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AN Infant with Poland Syndrome and Coexisting Beta Thalassemia Trait: A Case Report

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Abstract
Poland syndrome is a rare congenital anomaly characterized by unilateral chest wall hypoplasia with ipsilateral upper limb malformations. It has incidence of 1 in 36,000-50,000 live births with a male preponderance. Its presentation is highly variable, and it is rare to find all the features of this syndrome in a single patient. Poland syndrome is mostly unilateral, classically affecting the right side (60-75%). There have been no previous reports of co-existence with haemoglobinopathies. We present a case of an infant with Poland syndrome and co-existing beta thalassemia, which happens to be the first documented case of Poland syndrome in an infant in Ghana. It is also the first reported case of Poland syndrome with co-existing thalassemia, to the best of our knowledge.

Keywords: Poland Syndrome, Chest Wall Deformity, Limb Anomalies, Infant Thalassemia, Case Report

Key Clinical Message
Poland syndrome is a rare congenital anomaly with variable presentation. We report the first documented case of its coexistence with beta thalassemia. While this association may be coincidental, it highlights the need for thorough evaluation in congenital anomalies and consideration of hematologic disorders in affected patients.

Introduction
Poland syndrome is a rare congenital anomaly characterized by unilateral absence of the pectoralis major muscle and a wide spectrum of associated ipsilateral chest wall and upper extremity anomalies [1]. It is sporadic with variable presentations [2,3]. Depending on the degree of penetrance, the chest wall defect ranges from complete pectoralis muscle absence with severe rib abnormalities to partial absence of the sternocostal head of the pectoralis muscle [2,4]. In some cases, the latissimus dorsi, external oblique, and serratus anterior muscles are affected. The breast tissue is usually hypoplastic on the affected side. The nipple and areola may be completely absent or hypoplastic.1, 2The upper limb malformation may vary from mild syndactyly to severe brachysyndactyly [4,5]. It affects 1 in 30,000-80,000 live births with a male preponderance [2,6]. It is mostly unilateral, classically affecting the right side, although bilateral cases exist [2,5].

The exact etiology of Poland syndrome is unknown, although it is thought to result from disruption in the development of the subclavian artery during the 6th week of gestation [1,2] This 'subclavian artery supply disruption sequence' explains the wide phenotypic variations in Poland Syndrome and some associated anomalies such as M¨obius and Klippel-Feil syndromes [1,2]. Other etiological theories include prenatal exposure to noxious environments such as maternal smoking, cocaine, viral infections, and attempted abortion with misoprostol [1,2,7-9].

Besides chest wall and upper limb abnormalities, there may be cardiac, gastrointestinal, neurologic, pulmonary, renal, and dermatologic manifestations [1-3,10]. Also, cases of Poland syndrome with leukemia, non- Hodgkin's lymphoma, carcinoma of the hypoplastic breast, leiomyosarcomas, lung, and cervical carcinomas suggest a relationship with neoplasms [1]. However, no previous report of coexistence with hemoglobinopathies is documented, to the best of our knowledge. We present the first case of Poland syndrome with beta thalassemia trait in an infant.

Case History and Examination

Patient Information: A 6-month-old male presented to our pediatric clinic in 2019 with a flattened right anterior upper chest wall and shortness of the fingers of the ipsilateral hand, noticed at birth. He was born at term to a 28-year-old mother and a 30-year-old father. The pregnancy was uneventful. His mother is a non-smoker and did not use over-the-counter or illicit drugs during pregnancy. He is the second child from a non-consanguineous marriage. There are no congenital malformations in the immediate family.

Clinical Findings: On examination was a healthy-looking male infant with anthropometric measures within normal limits for age and sex. The superior third of the right anterior chest wall was depressed and moved paradoxically with respiration. The right breast was hypoplastic with a rudimentary nipple. The underlying subcutaneous tissue was atrophic with no right anterior axillary fold as shown in Figure 1. The ipsilateral scapular and latissimus dorsi were normal. All ribs were palpable, but those on the right were unusually crowded.

Examination of his right-hand revealed syndactyly and shortening of the 2nd to 5th fingers as shown in Figure

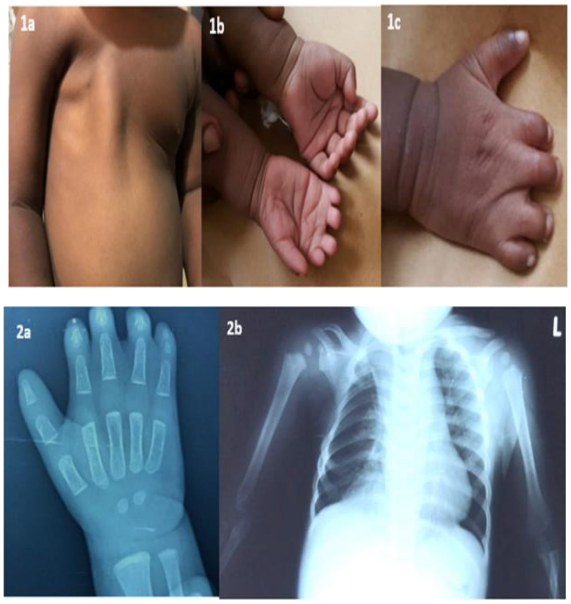

Figure 1: Anterior Chest and Right-Hand Anomalies Seen in the Patient

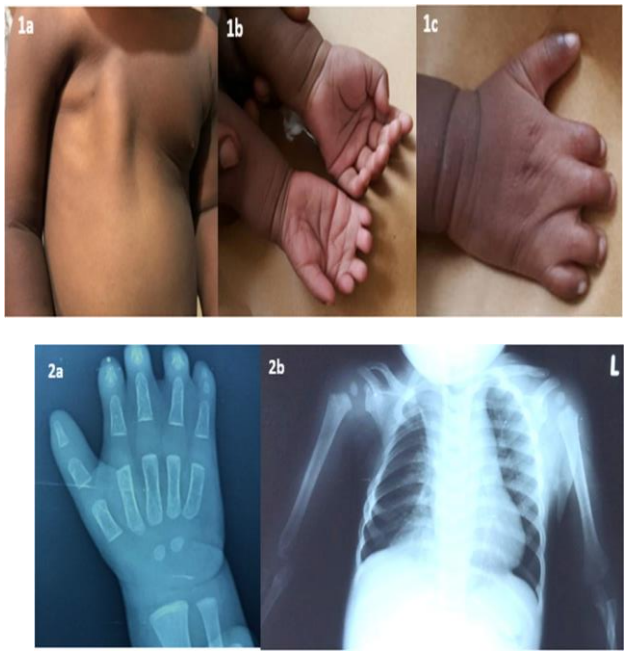

Figure 2: X-Rays of the Right Hand and the Chest

1. His arm span was 58 cm, with the right side contributing 26 cm and the left 32 cm (measured from the mid-point of the sternum). The rest of the right upper limb was normal. The contralateral hemithorax, left upper limb, lower limbs, and vertebrae were normal.

He had no unusual craniofacial findings. His air entry was reduced in the right lower zone. His hearing, vision, and all other systemic examinations were normal.

Differential Diagnosis, Investigations, and Treatment

An x-ray of his right hand showed soft tissue fusion of the 2nd to 5th digits with normal bony architecture and mineralization. His chest x-ray showed hyperlucent right lung fields with crowding of the right ribs. There were areas of atelectasis in the right lower zone. The left lung field was clear with normal ribs. The cardiac shadow and both hemidiaphragms were normal as shown in Figure 2. He had normal echocardiogram and abdominal ultrasound scans.

His complete blood count revealed mild anemia with microcytic hypochromic red cells (HB 11.0 g/dL, MCV 61.9 fL, MCH 22.7 pg) and increased red cell distribution width (RDW-CV 19.0%). RBC count was 4.85 x 103/µL with Mentzer's index of 12.7, indicating a possible thalassemia trait. His WBC and platelet counts were normal.

A peripheral blood film showed moderate microcytic hypochromic RBCs, increased target cells, and a few platelet clumps. An impression of hemoglobinopathy was made, but the sickling test was negative. A high-performance liquid chromatography (HPLC) showed low hemoglobin A and high hemoglobin A2 concentrations, suggesting beta thalassemia minor as shown in Table 1.

	PATIENT	**MOTHER**	FATHER
Hb A	49.0% (L)	64.0% (L)	97.0%
Hb A2	43.6% (H)	35.5% (H)	3.0%
Hb F	6.4%	0.5%	0%

Table 1: HPLC Hemoglobin Analysis of the Patient, his Mother, and the Father Showing a High Hemoglobin A2 and Low Hemoglobin A for Both the Patient and Mother, Indicative of Beta Thalassemia Minor. That of the Father is Normal.

Sickling tests for both parents were negative. The father's HB electrophoresis revealed AA genotype. HPLC of the mother detected 35.5% hemoglobin A2, indicating heterozygous beta thalassemia trait in the mother as shown in Table 1.

Outcome and Follow-Up

Ηε ωας διαγνοσεδ ωιτη Πολανδ σψνδρομε ωιτη ϛοεξιστινγ β-τηαλασσεμια τραιτ, ενρολ- λεδ ιν τηε πεδιατρις ηεματολογψ ϛλινις, ανδ ρεφερρεδ το πλαστις συργερψ φορ φυρτηερ μαναγεμεντ.

Discussion

Poland syndrome is a rare congenital anomaly with several suggested etiologies. Our patient was born from a non-consanguineous marriage and is the first in his family to have this syndrome, suggesting a sporadic occurrence. Autosomal dominant mutations are typically associated with increased parental (especially paternal) age, but our patient has young parents [11]. The pregnancy was uneventful with no history of attempted abortions, alcohol, or illicit drug use.

The diagnosis of Poland syndrome may be made at birth based on the presence of the classical physical findings leading to timely therapeutic interventions to improve the quality of life of patients and their families [12]. Unfortunately, this does not occur in some cases, possibly due to the lack of homogeneity of the diagnostic work-up. In an Italian survey, the average age at diagnosis was around 14 years; only 31.58% of cases were diagnosed at birth even though the classical symptoms of the syndrome were present at an early age [12]. In our case, though the deformities were noticed at birth, the child was not appropriately referred, delaying the diagnosis till age 6 months old.

Poland syndrome is mostly unilateral, with the majority being right-sided, though left-sided and bilateral cases have been reported [1,2,13-15]. Our patient had right chest wall hypoplasia and ipsilateral symbrachydactyly in keeping with the classic presentation of Poland syndrome. Ideally, a cross-sectional CT scan or MRI of the chest wall will clearly show the unilateral absence of the pectoralis major and minor muscles. Unfortunately, this was not done in our case, and we state it as a limitation even though clinical examination sufficiently demonstrates Poland syndrome in this case.

Paradoxical motion of the chest wall is usually seen in patients with lung herniation or pulmonary atelectasis [3]. Anteroposterior chest x-ray in individuals with Poland syndrome typically demonstrates a hyperlucent lung on the ipsilateral side [6]. There may also be mediastinal shift towards the affected side. Our patient had paradoxical chest wall motion with no lung herniation but some atelectasis in the right lower lung zone. There was, however, no functional cardiopulmonary impairment.

Cases of Poland syndrome with leukemia and other carcinomas suggest a relationship between developmental anomalies and neoplasms [1]. A case of Poland syndrome and bleeding diathesis has been reported, suggesting a possible link with hematological disorders.

Beta thalassemia has a wide phenotypic variability due to the over 200 beta-globin gene mutations identified [16]. It has a high prevalence in West Africa and other malaria-endemic areas, as explained by the Haldane's malaria hypothesis [4]. Its prevalence in northern Ghana is reported to be 5% [10]. Due to this high prevalence, the association with Poland syndrome in our patient may just be a coincidence. It is, however, worth reporting this as the first case of Poland syndrome with coexisting β-thalassemia, although no experimental methods were used to elucidate such a link.

Conclusion

We have presented a rare case of Poland syndrome with coexisting beta thalassemia trait, the coexistence of which may be a coincidence owing to the high prevalence of hemoglobinopathies in our part of the world. Further research is required to elucidate the link between hemoglobinopathies and this syndrome [17].

Authors' Contributions

SKB and AOJ conceptualized the study, wrote the original draft, completed the literature search, and summarized the case. PAO contributed to the original draft. PAO and VP provided critical comments on the hematological aspect of the case report. OKO reviewed and provided critical advice on all radiological investigations. CKH was involved in the study conception, supervised the case summary and literature review, and provided critical revision to the manuscript. SKF was involved in the review, critical revision, and submission of the manuscript. All authors have read and agreed to the final version of this manuscript.

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Declaration of Conflicting Interests

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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Informed Consent

Written informed consent was obtained from the patient's parents for their anonymized information to be published in this article.

Data Availability Statement

The data that support the findings of this case series are available from the corresponding author upon reasonable request. Due to patient confidentiality and privacy concerns, some restrictions may apply to the availability of certain data points. Requests for access to the data should be directed.

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