Limb body wall complex/Body stalk anomaly – different spectrum of the same pathology?

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Objective
To analyse sonographic features of limb-body-wall complex (LBWC) with regard to different phenotypes proposed by Russo et al. (1997) and overlap with body stalk anomaly and amniotic band syndrome.

Methods
We conducted a retrospective study on fetuses with LBWC that were evaluated in our Ultrasound Department in a tertiary referral hospital in Poland between 1997 and 2015. Our electronic database was searched for cases concordant with the criteria proposed by van Allen et al. in 1997 (i.e. eversion, acrania/encephalocele with or without facial clefts, and limb anomalies). Cases were enrolled in the study if two out of three were present. Sonographic features as well as karyotypes were analysed. LBWC type 1 was diagnosed in the presence of acrania/encephalocele with or without facial clefts. Otherwise the fetus was categorized as having LBWC type 2. Body stalk anomaly was defined as severe thoracoabdominal wall defect along with kyphoscoliosis and a short or absent umbilical cord with or without limb anomalies. The study group consisted of 118 fetuses. The mean maternal age at the diagnosis was years 27.6 (SD 6.1 years). The median gestational age at the diagnosis was 15 weeks (Q1=13; Q3=21 wks).

Results
The majority of cases were categorized type 2 LBWC (70.3%; N=83). The most common sonographic finding was abdominoschisis (92.4%; N=109; comprising liver in 98 cases) followed by kyphoscoliosis (66.1%; N=78), limb anomalies (55.9%; N=66), myelomeningocele (22.0%; N=26), acrania (21.2%; N=25) and encephalocele (8.5%; N=10). In 33 fetuses there were other less common anomalies present. Severe thoracoabdominal wall defect with kyphoscoliosis was present in 78 fetuses (66.1%), however the umbilical cord was assessed as absent or short in 53 cases (67.9%; 53/78) and all features characteristic for body stalk anomaly were present in only 25 cases (32.0%; 25/78). Amniotic bands were seen in 6 cases overall (0.05%; 6/118).

Conclusion
To our knowledge this is the largest cohort of fetuses with LBWC reported up to date in the literature. LBWC is a heterogenous group of anomalies comprising body stalk anomaly with probably different pathophysiology.