Multiple Cardiac Tumors in the Fetus

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An abnormal fetal heart rhythm was auscultated on routine examination in a 33-year-old mother whose past medical history was unremarkable.

A first echocardiogram at 22.5+2 weeks noted a thickened nuchal fold and a mild to moderate amount of pleural effusion and ascites. The fetus was found to have a slow heart rate of 110 bpm and episodes of bigeminy. Several echogenic masses were noted (Figure). The Doppler study confirmed that there was no obstruction to ventricular inflow or outflow or to the foramen ovale.

These masses appeared similar to the rhabdomyomas seen in the newborn, which are large, hyperechogenic, homogeneous, well delineated, often multiple, and sometimes associated with dysrhythmias.

Rhabdomyomas are hamartomatous proliferative lesions associated with tuberous sclerosis in 80% of cases. In this fetus, no cerebral or other abnormalities could be detected by echography. The situation was discussed with the patient and her husband, who indicated that they would prefer the pregnancy to continue. At 36 weeks gestation, a new examination showed polyhydramnios, ascites, and intrauterine fetal death. Fetal karyotype was normal. No immunological or infectious cause for the tumors was found.

The definitive diagnosis was confirmed at autopsy, which showed multiple macroscopic and microscopic rhabdomyomas in the myocardium, associated with multifocal cortical tubers in the brain, which is consistent with tuberous sclerosis. No cardiac flow obstruction was found at autopsy, and fetal polyhydramnios was thought to be secondary to transient severe cardiac dysrhythmia.

Tuberous sclerosis is most often due to sporadic mutations at 2 gene loci (TSC1 and TSC2). Because to date there is no prenatal diagnosis for these mutations, meticulous description of echocardiographic images is essential for counseling parents. Rhabdomyoma should be differentiated from other tumors and cardiac foci that have a very different prognosis.

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