In Utero Presentation of Cri-Du-Chat Syndrome

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Abstract

- Cri-du-Chat syndrome is a genetic disease caused by a partial deletion of the short arm of chromosome 5 characterized by a high-pitched cry and other phenotypic variations. This case represents an atypical constellation of ultrasound findings and genetic abnormalities with a guarded neonatal prognosis.
- This case emphasizes the association between fetal karyotypic abnormalities and multi-organ system complexities. Collaborative healthcare and effective patient communication play crucial roles in determining maternal and neonatal outcomes.

Background

- Cri-du-Chat is a rare inherited syndrome caused by a chromosome aberration with an incidence of 1 in 15,000 to 50,000 newborns [1]. Most cases are caused by de novo mutations in early embryonic development [2].
- Prenatal ultrasound findings may include cerebellar hypoplasia, cardiac abnormalities, ventriculomegaly and hydrops fetalis, though none are specific to this disease [3].
- Deletions of variable sizes of the short arm of chromosome 5 produce genotypic and phenotypic variations among patients.

Case

- A 36-year-old female, G4P1112 was referred to our Maternal-Fetal Medicine clinic at 25 weeks EGA for consultation regarding abnormal fetal heart views.
- Prenatal lab screening revealed negative NIPT, AFP, and carrier screening for cystic fibrosis. Ultrasound revealed cerebellar hypoplasia, mega cisterna magna and hypertrophic cardiomyopathy with SVT and a rate of 277 BPM (figure 1). Placentomegaly was appreciated.
- The patient was admitted to MUMC for treatment of fetal tachycardia. Pediatric cardiology consultants recommended oral digoxin and flecainide. Fetal ascites and a pericardial effusion (figure 2) were noted the following day.

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Case (continued)

- Amniocentesis microarray studies returned consistent with Cri-du-Chat syndrome.
- Patient left the hospital and cardiac medication treatment was interrupted for a few days. After readmission and re-initiation of digoxin and flecainide the ventricular tachycardia resolved. Ascites remained present (figure 3). The fetus was monitored as an outpatient for 20 days.
- At 32 weeks EGA, the patient ruptured membranes. Due to vaginal bleeding and transverse lie, a low transverse C-section was performed.
- The neonate weighed 1734 g with Apgar scores of 1,6, 7. Aggressive NICU cardiopulmonary support was required.

Figure 1. Fetal SVT, rate of 277 bpm. 25 weeks EGA

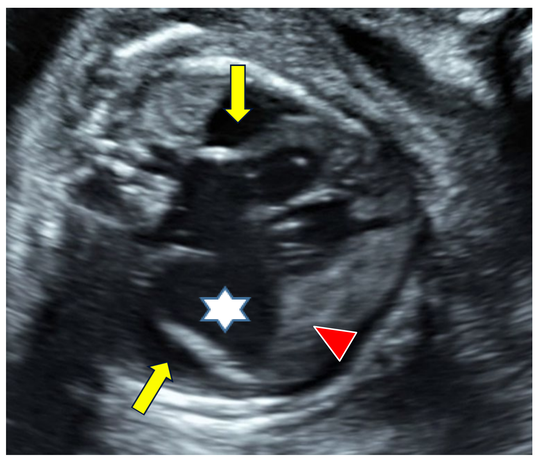

Figure 2. Fetal ultrasound at 28 weeks EGA. Dilated right atrium (✡), ventricular wall hypertrophy (▲), pericardial effusion (⇧)

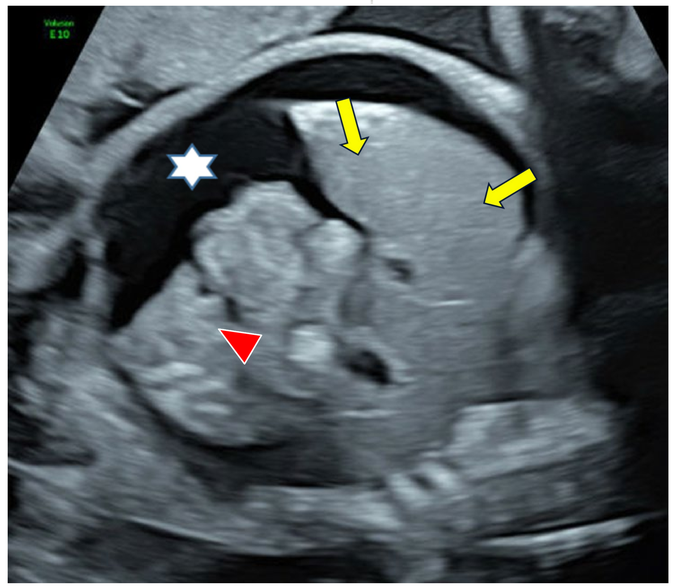

Figure 3. Hydrops fetalis at 28 weeks EGA. Ascites (✡), liver (⇧) and small intestine (▲).

Conclusion

- This case was initially diagnosed via microarray analysis from amniocentesis after observing several fetal structural on ultrasound. Collaborative healthcare and effective patient communication play crucial roles in determining outcomes for mothers and newborns. This case emphasizes the association between fetal karyotypic abnormalities and multi organ system complexities.

References

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