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Recognizing Poland syndrome at birth: Chest wall and hand manifestations

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Abstract

Background: Poland syndrome is a rare congenital anomaly defined by unilateral agenesis or hypoplasia of the sternocostal head of the pectoralis major, with possible involvement of the pectoralis minor and variable ipsilateral chest wall and upper-limb abnormalities, including syndactyly. The presentation is typically unilateral, more often affecting the right hemithorax, while bilateral involvement is uncommon.

Purpose: To report a neonatal case of Poland syndrome and describe the clinical manifestations and early management to support timely intervention and appropriate care planning.

Method: A case study was conducted at Dumai Hospital involving a preterm neonate delivered to a 27-year-old woman (G2P1A0) at 27 weeks of gestation who presented with uterine contractions and stable vital signs. Prenatal ultrasonography was performed prior to delivery. After birth, the neonate underwent a structured physical examination focusing on the chest wall and upper extremities to identify congenital anomalies, followed by initial stabilization and referral based on clinical findings.

Results: The patient was a female preterm neonate with findings consistent with Poland syndrome, including unilateral absence of the pectoralis major and pectoralis minor on the affected side and ipsilateral hand syndactyly. The infant was referred to the perinatology unit for ongoing monitoring and multidisciplinary management, while the mother received routine postpartum care according to hospital protocol.

Conclusion: Poland syndrome may be recognizable at birth through unilateral thoracic muscle absence with ipsilateral limb anomalies, requiring careful newborn examination and early coordinated care.

Keywords: Newborn; Pectoralis Major Agenesis; Pectoralis Minor Agenesis; Poland Syndrome; Syndactyly.

INTRODUCTION

Poland syndrome traces back to Alfred Poland's 1841 report describing a unilateral absence of the pectoralis major in an adult male, which became the historical cornerstone for recognizing a distinct congenital pattern of anomalies (Poland, 1841). In modern clinical usage, the condition is defined primarily by agenesis or hypoplasia of the sternocostal head of the pectoralis major, with a variable set of associated ipsilateral thoracic and upper-limb findings that justify viewing it as a spectrum rather than a single defect (Tafti & Cecava, 2023). Consensus-based guidance further emphasizes structured phenotyping because the

"core" pectoral defect can coexist with diverse chest wall, rib, soft-tissue, and limb manifestations that differ markedly between individuals (Baldelli, Baccarani, Barone, Bedeschi, Bianca, Calabrese, Castori, Catena, Corain, Costanzo, De Paoli Barbato, De Stefano, Divizia, Feletti, Formica, Lando, Lerone, Lorenzetti, Martinoli, Mellini, Nava, Porcellini, Puliti, Romanini, Rondoni, Santi, Sartini, Senes, Spada, Tarani, Valle, Venturino, Zaottini, Torre, & Crimi, 2020).

Epidemiologically, Poland syndrome is consistently categorized as rare, with commonly cited estimates around 1-9 per 100.000 live births

and an incidence near 1 per 30.000 live births, depending on ascertainment and diagnostic thresholds (Singh & Gupta, 2022). These numbers likely underestimate the true burden because mild phenotypes especially those limited to subtle pectoral hypoplasia may be missed in routine newborn assessment and only recognized later when asymmetry becomes more apparent (Hashim, Quek, & Chandran, 2021). Most cases are described as sporadic, and clinical summaries repeatedly note a predominance of involvement of the right hemithorax, which shapes expectations during early-life screening and documentation (Tafti & Cecava, 2023).

A newborn-focused approach matters because the earliest clinical “signals” can be easy to overlook unless the examiner is actively looking for asymmetry of the anterior chest wall and ipsilateral soft-tissue deficiency (Baldelli et al., 2020). In neonates, the manifestations most likely to be observable include unilateral flattening or depression of the chest, absence or hypoplasia of the pectoral contour, rib or chest wall anomalies, and variable hypoplasia of the nipple-breast complex, although the latter may become more obvious with growth (Tafti & Cecava, 2023). Clinical reports that pair physical examination with imaging highlight how radiologic evaluation can clarify thoracic anatomy and better delineate the extent of muscle and rib involvement when findings are subtle at birth (Chait, Bahloul, Chehrastane, Mrani Alaoui, Boughaleb, Zouheir, Tarik, Allali, El Haddad, & Chat, 2023). This variability at the bedside is exactly why “clinical manifestations in newborns” cannot be reduced to one hallmark sign, even though pectoralis major involvement remains central to the diagnosis (Agranovich, Komolkin, & Dimitrieva, 2017).

Upper-limb involvement further broadens the neonatal presentation, ranging from no apparent limb anomaly to classic hand findings such as syndactyly, brachydactyly, or symbrachydactyly on the affected side (Agranovich et al., 2017). Because hand differences are often more visible than mild chest asymmetry in the nursery, classification systems have been proposed to standardize description and avoid inconsistent labeling across reports and clinical settings (Al-Qattan, 2001). Additional proposals focusing on hand and upper-limb patterns aim to improve comparability between cases, which is

particularly useful when the thoracic phenotype is mild or when limb findings are atypical (Catena, Divizia, Calevo, Baban, Torre, Ravazzolo, Lerone, & S  n  s, 2012). From a newborn perspective, this structured reporting helps reduce diagnostic drift where some infants are recognized only because of hand anomalies, while others with “thorax-only” findings remain uncoun  ted (Baldelli et al., 2020).

Case reporting remains urgent because the literature is intrinsically skewed: severe neonatal presentations draw immediate attention, while milder forms are more likely to be missed, reinforcing underdiagnosis and distorting apparent prevalence and phenotype frequencies (Hashim et al., 2021). Rare but clinically important neonatal presentations such as lung herniation at birth illustrate how thoracic involvement can occasionally have acute consequences, making early recognition more than a cosmetic concern in selected cases (Chandran, Revanna, Ari, & Rana, 2013). Uncommon neonatal variants, including presentations with axillary pterygium, also expand the clinical spectrum and underscore why “textbook” expectations do not capture every newborn presentation (Dos Santos Mendo, Almeida, Maria, & Tuna, 2021). Although vascular disruption theories are often cited to explain the unilateral pattern and associated defects, their main relevance to a newborn-oriented paper is practical: they reinforce the expectation of segmental, ipsilateral findings that should be actively sought and carefully documented at birth. (Bavinck, Weaver, Opitz, & Reynolds, 1986) Within this context, the present case report of a female newborn at RSUD Dumai is positioned to add to national and international evidence and to raise clinical vigilance so that subtle newborn cases especially those without obvious limb malformations are less likely to be overlooked.

RESEARCH METHOD

The *descriptive case study* design to document and interpret the clinical manifestations of Poland syndrome in a newborn through systematic clinical observation and detailed case recording (Yin, 2018; Creswell & Creswell, 2017). The participant was a *premature* female infant born to a 27-year-old mother with an obstetric status of G2P1A0 at 27 weeks of gestation. The study took place at Dumai Hospital, covering the labor unit and the *perinatology*

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department, where both maternal assessment and neonatal care were provided in accordance with routine clinical pathways.

The procedure began when the mother presented with uterine contractions; initial maternal evaluation included a vital-sign assessment, which remained within normal limits. Prior to delivery, fetal assessment was performed using *ultrasonography* to evaluate fetal condition and screen for congenital abnormalities that could be identified antenatally. After birth, the infant underwent a comprehensive physical examination by the clinical team, with specific attention to the chest wall and upper extremities to identify congenital anomalies consistent with Poland syndrome. Clinical documentation consisted of structured recording of physical findings, including assessment of the pectoralis major and pectoralis minor (presence, absence, or hypoplasia) and careful examination of the hands to detect syndactyly (Yin, 2018). Clinical

photographs were obtained in line with hospital procedures to support accurate case documentation and reporting (Yin, 2018).

Following the initial assessment, the infant was referred to the *perinatology* department for further management and ongoing observation, where routine monitoring included evaluation of respiratory function, physical growth, and clinical stability appropriate to prematurity (Patton & Cochran, 2002). Data collected included maternal characteristics (age, parity, pregnancy status), the infant's condition at birth, and the results of congenital anomaly assessments, without a *control group* because the study was *observational* and descriptive in intent. Uncontrollable factors primarily involved the infant's prematurity and the influence of standard postnatal medical care, therefore the analysis emphasized careful description of phenotypic findings and their clinical implications for early care planning (Yin, 2018; Patton & Cochran, 2002).

RESEARCH RESULTS

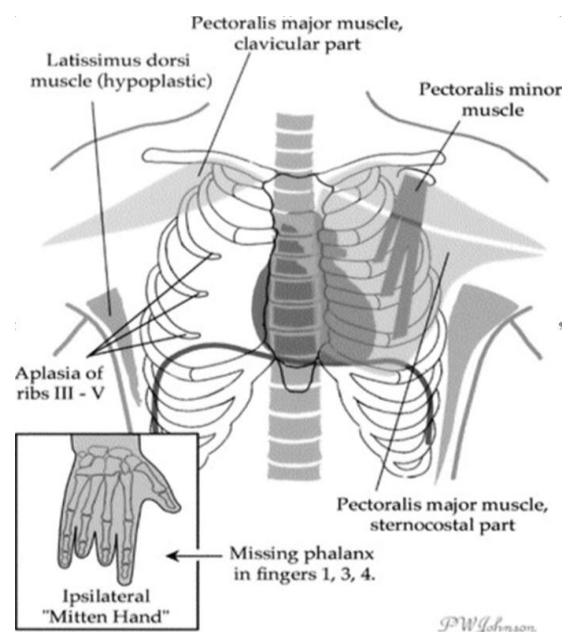


Figure 1. Manifestations of Poland Syndrome

Figure 1 shows the main clinical features of Poland syndrome in a neonate, characterized by chest wall asymmetry due to absence of the pectoralis major and pectoralis minor on the affected side. These findings are accompanied by limb anomalies, including syndactyly/hypoplastic fingers and a lower-limb abnormality, supporting a severe phenotypic spectrum.

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Figure 2. Chest Wall and Lower Extremity Abnormalities

Figure 2 highlights chest wall asymmetry and soft-tissue deficiency of the affected hemithorax, consistent with absence of the pectoralis major–minor complex. A lower-extremity anomaly is also visible, namely an incompletely formed ankle, presented as an associated finding.



Figure 3. Finger Abnormalities

Figure 3 shows on digital anomalies, showing syndactyly with hypoplasia resulting in incompletely formed fingers. This ipsilateral upper-limb finding is commonly reported in Poland syndrome and supports the clinical diagnosis.

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Table 1. Maternal and antenatal assessment

Variable	Results
Maternal and pregnancy history	
Maternal age	27 years
Gravida / Parity	G2P1A0
Gestational age at admission	27 weeks
Chief complaint	Uterine contractions for 5 hours
Bloody show	Present
Premature rupture of membranes	No
Heavy bleeding	No
Antenatal care and supplementation	
Antenatal care (ANC)	Never attended ANC
Pregnancy supplementation	Never received folic acid and ferrous sulfate
Prenatal ultrasonography findings	
Fetal presentation	Cephalic
Estimated fetal weight (EFW)	1200 grams
Fetal heart rate (FHR)	145 bpm

Table 1 summarizes maternal characteristics and the antenatal assessment prior to delivery. The mother was 27 years old (G2P1A0) and presented at 27 weeks' gestation with uterine contractions; prenatal ultrasonography documented a cephalic presentation, fetal heart rate of 145 bpm, and an estimated fetal weight of 1200 g. The table also highlights the absence of antenatal care and supplementation history, which is relevant to contextualizing overall pregnancy monitoring.

Table 2. Neonatal findings, early management, and outcome

Variable	Results
Neonatal characteristics at birth	
Sex	Female
Birth weight	1100 grams
Apgar score (1 min / 5 min)	3 / 4
General condition	Premature, respiratory depression
Clinical manifestations	
Chest wall	Chest asymmetry; absence of pectoralis major and pectoralis minor on the affected side
Upper extremities	Incomplete fingers (syndactyly, hypoplasia)
Lower extremities	Ankle not formed
Overall impression	Findings consistent with severe Poland syndrome
Care pathway and outcome	
Place of care	Perinatology department
Interventions	Initial stabilization; intensive observation; premature neonatal care
Survival duration	2 days
Outcome	Deceased

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Table 2 highlights the newborn's key clinical findings consistent with Poland syndrome, notably unilateral absence of the pectoralis major and minor with chest wall asymmetry and associated limb anomalies, including syndactyly/hypoplastic fingers and lower-extremity abnormality. It also outlines the immediate care pathway through referral to the perinatology unit for stabilization and monitoring. The early clinical course and outcome are included to provide a complete picture of management needs and short-term prognosis in a preterm neonate.

DISCUSSION

The present newborn showed a pattern that fits Poland syndrome, with unilateral absence of the pectoralis major–minor complex and ipsilateral limb anomalies documented in Figure 1 and summarized in Table 2. Chest wall asymmetry and soft-tissue deficiency are clearly illustrated in Figure 2, while Figure 3 focuses on syndactyly with digital hypoplasia, supporting a severe end of the phenotypic spectrum in this neonate. Although Poland originally described unilateral pectoral muscle deficiency, current developmental interpretations place the key disruption during the 6th–7th gestational weeks, when thoracic wall and limb structures are highly sensitive to early patterning errors (Poland, 1841; Fokin & Robicsek, 2002). In this case, the maternal and antenatal profile in Table 1 provides essential context for pregnancy monitoring, while Table 2 consolidates the newborn's phenotype, early stabilization, and short-term outcome.

A leading explanatory framework for the combined thoracic and limb findings is the subclavian artery supply disruption sequence, in which reduced flow to the developing subclavian system can affect both chest wall tissues and the ipsilateral upper limb in a single event (Bavinck et al., 1986). In practical terms, this model links the “one-sided” nature of the syndrome to a mechanism of early regional hypoperfusion rather than separate defects arising independently (Robb, Bhalla, & Raptis, 2022). Physiologic observations comparing upper-limb vascularization in affected individuals have also been used to support a vascular contribution, even if individual cases remain difficult to prove mechanistically (Bouvet, Leveque, Bernetieres, & Gros, 1978). For the present newborn, the co-occurrence of chest muscle absence and hand

anomalies seen in Figures 1-3 is consistent with a single early insult affecting adjacent developmental fields.

Environmental and teratogenic factors are often discussed because several exposures have been proposed to disturb placental or fetal perfusion, including maternal smoking and cocaine exposure, while misoprostol exposure has been explored epidemiologically in relation to congenital anomaly patterns (Ibrahim, Ramatu, & Helen, 2013; Orioli & Castilla, 2000). This case did not document such exposures, so the discussion is best limited to plausibility rather than attribution (Ibrahim et al., 2013). What is clearly documented is the absence of antenatal care and supplementation (including folic acid) in Table 1, which matters because periconceptional folate is a well-established preventive factor for neural tube defects and a marker of broader nutritional risk mitigation in pregnancy (Czeizel & Dudas, 1992). While folate deficiency is not a proven cause of Poland syndrome, inadequate supplementation and limited prenatal follow-up reduce opportunities for counseling, early risk identification, and targeted fetal assessment when anomalies are suspected (Krasovec & Anderson, 1991).

Epidemiologically, Poland syndrome is rare and more frequently reported in males and on the right hemithorax, which can bias clinician expectations during newborn assessment (Singh & Gupta, 2022). The present patient was female, reinforcing that the diagnosis should remain on the differential regardless of sex and that female presentations are well documented across broader case series (Lin, Ang, Xiao-jun, & Zhi-fei, 2012). Phenotypic variability ranges from minimal “thorax-only” involvement to more extensive defects across multiple structures, and this case combining thoracic muscle absence with digital anomalies and a lower-limb abnormality fits a severe, multi-structure presentation recorded in Table 2 and visualized in Figure 1. Classification approaches for limb findings help standardize reporting and improve comparability across cases, particularly when the hand phenotype is complex or atypical.

Prematurity substantially shaped prognosis in this case, because delivery at 27 weeks with very low birth weight and low Apgar scores indicates a high baseline risk of respiratory failure and early neonatal

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mortality independent of congenital anomalies (Lemons, Bauer, Oh, Korones, Papile, Stoll, Verter, Temprosa, Wright, Ehrenkranz, Fanaroff, Stark, Carlo, Tyson, Donovan, Shankaran, Stevenson, & NICHD Neonatal Research Network., 2001). The newborn's documented respiratory depression and death within two days in Table 2 align with known challenges of viability and respiratory support at the limits of prematurity (Koc & Unal, 2025; Pramanik, Rangaswamy, & Gates, 2015). Severe thoracic wall deficiency can plausibly worsen respiratory mechanics by reducing effective chest stability, which becomes particularly relevant when ventilatory support is needed in very immature lungs (Emil, 2019; Fokin & Robicsek, 2002). Reports of neonatal Poland presentations with major thoracic complications, including lung herniation at birth, underscore that thoracic involvement can have immediate physiologic consequences in selected newborns (Chandran et al., 2013).

The antenatal ultrasonography recorded in this report mainly documented fetal presentation, estimated fetal weight, and heart rate in Table 1, which is typical of routine assessment and may not capture subtle chest muscle deficiency unless specifically sought. (Chait et al., 2023). This supports the clinical reality that recognition of Poland syndrome in neonates still relies heavily on careful postnatal physical examination, followed by imaging when needed to define chest wall and rib involvement. (Fokin & Robicsek, 2002). In very severe cases, rare associated thoracoabdominal defects have been described such as intercostal liver herniation illustrating why imaging can be important when chest wall integrity appears compromised (Jimbo, Hori, Ushiyama, & Masumoto, 2022). The presence of neonatal variants such as axillary pterygium also highlights that the newborn phenotype can deviate from classic teaching points, making structured documentation of "what was seen" clinically valuable.

Management priorities in the neonatal period are stabilization of vital functions and coordinated monitoring, while definitive reconstructive procedures for chest wall or breast asymmetry are generally deferred to later childhood or adolescence (Baldelli et al., 2020; Di Lorenzo, Corradino, & Cherubino, 2020; Da Silva Freitas, Tolazzi, Santos, Graf, & Cruz, 2008). In this case, referral to perinatology for

intensive observation and prematurity-focused care is consistent with the expected immediate pathway for a 27-week neonate with respiratory depression, as summarized in Table 2. The main clinical challenge is that severe prematurity and thoracic wall deficiency may interact to complicate ventilation and overall physiologic reserve, limiting what can be achieved beyond supportive management in the first days of life (Emil, 2019; Koc & Unal, 2025). For the broader literature, detailed neonatal case reporting remains useful because it improves recognition of severe and atypical presentations and helps refine how clinicians document phenotype, anticipate complications, and plan follow-up in survivors (Romanini, Calevo, Puliti, Vaccari, Valle, Senes, & Torre, 2018).

CONCLUSION

Poland syndrome in newborns is marked by unilateral chest muscle absence with associated limb anomalies; early recognition and coordinated neonatal management are essential, and case reporting improves detection and follow-up planning.

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Recognizing Poland syndrome at birth: Chest wall and hand manifestations

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