Isolated fetal ascites is a rare ultrasound finding and should always be considered an abnormal sign. It can occur due to chromosomal anomalies, intrauterine infections and structural anomalies and should be thoroughly investigated.

Cri du Chat (CdC) syndrome is a rare genetic disorder caused by variable size deletions of the short arm of chromosome 5 and has an incidence of approximately 1 in 45,000 liveborn infants. The most recognizable phenotype is characterized by a high-pitched cry, dysmorphic features, psychomotor and developmental impairment.

The aim of this study is to review the obstetric particularities of a case of CdC syndrome presenting with fetal ascites, performed in Divino Espírito Santo Hospital, Ponta Delgada, Azores, Portugal (HDES).

A 43-year-old woman, G4P3A0, blood group O Rh-positive, with no relevant personal or familial medical history, was referred to HDES due to maternal age.

1st trimester ultrasound
- 12 weeks + 3 days CRL 58.7 mm
- NT 0.8 mm
- Moderate fetal ascites

Non Invasive Prenatal Testing
- Low risk of trisomy 21, 18 and 13.

Infection diseases screening
- Negative for Syphilis, Toxoplasma gondii, Rubeolla, Herpes simplex, Cytomegalovirus, Parvovirus B19, HIV and HCV.

Amniocentesis
- 46,XX,del(5)(p15.2) Cri du Chat syndrome

After extensive counseling the couple opted for termination of the pregnancy, at 19WG.

Fetal Autopsy
- A female fetus weighing 209g was delivered without dysmorphic features.
- Fetal autopsy evidenced bilateral temporal lobe atrophy, coarctation of the aorta and extra-hepatic cyst.

Due to the variety of etiologies of fetal ascites and its poor prognosis, a systematic fetal medicine investigation should be followed to ensure reaching a proper diagnosis. Fetal autopsy is indicated in all the cases of fetal demise and abnormal ultrasound findings in order to find out the cause and make the complete diagnosis.