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Dissertation June 27 2003

First trimester screening for Down syndrome

Abstract

The aim of the present study was to evaluate the efficacy of the first trimester screening for Down syndrome (DS) in an unselected low-risk Finnish population. The study involved 4,617 women who attended screening between the 8th and 14th weeks of pregnancy in 1998-2000. They gave a blood sample for the measurement of pregnancy associated plasma protein A (PAPP-A) and free beta human chorionic gonadotrophin (β -hCG). Of these women, 3,178 also had an ultrasound examination for the measurement of fetal nuchal translucency (NT). The risk figure for every screened woman was calculated using a computerized risk figure program. The risk 1 in 250 was used as a cut-off. The subgroup of screen positives comprised 5.8% of the study group.

There were 16 DS cases. The combined method (maternal age, NT and the biochemical markers) detected 77% of the affected pregnancies. NT combined with maternal age gave a detection rate of 69%. Serum markers without NT combined with maternal age found 75% of the Down's.

In 49 consecutive singleton in-vitro-fertilization (IVF) pregnancies, the β -hCG value was more often elevated compared to spontaneous pregnancies, increasing the false positive rate. In 67 twin pregnancies, the serum marker levels were approximately double those in singletons. Smoking reduced PAPP-A by 20% making the smokers more likely to get a positive screening result.

To determine the impact of the screening on the live born incidence of DS, two historical populations were compared. The first group was screened by second trimester serum samples (β -hCG and AFP) and the second group by first trimester ultrasound examination. When detection rates were at the same level, the second trimester screening reduced the number of live born Down's children more effectively.

In conclusion, the first trimester combined method (maternal age, NT, β -hCG and PAPP-A) for Down syndrome screening is efficient in an unselected low risk population. The biochemical screening is not recommended in IVF-pregnancies.