Objective
To present the prenatal ultrasound findings in a case of Pentalogy of Cantrell.

Methods
This is a case report.

Results
Pentalogy of Cantrell is a rare syndrome with a reported incidence of 5-15 per million births. Ultrasound (US) diagnosis is feasible from the first trimester and outcomes depend on severity, type and associated abnormalities with cardiac anomalies considered the most important prognostic predictor. A 25-year-old G2P0A1 woman with an unremarkable history, at 26th weeks’ gestation was referred to our hospital for suspected large abdominal wall defect with ectopia cordis and exomphalos. The maternal serological tests for toxoplasmosis, rubella, and cytomegalovirus were negative. At detailed US evaluation, a giant ectopia cordis and exomphalos with bowel and liver herniation, absent sternum and diaphragm were noted. Truncus arteriosus was also seen on fetal cardiac assessment. Because of the lethality of this findings, termination of pregnancy was offered and accepted.

Conclusion
Pentalogy of Cantrell is a rare syndrome presenting with five classic features: deficiency of the anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, congenital intracardiac abnormalities, and a defect of the lower sternum. Association of ectopia cordis with exomphalos is the best diagnostic clue of this syndrome. Most cases are sporadic. Chromosomal, cardiac, craniofacial and vertebral anomalies can be associated and genetic testing and fetal echocardiography are recommended. Differential diagnosis are isolated exomphalos, body stalk anomaly, isolated ectopia cordis, and amniotic band syndrome. Prognosis depends on severity of the lesions, and survival is correlated to the type and extent of the thoracoabdominal defects and associated cardiovascular malformations.