

Congenital unilateral amastia: An unusual yet unique presentation associated with breech delivery

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Received - 10 May 2020

Initial Review - 26 May 2020

Accepted - 04 June 2020

ABSTRACT

Absence of the breast tissue is the least frequent of all the congenital breast anomalies. Amastia can be unilateral or bilateral and may have a syndromic association. Isolated congenital unilateral amastia is a very rare condition that is often misdiagnosed as Poland syndrome. Poland syndrome is a disease more common among males and often associated with other limbs, chest wall, and other digits anomalies, which makes surgical correction more tedious. Unilateral amastia in the absence of other anomalies may have a different etiology, and hence, it is worthwhile to explore other causes for the same. Herein, we present a rare case of a newborn with congenital unilateral right-sided amastia born through cesarean section in view of breech presentation.

Key words: Breech presentation, Congenital amastia, Poland syndrome, Unilateral amastia

Amastia is a rare condition wherein the normal growth of the breast and nipple does not occur. Mammary glands develop along the mammary ridge at 6 weeks of gestation. Any insult, genetic or environmental during this period may arrest the development of the mammary gland [1]. It can be unilateral or bilateral and can be associated with a syndrome or in rare cases be an isolated finding. The unilateral variety is usually associated with an absence of pectoralis muscles, costal cartilage and rib defects, hypoplasia of subcutaneous tissues of chest wall, and defects of hand and digits also known as Poland syndrome [2,3]. Poland's syndrome can be often confused with isolated congenital unilateral amastia, unless differentiated by thoracic ultrasound to rule out pectoralis muscle abnormalities (which is essential in diagnosing Poland's). The literature shows a few instances wherein researchers have suggested considering isolated congenital unilateral amastia as a separate entity. Some have mentioned it as "anterior thoracic hypoplasia" [4]. The main pathophysiology of isolated congenital unilateral amastia is related to in utero vascular compromise to the developing thoracic structure and/or compression/trauma to the breast bud causing destruction of the same [5,6].

The latter mentioned etiology seems to be the most likely explanation for our case of isolated congenital unilateral amastia which presented with an abnormal fetal uterine lie (breech). Breech and other abnormal fetal positions are often associated with compression and fetal deformations [7,8].

CASE REPORT

A female child, born to a 30-year-old multiparous lady, was associated with congenital unilateral right-sided amastia.

The baby was delivered through a cesarean section in view of breech presentation and non-progress of labor. The baby cried immediately after birth and did not require resuscitation. The APGAR scores at birth were 9/10 and 9/10 at 1 and 5 min, respectively. The mother had no antenatal complications or past medical illnesses. Anomalies scan done at 20 weeks were reported to be normal. She had no history of drug intake during pregnancy other than prenatal vitamins. There was no history of consanguinity among parents (which is a rare cause for amastia). There was no family history of similar illness. The newborn was exclusively breastfed.

On clinical examination, the body weight was 2.6 kg and head circumference was 36 cm. The areola, nipple, and breast tissue were not palpable on the right side (Fig. 1). The rib cage felt normal clinically. No limb anomalies or limb length discrepancies were seen. The neurological examination was normal. External genitalia were normal. No other anomalies were noted on clinical examination.

Blood investigations were normal. Thoracic radiography and ultrasound were done. We confirmed that there was only the absence of the breast tissue and pad of fat, but pectoralis muscles and chest wall/rib cage were absolutely normal (Figs. 2 and 3). A multidisciplinary approach involving neonatology, pediatric surgery, plastic surgery, radiology, and genetics was done and the management approach finalized.

The parents were counseled about the nipple reconstruction and breast augmentation surgeries during adolescence and the need for child and parental psychological counseling during the stages of growth of the baby. At a follow-up visit, the child was gaining weight adequately and had no complaints.



Figure 1: The right-sided unilateral amastia with partial unilateral thoracic hypoplasia

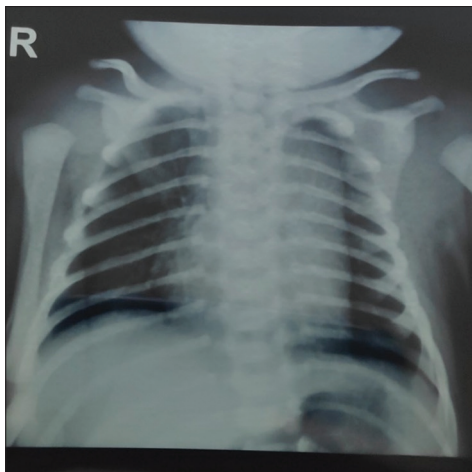


Figure 2: Chest radiograph with no ribcage anomaly

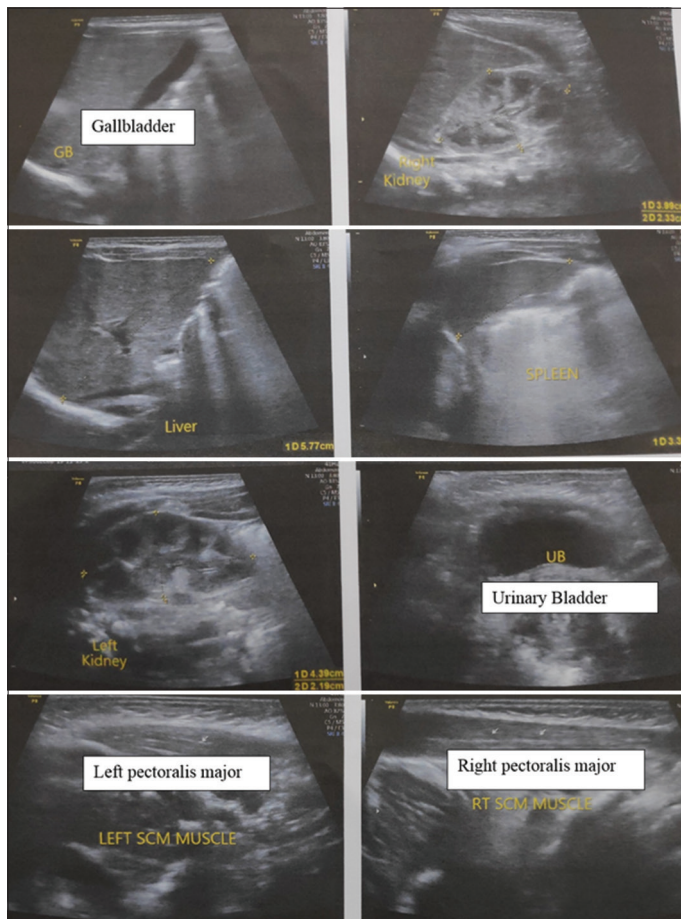


Figure 3: Lung and abdominal ultrasound showing presence of pectoralis major and no anomalies

DISCUSSION

We believe that this is a case of isolated congenital unilateral amastia which in itself is a separate diagnosis and should not be diagnosed under Poland syndrome. Isolated congenital unilateral amastia with normal pectoralis muscle without limb anomalies represents a separate entity, which when diagnosed correctly can be treated with excellent results.

Poland syndrome was first described in 1841. It consists of unilateral absence or hypoplasia of pectoralis muscles, absence of the breast tissue, chest wall anomalies, and digit abnormalities. Other conditions seen to be associated with congenital amastia include hemivertebrae, renal anomalies, dextrocardia and Sprengel deformity, absent phalanges or digits, and hypoplasia of the forearm, wrist, and hand, and ulnar mammary syndrome, which are mentioned in the literature [9,10].

Breech presentation is often associated with an increased incidence of congenital anomalies. *In utero* compression of the breast bud and anterior thoracic wall can be a cause for this kind of presentation in the absence of any other anomalies [8]. We ruled out all other possible congenital anomalies associated with known syndromes and have come to the diagnosis of isolated congenital unilateral amastia. Similar case study has been reported by Spear *et al.* [4]. No other case of isolated congenital unilateral amastia in breech presentation has been reported before.

Cases of bilateral amastia [11], amastia with pre-auricular sinus [12], and amastia in a child born to a rape victim [13] have been described previously. This case is one of the kinds due to the fact that no other case of isolated congenital unilateral amastia has been found in the literature to have been associated with breech presentation and so gives us further insight.

Although Poland syndrome does constitute a major differential diagnosis for amastia, cases similar to our patient should be thoroughly evaluated to rule out the same to avoid misdiagnosis. Other reasons for breast tissue absence should be sought, for example, abnormal fetal lie, maternal drugs, and medical conditions. The importance of identifying this case to be distinctly different from Poland syndrome lies in the fact that an absence of rib cage, skeletal, and other abnormalities makes this diagnosis a better prognosis. Treatment for such cases can be done on the lines of post-oncological breast reconstruction, which is a safer and efficacious compared to surgery for Poland's syndrome (which requires surgery of pectoralis muscle and rib cage).

Surgery for cases similar to our patient is ideally divided into nipple reconstruction done in childhood/adolescence (by transverse rectus abdominis myocutaneous flap) and breast augmentation which is also done in adolescence (with a tissue expander inserted at the time of muscle transposition). These surgeries are better and efficacious than surgeries for Poland syndrome. Parental and child anxiety is also alleviated by psychological counseling during different phases of growth, especially in adolescence [14].

CONCLUSION

Unilateral amastia in the absence of other anomalies may have a different etiology, and hence, it is worthwhile to explore other causes for the same.

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Funding: None; Conflict of Interest: None Stated.

How to cite this article: James V, Rajan J. Congenital unilateral amastia: An unusual yet unique presentation associated with breech delivery. Indian J Case Reports. 2020;6(6):329-331.

Doi: 10.32677/IJCR.2020.v06.i06.015