

Detection of Intracranial Anomalies on *in utero* MRI in Ultrasound Diagnosed Agenesis of Corpus Callosum

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Objectives

Agenesis of the corpus callosum (ACC) in isolation is associated with neurodevelopmental problems in around 25% of cases. ACC particularly if associated with additional abnormalities may be associated with pathogenic genomic changes in over 30% of cases. Therefore detection of ACC and confirmation as to whether it is isolated or associated with anomalies is key to aid antenatal counselling.

In utero Magnetic Resonance Imaging (iuMRI) has been shown to detect additional structural anomalies in around 10% of cases of ACC.

This study aimed to establish the incidence of additional findings in fetuses with ultrasound (USS) suspected ACC following assessment in a tertiary fetal medicine centre.

Study Design & Methods

Study Design: Retrospective cohort study

Methods: All fetal brain iuMRI studies performed for ultrasound suspected isolated ACC between January 2019 to end December 2020 at request of a tertiary fetal medicine unit were included in the study. 17 cases had suspected isolated ACC on USS; in 3 cases further neuro-anatomical complications were suspected on USS.

Results

Twenty fetal iuMRI studies were performed for suspected ACC on USS between January 2019 to December 2020.

Median gestation at MRI: 24 weeks (Range: 19 to 34)
Median maternal age: 31 years (Range: 19 to 43)

The corpus callosum was found to be intact in 2 cases on iuMRI. ACC was confirmed in the remaining 18 cases (including 1 partial absence of ACC and 1 probable absence with quality degraded by artefact).

In 2 cases of USS suspected isolated ACC 2/15 (13.3%), further anomalies were found on iuMRI imaging: 1 fetus with septo-optic dysplasia and a fetus with a posterior cerebral artery territory infarction.

USS suspected ACC and additional abnormalities were confirmed in 2/3 (66%) cases: 1 fetus with a neuronal migration disorder and 1 case involving multiple anomalies including heterotopia, cerebellar hypoplasia and loss of the medial frontal lobe cortices.

Conclusions

In this study, USS has been found to be accurate at assessing for ACC in patients referred for fetal iuMRI, with 90% suspected ACC confirmed on iuMRI. Additional anomalies were found on iuMRI in over 10% of cases where isolated ACC was diagnosed USS. Our findings are broadly in line with recent studies. Where complex cases are suspected on USS, there is a high incidence of iuMRI confirmed anomalies. In cases of USS diagnosed ACC, dedicated neurosonography and iuMRI should be performed to establish the absence or presence of coexisting anomalies, given the implications on further genomic testing, prognosis and resultant antenatal counselling.

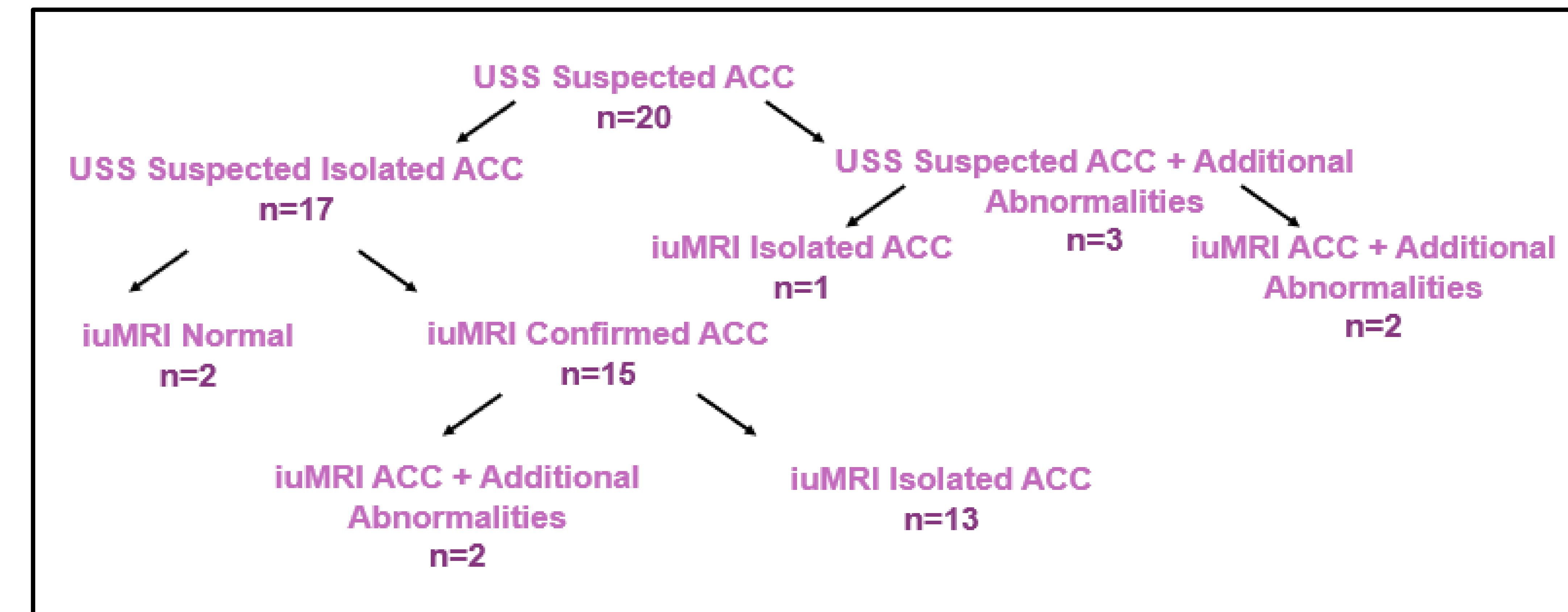


Figure 1 (above). Summary of findings between USS and iuMRI

Figure 2. Agensis of Corpus Callosum USS (left), Agensis of Corpus Callosum iuMRI (right)