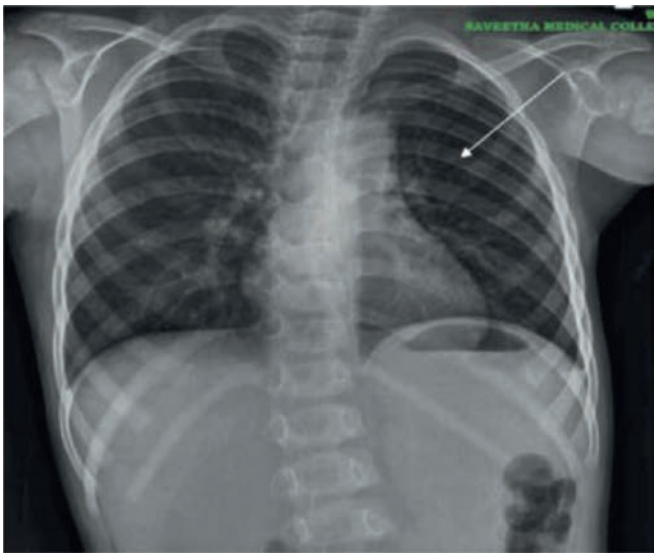


FIGURE 2. Chest radiograph posteroanterior view showing unilateral (left) hyper lucency of chest with absent pectoralis major muscle as indicated by the white arrow

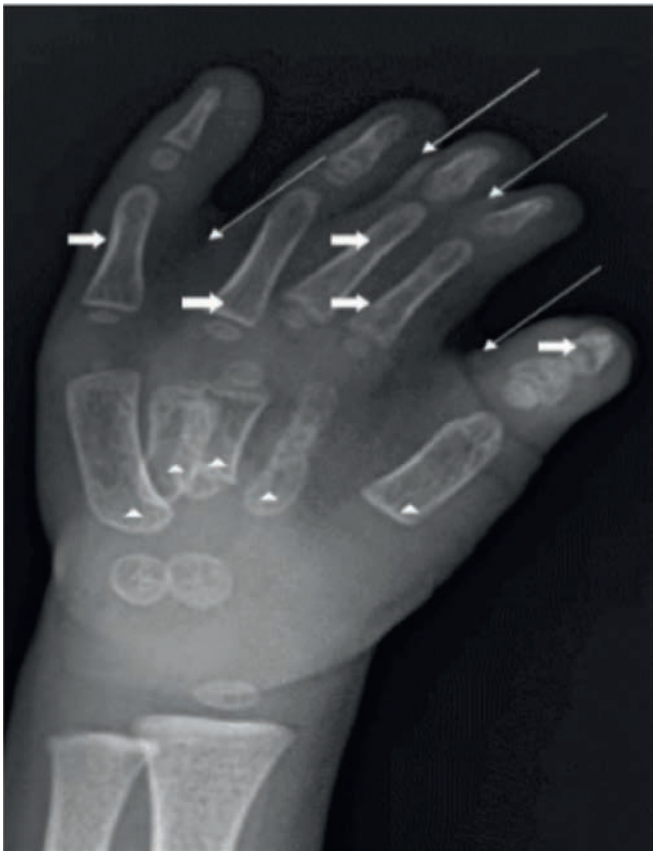


FIGURE 3. Radiograph anteroposterior view (AP) of the left-hand shows ahort and tubular metacarpals as indicated by a white arrowhead with the appearance of all corresponding primary ossification centers at physeal ends without evidence of fusion; ahort fingers with absent middle phalanx as indicated by the white thick short arrow. Suspicion of soft tissue syndactyly between the first and second digit (thumb and index finger), and between the second, third, and fourth digits (index, middle, and ring fingers) with relatively separate/longer fifth digit (little finger) as indicated by the white long arrow. No evidence of osseous syndactyly of the left hand

were noted to be short and broad with digits showing a bi phalangeal structure. No evidence of osseous syndactyly (bony fusion) was found as shown in (Figure 3). MRI scans confirmed the X-ray findings and provided detailed imaging of soft tissues and bones. It revealed the absence of the ossification center in the second digit's metacarpal head, with visible cortical irregularities. Additionally, soft tissue syndactyly was evident, with fused skin and underlying tissues between digits. The lack of ossification in the middle and distal phalanges indicated disruption in normal bone formation. MRI was crucial for surgical planning, offering precise anatomical details necessary for addressing the patient's specific anomalies (Figure 4). The child underwent multi-staged plastic surgical correction for the hand deformity. Interval corrective surgeries have been done for this patient for over 1 year as shown in (Figures 5 A-C) and (Figures 6 A, B), and been on regular follow-up with rehabilitation.

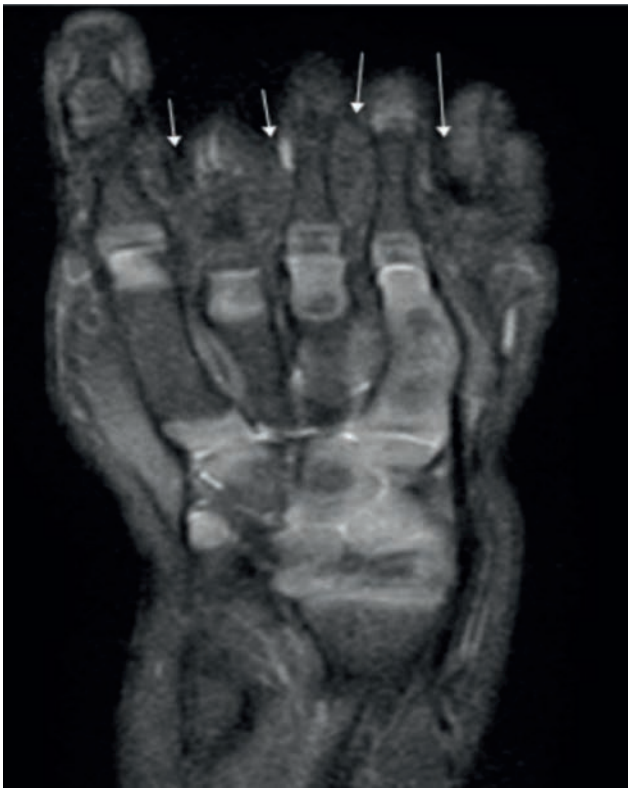
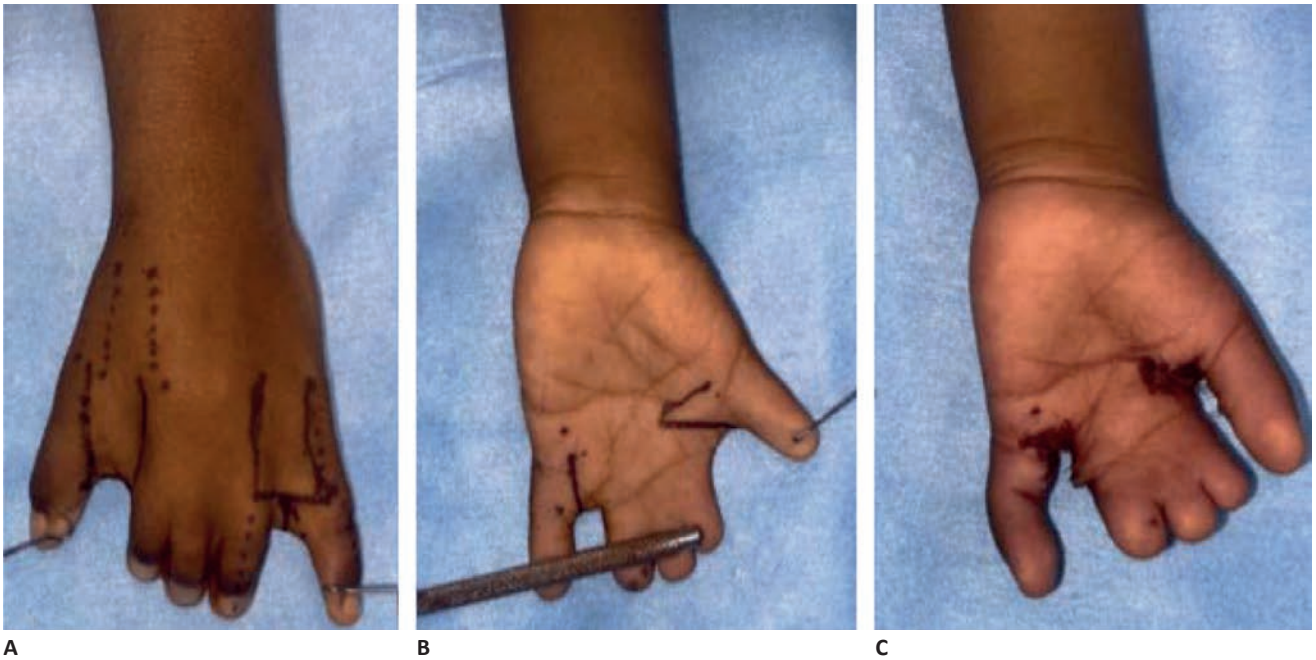


FIGURE 4. PD SPAIR sequence of MRI representing the soft tissue changes of 2nd-3rd-4th-fingers syndactyly. Proton Density (PD)Spectral Attenuated Inversion Recovery (SPAIR) with findings corresponding to x-ray

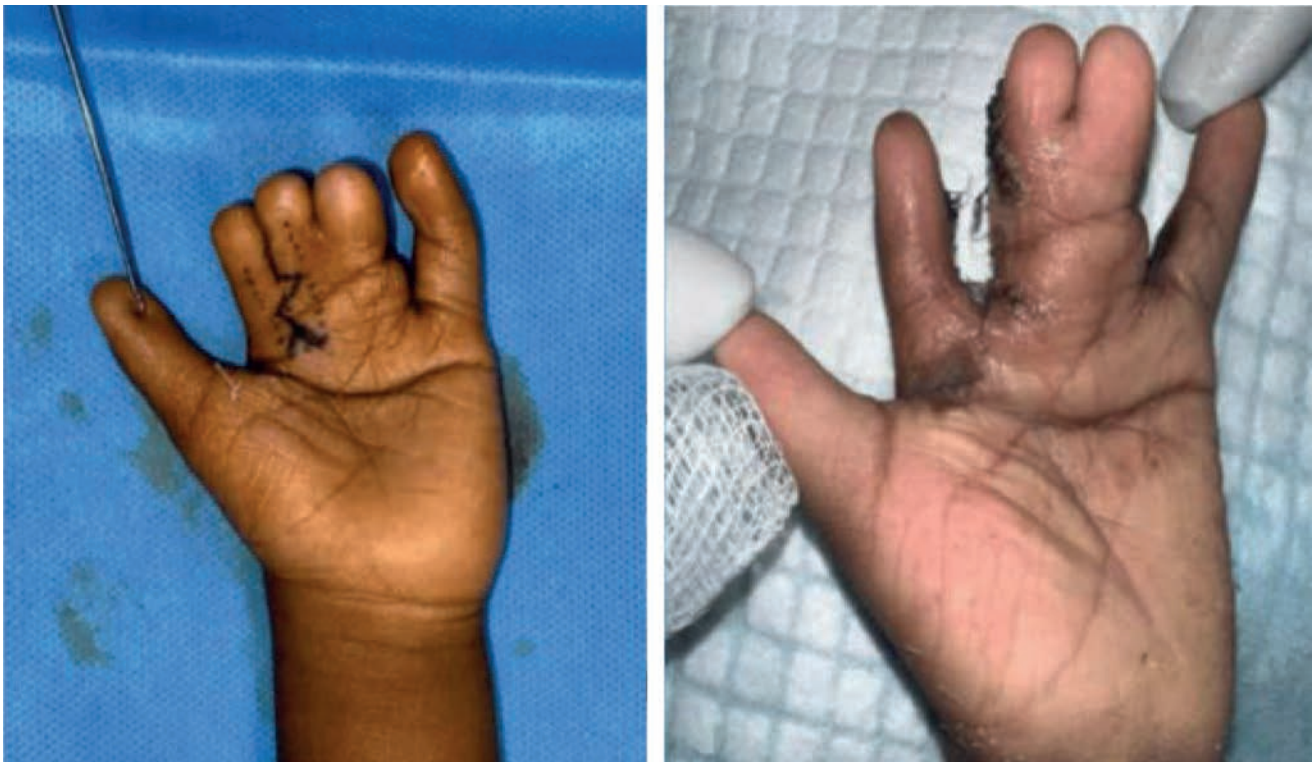
DISCUSSION

The development of the upper limb bud and adjacent chest wall is believed to be critical during the sixth week of gestation. Poland syndrome may result from an interruption in the embryonic blood supply during this time. Vascular problems – more especially, those affecting the subclavian artery or its branches – may cause hypoplasia of the ipsilateral subclavian artery. Supporting evidence includes observations of reduced subclavian artery diameter and decreased flow velocity, which may influence the extent of resultant anom alies. For example, the sternocostal region of the pectoralis major muscle may disappear due to hypoplasia of the internal thoracic artery, while hand abnormalities may result from hypoplasia of the brachial artery. The subclavian artery can sustain damage from a mesodermal alteration, which may result in a vari-



A B C

FIGURE 5. First stage release of soft tissue syndactyly between thumb and index finger and between 4th and 5th finger as marked in blue pen



A B

FIGURE 6. Soft tissue release between the 2nd and 3rd fingers as marked in blue pen and the post-operative follow-up

ety of developmental alterations in the regions it supplies. The SASDS (Subclavian Artery Supply Disruption Sequence) is a phenomenon first reported by Dr. Alfred Poland. It is believed to occur when the ribs grow into an abnormal position that causes the subclavian artery to become hypoplastic. Tumors, amniotic bands, abnormal muscle growth, surrounding tissue edema, and mechanical problems like avascular thrombosis or embolism in the subclavian artery can all be contributing factors to this condition. The degree of arterial blockage and the precise site of the vessel involvement affect the resulting musculoskeletal abnormalities. Generally, more proximal blockages result in more severe clinical symptoms.

One theory suggests that the observed defects could be caused by disruption of the lateral plate mesoderm, the source of the pectoralis muscle, between 16 and 28 days after fertilization. Additional theories regarding the causes of Poland syndrome include teratogenic effects, viral infections, trauma, autosomal dominant inheritance, genetics, and intrauterine insults. Individuals suffering from Poland syndrome often lack both the pectoralis minor and the sternocostal head of the pectoralis major muscle. Some patients (including athletes) may not experience functional impairment because they lack the pectoralis major muscle. In certain instances, it may also be observed that other muscles, comprising the serratus anterior, external oblique, as well as latissimus dorsi, are involved. Chest wall deformities, including unilateral chest wall depression and rib hypoplasia, are common. The ribs on the affected side may be thin, deformed, or hypoplastic, leading to a characteristic chest appearance. The asymmetry of the pectus carinatum may also be caused by the sternum rotating toward the afflicted side. Rib defects often progress during growth spurts. One kind of Poland syndrome that is thought to exist is isolated pectoral hypoplasia. In familial cases, there may be one person with hand and pectoral deformities and another with isolated pectoral hypoplasia. Diagnostic criteria should include isolated pectoralis major absence with associated breast hypoplasia, a feature often seen in women seeking breast augmentation due to asymmetry.

The hand abnormalities associated with Poland syndrome are usually unilateral as well as can vary from complete absence of the hand (ectrodactyly) to shortened middle phalanges with webbed skin (syndactyly) [4-5]. It is uncommon for hand anomalies to occur contralaterally to the chest deformity. Estimates suggest that around 10% of syndactyly patients also have Poland syndrome. Poland syndrome may co-occur with other congenital conditions, such as Möbius syndrome, characterized by the paralysis of the eye abductor muscles and bilateral congenital facial nerve palsy, or Klippel-Feil syndrome, marked by a short neck due to cervical vertebrae fusion. When unilateral renal agenesis or other renal abnormalities are coupled with pectoralis major muscle aplasia, an acro-pectoral-renal field defect re-

sults. This combination can cause renal hypertension, which means that all patients with pectoralis major aplasia need to have renal evaluations. Poland syndrome has also been associated with tumors and other developmental defects, possibly due to abnormalities in homeobox and tumor suppressor genes. Despite breast hypoplasia, patients with Poland syndrome are still at risk for pathological processes in mammary tissue, including invasive ductal carcinoma, highlighting the importance of cancer surveillance.

The use of diagnostic imaging methods like CT (Computed Tomography) scans as well as MRI (Magnetic Resonance Imaging) is essential for identifying and evaluating the degree of muscle and bone abnormalities in Poland anomaly. MRI is particularly valuable as it provides detailed images of soft tissues, allowing clinicians to evaluate the presence, size, and state of the muscle known as the pectoralis major. An MRI usually shows that this muscle is absent or severely hypoplasia in cases of Poland anomaly [6]. CT scans complement MRI by providing detailed cross-sectional images of the chest wall as well as the rib cage. This imaging modality is useful for assessing rib aplasia or hypoplasia, chest wall deformities, and associated bony abnormalities. CT imaging can also help visualize any compensatory changes in adjacent structures. Several musculoskeletal conditions can mimic the clinical presentation of Poland anomaly, including congenital deformities of the chest wall along with upper limb conditions. Diagnostic imaging aids in differentiating Poland anomaly from syndromic conditions with similar chest wall deformities, such as Jeune syndrome or congenital rib anomalies, and Complex Muscular Conditions such as congenital muscular dystrophy or other muscle-wasting disorders may present with similar muscle deficiencies. An MRI confirms the specific absence of the pectoralis major muscle, which is the hallmark of the Poland anomaly.

Poland anomaly may be associated with vascular anomalies [7]. Advanced imaging techniques like CT angiography or MRI angiography are essential to identify and differentiate vascular abnormalities, such as variations in the blood supply to the affected area, which may impact surgical planning. Imaging is crucial in ruling out tumors or other lesions that could present with similar clinical symptoms. MRI and CT scans can differentiate between benign and malignant masses and other pathologies, ensuring accurate diagnosis and appropriate management. Detailed imaging is indispensable for preoperative planning. It helps in mapping out the precise anatomical details, such as the extent of muscle deficiency, rib deformities, and any associated anomalies. This information guides the surgical approach, whether it involves muscle flap transposition, rib reconstruction, or breast augmentation. Imaging is also useful for assessing surgical outcomes and monitoring any potential complications. Postoperative MRI or CT scans can help evaluate the success of muscle flap integration, the stability of rib reconstructions, and the overall alignment of the chest wall. Regular imaging over time can monitor the progression or resolution of the anomalies as the patient grows. This is particularly important in pediatric patients, where developmental changes might impact the effectiveness of initial treatments and may require additional interventions. Long-term follow-up imaging helps in detecting any recurrence of anomalies or complications arising from surgical interventions. This ongoing assessment ensures timely intervention if any issues arise.

In summary, diagnostic imaging is fundamental in the comprehensive evaluation of Poland anomaly, aiding in accurate diagnosis, differentiation from other conditions, effective preoperative planning, and ongoing monitoring. The use of MRI and CT scans provides critical insights into the anatomical and pathological aspects of the anomaly, facilitating better management and treatment outcomes.

Anomalies associated with Poland syndrome can result from vascular disruptions and abnormal embryological development of associated structures, including skeletal elements [8]. Genetic research suggests that mutations in the IHH (Indian hedgehog) gene on chromosome 2q35–36 and a locus on chromosome 5p13.3–p13.2 may cause Poland syndrome [9,10].

The severity of the abnormality, the patient's age, as well as their sex, should all be taken into consideration when treating Poland syndrome. The surgical repair can be conducted in either 1 or 2 stages. In cases of severe rib aplasia and significant chest wall depression in children, a two-stage correction is recommended. Initially, the rib defects are repaired, followed by muscle flap transposition after puberty. For female patients, this procedure is combined with breast augmentation. Surgery should be put off until after puberty for children with milder forms, which are defined by breast hypoplasia and the absence of pectoralis muscles. At that point, female patients can have breast augmentation and muscle flap transposition. Mesh patches, additional bony allografts or autografts, subperiosteally harvested rib split grafts from the side not injured, or a combination of these techniques can be used to stabilize the chest wall in cases of lung herniation and substantial rib defects. On the afflicted side, there might still be insufficient muscle and subcutaneous tissue despite these surgeries. In these situations, implanting a silicone breast prosthesis and transferring the latissimus dorsi muscle accomplish the necessary

correction. Additionally, to correct the sternal rotation, a "reversed" figure-eight wire suture and a transverse sternotomy should be done simultaneously. It is possible to strengthen the underdeveloped pectoralis major muscle with prosthetic implants or musculocutaneous flaps. After puberty, augmentation should be performed to match the contralateral breast's size that has reached full development. Because subcutaneous tissue and skin are usually weak and thin, muscle-cutaneous flaps are used. The latissimus dorsi musculocutaneous flap can also be used to replace the anterior axillary fold and the lost muscle mass. Endoscopically assisted minimally invasive techniques have been proposed recently to lower donor site morbidity.

When the latissimus dorsi is hypoplastic, it can cause an avascular fibrous sheet and a posterior axillary fold that looks normal. Accurate diagnosis requires imaging techniques such as MRI. In cases where the latissimus dorsi muscle is hypoplastic or absent, microsurgical transfer of the contralateral muscle can be performed. Other options include using the internal thoracic vessels to perform microvascular-free TRAM (transverse rectus abdominis muscle) flaps or upper gluteal flaps. Angiography is done pre-operatively to assess vascular supply before tissue transfer. In patients with extensive rib defects as well as muscle Hypoplasia, a prefabricated chest wall implant could be used. Bone allografts and mesh patches were used in some cases, with the addition of latissimus dorsi muscle flaps. This can correct concave deformities, stabilize the chest wall, along with prevent lung herniation. Post-operative physical rehabilitation increases the results by promoting hypertrophy of the transformed muscle flaps.

Surgical management of Poland syndrome with syndactyly requires a coordinated, staged approach to optimize hand functionality and aesthetic outcomes. The patient underwent several corrective surgeries over a year: The primary surgical goal was to separate the fused digits to improve function and appearance. Syndactyly release utilized the Z-plasty technique, which involves Z-shaped incisions to divide the digits while minimizing tension on the skin. Full-thickness skin grafts were harvested from donor sites, such as the groin or inner arm, to cover the newly exposed areas between the digits, ensuring adequate coverage and flexibility [11]. The short and broad appearance of the digits necessitated lengthening procedures to enhance hand function. Distraction osteogenesis, which involves gradually stretching the bone after it has been cut, was employed to lengthen the affected digits. This method promotes new bone growth in the gap, requiring careful monitoring and adjustments to ensure proper alignment and avoid complications like joint stiffness. Cortical irregularities and the absence of ossification centers were addressed through reconstructive procedures on the metacarpals and phalanges. Bone grafting was used in areas with significant bone deficits, with autologous bone harvested from the iliac crest providing a stable structure for the affected digits. For cases involving tendon or nerve damage, microvascular techniques were used for repair or reconstruction. Tendon transfer procedures were also considered to restore digit function. These complex procedures are essential for maximizing hand functionality.

Postoperative rehabilitation is critical in managing Poland syndrome, particularly in cases involving syndactyly correction. Rehabilitation supports surgical outcomes and enhances overall hand function. Post-surgery, gentle range of motion exercises were introduced to prevent joint stiffness and enhance flexibility, focusing on newly separated digits to ensure full movement without stress on healing tissues. As healing progressed, strengthening exercises targeting intrinsic hand muscles were incorporated to improve grip strength and dexterity, vital for daily activities. Occupational therapy involves play-based activities designed to improve fine motor skills and encourage hand use in daily tasks such as feeding and writing [12]. Adaptive tools and devices were introduced to assist with hand function, including modified utensils, writing aids, and toys to support fine motor skills and bilateral hand use.

Counseling services were provided to support the child's self-esteem and social interactions, offering emotional support and coping strategies for navigating the challenges of the condition and its treatment. The family was encouraged to connect with support groups for Poland syndrome to share experiences and gain insights from others facing similar challenges.

Although prenatal diagnosis is uncommon, if a hand abnormality is found, suspicions might be raised.

CONCLUSION

This case of Poland syndrome in a 3-year-old boy highlights the complexity of diagnosing and managing the condition. The successful outcome underscores the importance of a multidisciplinary approach, involving plastic surgeons, radiologists, therapists, and psychological support services. Early intervention and tailored treatment strategies are critical in optimizing functional and aesthetic outcomes for patients with Poland syndrome. Future research should focus on exploring the genetic and embryological underpinnings of the syndrome to improve early detection and intervention strategies.

Informed consent:
The authors obtained the written consent of the identified persons or their legal guardians for the presentation of the cases within the present scientific paper.

Conflict of interest:
The authors declare they do not have any financial or personal relationships that might bias the content of this work.

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Statement on human and animal rights:
The authors delare that the research conducted complied with the ethical standards in accordance with Helsinki Declaration (of 1975, revised in 2013), as well as national regulations in the field.

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