Keywords
Body stalk anomaly; limb-body wall complex; abdominal wall defect; prenatal ultrasound; fetal anomaly; case report

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Received:14-06-2026
Revised: 20-07-2026
Accepted: 28-07-2026
Doi:10.1922/ejprd.v34i7s.1636

European Journal of Prosthodontics and Restorative Dentistry (2026) 34(7s), 433–438

Disappearing Umbilical Cord, Disrupted Body Wall: Late Sonographic Diagnosis of Body Stalk Anomaly—A Case Report and Literature Review

Abstract

Background: Body stalk anomaly (BSA), also known as limb-body wall complex (LBWC), is an extremely rare and almost uniformly lethal disruption of early embryonic folding. Because of its lethality, the central clinical challenge lies in early identification and accurate diagnosis, which are essential for appropriate antenatal counselling and management.

Case Presentation: We report a 27-week gestation in a 2nd-gravida woman referred for a suspected fetal abdominal wall defect detected on antenatal ultrasound. Sonography demonstrated a large ventral wall defect with herniation of abdominal contents, ventriculomegaly, kyphoscoliosis, and an abnormally short umbilical cord with direct placental attachment, consistent with body stalk anomaly. Because the condition was incompatible with extrauterine survival, the pregnancy was terminated. The male neonate, born vaginally, weighed 1000 g with an Apgar score of 1/0 and showed a giant closed omphalocele, spina bifida, anorectal malformation, low-set ears, and bilateral congenital talipes equinovarus (CTEV); the herniated viscera remained covered by amniotic membrane and were directly continuous with the placenta.

Conclusion: Body stalk anomaly must be distinguished early from other, more survivable, anterior abdominal wall defects such as gastroschisis and omphalocele, because its prognosis and counselling implications differ substantially. First-trimester ultrasound remains the cornerstone of diagnosis, enabling timely, informed decision-making for affected families.

INTRODUCTION

Body stalk anomaly (BSA), also termed limb-body wall complex (LBWC) or body stalk syndrome, is an exceptionally rare and severe congenital disruption sequence first systematically described by Van Allen and colleagues in 1987.1 Estimates of its prevalence vary widely depending on whether early pregnancy losses are captured: figures range from 1 in 14,000 to 1 in 42,000 pregnancies overall, rising to approximately 1 in 7,500 fetuses examined sonographically between 10 and 14 weeks of gestation, while the prevalence among registered live and stillbirths is far lower, at roughly 0.12 to 0.32 per 10,000 births.2,3

BSA results from a profound failure of normal embryonic folding during the fourth and fifth weeks of gestation, when four contiguous body folds—cephalic, caudal, and two lateral—normally transform the flat trilaminar embryonic disc into a cylindrical fetus while simultaneously separating the intra- and extra-embryonic coelomic cavities and forming the umbilical cord.4 When this process fails, abdominal and/or thoracic viscera remain outside the body cavity, tethered directly to the placenta by a short or absent umbilical cord.4

Several classification schemes have been proposed to bring order to this heterogeneous condition. Van Allen's original criteria require at least two of three features—craniofacial defects (exencephaly/encephalocele with facial clefting), thoraco-abdominoschisis, and limb defects—for the diagnosis.1

Because BSA is invariably or near-invariably fatal, and because its sonographic appearance overlaps with several more survivable abdominal wall defects, early and accurate prenatal diagnosis is essential, both to avoid futile intervention and to allow parents time for informed, supported decision-making.5,8 We report a case of body stalk anomaly diagnosed by antenatal ultrasonography in the third trimester after late presentation, confirmed postnatally, and discuss it in the context of the current diagnostic and counselling literature.

METHODS

This is a descriptive, retrospective single-case study based on secondary data extracted from the patient's medical records, antenatal ultrasound archive, and delivery and neonatal documentation at Dr. Soetomo Academic General Hospital, Surabaya, Indonesia, following the patient's referral from a secondary-level hospital. The case report was prepared in accordance with the CARE (CAse REport) guidelines for the structured reporting of clinical cases.9 All identifying information, including the patient's name, date of birth, and medical record number, has been removed from the clinical images. Written informed consent for the use of clinical data and images for publication was obtained from the patient; this study was conducted in accordance with the Declaration of Helsinki and did not require separate ethics committee approval as it concerns the retrospective description of routine clinical care of a single patient.

CASE PRESENTATION

A 27-year-old woman, gravida 2 para 1 (G2P1001), at 27 weeks of gestation by last menstrual period, was referred to Dr. Soetomo Academic General Hospital from a secondary-level hospital, with a high-risk pregnancy attributed to a suspected fetal congenital abnormality. Her obstetric history included one previous caesarean delivery complicated by impending eclampsia; her menstrual cycles were otherwise regular. She had received six antenatal care visits at her local primary health centre and, between November and December 2023, attended the referral hospital's antenatal clinic three times, where she underwent laboratory testing and serial ultrasound examinations, was prescribed prenatal vitamins and low-dose aspirin (Aspilet 80 mg once daily), and was advised to continue regular follow-up.

Antenatal ultrasonography at referral demonstrated ventriculomegaly and a 6 cm cystic-solid mass, initially considered for sacrococcygeal teratoma with megacystis and an abnormal cyst as differential diagnoses; the fetal heart rate was normal. Subsequent detailed evaluation at our centre revealed findings consistent with a body stalk anomaly, including abdominoschisis and kyphosis of the fetal spine (Figures 1 and 2), in a fetus without chromosomal testing performed, following a previous caesarean delivery less than two years earlier. Because the constellation of findings was considered incompatible with extrauterine survival, the multidisciplinary team counselled the family and termination of pregnancy was carried out.

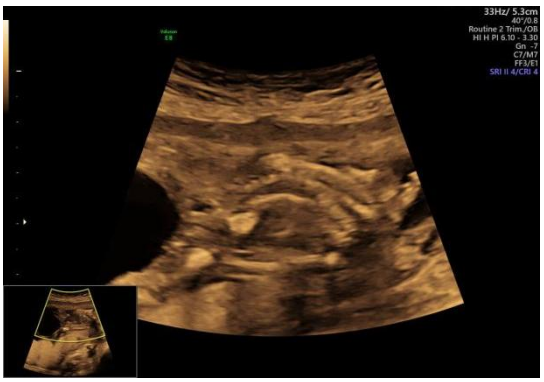

Figure 1. Antenatal ultrasound demonstrating severe kyphosis (over-curvature) of the fetal spine, arrowed, one of the diagnostic hallmarks of body stalk anomaly. (Identifying header information has been redacted.)

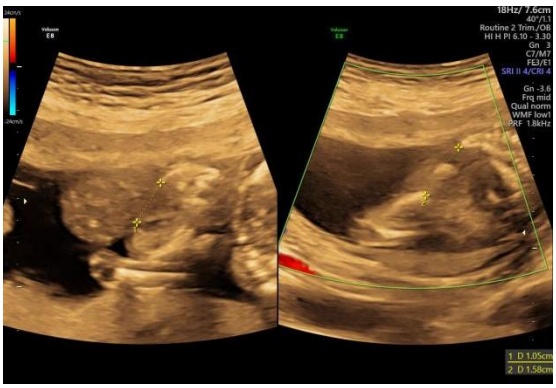

Figure 2. Antenatal ultrasound demonstrating a giant omphalocele, arrowed, with protrusion of the liver and gastrointestinal organs into the extra-coelomic space. (Identifying header information has been redacted.)

Delivery occurred vaginally. The male neonate had an Apgar score of 1/0 and a birth weight of 1000 g. On examination, multiple congenital anomalies were identified, including a giant, closed omphalocele, spina bifida, anorectal malformation, low-set ears, and bilateral congenital talipes equinovarus (CTEV) (Figures 4 and 5). The herniated abdominal organs remained enveloped by an intact amniotic membrane and were directly continuous with the placenta (Figure 3), a finding characteristic of the absent or rudimentary umbilical cord seen in body stalk anomaly.

Figure 3. Gross specimen of the placenta showing direct continuity between the placental mass and the herniated fetal viscera, consistent with an absent or rudimentary umbilical cord.

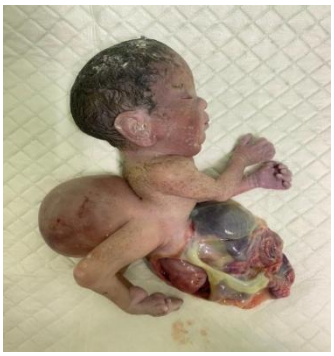

Figure 3. Gross specimen of the placenta showing direct continuity between the placental mass and the herniated fetal visceFigure 4. Stillborn male neonate showing a giant omphalocele with herniated viscera covered by amniotic membrane, in continuity with the placental massra, consistent with an absent or rudimentary umbilical cord.

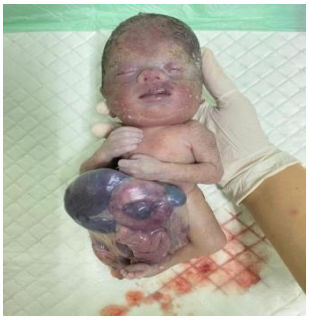

Figure 5. Clinical examination of the neonate demonstrating the body-wall and limb anomalies, including low-set ears and bilateral congenital talipes equinovarus, alongside the giant omphalocele.

DISCUSSION

Body stalk anomaly occurs in approximately 1 in 7,500 fetuses scanned between 10 and 14 weeks of gestation, a figure derived from a large multicentre series in London, and arises from a fundamental disruption of embryonic folding during the fourth to fifth gestational week.5,10 The resulting phenotype combines a frontal or lateral body wall defect with extra-coelomic protrusion of thoracic and/or abdominal organs, severe kyphoscoliosis, an absent or markedly shortened umbilical cord, and limb deformities.5

The pathogenesis remains incompletely understood. Leading hypotheses include early amnion rupture with mechanical tethering by amniotic bands, vascular disruption affecting the early embryonic circulation, and a primary defect of the germinal disc itself; a contribution from genes governing embryonic folding has also been postulated.2,10 BSA is classically subdivided into two principal phenotypes according to which body folds are most affected: a craniofacial (type I) phenotype with exencephaly or encephalocele and facial clefting, and a ventral wall (type II) phenotype dominated by thoraco-abdominoschisis.6 Building on anatomical work in a porcine model, Martín-Alguacil and Avedillo more recently proposed a refined classification incorporating up to eight subtypes, distinguished by the combination of spinal and umbilical cord defects, abdominoschisis versus thoraco-abdominoschisis, anal atresia or other visceral defects, and the presence of structural versus non-structural limb anomalies; this framework, while developed in animals, offers a useful anatomical vocabulary for describing the spectrum seen in human cases such as ours.7,11

Diagnosis rests principally on ultrasound, ideally performed at the end of the first trimester, when the diagnostic triad of an extra-coelomic location of fetal viscera, a short or absent umbilical cord, and severe kyphoscoliosis can already be demonstrated with first- or early second-trimester transabdominal or transvaginal sonography.5,12 In our case, the abnormality was not recognised until the third trimester, illustrating that BSA can occasionally escape detection on routine early scans, particularly outside tertiary centres, and underscoring the importance of a structured anatomical survey of the fetal body wall, spine, and cord insertion at every opportunity.13

Because several other ventral wall defects can mimic the sonographic appearance of BSA, a systematic differential diagnosis is essential (Table 1). Gastroschisis typically presents as a small, right-paraumbilical defect with a normal cord insertion and no covering membrane, and carries a comparatively favourable prognosis after surgical repair.10,14 Omphalocele produces a membrane-covered, midline defect of variable size with the cord inserting at the apex of the sac, and may be isolated or associated with chromosomal anomalies, again with a generally better outlook than BSA.10,14 Pentalogy of Cantrell combines a supraumbilical defect with a sternal cleft, ectopia cordis, and diaphragmatic/pericardial defects, while the OEIS complex (omphalocele, bladder or cloacal exstrophy, imperforate anus, and spinal defects) is distinguished by its infraumbilical location and urogenital involvement.10,14 The single most discriminating feature favouring BSA over these alternatives is the absent, rudimentary, or markedly shortened umbilical cord with viscera lying free within the extra-embryonic coelom rather than being neatly contained within a membranous sac with a normally inserted cord—precisely the configuration observed in our patient's placental specimen (Figure 3).10,14

Table 1. Differential diagnosis of fetal anterior abdominal wall defects

Condition	Abdominal wall defect	Umbilical cord	Associated findings	Prognosis
Body stalk anomaly (LBWC)	Large thoraco-/abdominoschisis with most viscera outside the coelom	Absent, rudimentary, or markedly short umbilical cord; direct fetoplacental fusion	Severe kyphoscoliosis; limb defects common	Uniformly lethal
Gastroschisis	Small, usually right-paraumbilical defect	Normal cord insertion, normal length	Spine usually normal; no membrane covering bowel	Generally good with surgical repair
Omphalocele	Variable-sized midline defect, membrane-covered	Cord inserts at apex of the sac	Spine usually normal; may coexist with chromosomal anomalies	Variable; depends on associated anomalies
Pentalogy of Cantrell	Supraumbilical defect with sternal cleft	Normal or mildly abnormal cord	Ectopia cordis, diaphragmatic and pericardial defects	Poor, but case-dependent
OEIS complex	Infraumbilical omphalocele	Variable	Bladder/cloacal exstrophy, imperforate anus, spinal defects	Survivable with staged surgery

Our patient's neonate additionally demonstrated spina bifida, anorectal malformation, low-set ears, and bilateral congenital talipes equinovarus—a combination of axial, urogenital, and limb anomalies that is well documented in

BSA case series and is thought to reflect the same primary folding defect rather than a separate, independently acquired anomaly.8,15 Comparable combinations of cranial, urogenital, and limb findings have been reported in other recent case series, reinforcing that the phenotype, although individually variable, recurrently clusters around defects of the body wall, spine, umbilical cord, and limbs.15,16 Notably, BSA is generally not associated with aneuploidy, and invasive genetic testing, when offered, is frequently declined by parents once the lethal prognosis has been explained, as the result is unlikely to alter management.13

Institutional experience with this anomaly remains limited even at tertiary referral centres: our hospital encountered only two prior cases of body stalk anomaly over the preceding five years, both managed by caesarean delivery, which is consistent with the rarity reported elsewhere in the literature.8,16 A useful comparison is a similar case managed at our institution a few months earlier, in a 35-year-old woman with one previous caesarean delivery, in whom body stalk anomaly coexisted with amniotic band sequence; because of a preterm gravid uterus with anterior corpus bulging and FIGO grade 1 placenta accreta, delivery was achieved by caesarean section with a modified uterine compression suture, illustrating that the mode of delivery in BSA must be individualised according to coexisting obstetric risk factors rather than dictated by the fetal anomaly alone.6,17

Once the omphalocele or thoraco-abdominal defect remains intact in utero, postnatal evaluation is generally deferred until after birth; however, when the abdominal wall defect is narrow relative to the size of the herniated viscera—for example, a defect under 1.5–2 cm containing a 5 cm protrusion—visceral incarceration and obstruction can occur, and urgent surgical ring release may be required even in a fetus whose overall prognosis remains lethal, in order to relieve acute distress.18 Because no curative treatment exists for the underlying anomaly and most affected neonates die within hours to days of birth, perinatal management is principally oriented around accurate diagnosis, multidisciplinary counselling, and supportive or comfort-focused care for the family, ideally provided by a coordinated team spanning maternal-fetal medicine, neonatology, clinical genetics, and palliative care.5,8,19 Early and accurate diagnosis therefore serves less a therapeutic purpose than a counselling one: it allows parents to make informed decisions about pregnancy continuation or termination, plan the circumstances of delivery, and access psychological and palliative support before birth rather than in the acute aftermath of an unexpected diagnosis.13,19

This report has the inherent limitations of a single-case description: chromosomal and molecular testing were not pursued, and detailed autopsy or histopathological correlation was not obtained, which limits the precision with which this case can be classified within emerging anatomical taxonomies of BSA.7,11 Nonetheless, the case adds to the relatively small body of published BSA experience from Southeast Asia and reinforces several recurrent themes from the international literature: the central diagnostic role of ultrasound, the importance of distinguishing BSA from its mimics, and the need for early, sensitive counselling once the diagnosis is suspected.

CONCLUSION

Body stalk anomaly is a rare, sporadic, and almost invariably fatal disruption of embryonic folding that must be carefully distinguished from gastroschisis, omphalocele, and other ventral wall defects, given the markedly different prognosis and counselling implications of each. The hallmark combination of a large body wall defect, severe kyphoscoliosis, and an absent or rudimentary umbilical cord directly continuous with the placenta—as demonstrated in this case—should prompt immediate consideration of BSA on antenatal ultrasound. Because no effective fetal therapy exists, the principal clinical value of early diagnosis lies in enabling timely, well-supported decision-making for affected families and coordinated multidisciplinary perinatal care.

DECLARATIONS

Ethics approval and consent to participate: Not applicable (retrospective single-case description of routine clinical care).

Consent for publication: Written informed consent for publication of clinical details and de-identified images was obtained from the patient. [Authors: please confirm and retain documentation prior to submission.]

Availability of data and materials: The de-identified data supporting this case report are available from the corresponding author upon reasonable request.

Competing interests: The authors declare that they have no competing interests.

Funding: This case report received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

Authors' contributions: YNN collected the clinical data and drafted the manuscript. NICM and MPW supervised clinical management, critically revised the manuscript, and approved the final version. All authors read and approved the final manuscript.

Acknowledgements: The authors thank the patient and her family, and the staff of the Department of Obstetrics and Gynaecology, Dr. Soetomo Academic General Hospital, for their care of this patient.

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