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Bio 294  
Assignment #3

Review: Role of Second-Trimester Genetic Sonography After Down Syndrome Screening

The objective of this study was to estimate the effectiveness of second-trimester genetic sonography in modifying Down syndrome screening test results, hoping to determine if sonography could increase detection rates beyond the current genetic screening processes. Midtrimester Down syndrome risks were estimated for five screening test policies: first-trimester combined, second-trimester quadruple, and testing sequentially by integrated, stepwise, or contingent protocols. When the results of these five screening tests come back, if they’re significantly concerning there is generally the next step of invasive procedures to determine if the fetus does indeed have down syndrome. The purpose of this study was to estimate how much more effective the second-trimester sonography could be so that there wouldn’t be a need to jump to invasive procedures.

To determine the significance of adding sonography to the screening process the trial participants were studied from 13 centers where a 15- to 23-week genetic sonogram was performed in the same center. Midtrimester Down syndrome risks were estimated for five screening test policies: first-trimester combined, second-trimester quadruple, and testing sequentially by integrated, stepwise, or contingent protocols. The maternal age-specific risk and the screening test risk were modified using likelihood ratios derived from the ultrasound findings. Separate likelihood ratios were obtained for the presence or absence of at least one major fetal structural malformation and for each “soft” sonographic marker statistically significant at the P<.005 level. Detection and false-positive rate were calculated for the genetic sonogram alone and for each test before and after risk modification.

Throughout their trials they used sonography and data collected from those sonograms to identify any abnormalities and soft markers from Down Syndrome tests to determine if there was substantial evidence to verify the presence of the syndrome. They then compared that to the results from the previously mentioned screening tests to see how accurately they were detecting the syndrome. Through this they found that the use of the genetic sonogram to look for Down syndrome risk after a second-trimester quadruple test can substantially increase the detection rate, reducing the need for invasive procedures. They also showed that the first-trimester combined test results can be substantially improved even when only women with borderline risks have the procedure. In contrast, the sequential screening results were only modestly improved with the addition of the genetic sonogram.

Their study concludes that each person’s pregnancy should be looked at individually, but that the data supports combing sonograms with the screenings to draw conclusions without the need for invasive testing in a considerable number of cases. They caution though that the sonograms should only be done at proper facilities, and that they should always be combined with the screenings as well to ensure accurate prognoses without use of the invasive procedures.

Citation:   
Aagaard-Tillery, K. M. et al. Role of second-trimester genetic sonography after Down syndrome screening. *Obstet Gynecol* 114, 1189–1196 (2009). https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4824304/