*Fetal Echocardiogram is Useful for Screening Fetuses with a Family History of Cardiomyopathy*

Nicholas Zaban, MD (fellow), Robert Darragh, MD (attending), John Parent, MD (attending)

Level of training: fellow

Background:

Cardiomyopathy is highly heritable with familial forms accounting for 30-40% of cases. Cardiomyopathy can present in the fetus and be detected by fetal echocardiogram. Data on the utility of fetal echocardiograms in patients with a family history of cardiomyopathy is limited. We investigated how useful fetal echocardiography is with a family history of cardiomyopathy.

Methods:

We screened all fetal echocardiograms performed at our institution for the past 5 years for the indication of family history of cardiomyopathy. We excluded patients with a family history of hypertrophic cardiomyopathy, since non-syndromic forms are not typically clinically significant in the newborn period.

Results:

Twenty-six patients were identified who had fetal echocardiograms performed due to a family history of cardiomyopathy. Three out of 26 patients (11.5%) had findings of decreased ventricular function and dilation consistent with cardiomyopathy.  All who had cardiomyopathy on fetal echocardiogram had parents with genetic mutations (2 maternal, 1 paternal), including one mother who had a cardiac transplant at age 8 for dilated cardiomyopathy. All 3 affected infants had prenatal planning for high level care and were transferred to our facility immediately after birth for cardiology evaluation and management. 2 patients required inotropic support in the newborn period. One patient was transplanted at age 2 months. One patient required ECMO support for one week but recovered. None of the patients with normal function on fetal echocardiogram had evidence of cardiomyopathy on newborn echocardiogram.

Conclusions:

In this study we found 11.5% of fetal echocardiograms performed for family history of cardiomyopathy had findings of cardiomyopathy on fetal echocardiography significant enough to require prenatal planning. We recommend patients with a strong family history of cardiomyopathy in either parent, especially those with known genetic mutations associated with cardiomyopathy, have fetal echocardiograms performed. Fetal echocardiograms in this population are useful for monitoring of the fetus and for postnatal planning.