First Trimester Nuchal Translucency Screening: Should It Be Standard of Care?

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Screening for Down syndrome has evolved significantly over the last number of years. Much research has been presented describing sonographic features that may be useful for the prenatal detection of Down syndrome, ranging from second trimester “soft markers” such as short femur, nuchal fold enlargement, or echogenic intracardiac foci, to first trimester features such as increased nuchal translucency or absent fetal nose bone.

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Screening for Down syndrome has evolved significantly over the last number of years. Much research has been presented describing sonographic features that may be useful for the prenatal detection of Down syndrome, ranging from second trimester “soft markers” such as short femur, nuchal fold enlargement, or echogenic intracardiac foci, to first trimester features such as increased nuchal translucency or absent fetal nose bone. Despite the presence of a multitude of studies documenting the association between these various sonographic features and Down syndrome, the decision on what screening tests should be organized and implemented on a population basis is much less clear.

This presentation will document the performance characteristics of a range of screening tests available during the first and second trimesters of pregnancy. Nuchal translucency is apparently a powerful tool for the detection of Down syndrome. However, there is considerable disagreement in the literature as to the precise Down syndrome detection rate that can be expected with this form of sonography. The main reason for this disagreement seems to focus on differences in quality of nuchal translucency sonography. Several studies have documented that nuchal translucency sonography, when performed in an uncoordinated manner, without proper attention to quality, will yield very poor Down syndrome detection rates. This would be a considerable disservice to our patients.

Studies of nuchal translucency screening published to date in which high Down syndrome detection rates have been described, emphasize the need for careful attention to sonographer training and to methods of ongoing quality control. How to achieve such ongoing quality control with nuchal translucency sonography on a widespread basis is still unclear. Other practical issues regarding nuchal translucency sonography that remain to be clarified include whether to use population medians or sonographer specific medians for interpretation of nuchal translucency size, and what the resource implications would be of a national nuchal translucency sonography training and quality assurance program.

Studies that have been presented documenting apparently high Down syndrome detection rates for nuchal translucency sonography are difficult to compare with studies evaluating existing screening tools for Down syndrome. The only methodologically correct way in which to compare the range of Down syndrome screening tests, is to evaluate such tests in a single population by means of a non-interventional trial. The results of two such trials will be available in the near future: the SURUSS Trial in the United Kingdom and the FASTER Trial in the United States. The value of these studies is that they will document Down syndrome detection rates for all forms of first and second trimester screening, including nuchal translucency, absence of fetal nose bone, PAPP-A, hCG, AFP, unconjugated estriol, inhibin-A, second trimester “soft markers”, and fetal cells in the maternal circulation. Given that all measurements have been derived from single large populations without intervention before screening is complete, for the first time precise comparative performance of all screening tests will be available.

Precise comparative detection rate data is however only one issue that needs to be considered before making recommendations for national aneuploidy screening policies. Just as important as Down syndrome detection rates are screen positive rates. An analysis recently by Wald of currently
available data on a variety of screening tests would suggest that nuchal translucency is the least efficient of the screening strategies currently available (Wald, J Ultrasound Med 21:481, 2002): It is clear from this table, that from a population screening perspective, it would be quite unwise to implement nuchal translucency based screening sonography in isolation. While there is little debate that nuchal translucency is a potentially useful screening tool, the remaining debate now is in what combination of other tests should it be implemented. There should also be little debate that nuchal translucency screening, if implemented in isolation, is an inefficient method of screening. Efficient alternatives to nuchal translucency based screening also exist, without the need for sonographic screening. The main debate remaining is whether the addition of nuchal translucency screening justifies the resource implications required to perform this type of ultrasound and to maintain adequate quality control over this type of ultrasound.

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