

Research Letter

Pathological characterization of a malformed umbilical cord associated with body stalk anomaly

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Accepted 20 November 2010

A 26-year-old primigravid woman was referred to the hospital at 18 weeks of gestation for genetic counseling because of structural abnormalities in the fetus. Prenatal ultrasound manifested a singleton fetus equivalent to 15 weeks of gestation, normal amount of amniotic fluid, normal skull and extremities, scoliosis, abdominal placental attachment, and a short umbilical cord. A diagnosis of body stalk anomaly was made. Amniocentesis revealed a karyotype of 46,XY. The pregnancy was subsequently terminated. An 89-g male fetus was delivered with severe kyphoscoliosis; a sternal defect; abdominal wall defects with eviscerated heart, stomach, liver, and intestines; and a 10-cm malformed umbilical cord attached to the placenta (Fig. 1). The skull, digits, limbs, external genitalia, and anus were normal, and no amniotic band could be found. The umbilical cord was covered by amnion and contained a single umbilical artery (Fig. 2). Pathological examination of the umbilical cord showed that the umbilical vessels over their entire length were embedded in the wall of an amniotic tunnel, and the residual connecting stalk connected the umbilical cord to the placenta (Fig. 3). The connecting stalk was covered on the side by the amniotic epithelium in continuous with that of the amniotic membrane and the other side facing the persistent extraembryonic coelomic cavity.

Body stalk anomaly has an incidence ranging from 1:14,000 to 1:42,000 pregnancies [1,2]. Among 106,727 singleton pregnancies with live fetuses at the 10–14-week scan, Daskalakis et al [3] found the diagnosis of body stalk anomaly in 14 cases (a prevalence of 1:7,500 pregnancies). Body stalk anomaly is characterized by abdominal wall defects, an absent or short umbilical cord, and sharp angulation of the spine but is not associated with craniofacial malformations or limb anomalies [4,5]. There is phenotypic overlap between body stalk anomaly and limb-body wall complex which is characterized with lateral body wall defects, limb reduction anomalies, and/or craniofacial defects [6–8]. Possible pathogenesis of body stalk anomaly includes maldevelopment of the body folds when a trilaminar embryo is transformed into a cylindrical embryo [4] and mechanical teratogenesis following rupture of the chorion or the yolk sac [9].

At 2 weeks postconception, the epiblastic cells differentiate into amnioblasts and form the amnion. At 3 weeks postconception, the amnion and chorion are separated by extraembryonic coelom. Afterward, the amniotic cavity expands rapidly, and the amnion is tightly apposed to the body stalk, and the extraembryonic coelomic cavity will reabsorb through rapid expansion of the amnion. At 12 weeks of gestation, the amnion fuses with the chorionic plate following completely replacing all the coelomic cavities. A normal umbilical cord is formed through compression of the connecting stalk and the yolk sac by the expanding amniotic cavity. The umbilical cord is finally covered by amnion along with the residual yolk sac [10].

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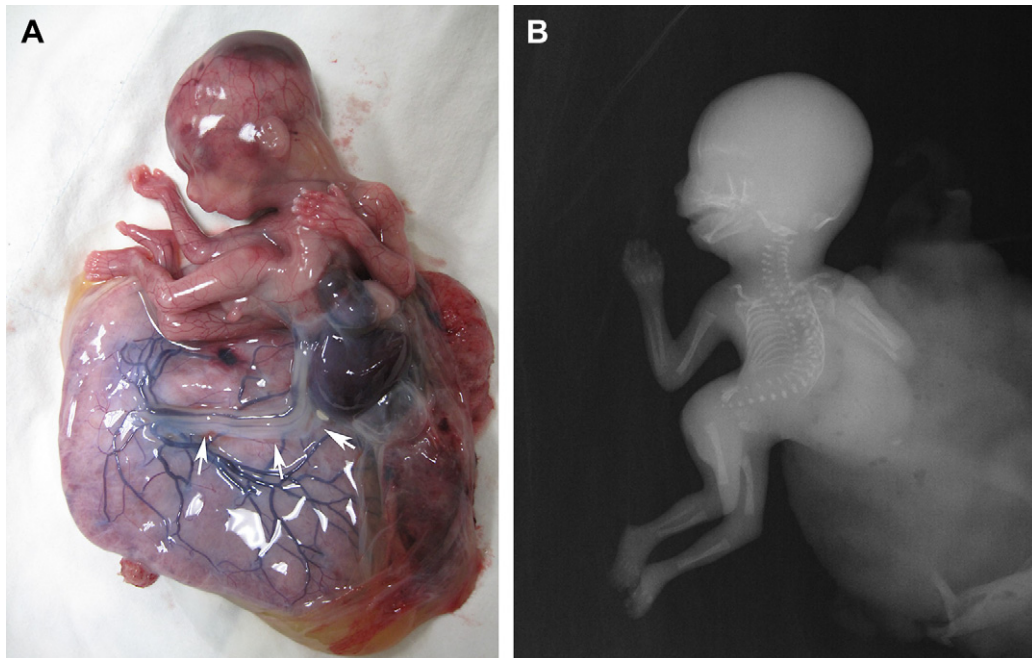


Fig. 1. (A) A fetus with body stalk anomaly and a malformed umbilical cord (arrows). (B) X-ray shows severe kyphoscoliosis.

The initiating event of limb-body wall complex and body stalk anomaly is likely to occur between the third and sixth weeks postconception before the fusion of amnion with chorion and consequently results in a malformed short umbilical cord or a nonexistent umbilical cord of which the

exerted traction force causes scoliosis in the fetus [10]. As shown in this presentation, the malformed umbilical cord is embedded and bunched by an amniotic tunnel and connects to the placenta via the persistent extraembryonic coelomic cavity [11].

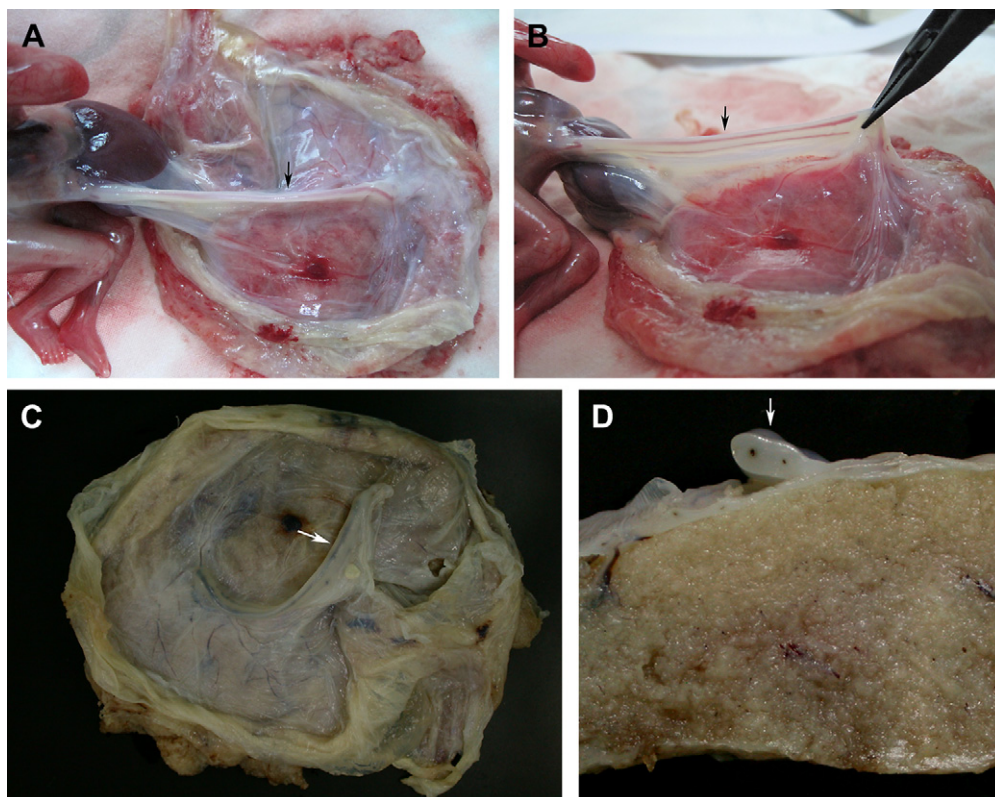


Fig. 2. (A–D) The umbilical cord (arrow) is malformed and contains two umbilical vessels that are bunched and embedded in an amniotic tunnel containing the connecting stalk.

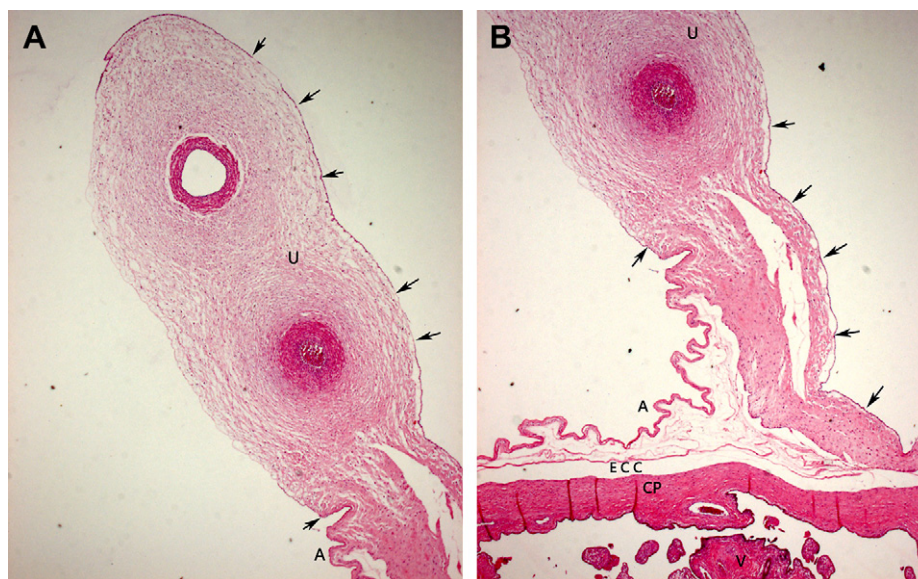


Fig. 3. (A and B) The umbilical vessels of umbilical cord remain in the connecting stalk, which was covered on one side by the amniotic epithelium (arrows) in continuous with that of the amniotic membrane and the other side facing the persistent extraembryonic coelomic cavity (Hematoxylin and eosin, 20 \times). A = amniotic membrane; CP = chorionic plate; ECC = extraembryonic coelomic cavity; U = umbilical cord; V = villi.

Acknowledgments

This work was supported by research grants NSC-96-2314-B-195-008-MY3 and NSC-97-2314-B-195-006-MY3 from the National Science Council and MMH-E-99004 from Mackay Memorial Hospital, Taipei, Taiwan.

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