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Background:

Prenatal diagnosis of Sotos syndrome is difficult. While CNS anomalies are reported, cortical features remain unclear. This study explores its prenatal neuroimaging phenotype.

Methods:

Retrospective study
4 fetuses Sotos syndrome
 (NSD1 variant, WES)

- Neurosonography
- Detailed US
- MRI

Conclusions

Novel neuroimaging features in Sotos syndrome are described, potentially aiding prenatal diagnosis. Further validation is required.

Results:

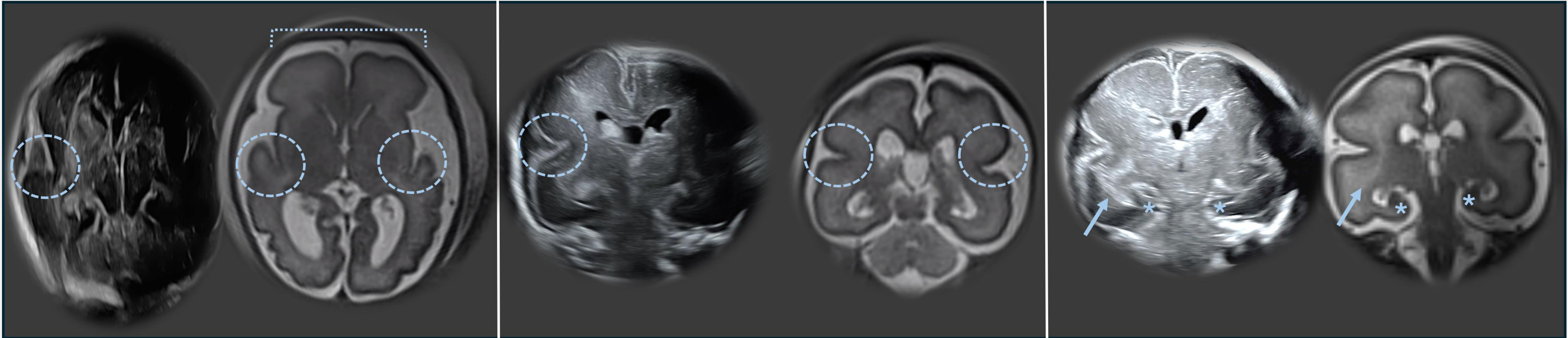
CNS “classic”

- Head overgrowth (4/4)
- Increased subarachnoid space (4/4)
- Mild ventriculomegaly (2/4)
- Corpus callosum anomalies (2/4)

Extra-CNS

- Renal anomalies (4/4)
- LGA (2/4), SGA (2/4)
- Pointed chin (4/4)
- ARSA (1)
- No polyhydramnios

CNS “New” Phenotyping



Posterior opercular shape with “pyramidal or pointed” (circle) # Incomplete hippocampal rotation (narrow)

Squared frontal lobes

WM hiperintensity (*)