Case report

Physiotherapeutic approach in Alfred Poland syndrome: a case report

Abordagem fisioterapêutica na síndrome de Alfred Poland: um relato de caso

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ABSTRACT | INTRODUCTION: Alfred Poland Syndrome (APS) is a rare congenital anomaly characterized by the total absence of the pectoralis major, pectoralis minor, serratus muscles, and the breast, as well as the areolar-nipple complex, with less than 1% risk of familial recurrence. It can also present with defects in cartilage and ribs, hypoplasia of subcutaneous tissues in the chest wall, and alopecia in the axillary and nipple regions. Poland was the first to describe an anomaly involving the absence of the pectoral muscle associated with hand deformities, which was later further described by Froriep. **OBJECTIVE:** This study aims to document the physiotherapeutic approach in a 6-year-old female child with normal neuropsychological development and atypical motor development, in order to assist the academic community in managing this rare syndrome. **METHOD:** The methodology refers to a case report, based on available medical examinations and the evaluation of the pediatric patient. The evaluation consisted of a biomechanical analysis of the scapular girdle. Following this investigation, therapeutic exercises were established. Reassessments were conducted approximately every 30 days, targeting the pectoralis major, pectoralis minor, and rhomboid muscles. **RESULTS:** There was an increase of one strength grade in the following muscles: deltoid, teres major and minor, supraspinatus, infraspinatus, subscapularis, rhomboids, and pectoralis minor. These muscles initially presented grade 3 muscle strength and progressed to grade 4. No strength improvement was observed in the pectoralis major. The child exhibited improved body awareness, reduced muscular compensations, and better postural alignment. **CONCLUSION:** This case report demonstrated that physiotherapy improved the motor development of a child with Alfred Poland Syndrome, particularly in terms of muscle strength, posture, and body awareness. Family support and encouragement, especially from the parents, contributed to the child's sense of security and motivation during treatment, fostering greater engagement. The family's active involvement integrated the treatment into the child's daily activities, helping her apply what she learned in therapy sessions and resulting in improvements in scapular girdle muscle strength, postural alignment, and body awareness.

KEYWORDS: Poland Syndrome. Physical Therapy. Pediatrics.

RESUMO | INTRODUÇÃO: A síndrome de Alfred Poland (SAP) é uma anomalia congênita rara com ausência total dos músculos peitoral maior, peitoral menor, serrátil e da mama. Além do complexo areolocapilar, com risco de menos de 1% de recorrência familiar, pode ser encontrado também defeitos em cartilagens e costelas, hipoplasia de tecidos subcutâneos da parede torácica e alopecia das regiões axilar e mamilar. Poland foi o primeiro a descrever uma anomalia com ausência do músculo peitoral associada à deformidade das mãos, mais tarde, essa mesma síndrome foi descrita por Froriep. **OBJETIVO:** O presente estudo tem como objetivo registrar a abordagem fisioterapêutica em uma criança do sexo feminino, de 6 anos de idade, com desenvolvimento neuropsicológico normal e motor atípico. O foco do estudo é poder auxiliar a comunidade acadêmica no manejo da síndrome supracitada. **MÉTODO:** A metodologia apresentada refere-se a um relato de caso, realizada a partir dos exames disponíveis e da avaliação da paciente pediátrica. Esta consistiu em uma análise biomecânica da cintura escapular. Após esta investigação, estabeleceu- se os exercícios terapêuticos. A reavaliação foi realizada a cada 30 dias, em média. Tendo como alvo os músculos peitoral maior, peitoral menor e romboides. **RESULTADO:** Houve ganho da força muscular em um grau dos músculos: deltoide, redondo maior e menor, supra espinhal, infra espinhal, subescapular, romboides e peitoral menor. Estes músculos possuíam grau 3 de força muscular e evoluíram para grau 4 de força muscular. Não houve ganho de força muscular em peitoral maior. A criança apresentou melhor consciência corporal e, consequentemente, diminuição das compensações musculares e melhor alinhamento corporal. **CONCLUSÃO:** Este relato de caso mostrou que a fisioterapia melhorou o desenvolvimento motor de uma criança com síndrome de Alfred Poland, especialmente em força muscular, postura e consciência corporal. O apoio e incentivo familiar, especialmente dos pais, contribuíram para a segurança e motivação da criança durante o tratamento, promovendo maior engajamento. A colaboração ativa da família integrou o tratamento às atividades diárias, ajudando a criança a aplicar os aprendizados nas sessões, resultando em melhorias na força muscular da cintura escapular, alinhamento postural e consciência corporal.

PALAVRAS-CHAVE: Síndrome de Poland. Fisioterapia. Pediatria.

Submitted Oct. 12th, 2025, Accepted Mar. 10th, 2025,
Published Apr. 7th, 2025
J. Physiother. Res., Salvador, 2025;15:e5969
http://dx.doi.org/10.17267/2238-2704rpf.2025.e5969 | ISSN: 2238-2704
Assigned editor: Marina Makhoul

How to cite this article: Lopes SL, Flor C. Physiotherapeutic approach in Alfred Poland syndrome: a case report. J Physiother Res. 2025;15:e5969. http://dx.doi.org/10.17267/2238-2704rpf.2025.e5969

1. Introduction

The Poland Syndrome (PS) is a rare congenital anomaly characterized by the complete absence of the major pectoral muscles, minor pectoral muscles, serratus, and breast tissue. PS is non-progressive, and in the absence of severe thoracic malformations, its survival rate is comparable to that of the general population.[1] It may present with bone defects such as scoliosis, as well as pulmonary hernia and dextrocardia.[2] The risk of familial recurrence is less than 1%. This syndrome can present with either partial or complete absence of the areola-papillary complex.[1] It may also include defects in the cartilage of the ribs and clavicles, hypoplasia of subcutaneous tissues in the thoracic wall, and alopecia in the axillary and mammary regions.[1-3]

Poland was the first to describe an anomaly with the absence of the pectoral muscle associated with hand deformities, which was later described by Froriep.[3] According to Tsung-Han Ho & Chun-Chieh Wang, a probable interruption in the embryonic development of the subclavian artery leads to PS4 at the end of the sixth week of gestation. This can cause hypoplasia of the developing subclavian artery or its branches, potentially leading to deformities in the upper extremities and manifestations in the hands.[4-7] Rupture of the developing internal thoracic artery may result in hypoplasia or aplasia of the major pectoral muscle and the thoracic wall. Based on the embryonic development of PS and its resulting characteristics, functional impacts can be observed from performing daily life activities (eating, writing, and self-care) to respiratory changes (reduced lung capacity). Bazzi Junior JL, Matta ES et al.[8] report a case of spontaneous pneumothorax due to the malformation of the subclavian artery in a patient diagnosed with PS.

A very important aspect to consider is the alteration of body image, particularly during adolescence, which can lead to reduced self-esteem.[9] In some cases, especially in women, surgical treatment for breast reconstruction is recommended.[3,4]

Diagnosis is made through physical examination, chest X-ray[5], ultrasound (US), computed tomography (CT), and magnetic resonance imaging (MRI). US represents the first-line diagnostic imaging method to support or confirm the clinical diagnosis of PS due to its accessibility, cost-effectiveness, and radiation-free nature. In selected cases of severe thoracic malformation, CT and/or MRI are appropriate for a complete preoperative evaluation. Both CT and MRI clearly show the absence of the major pectoral muscle and allow for better evaluation of other associated musculoskeletal and internal organ anomalies. However, considering the radiation exposure of CT and the need for anesthesia in younger pediatric patients undergoing MRI, a test that is not inexpensive, despite its effectiveness, should not be indicated as the first approach.[9] Early diagnosis can contribute to better pediatric, orthopedic, and aesthetic management (in the case of women with absent breasts).[8]

In Brazil, studies show not only the low incidence of PS1 (1:30,000 people) but also genetic influences and chromosomal abnormalities.[5,10] This syndrome is more common in men than in women and is rarely bilateral.[8,10]

Poland Syndrome is not a progressive disease, and in the absence of severe thoracic malformations, its survival rate is similar to that of the general population.[9]

The child in this case study is six years old, female, with normal neuropsychological development and atypical motor development. Imaging examination through ultrasound did not reveal the presence of the major and minor pectoral muscles on the right side. This work is justified by the lack of literature reports on physiotherapeutic treatment for this syndrome. Therefore, this study aims to document the physiotherapeutic treatment to assist the academic community in managing the aforementioned syndrome. Since physiotherapy aims to improve muscle function and mobility, promoting better quality of life and adaptation to daily life.

2. Method

A child, FFB, 6 years old, diagnosed with Alfred Poland Syndrome, participated in the study with the voluntary authorization of the parents, who were properly informed about the study's objectives, procedures, and possible implications. They provided their free and informed consent by signing the Consent Form. This article presents a case report approved by the Research Ethics Committee of the Couto Maia Hospital/SES/BA - Plataforma Brasil, in accordance with current ethical guidelines. The methodology presented refers to a case report.

- Data collection: Data was obtained from the medical exams provided by the child's parents, a kinesiological analysis of the right and left upper limbs, and a physical examination through palpation and muscle strength testing of the child in the study.

- Nosological diagnosis: Unilateral and right-sided Alfred Poland Syndrome was verbally communicated to the parents by the attending physician after an ultrasound examination of the right pectoral region. The report describes the absence of pectoral muscle remnants on the right side but does not specify which portion of the muscle is missing.

- Physiotherapeutic intervention: A therapeutic plan was developed with two 45-minute sessions per week, including playful activities to strengthen the muscles, improve postural alignment, and enhance body awareness. Isometric exercises, active kinesiotherapy, throwing activities, and proprioceptive movements were performed, resulting in increased range of motion, better alignment, and absence of pain.

It was not possible to subject the child, FFB, to a computed tomography scan for a deeper evaluation, as it requires sedation and contrast use. Thus, the physician opted not to proceed with it.

3. Patient information

This is a female child, 06 years old, with normal neuropsychological and atypical motor development. She had a clinical diagnosis of Alfred Poland Syndrome on the right. An ultrasound report of the anterior region of the shoulder girdle showed no evidence of the major or minor portions of the pectoral muscle.

4. Clinical findings

An analysis of the movements of the right shoulder complex was carried out and it was noted that some movements were performed in a dysfunctional manner. The child had passively free ranges of movement. When asked to internally rotate the right shoulder in the supine position, the child rotated the trunk to the same side. When asked to flex the right shoulder in the supine position, the child flexed the right shoulder up to 180 degrees. To achieve this range of right shoulder flexion, the child starts the movement with a shoulder-ear approach and internal rotation of the right shoulder, showing a winged scapula during the movement. Horizontal adduction in sedation begins with the shoulder flexed to 90 degrees, performing the movement without compensations. When performing horizontal abduction of the right shoulder with it flexed to 90 degrees, when it reaches 60 degrees of external rotation, the trunk begins to tilt to the right, activating the trunk muscles on the same side. When trying to perform the right shoulder extension movement, she tilts her trunk anteriorly and throws her right upper limb further back with elbow flexion. In the right shoulder abduction movement, it was observed that the child begins the movement with an elevation of the assessed right shoulder and then begins the shoulder abduction movement with a trunk tilt to the left side. When performing adduction of the same shoulder, the child initially flexed the elbow and brought the upper limb closer to the body. The scapula remains straight in adduction of the right shoulder. Bilateral winged scapulae were observed, more evident on the right. A kinesiological analysis revealed bilateral grade 03 rhomboid muscle weakness. There was no trace of contraction of the pectoralis major muscle on the right and grade 02 muscle strength in a small portion of the pectoralis minor on the right. In addition to possible muscle weakness in the serratus anterior. When asked to move the MSD to any plane, the child shows a significant change in the center of gravity, with lumbar hyperlordosis, abdominal protrusion and trunk inclination to the left.

The child reports pain in the anterior region of the right shoulder, grade 4 on the numerical pain scale (0-10). These pains are rarely associated with using the right upper limb in sporting activities, such as throwing a ball or hula hoop.

Table 1. Kinesiological analysis of the right shoulder

internal rotation in supine position	shoulder flexion in sitting position	Horizontal adduction in sitting position	Horizontal abduction in sitting position	shoulder extension	shoulder abduction	shoulder adduction
tilt of the trunk to the same side	It reaches 180 degrees with compensation s , bringing the humerus closer to the ear	With right shoulder at 90 degrees, movement without compensations	Movement performed with the shoulder at 90 degrees. The movement begins without compensation. Upon reaching 60 degrees of external rotation, the trunk tilts to the right.	Lean your trunk forward and flex your right upper limb at the elbow.	The child brings the humerus closer to the ear and tilts the trunk to the left side.	Flex your right elbow to bring your right upper limb closer to your trunk. Keep your shoulder blades winged during the movement.

Source: the authors (2025).

5. Diagnostic evaluation

Upon taking the family history, there is no report of previous cases of individuals with Poland Syndrome. The child in question does not have any other comorbidities. She is active and participates in various educational and social activities. No signs of low self-esteem or discouragement were noted by the child. The family reports that no treatment for Poland Syndrome has been carried out previously.

6. Results timeline

After conducting an ultrasound examination of the anterior scapular region, the absence of the pectoralis minor and major muscles was observed. The attending physician recommended that the family arrange a physiotherapy assessment to strengthen the musculature adjacent to the right pectoralis major and minor muscles.

A muscle strength test with resistance was performed, and traces of muscular contraction of the right pectoralis minor muscle were noted when compared to the same muscle on the left side.

Therapeutic intervention was then initiated with the aim of strengthening this minimal portion of the pectoral muscle and correcting the posture, which was beginning to show muscular compensations.

The evaluation of the pediatric patient was carried out on 08/10/2022. The physiotherapeutic approach started on 08/12/2022. Three follow-up evaluations were then conducted on 09/12/2022, 10/17/2022, and 11/29/2022. When re-evaluated on 11/29/2022, the patient was discharged from physiotherapy. It was recommended to continue activities such as swimming and rhythmic gymnastics.

Figure 1. Timeline of results found

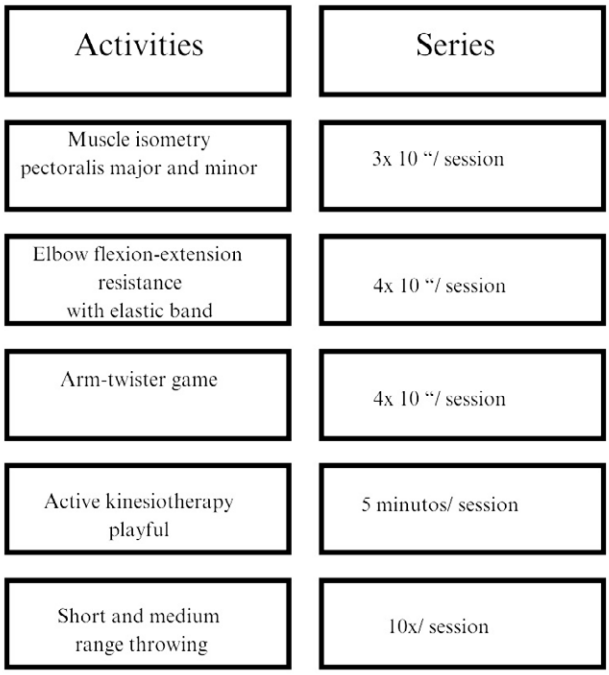
Source: the authors (2025).

7. Results

After evaluation, a therapeutic plan was outlined with two weekly sessions, each lasting 45 minutes.

Physiotherapeutic Intervention: The activities proposed in the therapeutic plan included playful isometric exercises for the pectoralis major and minor muscles, consisting of 3 sets of 10 seconds of squeezing a ball against the hands or pushing the ball against the wall, with the goal of strengthening the pectoral muscle portion and strengthening the rhomboids. Resistance exercises using an elastic band as a rope, with the child playing a tug-of-war to see who can pull the rope the most, were done four times per session, strengthening the flexors and extensors of the elbow as well as the rhomboids. Additionally, an arm wrestling game was included for active-resistant movements of internal shoulder rotators. The therapist resisted for 10 seconds and then allowed the child to overcome the resistance. This way, the child could gain strength and muscular endurance. This activity was done in four sets of 10 seconds. Active kinesiotherapy was also incorporated with playful activities such as: "clap your hands," "ABC," "Popeye," and "Babalu," each lasting 5 minutes, alternating with two games per session. Throwing activities at short and medium distances, such as hopscotch and basketball, were performed with the goal of working on active movement of the supraspinatus, deltoid, and rhomboids. As the child gained greater body awareness, shoulder circumduction exercises for the right shoulder with the aid of a hula hoop were introduced.

Figure 2. Descriptive Summary of the Interventions



Source: the authors (2025)

The child in the study showed improvement in the muscle strength of the stimulated muscles, as shown in the table below:

Table 2. Muscle Strength Presented and Its Evolution

Muscle	Deltoid	teres major	teres minor	Supra spinal	Infraspinatus	Subscapularis	Rhomboid	Pectoralis major	Pectoralis minor
Muscle Strength before approach	3	3	3	3	3	3	3	0	2
Muscle Strength after approach	4	4	4	4	4	4	4	0	3

Source: the authors (2025).

Concomitant with the improvement in muscle strength, the child showed better body awareness and, consequently, a reduction in muscle compensations and improved body alignment.

After achieving proper alignment, proprioceptive exercises were performed, such as pressing the ball against the wall and pressing the ball against the body. These exercises were important in helping the child adjust their body to its center of gravity.

This entire approach allowed the child to perform joint movements with a better range of motion and no pain in the anterior right shoulder region, graded as 0 on the numerical scale (0-10), thereby achieving the established goals.

8. Discussion

The main finding of this article is the relevance and effectiveness of an early, comprehensive physiotherapeutic approach in the functional treatment of Poland Syndrome (PS). Despite the lack of consensus on the use of physiotherapy for the functional management of the syndrome, this study demonstrates that this approach improves muscle strength in the shoulder complex muscles, enhances body awareness, minimizes muscle compensations, and promotes better biomechanical alignment. This makes it a viable alternative, even in the absence of surgical treatment. This is the first case report to highlight these benefits, with clinically relevant results.

Poland Syndrome (PS), also known as "Poland anomaly," was described by Alfred Poland in 1841. Patients affected by this syndrome can be identified at birth, and most cases are reported as sporadic.[11] Once diagnosed, it should be closely monitored, with particular attention to respiratory and muscular functions, especially in the upper limbs, as well as breast development.[10] The first-line examination for clinical diagnosis is ultrasound due to its easy access and better cost-effectiveness. In cases of suspected thoracic malformations, the PS consensus recommends chest X-rays and, in more severe cases, MRI or CT of the chest.[11]

PS is more common in males[11], in line with the findings of this case report, which involved a female patient. According to the same source[11], most cases are diagnosed in childhood.

The patient in this study showed decreased muscle strength in the right upper limb for internal and external shoulder rotation, as well as horizontal adduction and abduction of the shoulder of the same limb. This finding is consistent with the study by Schippers SM et al.[6], where 16 patients participated, and muscle strength tests were conducted comparing with the contralateral limb. All of these patients had a diagnosis of absent pectoral muscles.

According to the PS consensus, there are no specific surgical or medical therapies to treat PS or its main symptoms. However, the consensus recommends surgical treatment for major abnormalities and medical complications. In this consensus, there is no indication for physiotherapy treatment for the functional aspects of Poland Syndrome. However, this work demonstrates the importance and effectiveness of a comprehensive physiotherapeutic approach in treating Alfred Poland syndrome.

The PS consensus indicates physiotherapy treatment after surgical intervention. Surgical treatment is recommended when the patient has anomalies with functional deficits, such as syndactylia, severe thoracic malformations (pectus excavatum, carinatum, or both, rib agenesis), and breast reconstruction.

However, our physiotherapeutic approach focused on addressing the main muscle dysfunctions of the studied child, resulting in better body center of gravity alignment and improved biomechanical posture. This study shows the importance of an early, comprehensive physiotherapeutic approach, improving body awareness and minimizing muscle compensations.

Schippers SM et al.[6] highlighted that individuals with PS diagnoses have reduced capacity for work and sports, as well as greater difficulty playing musical instruments, compared to the general population. This contrasts with the findings of the present study, where the child was actively encouraged by her family to engage in sports activities, such as swimming and ballet, and also participated in music classes where she played the xylophone.

In the article by Schippers SM et al.[6], hand grip and pinch strength were assessed using a dynamometer. This was a limitation in our study, as we did not have access to this evaluation equipment.

There are several strengths in this study. It is the first case report of a physiotherapeutic approach in patients with Poland Syndrome, with clinically relevant results.

It will be important to extend the discussion and the acceptability of our recommendations to a broader community of physiotherapists, physicians, and families.

The report may directly contribute to patients or groups of individuals in similar situations by providing knowledge about a therapeutic approach that has not yet been reported in the literature.

In some sessions, it was not possible to complete the planned 45 minutes due to the child's fatigue from other daily activities. This unexpected issue required adjustments, such as reducing the time or adapting activities to respect the child's limits and ensure her well-being and adherence to the therapeutic process.

One of the main limitations of this study is that it is a case report involving only a single child with Poland Syndrome, which limits the ability to generalize the data to other individuals with the same condition. However, given the rarity of this syndrome, this study represents a relevant contribution, providing important information on a therapeutic approach that is still little explored, which could serve as a foundation for future studies and guide clinical practices.

9. Conclusion

This case study set out to demonstrate how the physiotherapeutic approach can contribute to motor development in a child diagnosed with Alfred Poland syndrome, who was diagnosed at the age of six.

The presence and encouragement of the family, especially the parents, helped the child to feel more secure and motivated during the sessions, facilitating engagement in the treatment. The integration of the treatment with the child's daily activities, such as playing, going to school, swimming or interacting with friends was made easier because the family actively collaborated, helping the child to apply what they learned in the sessions, with regard to improving the strength of the muscle groups that involve the shoulder girdle, as well as better postural alignment and improved body awareness.

More studies are needed, including with a larger population, in order to confirm or not the effectiveness of the approaches described in this study.

Authors' contributions

The authors declare that they have made substantial contributions to the work in terms of the conception or design of the research; the acquisition, analysis or interpretation of data for the work; and the writing or critical review of relevant intellectual content. All authors have approved the final version to be published and have agreed to take public responsibility for all aspects of the study.

Conflicts of interest

No financial, legal or political conflicts involving third parties (government, companies and private foundations, etc.) have been declared for any aspect of the submitted work (including but not limited to grants and funding, participation in advisory boards, study design, manuscript preparation, statistical analysis, etc.).

Indexers

The Journal of Physiotherapy Research is indexed by DOAJ, EBSCO, LILACS and Scopus.

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