Congenital high airways obstruction syndrome (CHAOS): Case report of two women without classical ultrasound finding

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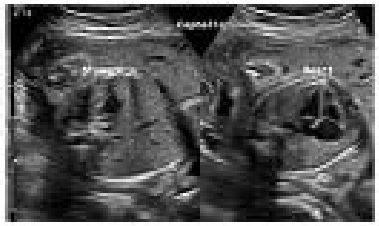
Abstract:

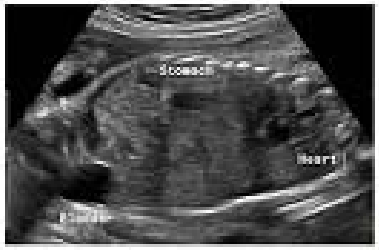
Congenital high airways obstruction syndrome (CHAOS) is a rare fetal anomaly. The underlying cause is congenital complete or incomplete obstruction of fetal upper airway tracts during embryogenesis, resulting in a spectrum of characteristic radiological feature diagnosed prenatally by ultrasound at time of routine anatomy scan. In many cases the diagnosis will be delayed to postnatal period at time of delivery when the newborn fail to open his/ her airway immediately after delivery.

We report our experience of two cases of CHAOS diagnosed only in the postpartum examination when there were no characteristic findings on detailed ultrasound examination.

Case 1

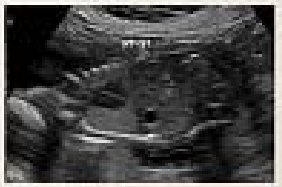
A 24-years old, was referred early at 11 weeks for dating ultrasound scan. During her antenatal follow up, the fetus was diagnosed to has absent right kidney, dilated bowel loop and he developed fetal growth restriction(FGR).Cesarean section was performed at 37 weeks' gestation for breech presentation and FGR. The baby delivered alive, has dysmorphic features with abnormal micro otia, micrognathesia and high arch palate and bifid tongue, trial of intubation failed as no laryngeal opening on examination. Infant died post CPR for 38 minutes after birth.
Neonatal whole exome sequencing revealed, FRAS1 gene with variant of unknown significance, the clinical features of the newborn is revise be medical genetics and they strongly suspecting Fraser syndrome (FRAS1 gene).

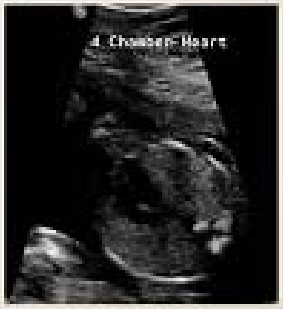




Case 2

A 33-years old was referred early at 15 week and anatomy scan was normal apart from echogenic lungs but normal cardiac axis, mediastinum and diaphragm curvature. Growth scan was done at 34 weeks and showed polyhydramnios with AFI of 38cm. At time of delivery the baby was not crying and NICU team failed to intubate hem, bronchoscopy examination showed subglottic laryngeal atresia, tracheostomy was done and the baby died after 30 min of CPR. Neonatal blood was obtained for whole exome sequencing and the results showed that the baby was homozygous for (FRAS1) gene ch.4 c.2917T>G, p. Cys 973 Gly. The result was reported as a variant of unknown significance.





Conclusion:

The current diagnostic criteria are not applicable for all the cases and physicians need to have high index of suspicion for the diagnosis when the lungs got to be echogenic in the ultrasound.

CHAOS can be part of FRASER syndrome but we are reporting a new variant that has not been identified to be of clinical significance till the mean time.