Heart defects before birth: Why are most cases still being missed (and what can we do about it)

Heart defects occur in roughly 1 in 100 live births—far more than any other kind of birth defect. Over the past decade, it has become increasingly clear that babies with major forms of heart disease have must better chances of good outcomes when the diagnoses are made early, particularly before birth (so that the delivery can be at a center equipped to take care of babies with severe forms of congenital heart disease).

Unfortunately, even nearly every pregnant woman in the USA receives an ultrasound during the second trimester to evaluate for birth defects, including heart defects, the current approach to screening has failed—the vast majority of cases of major heart defects are missed at the time of the prenatal ultrasound.

Many investigators and clinicians around the world have been working hard to find ways to improve the rate of detection of congenital heart disease. Various approaches have included the proposal to increase number of specific views of the heart that should be required, adding special color Doppler technology to be able to detect abnormalities in blood flow, and even the use of 3D ultrasound. Despite all these efforts, detection rates remain low throughout most of the world. In the US, more than half of babies born with major heart disease are still being missed.

In this paper, the authors (who together have over a half century of combined experience performing cardiac screening of babies before birth), propose that the most immediate approach is the most obvious—rather than our guidelines simply asking for more and more views, we need to change our guidelines to stipulate that images need to be of a reasonable image quality. So many cases are missed not because specific views were not obtained, but because they were not obtained well. Of course, training and education of those who scan will be important, but the approach is straightforward. If implemented, we believe that prenatal detection rates for congenital heart disease will improve dramatically, and countless babies and their families will benefit for years to come.

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