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Beyond a Cosmetic Deformity: Poland Syndrome Presenting with Significant Restrictive Lung Disease - A Case Report

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ABSTRACT

Poland syndrome is a rare congenital anomaly characterised by unilateral absence or hypoplasia of the pectoralis major muscle with variable involvement of the chest wall and adjacent structures. It is typically considered a cosmetic deformity, and functional impairment is often under-recognised. We report a 17-year-old male who presented with long-standing asymmetry of the right chest wall, noted since early childhood. Clinical examination revealed flattening of the right hemithorax with preserved nipple–areolar complex and absence of skeletal anomalies. Pulmonary function testing demonstrated a severe restrictive ventilatory defect, with reduced forced vital capacity and preserved FEV1/FVC ratio. High-resolution computed tomography confirmed non-visualisation of the right pectoralis major and minor muscles, along with associated parenchymal changes. Cardiac evaluation was unremarkable. The patient was managed conservatively with incentive spirometry and chest physiotherapy and is under regular follow-up.

This case highlights that clinically significant pulmonary involvement, although uncommon, can occur in Poland syndrome even in the absence of major skeletal deformities. It underscores the importance of early functional assessment and longitudinal monitoring to identify subclinical respiratory compromise. Recognition of such presentations is essential to guide timely intervention and optimise long-term outcomes.

Keywords: Poland syndrome; Restrictive Lung Disease; Pectoralis Major; Agenesis.

INTRODUCTION

Poland syndrome is a rare congenital anomaly characterised by unilateral absence or hypoplasia of the pectoralis major muscle, frequently associated with variable abnormalities of the chest wall, breast, and ipsilateral upper limb[1]. First described by Alfred Poland in 1841, the condition demonstrates marked phenotypic heterogeneity, ranging from isolated muscular defects to more complex musculoskeletal involvement[2]. It occurs sporadically in most cases, with a reported male predominance and preferential involvement of the right hemithorax[2].

Pathogenesis is not fully elucidated, although the most widely accepted hypothesis implicates disruption of blood flow in the subclavian artery territory during early embryogenesis, leading to defective development of the pectoral region and adjacent structures. Owing to its variable presentation, diagnosis is primarily clinical,

with imaging used when required to delineate the extent of involvement[3].

Despite increasing recognition of the morphological spectrum of Poland syndrome, the functional implications of the condition remain insufficiently characterised, particularly in patients without overt skeletal anomalies. This gap in understanding may contribute to under-recognition of clinically relevant manifestations in routine practice.

In this report, we describe an adolescent with Poland syndrome presenting with significant functional involvement, highlighting the need for a more comprehensive clinical evaluation beyond structural assessment.

CASE REPORT

A 17-year-old male presented with a deformity of the right side of the chest, first noticed at 4 years of age by the patient and his parents. The deformity was non-progressive and not associated with pain, nipple discharge, retraction, or a history of trauma. Birth and developmental history were unremarkable. There was no history suggestive of respiratory distress in early childhood. He was the second child of a non-consanguineous marriage, with no similar complaints in family members.

At the presentation, he was studying in class 11, was interactive with peers, and had good scholastic performance. Anthropometric assessment revealed a weight of 31 kg (<3rd centile), a height of 152 cm (3rd–10th centile), and a body mass index of 13.4 kg/m², consistent with severe thinness.

On general examination, there were no facial dysmorphic features or hair abnormalities. Local examination of the chest revealed flattening and relative hollowness over the right anterior chest wall, with preservation of the nipple–areolar complex. Examination of the back demonstrated kyphosis **(Figure 1)**. Cardiovascular examination was unremarkable, with normal heart sounds and no murmurs. Respiratory examination revealed reduced expansion of the right hemithorax, with preserved vesicular breath sounds. Neurological and abdominal examinations were within normal limits.

Two-dimensional echocardiography showed situs solitus with levocardia, preserved left ventricular systolic function, and grade II diastolic dysfunction. Right ventricular function was normal, with no chamber dilatation or hypertrophy. Pulmonary function testing demonstrated a normal FEV1/FVC ratio (99.9%) with a reduced forced vital capacity (1.47 L), suggestive of a severe restrictive ventilatory defect. High-resolution computed tomography (HRCT) of the thorax revealed fibro-atelectatic bands in the right upper lobe and mosaic attenuation in the right middle and lower lobes. Notably, the right pectoralis major and minor muscles were not visualised, confirming the diagnosis of Poland syndrome **(Figure 2)**.

The patient was initiated on incentive spirometry and chest physiotherapy. Regular follow-up for pulmonary and cardiac function has been planned. Surgical reconstruction options were discussed; however, the patient and family opted for conservative management at present.

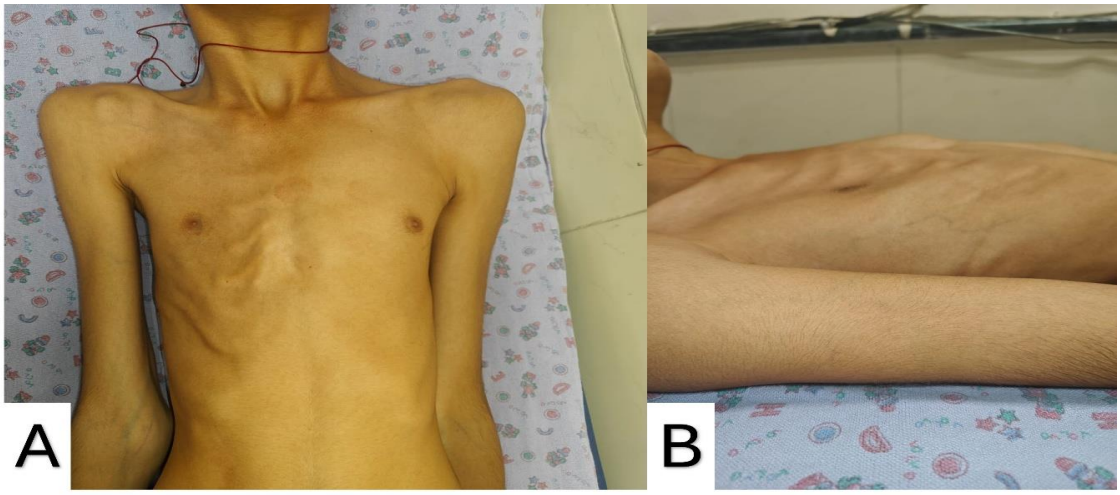

Figure 1. Clinical photographs demonstrating chest wall asymmetry.
(A) Frontal view showing flattening and relative hollowing of the right anterior chest wall with preserved nipple–areolar complex.
(B) Lateral view highlighting depression of the right hemithorax with reduced anteroposterior chest wall contour. Findings are consistent with the absence of underlying pectoral musculature in Poland syndrome.

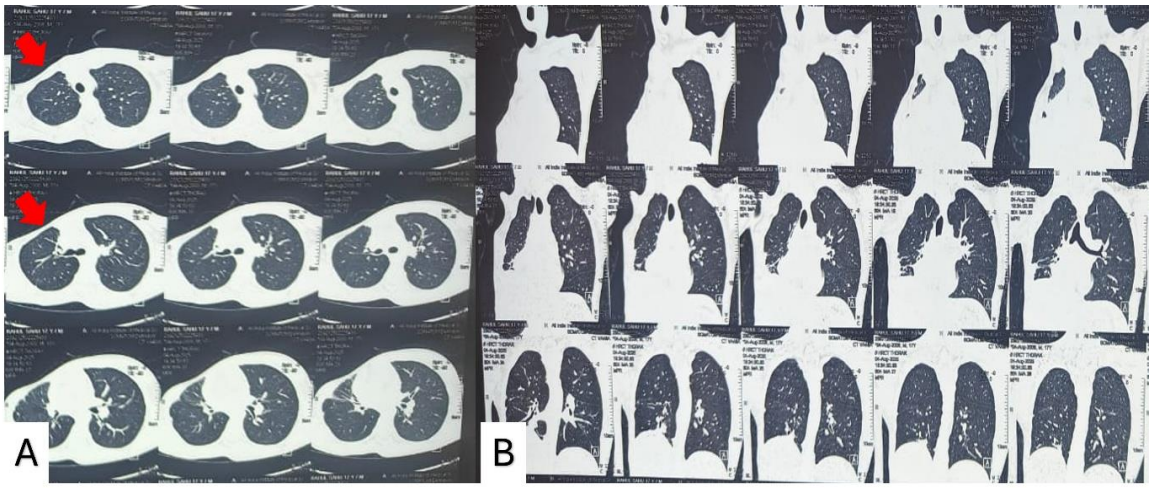

Figure 2. High-resolution computed tomography (HRCT) of the thorax.
(A) Axial lung window images demonstrating non-visualisation of the right pectoralis major and minor muscles (red arrows) with associated subtle parenchymal changes.
(B) Coronal reconstructions showing reduced lung volume on the right side with fibro-atelectatic changes, consistent with restrictive lung involvement secondary to chest wall deformity.

DISCUSSION

Poland syndrome is a rare congenital anomaly characterised by unilateral agenesis or hypoplasia of the pectoralis major muscle with variable involvement of the chest wall, upper limb, and adjacent structures [1,4]. Although the condition is typically described as a cosmetic deformity, its clinical spectrum is highly variable, and functional impairment may occur in selected cases.

The most widely accepted etiological mechanism is the subclavian artery supply disruption sequence, wherein a vascular insult during early embryogenesis leads to impaired development of the pectoral region and adjacent tissues[2]. While musculoskeletal manifestations are well recognised, respiratory involvement remains uncommon and is only sparsely documented in the literature.

Pulmonary complications in Poland syndrome are primarily related to chest wall hypoplasia and altered thoracic mechanics, resulting in restrictive ventilatory defects and reduced lung volumes. Only a limited

number of reports have described patients presenting with respiratory symptoms such as dyspnoea, chronic cough, or recurrent infections, often associated with radiological evidence of volume loss on the affected side[3,5]. In rare instances, thoracic deformity may contribute to hypoventilation or sleep-related breathing disorders[6]. However, most individuals remain asymptomatic or have minimal functional limitations.

In this context, our case is notable for demonstrating significant restrictive lung disease in the absence of major associated skeletal anomalies, such as Sprengel deformity or rib agenesis. This highlights that even an isolated absence of pectoral muscle can result in clinically meaningful pulmonary compromise. The preserved FEV1/FVC ratio, despite reduced forced vital capacity, in our patient supports a restrictive pattern attributable to impaired chest wall expansion rather than intrinsic pulmonary pathology.

These findings emphasise the need for heightened clinical awareness and early respiratory evaluation, even in patients who appear to have predominantly cosmetic deformity. Baseline and follow-up pulmonary function testing can help detect subclinical impairment. Early initiation of chest physiotherapy and lung expansion strategies, including incentive spirometry, may play a key role in optimising respiratory function and preventing progression.

CONCLUSION

Poland syndrome is typically considered a cosmetic chest wall anomaly; however, this case highlights that clinically significant pulmonary involvement, although rare, can occur even in the absence of major skeletal abnormalities. Early recognition and routine respiratory assessment are essential to identify restrictive lung disease. Timely institution of physiotherapy and lung expansion measures, along with longitudinal monitoring, may improve functional outcomes and prevent complications. This case underscores the importance of viewing Poland syndrome as a condition with potential functional implications rather than purely as an aesthetic concern.

Declaration by Authors

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

REFERENCES

1. Bansal A, Reddy K, Dinsmore K, Gonzalez-Estrada A. Poland syndrome: a case report. BMJ Case Rep. 2017;2017: bcr2017221564. doi:10.1136/bcr-2017-221564
2. Chait F, Bahlouli N, Chehrastane R, et al. Clinical and Radiological Features in Poland Syndrome: Report of 3 Cases and Review of Literature. Glob Pediatr Health. 2023; 10:2333794X231219388. doi:10.1177/2333794X231219388
3. Baldelli I, Baccarani A, Barone C, et al. Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet J Rare Dis. 2020;15(1):201. doi:10.1186/s13023-020-01481-x
4. Agarwal S, Arya A. Poland's syndrome. Indian J Dermatol Venereol Leprol. 2008;74(1):62-63. doi:10.4103/0378-6323.38415
5. Bhatia M, Balasubramanian G, Kumar V, Rajvansh P. Left-sided Poland Syndrome Presenting as Chronic Respiratory Symptoms without Hand Deformity: A Rare Case Report. Indian Journal of Respiratory Care. 2024;12(4):352-354. doi:10.5005/jp-journals-11010-1069
6. Sugiyama AI, Shiota S, Yamada T, Ito J, Suzuki K, Takahashi K. Sleep-related breathing disorders associated with the characteristics of underlying congenital rare diseases of Moebius syndrome and Poland syndrome. Respirol Case Rep. 2020;8(5): e00579. doi:10.1002/rcr2.579

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