**EP09.17 Prenatal ultrasound findings of an occipital encephalocele with an underlying duplication of chromosome 7q36.3 including the sonic hedgehog gene (SHH).**

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**Introduction**  
Encephaloceles are classified as neural tube defects. Chromosome duplications involving band 7q36.3 specifically are rare. The sonic hedgehog gene (SHH) at this locus is critical for neural tube patterning. Abnormal SHH signalling can result in abnormal brain development.

**Case**  
An 18year old NZ European mother presented for her routine 20 week anatomy ultrasound scan at 22+1 weeks. There was an underlying history of multiple miscarriages exacerbating maternal depression and anxiety. A fetal encephalocele including an abnormal posterior fossa were identified as an isolated finding. Amniocentesis at 24/40 weeks. Vaginal delivery at 39/40 weeks.

**Management**  
Successful surgical excision of encephalocele at birth. Ventriculomegaly ventriculoperitoneal shunt insertion. Abnormal brainstem, cerebellum and 4th ventricle: tectocerebellar dysraphism  
Ongoing neonatologist and paediatric assessments of significant developmental delays, IQ and development of large motor skills long term.

**Discussion**  
Full karyotype and microarray analysis revealed a de novo 7q36.3 duplication involving the SHH gene. A 7q36.3 duplication including SHH, associated with an occipital encephalocele is reported only once previously. Evidence suggests the duplication, may be the cause for the encephalocele. To understand further it is important to determine what proportion of occipital encephaloceles do in fact have this underlying genetic lesion.

**Conclusion**  
This is a unique case of a happy smiling now 2 year old female with a repaired encephalocele, likely due to a 7q36.3 duplication involving the SHH gene, that we are able to follow developmentally.

**References**