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Severe Ventriculomegaly; A Diagnostic and Prognostic Conundrum

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Background

- Ventriculomegaly is a common feature found sonographically in fetal life that has been associated with a poor outcome.
- Severe ventriculomegaly is defined as lateral ventricles measuring ≥15mm.
- Studies on pregnancy outcomes with isolated ventriculomegaly suggests that 80% of fetuses will survive, of which 40%have subsequent normal neurodevelopment.
- However, given many cases diagnosed at the time of morphology undergo termination, there is limited data on progression available to guide counselling for these patients, particularly if other abnormalities are present.

Case

- A 38 year old G3P1 low risk IVF pregnancy, presented at 19 weeks with sonographic findingsof bilateral severe ventriculomegaly (Right 12.5mm, Left 13-14mm) and communication of the anterior horns ultrasound. Head circumference >97%
- She had a low probability NIPT (Non-Invasive Prenatal Test).
- An MRI suggested severe hydrocephalus (Image 2) rather than holoprosencephaly spectrum. She had an amniocentesis and microarray which was normal, female fetus.
- Following extensive counselling on poor prognosis and outcomes, with a multidisciplinary team (MFM, Genetics, Paediatric Neurology, Neonates and Palliative care), a decision was made to continue the pregnancy.
- Repeat US and MRI in late third trimester (35/40) showed normalisation of the ventricles (Image 3), but hypoplastic frontal horns and hypotelorism suggestive of lobar holoprosencephaly (Image4).
- Patient had an uncomplicated vaginal delivery.
- Postnatal MRI confirmed appearances consistent with lobar holoprosencephaly with optic nerve hypoplasia, with poor long term neurodevelopmental prognosis.

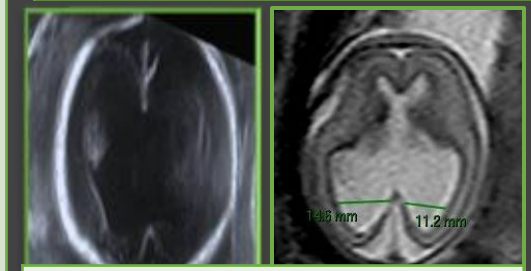

Image 1+2 (22 Weeks)

Severe bilateral ventriculomegaly with communication of the anterior horns of lateral ventricles

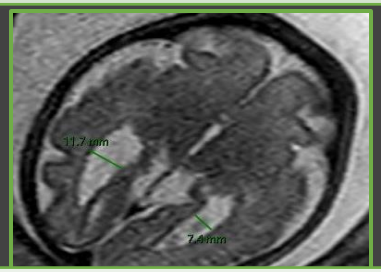

Image 3 (35 Weeks)

Mild left lateral ventriculomegaly, absent pellucidum, fusion of the anterior and inferior aspects of the frontal lobes

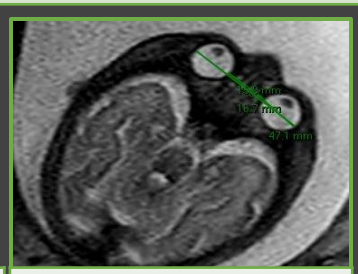

Image 4 (35 Weeks)

Hypoplastic frontal horns and hypotelorism

Discussion

- Holoprosencephaly occurs in 1:1300 fetuses at 12/40, but only 1:10,000 births.
- It arises due to midbrain cleavage abnormalities, and is a key differential for severe ventriculomegaly, particularly if anterior horn fusion is identified.
- In this case, the initial MRI was not suggestive of a holoprosencephaly spectrum, but as the pregnancy progressed it became more obvious.
- This case highlights the utility of both MRI and US in the assessment of severe ventriculomegaly.
- Long-term follow up of the baby will assist with future prognostic counselling for similar patients.

References

- C.Scala et al. Perinatal and long-term outcomes in fetuses diagnosed with isolated unilateral ventriculomegaly: systematic review and meta-analysis; ISUOG APRIL 2016.
- Solomon BD, Gropman A & Muenke M. Holoprosencephaly Overview. GeneReviews. August 29, 2013]