Infant with cutis aplasia congenita, encephalocele and syntelencephaly: a case report

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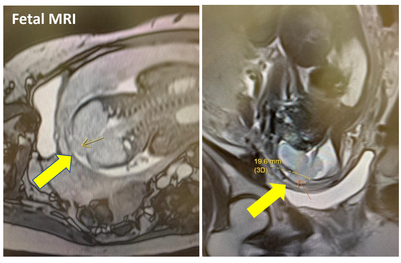
Background

Syntelencephaly is a rare variant of holoprosencephaly, both characterized by abnormal separation of midline structures but with different embryopathogenesis, resulting in different clinical presentations. We report a 3-month old girl with features of syntelencephaly.

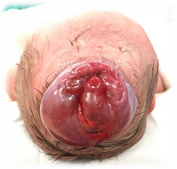
Case summary

- At 1 month gestation, a baby girl was incidentally noted to have a skull defect in antenatal structural ultrasound
- Fetal MRI: vertex encephalocele
- She is the first child of a Chinese-Polish couple. The couple decided to continue the pregnancy due to religious beliefs
- At 37 weeks gestation, delivered by emergency Caesarean section due to premature rupture of membranes



Neonatal physical examination

- 4.5cm cutis aplasia congenita with encephalocele and active CSF leak
- Spontaneous breathing maintained with active limb movements
- Fetal weight 2287g (5th percentile), Head circumference <3rd percentile
- APGAR score 9/10

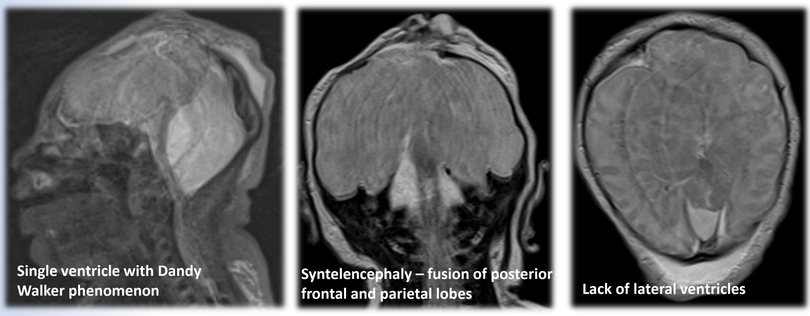


Management

- Emergency repair of encephalocele
- Necrotic brain tissue excision
- Dura and scalp wound closure

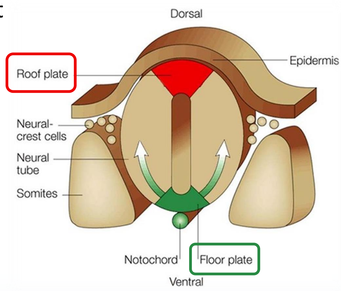
Post-natal MRI

- Failure of separation of posterior frontal and parietal lobes and deficient supratentorial midline structures, confirming diagnosis of Syntelencephaly.



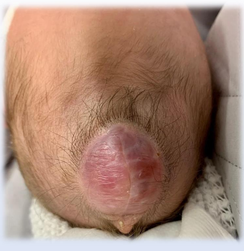
Embryopathogenic mechanism

- Holoprosencephaly (HPE) is characterized by incomplete separation of cerebral hemispheres and deep brain structures
- Classic HPE results from a defect in embryonic **floor plate** (ventral)
- Syntelencephaly is a variant of HPE, it results from a defect in **roof plate** (dorsal)[1]
  - Causing failure of separation of posterior frontal and parietal lobe
  - Caused by mutation of ZIC2 gene[2], located in dorsal telencephalon, while other genes identified in HPE are expressed predominantly in ventral regions
  - Less severe craniofacial anomalies
  - Spasticity and seizures are the most common clinical manifestations



Progress at 3 months old

- Living at home with parents, on Ryle's tube feeding
- Spontaneous limb movement with good tone
- Normal breathing
- No clinical seizure
- Body weight and length 25-50th percentile, head circumference <3rd percentile[3]



Discussion

- The decision to operate was made in view of the baby's ability to maintain spontaneous breathing with active limb movements
- After repair of open encephalocele, close monitoring of head circumference and ventricular size are important to detect hydrocephalus

Conclusion

Syntelencephaly, being a rare variant of HPE, has characteristic radiological findings that can aid early diagnosis and prognostication.

References

1. Rajalakshmi PP, Gadodia A, Priyatharshini P. Middle interhemispheric variant of holoprosencephaly: A rare midline malformation. *J Pediatr Neurosci*. 2015;10(3).
2. Brown LY, Odent S, David V, Blayau M, Dubourg C, Apacik C, et al. Holoprosencephaly due to mutations in ZIC2: Alanine tract expansion mutations may be caused by parental somatic recombination. Hum Mol Genet. 2001;10.
3. Fok TF, So HK, Wong E, et al. Updated gestational age specific birth weight, crown-heel length, and head circumference of Chinese newborns. *Arch Dis Child Fetal Neonatal Ed*. 2003;88(3).