

Poland Syndrome and Pregnancy

Poland Sendromu ve Hamilelik

Emre Zafer, Selda Demircan Sezer, Sümeyra Nergiz Avcıoğlu, Burcu Bıçakçı

Adnan Menderes University Faculty of Medicine, Department of Obstetrics and Gynecology, Aydın, Turkey



Abstract

Poland syndrome is a rare congenital disorder presenting with absence of unilateral thoracic muscles. Upper extremity, rib and connective tissue deformities usually accompany muscle agenesis or hypoplasia on the same side. Although its etiology remains unknown, inadequate perfusion is suspected for related regional tissue hypoplasia/aplasia. There is scarce information about possible cardiopulmonary function involvement due to abnormal thoracic muscle and tissues. A slight change in this function may be worsened in pregnancy affecting maternal and fetal health. Here we present a 30 weeks pregnant patient with Poland syndrome who developed severe shortness of breath and eventually had to have iatrogenic preterm delivery. Rare syndromes compatible with life and reproduction can be a challenge for clinicians especially during pregnancy due to unpredictable effects on gestational physiologic adaptation processes. Therefore, case reports or series regarding pregnancy complications of these rare syndromes provide valuable information for possible complications and their management options.

Keywords

Poland syndrome, pregnancy, preterm

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Address for Correspondence/Yazışma Adresi:

Emre Zafer MD,
Adnan Menderes University Faculty of
Medicine, Department of Obstetrics and
Gynecology, Aydın, Turkey
Phone : +90 530 435 89 09

E-mail : dr.emrezafer@gmail.com

ORCID ID: orcid.org/0000-0001-9262-2947

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Öz

Poland sendromu tek taraflı göğüs kas deformiteleri ile seyreden konjenital bir hastalıktır. Genellikle aynı taraf üst ekstremité, kaburga ve bağ dokusu deformiteleri, kas agenezileri veya hipoplazilerine eşlik eder. Etiyolojisi hala belirsiz olsa da etkilenmiş bölgelerin yetersiz ve defektif perfüzyonunun bölgesel doku hipoplazi ve aplazisine yol açtığı düşünülmektedir. Kardiyopulmoner fonksiyonun göğüs kafesi kas ve doku anormalliğine bağlı etkilenme düzeyi ile ilgili çok az bilgi vardır. Bu fonksiyondaki hafif sayılabilecek değişiklikler gebelik döneminde maternal ve fetal sağlığı etkileyebilecek düzeyde kötüleşebilir. Burada Poland sendromu olan 30 haftalık gebelikte şiddetli nefes darlığı şikayeti nedeniyle iyatrojenik preterm doğum olgusu sunulmaktadır. Hayatla bağdaşan ve üreme fonksiyonunun korunduğu sendromlar özellikle hamilelik dönemlerinde fizyolojik adaptasyon süreçleri üzerinde öngörülemez etkileri nedeniyle klinisyenler için zorluk oluştururlar. Bu nedenle nadir sendromların hamilelikte görülen komplikasyonlarına dair olgu sunumları beklenen olası sorunlar ve yönetim biçimleri hakkında değerli bilgiler sağlar.

Introduction

Poland syndrome (OMIM 173800), a rare congenital syndrome with incidence between 1/10.000-100.000, was first described by Alfred Poland in 1841 (1,2). It consists of unilateral aplasia/hypoplasia of pectoralis major muscle and accompanying defects of other thoracic muscles, ribs, and/or ipsilateral upper extremity at varying frequency

and degrees (2). Among these abnormalities, it is plausible to expect that some of these can cause respiratory function problems under certain conditions like anesthesia and pregnancy: dextrocardia, scoliosis, pectus carinatum, pectus excavatum, lung herniation, paradoxical respiration (3). Our pregnant case did not have neither of the latter, however her respiratory complaints were severe enough to cause iatrogenic preterm delivery.

Case Report

A written informed consent was obtained from patient for this presentation. An 18-yr-old gravida 1, parity 0 patient with a known diagnosis of Poland syndrome were presented at her 30th weeks of gestation with shortness of breath. Her medical history was unremarkable other than Poland syndrome. Her physical examination was normal except absent pectoralis major muscle, short forearm, hypoplastic hand, brachysyndactyly, second-third finger hypoplasia, and atrophic breast on the right, components of her Poland syndrome diagnosis (Figure 1, 2). Also, minimally decreased respiratory sounds noticed on auscultation on the affected side. Complete blood count and routine laboratory evaluation were in normal range. Karyotype analysis and florescent *in situ* hybridization (FISH) for chromosome 11q were also normal. Obstetric ultrasonography and umbilical artery Doppler evaluation were normal. During her follow up examination in one week, she complained about worsening dyspnea. Patient was hospitalized for close observation and detailed evaluation. Respiratory function tests, pulse oximetry readings, arterial blood gas analysis, electrocardiography, non-stress test were normal. There was minimal anterior septum hypokinesis, and first degree pulmonary valve regurgitation on echocardiography. For a possible preterm delivery, antenatal maternal steroid administration was achieved by two doses of betamethasone 12 mg (Celestone Chronodose; Merck Sharp&Dohme, New Jersey, USA) via intramuscular route. Patient's respiratory complaints progressed further, being orthopneic at 32 weeks of gestation and decision to intervene by cesarean delivery was made. After delivery, patient's complaints dramatically resolved with no complication.



Figure 1. Right hand anomaly of the patient with Poland syndrome



Figure 2. Unilateral right sided pectoralis major muscle agenesis and ipsilateral breast hypoplasia

Discussion

Etiology of Poland syndrome remains unknown. Subclavian artery disruption and abnormal perfusion related aplasia/hypoplasia of regional muscles and other tissues between 42-47 days of embryogenesis is common popular theory (4). However, this is challenged by a latest case report with intact thoraco-acromial arteries and muscle fascia, suggesting an alternative theory of paraxial mesenchyme developmental failure (5). Recently, monozygotic twins with this syndrome were found to have 11q12.3 deletion, pointing five candidate genes in the deleted region (6). The majority of Poland syndrome cases are sporadic however there are some familial clustered cases implying different inheritance patterns (7). Our case has normal karyotype and FISH result and she did not mention any relatives with similar findings. Majority of the surgical intervention reports cite cosmetic reasons for indication of surgery (2,8). Apart from scarce cases of lung herniation, recurrent pneumothorax and paradoxical respiratory movements, serious cardiac or pulmonary complaints have not been reported (3). In addition, no pregnant case reports could be spotted in

the PubMed search. Our case had a mild complaint of dyspnea in midtrimester and worsened progressively with advancing gestational age. Normal respiratory function tests, echocardiography and arterial blood gas analysis held us from earlier intervention even though patient's subjective complaint was impressive. Significant respiratory tract changes are expected during pregnancy. Functional residual capacity, residual volume, and expiratory reserve volume decreases. Diaphragm rises approximately 4 cm and thoracic cage girth increases 6 cm. In our case, with enlarging uterus, it is possible that elevation and limitation of diaphragm movement was more prominent and negatively affected stabilization of affected right hemithorax, caused significant respiratory discomfort. Even though we could not document respiratory dysfunction with tests, it seemed that we would see deteriorated test results if preterm delivery was held for longer. Our case presentation must alert clinical thinking in a way that even benign conditions like Poland syndrome may complicate pregnancy with their unexpected effect on normal pregnant physiologic adaptations. Poland syndrome can cause significant respiratory discomfort during pregnancy that may not be reflected on respiratory function tests and can alter obstetric management. Therefore, early baseline respiratory function tests may be indicated in pregnant patients with Poland syndrome in order to discern worsening results (even in the normal range) with advancing gestational age.

Ethics

Informed Consent: A written informed consent was obtained from patient for this presentation.

Peer-review: Externally and internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: E.Z., B.B., Concept: E.Z., S.D.S., Design: E.Z., Data Collection or Processing: B.B., Analysis or Interpretation: E.Z., S.N.A., Literature Search: E.Z., S.D.S., Writing: E.Z., S.N.A.

Conflict of Interest: No conflict of interest was declared by the authors.

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