

Prenatal diagnosis of tuberous sclerosis complex: a case report

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DOI: 10.36129/jog.2022.S03

Objective. Tuberous sclerosis is a rare genetic condition that causes hamartomas to develop in different parts of the body. It has an autosomal dominant inheritance pattern with variable penetrance, and it can adversely affect maternal and fetal outcome.

Cardiac rhabdomyomas are an early manifestation of tuberous sclerosis. They can grow in size and number during fetal development, but they usually regress within the first years of life. However, the presence of a rhabdomyoma in the right atrium could lead to cardiac arrhythmias in both the antenatal and postnatal periods.

Materials and Methods. A 21-year-old primigravida performed an ultrasound at 20 + 2 gestational weeks and the fetus was diagnosed with an apical rhabdomyoma of the interventricular septum and a right atrial rhabdomyoma between the superior vena cava orifice and the posterior wall.

Results. A genetic examination was performed on the amniotic fluid revealing a paternal mutation for TSC1. The genetic investigations and the fetal ultrasound findings suggested the fetus was suffering from tuberous sclerosis. Therapeutic termination of pregnancy was performed at 21 + 1 weeks.

The pathological examination confirmed the presence of the rhabdomyomas.

Conclusions. Tuberous sclerosis occurs due to the deletion, rearrangement, and inactivating mutation of tumor suppressor genes TSC1 or TSC2 leading to the formation of hamartomas in many and different organs.

The early manifestation of tuberous sclerosis can be detected in prenatal screening and most commonly involves the heart and brain. The cardiac localization requires a careful evaluation of organ function in the postnatal period.