

INTRODUCTION

Diencephalic–mesencephalic junction dysplasia (DMJD) is a rare and severe brain malformation, with early prenatal diagnosis remaining challenging despite advances in fetal neuroimaging.

OBJECTIVE

To report early ultrasound detection of type A DMJD in two male siblings with genetically confirmed L1CAM mutations.

MATERIALS AND METHODS

A retrospective neurosonographic review of two affected fetuses was performed, with genetic confirmation by whole-exome and Sanger sequencing.

RESULTS

In the first fetus, DMJD was diagnosed at 26 weeks, and the mother was identified as an X-linked carrier. In the second pregnancy, targeted neurosonography demonstrated features of type A DMJD as early as 14 weeks, including ventral clefing, thalamic fusion (diencephalosynapsis), dorsoventral midbrain elongation, and aqueductal narrowing.

CONCLUSION

Recognition of axial-plane markers enables early detection of L1CAM-associated DMJD in the early second trimester. This diagnosis at 14 weeks, among the earliest reported, highlights the value of targeted neurosonography for prenatal diagnosis and counseling.

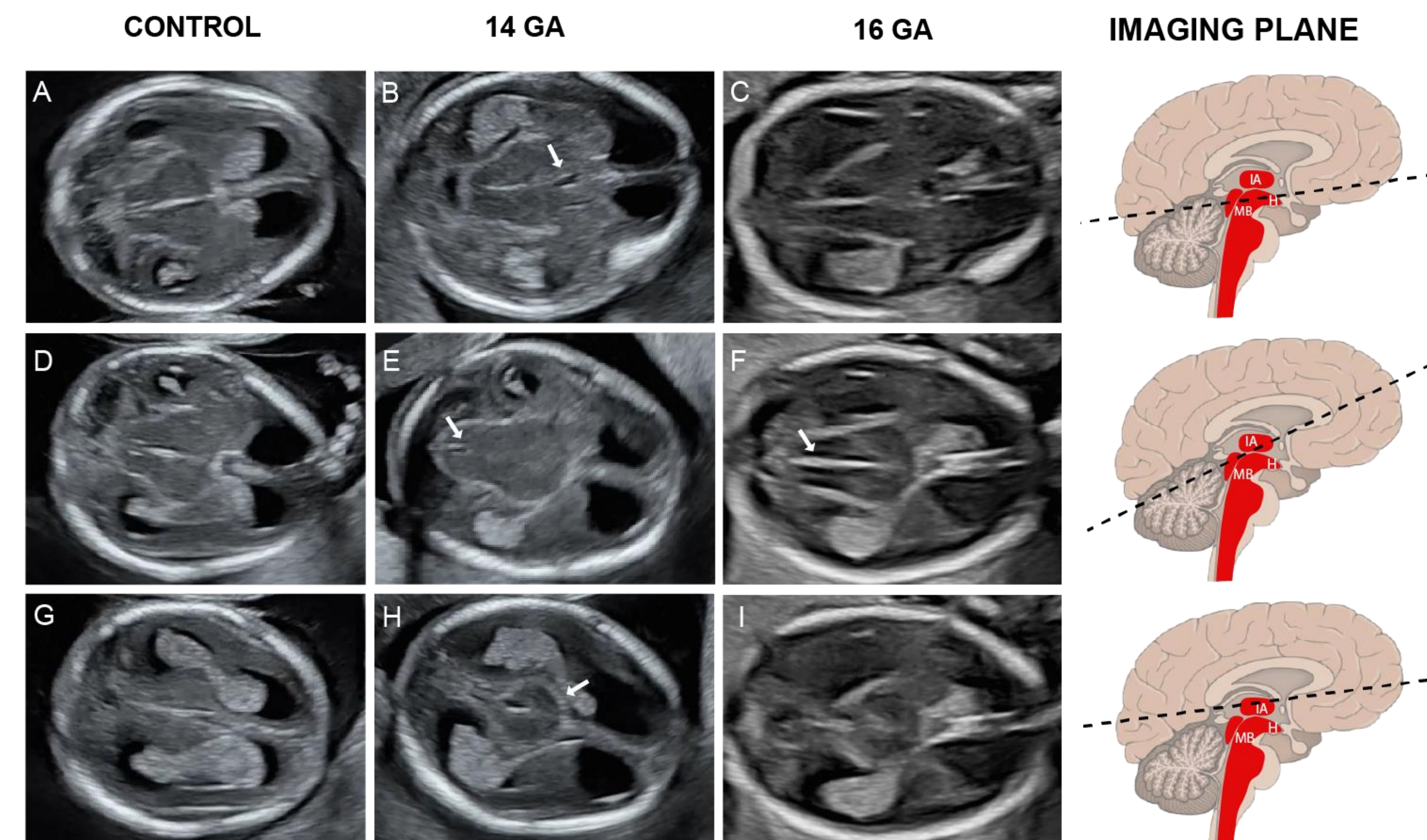


Figure: Comparison of axial-plane ultrasound images between a normal fetal brain (Control) and the brain of the second fetus with DMJD at 14 and 16 weeks of gestational age (GA).

Note: A-D-G: Control ultrasound images of a normal fetus at 14 weeks of gestation, B-C: “Butterfly sign” at the level of the lower thalami, with visualization of the midbrain on low axial planes; a subtle midline cleft is appreciable, E-F: Dorsoventral elongation of the midbrain with an abnormally narrowed aqueduct at the level of the thalami, H-I: Fused thalami at the level of the lateral ventricles with interruption of the interhemispheric fissure. The imaging directions on axial plane were revealed as the dash lines.