

Martha D. Bulus ¹, Pedro T. Castro ¹, Celso Hygino ¹, Tatiana Fazecas ¹, Ana Paula P. Matos ^{1,2}, Pedro Daltro ¹, Edward Araujo Junior ³, Heron Werner ^{1,2}

1. Biodesign Lab Dasa/PUC-Rio, Rio de Janeiro, Brazil

2. Universidade Federal do Rio de Janeiro, Rio de Janeiro, RJ, Brazil

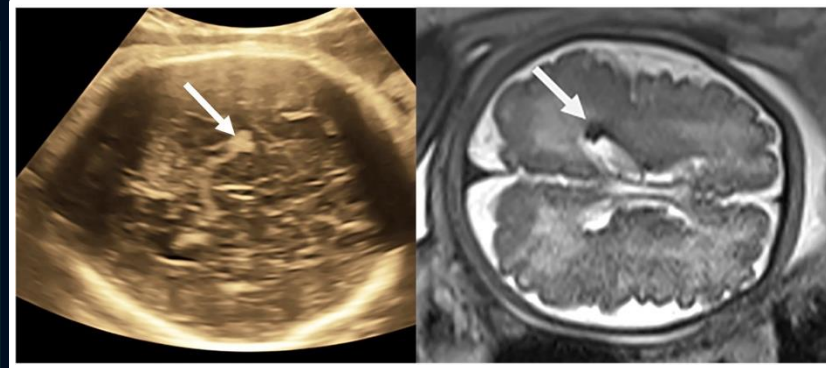
3. Universidade Federal de São Paulo, São Paulo, SP, Brazil

Objective: Tuberous sclerosis complex (TSC) is a multisystem neurocutaneous disorder caused by various genetic mutations. It can lead to tumor development in the heart, brain, kidneys, eyes, and lungs. Prenatal diagnosis is crucial for optimizing perinatal care and planning appropriate follow-up assessments. This study aims to describe the key lesions identified in fetuses diagnosed with TSC.

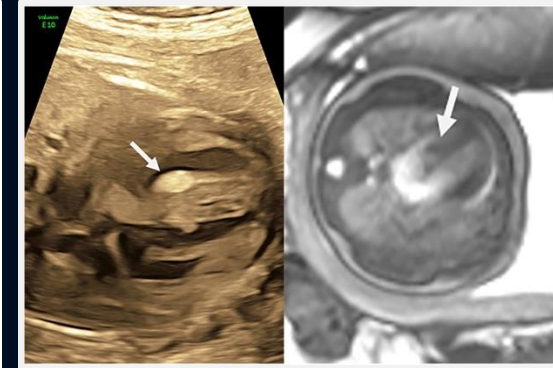
Methods: We conducted a retrospective analysis of TSC cases evaluated via ultrasound (US) and magnetic resonance imaging (MRI) between 2022 and 2024, in fetuses between 22 and 35 weeks of gestation.

Results: Postnatal confirmation was obtained for 3 cases of TSC. The primary US findings included cardiac rhabdomyomas and echogenic cerebral lesions. MRI was performed in the same cases, revealing characteristic rhabdomyomas. Additionally, MRI identified typical cerebral lesions, including subependymal and periventricular nodules.

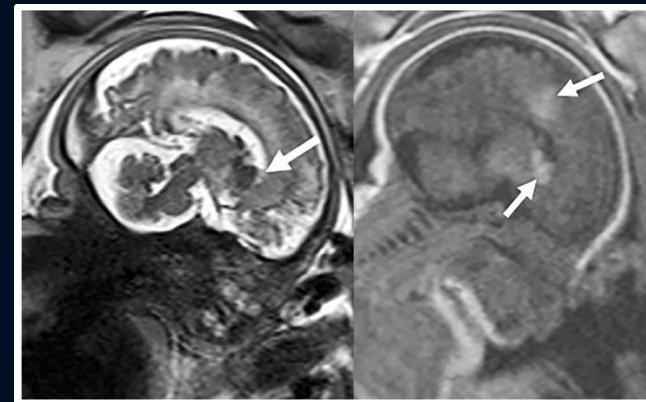
Conclusion: US remains the first-line screening tool for TSC, primarily detecting cardiac rhabdomyomas. MRI plays a complementary role in identifying cerebral lesions, particularly subependymal and periventricular nodules.



US shows brain cortical nodule. MRI (axial T2) showing a typical brain periventricular low-signal-intensity nodule (35 weeks) (arrows)



US and MRI image showing a cardiac rhabdomyoma in the interventricular septum (arrows)



Sagittal T2 (35 weeks) showing a typical brain subependymal low-signal-intensity nodule (arrow)

Sagittal T1 showing a brain subependymal and cortical high-signal-intensity nodule (arrows)



Coronal T2 showing a typical brain periventricular low-signal-intensity nodules (arrows)