

# Two Cases of the Prenatal Diagnosis of Body Stalk Complex

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## INTRODUCTION:

We present two cases of prenatal ultrasound diagnosis of body stalk anomalies (also known as limb body wall complex).

Body stalk anomalies are a group of massively disfiguring abdominal wall defects in which the abdominal organs lie outside of the abdominal cavity in a sac of amnioperitoneum with absence of or very small umbilical cord.

Various hypotheses proposed to explain the pathogenesis of limb body wall complex include early amniotic disruptions, embryonic dysplasia and vascular disruption in early pregnancy.

Body stalk anomalies occur in approximately 1/15 000- 30 000 births.

## CASE REPORT:

Ultrasound investigations in two pregnant women in 12th and 15th weeks of pregnancy show large thoracoabdominal wall defect with everted abdominal and thoracic organs, no-well-formed umbilical cord with severe kyphoscoliosis and limb anomalies. Both pregnancies were terminated.

Microarray analysis of fetus in case 1 was normal.

The next family rejected the prenatal diagnosis, but they were investigated because of habitual abortions without proven genetic cause.

The autopsy finding in fetus 2 shows large thoracoabdominal wall defect, prolapse of abdominal and thoracic organs, diaphragm defect, extreme left-sided kyphoscoliosis, amelia of right upper limb, shortening of the umbilical cord.

## RESULTS:

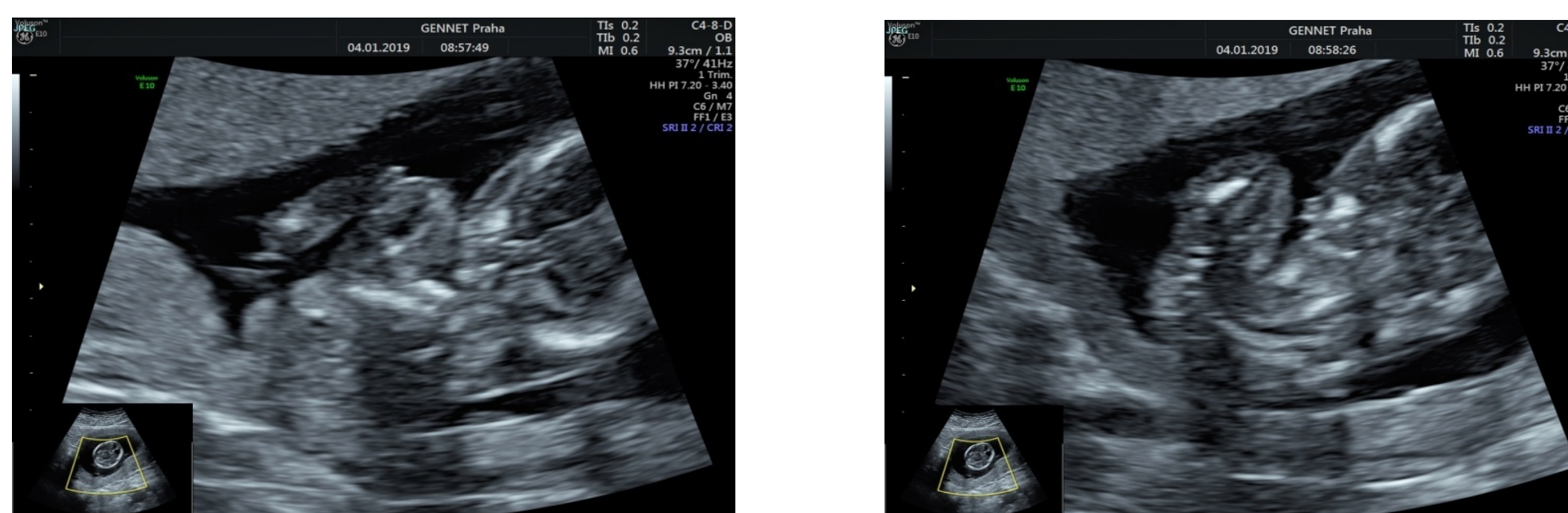
### Prenatal ultrasound - patient 1

Large thoracoabdominal wall defect, everted abdominal organs, severe kyphoscoliosis, agenesis a. umbilicalis.



### Prenatal ultrasound - patient 2

Amniotic band syndrome, large thoracoabdominal wall defect, everted abdominal and thoracic organs, severe kyphoscoliosis.



## CONCLUSIONS:

Body stalk anomalies are rare lethal polymalformative fetal syndrome. Our cases support the embryonal dysplasia theory of pathogenesis.

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