

Amniotic Band Syndrome – Case Report of a Rare Congenital Condition Causing Undue Suffering through Delayed Diagnosis

Dodampahala SH¹, Dodampahala SK², Dodampahala SD³, Meegahawatta AR⁴

¹Associate professor, Department of Obstetrics and Gynaecology, Faculty of Medicine, University of Colombo, Colombo, Sri Lanka

²Undergraduate Research Student

³Undergraduate Research Student

⁴Research Associate, Department of Obstetrics and Gynaecology, Faculty of Medicine, University of Colombo, Colombo, Sri Lanka

Abstract: *Amniotic band syndrome (ABS) is a rare congenital disorder that can lead to a wide range of physical abnormalities in the newborn infant. In this report we describe the case of a child with ABS who was diagnosed at 8 years of age, the circumstances leading to the delayed diagnosis and the effects of delayed diagnosis on the child and parents. The parents who had avoided pregnancy fearing recurrence of similar defects in a subsequent pregnancy were counselled on the condition. Later the mother of the child with ABS presented in her second pregnancy to the author and delivered a healthy baby by elective cesarean section following an uneventful pregnancy.*

Keywords: Amniotic band syndrome, Limb reduction defects, Limb Body-wall complex

1. Introduction

Amniotic band syndrome (ABS) is a rare congenital disorder that is associated with a wide range of physical abnormalities [1] in the newborn infant, some of which are significantly disabling and disfiguring in nature. The commonest abnormalities usually involve the limbs and can range from simple construction rings to complete amputation occurring at various levels [2]. Abdominal wall defects and abnormalities of the cranio-facial region such as cleft lip and cleft palate are also associated with ABS [2] while in the more complex cases, visceral defects such as renal agenesis [3] and rarely septo-optic dysplasia [2-4] are also known to occur. Various studies estimate the incidence of ABS to be between 1 in 1300 to 1 in 15000 though [4,5] the real figure is likely closer to the latter mark.

The most widely accepted theory on the etiology of the condition was put forward by Torpin in 1965 [2] and attributes the defects to constricting action of fibrous amniotic bands on fetal extremities following early rupture of the amnion with subsequent loss of amniotic fluid and extrusion of fetal parts in to the chorionic cavity [6]. The defects are thought to result from vascular compression and resultant necrosis secondary to the compressive effect of the bands [6] with the timing of amniotic rupture and the degree of compression being determinants of the extent of abnormalities seen at birth. However some of the abnormalities associated with ABS such as imperforate anus [7] and Septo-optic dysplasia [8] are difficult to explain through this theory. Other proposed etiologies for ABS include genetic predisposition [9], germ disc disruption [10] and vascular disruption [11]. Of these, vascular etiology seems the most likely with defects of the abdominal wall, cranio-facial region [9-11] and the limbs [12] being

attributed to underlying vascular defects by a number of studies.

The condition can be identified as early as the late first trimester through ultrasound scanning [1]. Today the defects caused by ABS can be identified and studied in detail through advanced techniques such as 3D ultrasonography early in the pregnancy [1]. The mainstay of treatment in ABS is plastic and reconstructive surgery [13]. However in-utero surgeries are becoming increasingly popular with the advent of advanced imaging techniques [13].

2. Case Report

An 8 year old child with missing fingers on bilateral upper limbs (Figure 1 - Amputations affecting fingers of both hands), below knee amputation on the left lower limb and missing toes on the right foot [Figure 2 – Below knee amputation of left lower limb 2A (prosthesis also pictured) and missing toes on the right foot 2B] presented with his 28 year old mother to a leading tertiary care Obstetrics unit in Sri Lanka.



Figure 1: Amputations affecting fingers of both hands



Figure 2: 2A Below knee amputation of left lower limb (with prosthesis), 2B Missing toes on the right foot

The mother had been directed to the unit by a geneticist for counselling regarding the condition of her child and the next pregnancy.

The child was an only child and was delivered vaginally at 39 weeks of gestation with no maternal complications. The defects described were congenital and no diagnosis had been made at the time. Mother admits that no ultra-sound scans were performed during the antenatal period. The pregnancy had been uneventful until the time of delivery, save for the lack of adequate antenatal care. The child had received rehabilitative care from early childhood and had a left lower limb prosthesis at the time of his presentation. Despite his disabilities, he was attending a regular school and his

academic performances were excellent according to the parents. During the consultation the child was noted to be bright and alert with no signs of mental or cognitive deficiency. Motor deficits observed were limited to those occurring as a direct result of the congenital deformities. Since birth of the child, parents had been worried about the deformities and their etiology. They were also worried about the possibility of recurrence of similar disabilities in a subsequent pregnancy. However, despite multiple consultations with pediatricians, the parents had not received a clear diagnosis for the child's condition or on the possibility of recurrence in a subsequent pregnancy. This had led the parents to use barrier contraception since the birth of their first child. Eventually the parents had consulted a geneticist who had made a diagnosis of amniotic band syndrome based on clinical examination findings. He had then counselled the parents regarding the nature of the condition and the lack of evidence to suggest significantly increased risk of recurrence in subsequent pregnancy. The parents and the child were then directed to the Obstetric unit for further management.

The disabilities seen in the child were typical of Amniotic band syndrome and normalcy in his developmental domains helped confirm the diagnosis. The mother was further reassured and expressed willingness for pregnancy. She was given standard pre-pregnancy care and became pregnant within a month of initial consultation. In addition to routine antenatal care, careful, periodic ultrasound imaging were carried out starting from the end of the first trimester. No abnormalities were noted and the mother delivered a healthy baby by elective cesarean section at 39 weeks of gestation. (Figure 3)



Figure 3: A still image from the 4D ultrasound scan that was used to confirm normal morphology of the fetus

3. Discussion

Amniotic band syndrome is a rare disorder. It often results in congenital physical defects in the infant which are disabling and disfiguring. These can be a constant source of worry for the parents especially in the absence of a clear diagnosis as illustrated in the case we describe above. In this particular case, the parents of the child were from a lower middle income family and were convinced that another child with similar disability would mean that they would not be able provide optimum care to either child. This belief coupled with the delayed diagnosis meant that the poor parents suffered needless uncertainty for 8 long years and actively avoided pregnancy for the same duration. In this instances the parents were lucky to have had their first child at an early age, allowing them to complete a successful pregnancy despite the delay in diagnosis. However needless suffering as well as a significant amount of expenses borne for private consultation with various clinicians could have been avoided had the diagnosis been made earlier.

On a positive note, we must also highlight that the child had undergone early rehabilitation. This and the prosthesis supporting his left lower limb meant that he was capable of near-normal independent function. He was attending a regular school and despite the missing fingers on her right hand, was capable of carrying out school work satisfactorily. Antenatal care in Sri Lanka is freely available through the state sector and include routine imaging using ultrasound scans at predefined points during pregnancy. While this means that under proper antenatal care, the defects could have been identified in-utero, this would have had little effect on the eventual outcome of the child. There are currently no legal provisions for termination of pregnancy in Sri Lanka except in cases where the life of the mother is threatened by its' continuation meaning this is not an option even if the fetus has a severe defect incompatible with life. In addition facilities for in-utero surgeries are currently not available in the country. Plastic and reconstructive surgical facilities have limited availability and offer the best hope for a child with ABS.

References

- [1] Nardoza LMM, Araujo E, Caetano ACR, Moron AF. Prenatal Diagnosis of Amniotic Band Syndrome in the Third Trimester of Pregnancy using 3D Ultrasound. *J Clin Imaging Sci* [Internet]. India: Medknow Publications & Media Pvt Ltd; 2012;2:22. Available from: <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3352605/>
- [2] Cignini P, Giorlandino C, Padula F, Dugo N, Cafà EV, Spata A. Epidemiology and risk factors of amniotic band syndrome, or ADAM sequence. *J Prenat Med* [Internet]. CIC Edizioni Internazionali; 2012;6(4):59–63. Available from: <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3530965/>
- [3] Van Allen MI, Curry C, Walden CE, Gallagher L, Patten RM. Limb-body wall complex: II. Limb and spine defects. *Am J Med Genet.* 1987/11/01 ed. 1987;28(3):549–65.

- [4] Orioli IM, Ribeiro MG, Castilla EE. Clinical and epidemiological studies of amniotic deformity, adhesion, and mutilation (ADAM) sequence in a South American (ECLAMC) population. *Am J Med Genet A.* 2003/03/26 ed. 2003;118a(2):135–45.
- [5] Marino T. Ultrasound abnormalities of the amniotic fluid, membranes, umbilical cord, and placenta. *Obs Gynecol Clin North Am.* 2004/04/06 ed. 2004;31(1):177–200.
- [6] Torpin R. AMNIOCHORIONIC MESOBLASTIC FIBROUS STRINGS AND AMNIONIC BANDS: ASSOCIATED CONSTRICTING FETAL MALFORMATIONS OR FETAL DEATH. *Am J Obs Gynecol.* 1965/01/01 ed. 1965;91:65–75.
- [7] Graham Jr. JM, Higginbottom MC, Smith DW. Preaxial polydactyly of the foot associated with early amnion rupture: evidence for mechanical teratogenesis? *J Pediatr.* 1981/06/01 ed. 1981;98(6):943–5.
- [8] Harrison IM, Brosnahan D, Phelan E, Fitzgerald RJ, Reardon W. Septo-optic dysplasia with digital anomalies--a recurrent pattern syndrome. *Am J Med Genet A.* 2004/09/16 ed. 2004;131(1):82–5.
- [9] Hunter AG, Carpenter BF. Implications of malformations not due to amniotic bands in the amniotic band sequence. *Am J Med Genet.* 1986/08/01 ed. 1986;24(4):691–700.
- [10] Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. *Am J Med Genet.* 1987/11/01 ed. 1987;28(3):529–48.
- [11] Herva R, Karkinen-Jaaskelainen M. Amniotic adhesion malformation syndrome: fetal and placental pathology. *Teratology.* 1984/02/01 ed. 1984;29(1):11–9.
- [12] Van Allen MI, Siegel-Bartelt J, Dixon J, Zuker RM, Clarke HM, Toi A. Constriction bands and limb reduction defects in two newborns with fetal ultrasound evidence for vascular disruption. *Am J Med Genet.* 1992/11/15 ed. 1992;44(5):598–604.
- [13] Shetty P, Menezes LT, Tauro LF, Diddigi KA. Amniotic Band Syndrome. *Indian J Surg* [Internet]. India: Springer India; 2013;75(5):401–2. Available from: <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3824760/>

Author Profile



Professor Hemantha Dodampahala received the M.B.B.S. (Hons) and M.S (Obstetrics and Gynaecology) from University of Colombo, Sri Lanka. He also obtained FRCOG and FRCS from United Kingdom. He is also a member of International Representative Committee RCOG and a member of International Society of Ultrasound in Obstetrics and Gynaecology. He has over 211 citations for his research work in areas of fetal brain activity, fetal disorders, interventional fetal surgery, high risk Obstetrics, recurrent miscarriages, polycystic ovarian disease and subfertility. He ranks the 18th position in world ranking for research citations in fetal medicine.