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Unveiling Fraser syndrome: a prenatal ultrasound case report of rare congenital anomalies

A Ghanem ; H Ben Ahmed ; A Masmoudi ; D Chelli

Introduction

Fraser syndrome is a rare autosomal recessive disorder characterized by a constellation of congenital malformations, often severe and life-threatening.

Case

We report the case of a 27-year-old gravida 2 woman with a history of neonatal death due to Fraser syndrome. The current pregnancy progressed without complications until the second-trimester morphological ultrasound at 24 weeks of amenorrhea, which revealed multiple severe anomalies: pericardial effusion, ascites, bilateral renal agenesis, and a cleft lip and palate. Given these findings suggestive of Fraser syndrome, a medical termination of pregnancy (MTP) was performed. The fetal karyotype was normal, ruling out major chromosomal aberrations and supporting the hypothesis of autosomal recessive inheritance.

Fetopathological examination confirmed the ultrasonographic anomalies and further identified additional hallmark features of Fraser syndrome.

The diagnosis of Fraser syndrome is established by the presence of at least three of the four major criteria: syndactyly, cryptophthalmos, renal anomaly, and familial history. In this case, the coexistence of these diagnostic hallmarks allowed confirmation of the syndrome.

Conclusion

This case underscores the critical role of prenatal ultrasonography in detecting severe malformative syndromes. In the presence of suggestive familial history, targeted ultrasonographic monitoring, fetal karyotyping, and genetic counseling are imperative for optimizing patient management and guiding medical decision-making.



Figure 1. the second term ultrasound



Figure 2: Fetopathological examination

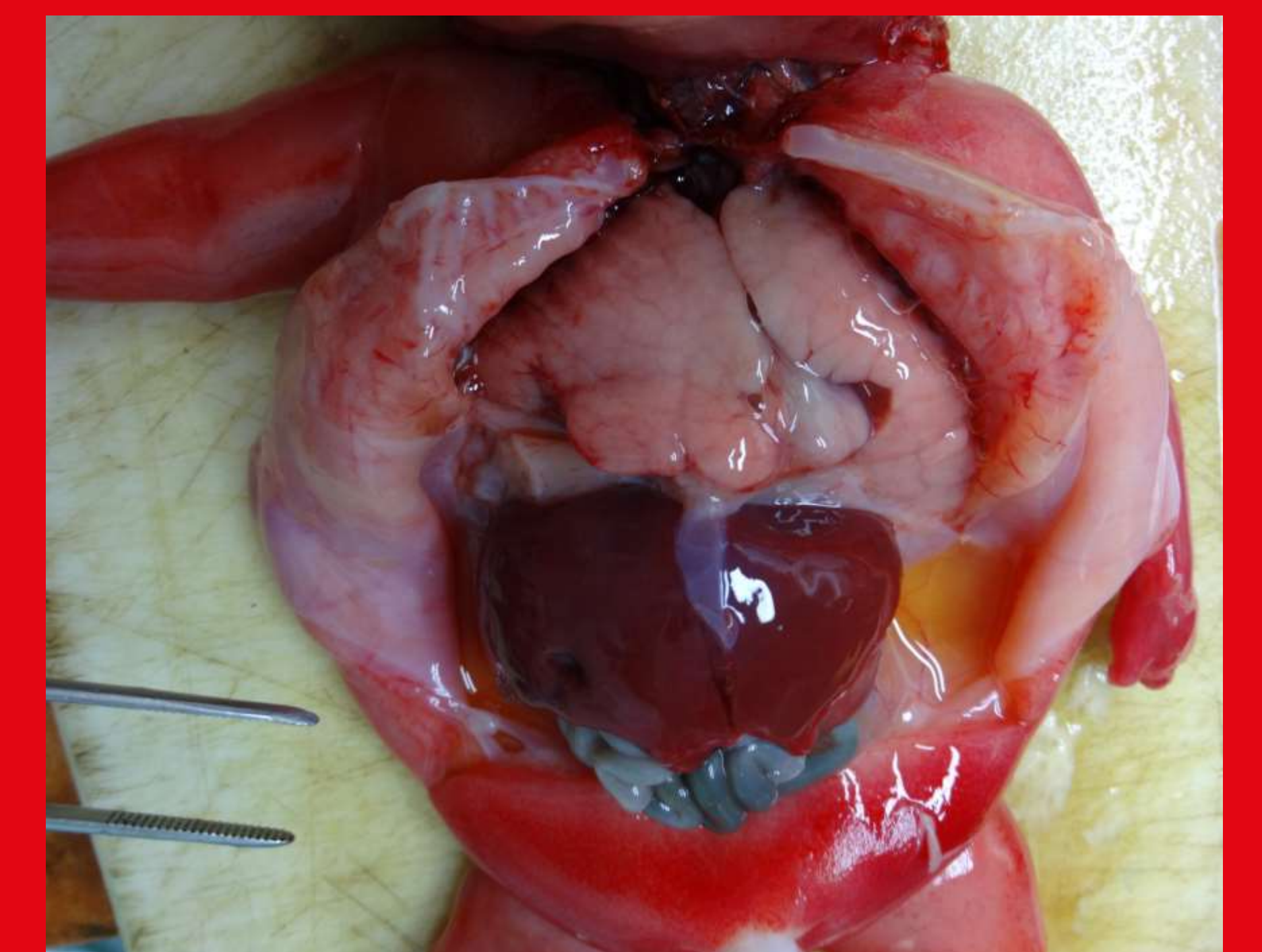


Figure 3: Fetopathological examination