

<b>Title:</b>	<b>Nuchal Translucency Measurement in the First Trimester of Pregnancy for Screening of Trisomy 21 and Other Autosomal Trisomies, May 2002</b>
<b>Agency:</b>	<b>Medicare Services Advisory Committee (MSAC) Mail Drop Point 107 Commonwealth Department of Health and Ageing GPO Box 9848, Canberra, ACT 2601, Australia</b>
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### **Aim**

To assess the safety, effectiveness and cost-effectiveness of nuchal translucency (NT) measurement in the first trimester of pregnancy for screening of trisomy 21 and other autosomal trisomies and under what circumstances public funding should be supported for the service.

### **Conclusions and results**

#### **Safety**

The literature on the safety of ultrasound in pregnancy is scant considering its widespread use and acceptance. Professional organisations have released recommendations about the use of specific types of ultrasonic imaging modalities and potentially adverse outcomes. Most advocate the prudent use and stress the importance of minimum output levels and exposure times.

#### **Effectiveness**

When used as a single modality, screening by measurement of NT in the first trimester has a detection rate (DR) for trisomy 21 of approximately 73-82 per cent at a false positive rate (FPR) of 5-8 per cent. For maternal biochemical screening (MBS), the comparable DR using double markers ((pregnancy-associated plasma protein-A (PAPP-A) and free beta human chorionic gonadotrophin (free  $\beta$ -hCG)) in the first trimester of pregnancy is 65 per cent at a FPR of 5 per cent. For MBS during the second trimester, the detection rate is approximately 67-69 per cent using triple marker testing (PAPP-A, free  $\beta$ -hCG and either alpha-fetoprotein (AFP) or unconjugated oestriol (uE3)) and approximately 70 per cent using the quadruple marker test (PAPP-A., free  $\beta$ -hCG, AFP and uE3). If used in combination in the first trimester, the detection rate with a 5 per cent FPR is 86 per cent for NT screening plus double markers, 87-88 per cent for NT screening plus triple markers and 88 per cent for NT screening plus quadruple markers.

#### **Cost-effectiveness**

The cost-effectiveness of NT ultrasound screening has been examined by comparing the expected cost of universal screening using each modality compared with no screening. The cost of a universal MBS program in the second trimester with 100 per cent uptake of pregnant women would be approximately \$21.3m. A combined screening program (incorporating NT ultrasound plus MBS in the first trimester) achieving a 20 per cent improvement in the DR of trisomy 21 compared with second trimester MBS, would cost \$26.7 million more per year, with 253 more trisomy 21 cases detected. This represents an incremental cost of \$105,484 per extra case detected.

If screening were restricted to women aged 30 years or over, then the cost per case detected would be one third less. If screening were restricted to women aged 35 years or over, then the cost per case detected would be halved. Sensitivity analysis around the main parameters of uncertainty suggested that, depending on the true DR in practice and the extent of substitution for current ultrasound and follow-up diagnostic tests in high risk pregnancies, the incremental cost per extra case detected would be between \$43,825 and \$141,664.

#### **Recommendation**

Public funding should not be supported for NT screening or NT screening in conjunction with first trimester MBS as stand alone services, due to their poor cost-effectiveness. Consideration should be given to public funding of NT screening or NT screening in conjunction with first trimester MBS by incorporating, as far as possible, provision of the services into existing services provided in early pregnancy.