

# Amniotic Band Syndrome: A Syndrome with Multiple Congenital malformations: A case Series

Sharma M\*, Chander B, Kaul R and Soni A

Department of pathology, Assistant Professor, DR RP Government Medical College, India

Submission: October 20, 2017; Published: November 14, 2017

\*Corresponding author: Manupriya Sharma, Department of pathology, Assistant Professor, DR RP Government Medical College, India, Tel: 91-8628000105; Email: manupriya.priyasharma@gmail.com

## Abstract

**Introduction:** Amniotic band syndrome (ABS) is an uncommon clinical entity which results in a wide spectrum of congenital malformations. These malformations may range from limb and digits amputations to serious life threatening craniofacial and abdominal deformities. Various theories have been proposed for this syndrome; however the etiology of the disease is still inconclusive. We hereby report three cases of ABS, highlighting the varied congenital malformations associated with the syndrome. All three cases had congenital defects in the extremities. An aberrant fibrous band was present in all three cases. Apart from limb deformities, the cases had a large spectrum of congenital malformations like anencephaly, gastroschisis, cleft lip, cleft palate, contractures in limbs and malformations in internal organs.

**Summary & conclusion:** The most common congenital abnormality associated with ABS is malformations in the extremities. We presume that the pathogenesis of ABS is multi factorial with all etiologies converging towards one final end point. The amputation in extremities can be explained by the formation of constriction bands in this disease. Congenital malformations like anencephaly and malformations in the internal organs support genetic factors as an etiology for ABS.

**Keywords:** Amniotic band syndrome (ABS); Limb body wall complex (LBWC); Membrane disruption; Contraction bands

**Abbreviations:** ABS: Amniotic Band Syndrome; ADAM: Amniotic Deformity Adhesion and Mutilation; NTD: Neural tube defects; LBWC: Limb Body Wall Complex

## Introduction

Amniotic band syndrome (ABS) is a heterogenous disease with a wide spectrum of clinical presentation [1]. ABS has many synonyms like amniotic disruption sequence, amniotic deformity adhesion and mutilation (ADAM) complex and limb body wall complex (LBWC). The incidence of the disease varies from 1 in 1200 to 1 in 15000 [2]. The wide range of congenital anomalies in this syndrome includes limb and digital amputations (most common) to lethal complications like craniofacial and abdominal defects. The etiology of the disease is still not completely understood. Various theories proposed for this syndrome include:

- I. Membrane disruption associated with development of amniotic band
- II. Vascular abnormality resulting in an ischaemic event
- III. Genetic factors [3].

We hereby report three cases of ABS, its varied spectrum of clinical manifestations and discussion about the various etiologies that may be responsible for the presentation.

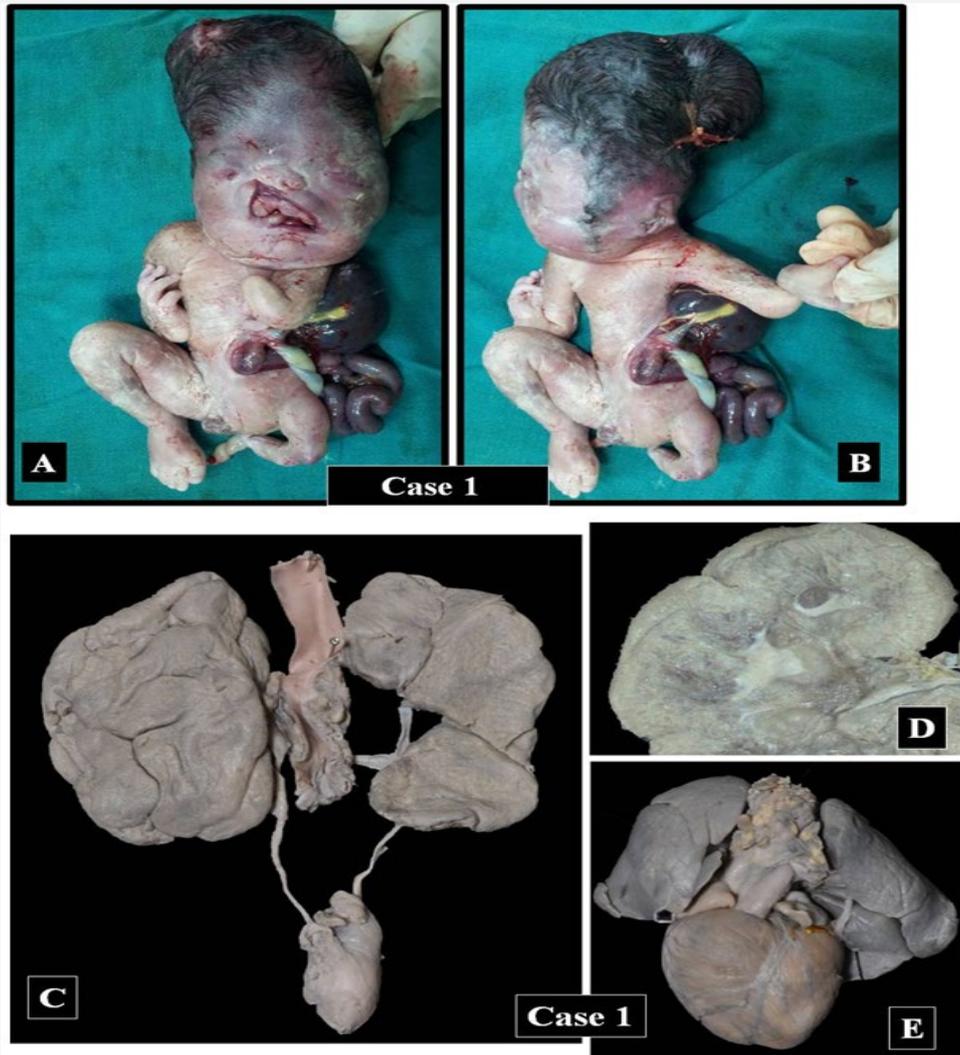
## Case Series

### Case 1

Twenty six year old female, primigravida, unbooked, unsupervised at POG32 weeks was admitted to the emergency labour room with spontaneous onset of labour. She delivered a 1700gms male fetus. On external examination, fetus had multiple congenital malformations. There was no history of consanguinity and no past family history of congenital malformations. The abnormal features included encephalocele, cleft lip and cleft palate and the nose was compressed, deformed with small nostrils (Case 1A). The left upper limb was nearly amputated with a constriction band and left lower foot revealed contractures (Case 1B). There was abdominal wall defect in

the form of gastroschisis with organs protruding out from the abdominal cavity. Aberrant fibrous tissue was attached to the head of the fetus. The umbilical cord was unremarkable and

normal sized with two arteries and one vein. Placenta was grossly unremarkable (Figure 1).



**Figure 1:**

- (A) Shows cleft lip and cleft palate and the nose was compressed with deformed with small nostrils. The left upper limb was nearly amputated with a constriction band and left lower foot was deformed with contractures.
- (B) Shows encephalocele with gastroschisis. The abdominal organs are protruding out.
- (C) Bilateral symmetrically enlarged kidneys. The capsular surface of kidneys showed many pin head sized cysts over the capsular surface.
- (D) Kidneys show sun ray appearance on the cut surface.
- (E) Hypoplastic lungs.

Autopsy revealed bilateral symmetrically enlarged kidneys (Case 1c). The capsular surface of kidneys showed many pin head sized cysts over the capsular surface and sun ray appearance on the cut surface (Case 1D). The lungs were hypoplastic (Case E). Thymus, heart, liver, spleen, stomach and intestine were unremarkable grossly.

### Case 2

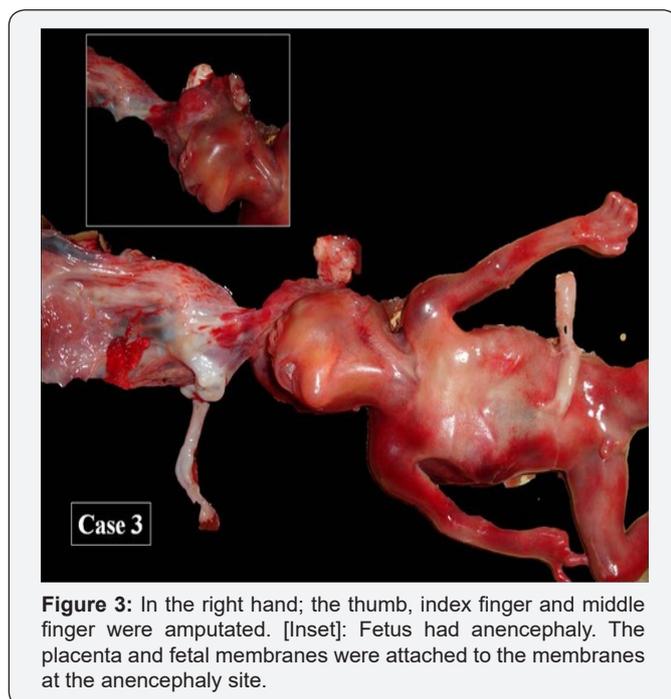
A 21 years old primigravida, after a non-consanguineous

marriage, presented to the Gynae OPD at 14 weeks of gestation with spontaneous onset of labour. The patient had not got any USG done during this period. She aborted a male fetus (Body weight: 800gms). On external examination, the fetus revealed a constriction band at left lower limb with congestion and deformed left foot (Case 2). A fibrous band was seen attached between the head and placenta. Placenta and umbilical cord were unremarkable. Autopsy did not show any abnormality in internal organs (Figure 2).



**Figure 2:** A constriction band was seen at left lower limb with congestion and deformed left foot. A fibrous band was seen attached between the head and placenta

**Case 3**



**Figure 3:** In the right hand; the thumb, index finger and middle finger were amputated. [Inset]: Fetus had anencephaly. The placenta and fetal membranes were attached to the membranes at the anencephaly site.

A 30 year old, multigravida G<sub>2</sub>P<sub>1001</sub>, presented at 20 weeks to the Gynae OPD with spontaneous onset of labour. A malformed female fetus was aborted with birth weight 1200gms. On external examination, the fetus had anencephaly (Inset: Case 3). The placenta and fetal membranes were attached to the membranes at the anencephaly site. In the right hand, the thumb, index finger and middle finger were amputated (Case 3). Umbilical cord and

placenta were unremarkable. No malformations in the internal organs were seen on autopsy (Figure 3).

**Discussion**

ABS is an uncommon clinical entity. It has a wide spectrum of clinical presentation of uncertain etiology. According to literature, the incidence of ABS ranges from 1:1200 to 1:15000 live births [2]. The congenital malformations of the disease range from amputations of the extremities or digits to life threatening craniofacial, thoracic and abdominal complications. Defects in extremities which have been reported include amputated limbs or digits, contractures in the limbs, club foot, syndactyly and absent limbs. Craniofacial abnormalities have been reported as encephalocele, anencephaly, cleft lip and cleft palate. Thoraco-abdominal complications include omphalocele and gastroschisis [4]. The presence of ABS in a fetus usually result in premature onset of labour and delivery. We support this belief as in our all three cases there was spontaneous onset of labour.

Defects resulting from ABS have been categorized into four groups-

- i. Neural tube defects (NTD)
- ii. Craniofacial abnormalities
- iii. Limb anomalies
- iv. Limb body wall complex (LBWC) [5].

The exact etiology of the disease is still uncertain. However, many theories describing the etiology of the disease have been proposed. These theories are mainly divided into two groups: Extrinsic theory and intrinsic theory. According to the extrinsic theory, ABS results due to the mechanical disruption of the amnion membrane. The membrane disruption can be an early event due to abnormal germinal disc development or later due traumatic disruption of membranes. Due to the rupture of amnion membrane, the amniotic fluid extrudes out of the cavity along with certain fetal parts which gets entrapped and are subjected to vascular compression by the constriction band created [6]. As a result, the defect will depend upon the location of bands like amputation of limbs or digits. We support this finding, as in all three cases we found congenital defects in extremities. The first case had amputated left upper limb and the third case had amputation of right hand digits. In second case, a clear constriction band was formed on the left lower limb with resulting marked congestion and deformity of left foot. Defects in extremities represent the most common deformity associated with ABS as in our cases.

We labeled the first case in our study as LBWC as this fetus had multiple congenital abnormalities in the form of encephalocele, cleft lip and cleft palate, gastroschisis, amputated upper limb and contracture deformity in the lower extremity. Internal organs revealed hypoplastic lungs and bilateral multicystic kidneys. The umbilical cord in this fetus was shortened. LBWC is defined

as a condition which fulfils the presence of at least two of the three anomalies including craniofacial defects, thoraco and/or abdominal defects and limb defects [7,8]. The first case fulfilled all the three required criteria for diagnosis. Amputated limb and gastroschisis can be explained by the theory of the amniotic band formation. But cleft lip and palate, contractures in the lower limb, hypoplastic lungs and multicystic kidneys suggests a genetic basis for the disease.

The second case in our series revealed constriction band at the lower limb with malformed left foot with marked congestion. This case also revealed amniotic band between placenta and the head of the fetus. Placentocranial adhesions have been rarely reported in literature [9-12]. These adhesions are considered as serious abnormality associated with considerable morbidity and mortality. Placentocranial adhesions are usually associated with spontaneous abortions as in our case. In literature, only 2 cases have been reported which survived for 48hrs [9-13]. Surgical correction of the adhesion band is needed to correct this defect. However, it is important to carefully evaluate the adhesion band for any bridging vessels between placenta and cerebral cortex or dural sinuses.

Third case revealed anencephaly and amputation of the digits. The internal organs on examination were unremarkable. A fibrous band was seen attached between the membranes at anencephaly and the placenta. To the best of our knowledge, we have not come across any case in literature describing the attachment of adhesion band between the placenta and the membranes at the anencephaly. In all the three cases described, we consistently found the presence of an aberrant fibrous band. Presence of the fibrous band supports the theory that membrane rupture is followed by the formation of an amniotic band and this is one of the underlying etiologies resulting in ABS. However, this theory does not support other malformations like cleft lip and cleft palate, anencephaly, club foot, contractures in limbs and internal organ abnormalities. These defects support a genetic basis of the disease. The study is limited by little number of cases. Secondly, no chromosomal analysis was done in these cases which would have helped us further in understanding the pathogenesis of the disease.

## Conclusion

To conclude, ABS is a heterogenous disease with a wide spectrum of clinical presentation. Defects in the extremities are the most common clinical presentation of the disease. We report extremely rare presentation of placentocranial adhesions in two of our cases. Out of these, one case revealed the fibrous band between placenta and the membranes at the anencephaly

site which has not been reported before. ABS usually leads to spontaneous onset of labour. Formation of amniotic bands resulting in various congenital malformations is definitely one of the etiologies of the disease as evident in our three cases. The presentation of disease will depend upon the location of constriction bands. Amputations in the extremities and abdominal wall defects can be explained by membrane disruption theory. However, malformations in internal organs, anencephaly, and club foot support a genetic basis of the disease. Hence, we support the fact that the etiology of the disease is multifactorial and all the factors converge towards one final end point.

## References

1. Bamforth JS (1992) Amniotic band sequence: Streeter hypothesis revisited. *Am J Med Genet* 44(3): 280-287.
2. Cignini P, Giorlandino C, Padula F, Dugo N, Cafa EV, et al. (2012) Epidemiology and risk factors of amniotic band syndrome, or ADAM sequence. *J Prenat Med* 6(4): 59-63.
3. Iqbal CW, Derderian SC, Cheng Y, Lee H, Hirose S (2015) Amniotic band syndrome: a single-institutional experience. *Fetal Diagn Ther* 37(1): 1-5.
4. Lowry RB, Bedard T, Sibbald B (2017) The prevalence of amnion rupture sequence, limb body wall defects and body wall defects in Alberta 1980-2012 with a review of risk factors and familial cases. *Am J Med Genet A* 173(2): 299-308.
5. Mian DB, Nguessan KL, Aissi G, Boni S (2014) Amniotic band syndrome (ABS): can something be done during pregnancy in African poor countries? Three cases and review of the literature. *Clin Exp Obstet Gynecol* 41(2): 226-232.
6. Torpin R (1965) Amniochorionic mesoblastic fibrous rings and amniotic bands: associated constricting fetal malformations or fetal death. *Am J Obstet Gynecol* 91: 65-75.
7. Van Allen MI, Curry C, Gallagher L (1987a) Limb body wall complex: I. Pathogenesis. *Am J Med Genet* 28(3): 529-548.
8. Hunter AG, Seaver LH, Stevenson RE (2011) Limb-body wall defect. Is there a defensible hypothesis and can it explain all the associated anomalies? *Am J Med Genet A* 155A(9): 2045-2059.
9. Merrimen JL, McNeely PD, Bendor-Samuel RL, Schmidt MH, Fraser RB (2006) Congenital placental- cerebral adhesion: an unusual case of amniotic band sequence - Case report. *Neurosurgery* 104(5): 352-355.
10. Chandran S, Lim MK, Yu VY (2000) Fetal acalvaria with amniotic band syndrome. *Arch Dis Child Fetal Neonatal Ed* 82(1): 11-13.
11. Ghritlaharey RK, Kushwaha AS, Chanchlani R, Nanda M (2007) Placento-cranial adhesion: a report of two cases. *J Indian Assoc Pediatr Surg* 12(2): 102-103.
12. Woyton J (1961) Encephalocele attached to the placenta. *Am J Obstet Gynecol* 81: 1028-1032.
13. Menekse G, Mert MK, Olmaz B, Celik T, Celik US, et al. (2015) Placento-Cranial Adhesions in Amniotic Band Syndrome and the Role of Surgery in Their Management: An Unusual Case Presentation and Systematic Literature Review. *Pediatr Neurosurg* 50(4): 204-209.



This work is licensed under Creative Commons Attribution 4.0 License

## **Your next submission with Juniper Publishers will reach you the below assets**

- Quality Editorial service
- Swift Peer Review
- Reprints availability
- E-prints Service
- Manuscript Podcast for convenient understanding
- Global attainment for your research
- Manuscript accessibility in different formats  
**( Pdf, E-pub, Full Text, Audio)**
- Unceasing customer service

**Track the below URL for one-step submission**

<https://juniperpublishers.com/online-submission.php>