

THE CHALLENGES OF PRENATAL DIAGNOSIS

Regarding a clinical case

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We all know that the prenatal diagnosis is sometimes very challenging and not very conclusive. We present an intriguing clinical case, challenging for ultra-sonographers, paediatric cardiologists, radiologists, and neonatologists. A multidisciplinary team was needed for this demanding case with a difficult diagnosis.

- 29-year-old
 - Pakistani
 - G2P1 (one child with craniosynostosis)
 - First cousins couple
- First Fetal Medicine appointment was at **31 weeks** of gestation, referred by Paediatric Cardiology.

OBSTETRIC ULTRASOUNDS

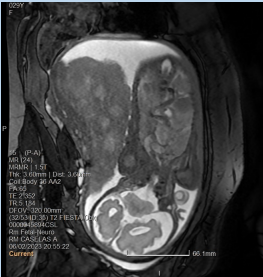
- First ultrasound at **17 weeks** with no prior screening or appointments
- **Multiple malformations** - cranio-encephalic abnormalities, limbs with long and thin bones with a different curved form, and hypospadias

FETAL ECHOCARDIOGRAMS

Cardiomegaly; Numerous vascular fistulas fed by a gigantic major aortopulmonary collateral artery (**MAPCA**) that arose from the aorta near the mesenteric artery, with hemodynamic consequences and elevated risk of high output **heart failure** with **pulmonary hypertension** immediately after birth.

FETAL MRI OF THE BRAIN

Deficient Sylvian fissure operculization, enlargement of subarachnoid space in supra and infratentorial regions, and a larger foramen magnum due to the coexistence of agenesis of the posterior arch of the atlas. Further MRI study of the cardiac abnormalities was inconclusive.



Multiple malformations were confirmed

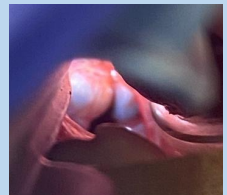


1. Fetal MRI showing a MAPCA

A persistent non-reassuring fetal heart tracing led to a C-Section



36 weeks and 6 days
2897g
Apgar Score: 1/5/7



Intubation was difficulted by a distortion in the airway structure and by a **pulsatile violaceous mass**. Death was declared nearly 2 hours after birth. Autopsy and blood samples were collected for further investigation; however, the parents did not consent to it.

It would have been crucial that an autopsy be performed to have a better understanding of this case. Unfortunately, post-mortem studies were not accepted by the couple because of religious issues. Honouring the parents' wishes is imperative. However, for effective genetic counselling of future pregnancies, as well as for the evolution of doctors' knowledge, it is very important to study these cases in full detail. As such, efforts should be made to sensitize parents to the importance of these studies.