

EP15.99 | Prenatal diagnosis of Fraser syndrome by ultrasound

Key sonographic features and perinatal outcome

Dr. Amel Kamil • Dr. Nabilah Mashharawi • Dr. Shalini Fernandes

Latifa Hospital • Dubai, UAE

BACKGROUND

Fraser syndrome is a rare autosomal-recessive disorder characterized by cryptophthalmos, syndactyly and renal agenesis. Prenatal diagnosis is challenging because phenotypic expression varies, yet recognition is essential for parental counselling and perinatal planning.

CASE PRESENTATION

A multigravida with second-degree consanguinity was referred for a second-trimester anomaly scan after first-trimester nasal-bone hypoplasia.

Detailed ultrasound demonstrated bilateral enlarged hyperechoic lungs with mediastinal compression, suggesting congenital high airway obstruction syndrome (CHAOS). Additional findings included unilateral renal agenesis, bilateral clenched hands, micrognathia, low-set ears, hypertelorism and partial lens opacification.

DIAGNOSTIC WORK-UP

Fetal MRI confirmed CHAOS secondary to laryngeal atresia. Amniocentesis with chromosomal microarray was normal. Whole-genome sequencing identified a pathogenic homozygous variant in *FREM2*, confirming Fraser syndrome.

MANAGEMENT & OUTCOME

Following extensive counselling regarding the poor prognosis, the parents continued the pregnancy with planned ex-utero intrapartum treatment (EXIT). Despite successful tracheostomy at birth, the neonate died shortly after delivery due to profound hypoxia.

DISCUSSION

Fraser syndrome results from mutations in *FRAS1*, *FREM2* or *GRIP1*, disrupting epithelial–mesenchymal interactions. Perinatal mortality is high, primarily because of renal agenesis or laryngeal obstruction. Prenatal ultrasound is pivotal in identifying suggestive features.

KEY MESSAGE

The combination of CHAOS, renal anomalies and orbital findings should prompt suspicion of Fraser syndrome. Early recognition enables timely genetic testing, multidisciplinary planning and informed parental counselling.

PERINATAL PHENOTYPE



Postnatal appearance demonstrating craniofacial and limb abnormalities consistent with the prenatal diagnosis.