SciMedCentral

JSM Clinical Case Reports

Case Report

Body Stalk Anomaly in One of the Dichorionic Diamniotic Twins Following *In vitro* Fertilization and Embryo Transfer

Krishna GR[1], Abdul Haium Abdul Alim[1]*, Kenneth Chang TE[2], Edwin Thia WH[3] and Bhavani Sriram[1]

[1]*Department of Neonatology, K K Women's and Children's Hospital, Singapore*
[2]*Department of Pathology, K K Women's and Children's Hospital, Singapore*
[3]*Department of Maternal Fetal Medicine, K K Women's and Children's Hospital, Singapore*

***Corresponding author**
Abdul Haium Abdul Alim, Consultant, Department of Neonatology, KK Women's and Children's Hospital, 100 Bukit Timah Road, Singapore 229899, Singapore, Tel: 65 90269663; Email: drabdulhaium@yahoo.co.uk

Submitted: 18 November 2014

Accepted: 05 June 2015

Published: 09 June 2015

Keywords

- Body stalk anomaly
- *In vitro* Fertilization- embryo transfer
- Limb body wall complex

Abstract

Body stalk anomaly/Limb body wall complex (BSA/LBWC) is a sporadic, rare and severe congenital anomaly with a poor prognosis. Association of this lethal malformation in multiple gestations following assisted reproduction techniques is even more uncommon and had been reported in case reports only. In-vitro fertilization embryo transfer (IVF-ET) is an effective treatment for various types of infertility and there is increased incidence of congenital anomalies when compared to natural pregnancy. We report a case of body stalk anomaly in one of the dichorionic diamniotic twins conceived after IVF-ET.

ABBREVIATIONS

BSA: Body Stalk Anomaly; BSA/LBWC: Body Stalk Anomaly/ Limb Body Wall Complex; IVF-ET: *In-Vitro* Fertilization Embryo Transfer

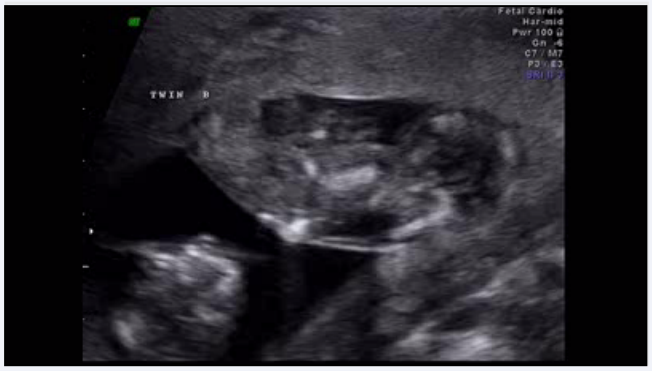
INTRODUCTION

Body stalk anomaly or limb body wall complex is a heterogeneous congenital anomaly with a series of characteristic features. It is characterized by facial clefts, major abdominal wall defects, and skeletal abnormalities such as kyphoscoliosis. The association of this anomaly with IVF-ET is very rare and to the best of our knowledge has been published in only one case report. We report a case of body stalk anomaly arising in one of the twin following IVF-ET technique to highlight the need for obstetricians, obstetric ultrasonographers and, neonatologists to be aware of this anomaly and its association in multiple gestation and artificial reproductive techniques.

CASE PRESENTATION

A 32 year old gravida-2 mother conceived with twin pregnancy following IVF-ET. The screening ultrasound scans at 13 weeks of gestation showed normal twin A and twin B showing gross fetal malformation consisting of disorganized fetal structures with cystic change below the fetal thorax, severe kyphoscoliosis, underdevelopment of lower limbs and fetal bowel in the celomic cavity (Video 1). These features were consistent with BSA.

In view of poor prognosis of this syndrome a decision for selective feticide was suggested by the obstetrician and was carried out at 18th week of gestation. Pregnancy was continued with the healthy twin till 28 weeks of gestation and both twins were



Video 1 Video Shows large cystic cavity below fetal thorax. Abdominal contents seen in coelomic cavity. Also noted severe kyphoscoliosis and under developed lower limbs.

delivered by Caesarian section in view of severe oligohydramnios in the healthy twin due to leaking liquor following feticide procedure. At delivery healthy twin was born with respiratory depression requiring resuscitation. Subsequently the healthy twin required intensive care and improved. The healthy twin was discharged home at corrected gestational age of 48 weeks and currently doing well on follow up. Except for the ultrasound finding of nephrocalcinosis of the left kidney there were no other major morbidities or congenital anomalies on the healthy twin.

On examination, the affected twin showed 2 cm lower anterior wall defect through which abdominal viscerae (mainly intestines) without any covering membranes were extruded. There was severe deformity of the spine. No cranial abnormalities or facial clefts were noted (Figure 1). Full autopsy was not conducted because of lack of parental consent.

DISCUSSION

BSA is a rare congenital anomaly with incidence of 0.32 per 100,000 births [1]. Most cases of body stalk anomaly are sporadic with unknown etiology. According to Van Allen et al and Russo et al, the diagnosis is based on presence of two out of three of the following anomalies [2,3].

1) Exencephaly or encephalocele with or without facial defects

2) Thoracic and or abdominal defects

3) Limb defects.

The exact mechanism of body stalk anomaly is still unclear and being debated. The theory of early amnion rupture resulting in amniotic bands by Torpin et al [4] and early vascular disruption theory around 4-6 weeks' gestation by Van Allen et al [2], failed to explain all the features of body stalk anomaly.

The theory of early embryonal dysplasia by Hartwig et al [5], describes the morphogenesis of BSA from a malfunction in the ectodermal placodes which results in defective closure of the embryonic abdominal wall and persistence of the extra embryonic celome communicating with abdominal cavity. This is currently regarded as the most widely accepted theory explaining most features of BSA.

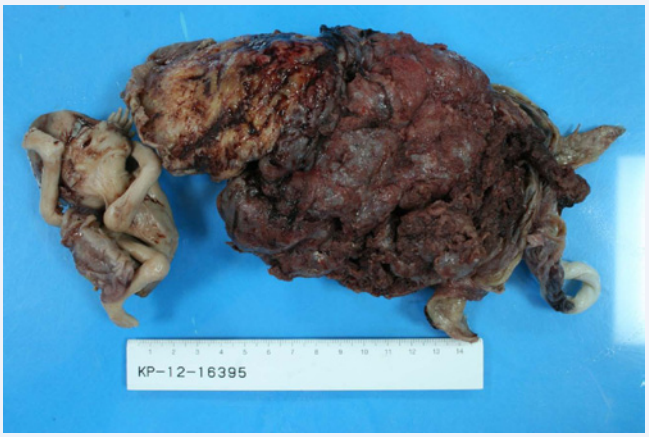


Figure 1 Shows macerated fetus attached to placenta with herniated abdominal contents, severe kyphoscoliosis and a short umbilical cord. There are no facial clefts or gross CNS malformation visible.

An interesting hypothesis put forth by Russo et al, explaining the two different entities seen in this anomaly may be due to two different pathogenesis [3]. The entity with craniofacial defects may be due to early amnion rupture with formation of amniotic bands and the entity with no craniofacial defects, a phenotype in reality representing BSA may be related to defective folding process of the embryonic disk. In our case the affected Twin had a lower anterior wall defect with extrusion of abdominal viscera and severe kyphoscoliosis. No cranial abnormalities or facial clefts were noted.

Body stalk anomaly is associated with wide range of internal anomalies. The common central nervous system anomalies include anenecephaly and alobar holoproscencephaly. Cardiovascular anomalies include septal defects and ectopia cardis. Wide spectrum of urogenital anomalies including renal aplasia/dysplasia, polycystic kidneys, genital and bladder anomalies had been reported with this condition. The commonly associated skeletal abnormalities include club foot, single bone, absent limbs kyphoscoliosis and arthrogryposis [2-6].

Antenatal ultrasound diagnosis can be made by identifying typical features like large abdominal defect, abnormalities in the axial skeleton such as kyphosis, scoliosis and a short or absent umbilical cord by the end of first trimester (11-13 weeks). Nuchal translucency measurement abnormality as one of the ultrasound features has been reported in literature. The maternal serum alpha fetoprotein is abnormal in second trimester in 100 percent of cases but not specific for this anomaly [7].

BSA is universally fatal and mortality is inevitable, recently there have been case reports of survival of fetuses with BSA [8]. Management of BSA/LBWC in twin pregnancies poses a challenge. The option of either selective feticide (dichorionic twins) or expectant management (monochorionic twins) will result in survival of healthy fetus [9]. The recent literature review favor the expectant management in both dichorionic and monochorionic pregnancies [10].

The occurrence of BSA is rare in multiple pregnancies than singletons, more frequently associated with monoamniotic pregnancies [11]. Their associations with multiple pregnancies following artificial reproductive techniques have been reported in only one case reports [12].

IVF-ET is a promising treatment for various types of infertility and associated with adverse neonatal outcome and complications. In a recent population based study by Ericson et al a three-fold increase in congenital malformation seen in babies born by IVF compared to natural pregnancies [13]. In another study by Koivurova et al, it was observed that the prevalence of heart malformations was four-fold in the IVF population than in the controls representing the general population, implicating that these artificial reproductive techniques are associated with higher rate of congenital malformations [14].

The BSA affecting one of dichorionic twin following IVF-ET technique, where one twin underwent feticide with survival of another twin makes this case rare and interesting.

It is important for obstetricians and neonatologists to be aware that the multiple gestations following artificial techniques may be prone for lethal malformations like BSA.

CONCLUSION

The BSA/LBWC is a rare and lethal malformation which is being reported in pregnancies after artificial reproductive techniques especially in multiple gestations. The awareness of these lethal malformations and early detection will be helpful in planning the antenatal counseling, timing and place of delivery, management of affected and healthy fetus.

REFERENCES

1. Chikkannaiah P, Dhumale H, Kangle R, Shekar R. Limb body wall complex: a rare anomaly. J Lab Physicians. 2013; 5: 65-67.
2. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. Am J Med Genet. 1987; 28: 529-548.
3. Russo R, D'Armiento M, Angrisani P, Vecchione R. Limb body wall complex: a critical review and a nosological proposal. Am J Med Genet. 1993; 47: 893-900.
4. Torpin R. Amniochorionic mesoblastic fibrous strings and amnionic bands: associated constricting fetal malformations or fetal death. Am J Obstet Gynecol. 1965; 91: 65-75.
5. Hartwig NG, Vermeij-Keers C, De Vries HE, Kagie M, Kragt H. Limb body wall malformation complex: an embryologic etiology? Hum Pathol. 1989; 20: 1071-1077.
6. Managoli S, Chaturvedi P, Vilhekar KY, Gagane N. Limb body wall complex. Indian Pediatr. 2003; 40: 891-894.
7. Smrcek JM, Germer U, Krokowski M, Berg C, Krapp M, Geipel A, et al. Prenatal ultrasound diagnosis and management of body stalk anomaly: analysis of nine singleton and two multiple pregnancies. Ultrasound Obstet Gynecol. 2003; 21: 322-328.
8. Kurosawa K, Imaizumi K, Masuno M, Kuroki Y. Epidemiology of limb-body wall complex in Japan. Am J Med Genet. 1994; 51: 143-146.
9. Daskalakis G, Sebire NJ, Jurkovic D, Snijders RJ, Nicolaides KH. Body stalk anomaly at 10-14 weeks of gestation. Ultrasound Obstet Gynecol. 1997; 10: 416-418.
10. Spiller E, Salvador L, Bogana G, Neri F, Ambrosini G, Cosmi E, et al. Body stalk anomaly: management of two dichorionic-diamniotic pregnancies. J Matern Fetal Neonatal Med. 2008; 21: 758-759.
11. Vidaeff AC, Delu AN, Silva JB, Yeomans ER. Monoamniotic twin pregnancy discordant for body stalk anomaly: case report with nosologic implications. J Ultrasound Med. 2005; 24: 1739-1744.
12. Hirokawa S, Uotani H, Futatani T, Sasaki Y, Ogawa J, Sakai M, et al. A case of body stalk anomaly arising in the second baby of a triplet pregnancy after in-vitro fertilization and embryo transfer. Pediatr Surg Int. 2003; 19: 223-225.
13. Ericson A, Källén B. Congenital malformations in infants born after IVF: a population-based study. Hum Reprod. 2001; 16: 504-509.
14. Koivurova S, Hartikainen AL, Gissler M, Hemminki E, Sovio U, Järvelin MR. Neonatal outcome and congenital malformations in children born after in-vitro fertilization. Hum Reprod. 2002; 17: 1391-1398.

Cite this article

Krishna GR, Abdul Alim AH, Kenneth Chang TE, Edwin Thia WH, Sriram B (2015) Body Stalk Anomaly in One of the Dichorionic Diamniotic Twins Following In vitro Fertilization and Embryo Transfer. JSM Clin Case Rep 3(1): 1077.