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Clinical Image
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Prenatal Diagnosis of Tuberous Sclerosis Complex

Tonni G[1], Grisolia G[2], Pisello M[2] and Bergamini V[3]*

[1]Department of Obstetrics and Gynecology; Institutes of Hospitalization and Scientific Care, Italy

[2]Department of Obstetrics and Gynecology, Carlo Poma Hospital, Italy

[3]Department of Obstetrics and Gynecology, Verona Integrated University Hospital, Italy

Clinical Image

Here we describe a primigravida with a Dichorionic Diamniotic (DC/DA) twin pregnancy following ART (Assisted Reproduction Technology) that was referred at 32 weeks for a thorough ultrasound examination. The couple was not consanguineous and the mother had an uneventful clinical history unless of infertility. Ultrasound apparatus used was a Samsung WS80A™ (Samsung, Seoul, South Korea) equipped with a 2.5 MHz to 5.0 MHz transabdominal probe. The fetus showed hyperechogenic foci located at the level of cerebral cortex (Figure 1A) as well at the level of the myocardium (Figure 1B). No other lesions were seen at the level of the fetal skin, lungs and other apparatus. A prenatal diagnosis of Tuberous Sclerosis Complex (TSC) was posed. Genetic examination demonstrated a mutation in the *TSC2* gene at chromosome 16p13. TSC is inherited as an autosomal dominant condition with variable penetration and wide genotype-to-phenotype correlation; moreover a *de novo* mutation may be seen in almost half of cases.

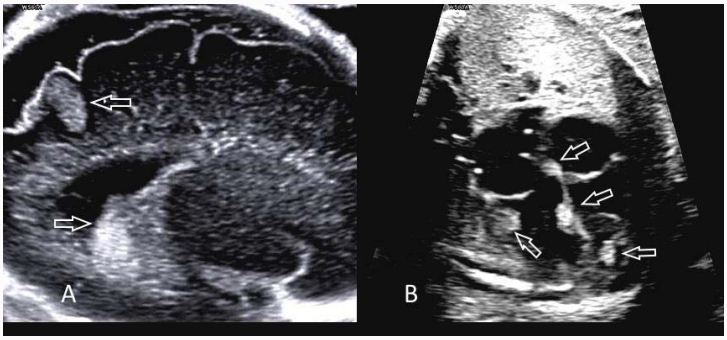

Figure 1: Prenatal transabdominal ultrasound performed at 32 weeks of gestation showing multiple tuberous involving either the fetal brain (open arrows) and located within the cortex and the cavum septum pellucidum in midsagittal plane. A) and the heart (myocardium, interventricular and interatrial septum) in a four-chamber view.

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Correspondence:
Gabriele Tonni, Department of Obstetrics and Gynecology, Institutes of Hospitalization and Scientific Care, AUSL di Reggio Emilia Via Amendola, 242100, Reggio Emilia, Italy, E-mail: tonnig@ausl.re.it

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