

Highly improved test performance for prenatal aneuploidy screening by the novel 'Advanced Firsttimester Screening'

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Abstract

Objective: The common background risk for trisomy 21 does not regard to the individual pregnancy and it was postulated to completely exclude the age depending component from firsttrimester screening. It was the aim of this study to verify a new algorithm, which is called 'Advanced Firsttrimester Screening' (AFS), disregarding the age.

Material and methods: In a multicenter study the measurement values of a firsttrimester screening were retrieved from 10,116 pregnancies. Risk assessment was performed by Nicolaides' method (FTS) and in addition an AFS score has been calculated. Statistical significance was tested by McNemar test. Testperformances were compared to each other by Receiver-operating-characteristics (ROC) and significance was tested by combined t-test.

Results: Whithin this population, 85 fetuses had an abnormal karyotype. By AFS the positive predictive value rises from 9.9% (FTS) to 12.7% (AFS). The test positive rate could be lowered by 138 cases (-21.1%) by AFS ($p < 0.0000001$), which is completely attributed to a reduction of the false negative cases. Simultaneously three more cases (+4.2%) of chromosomal defects were detected by AFS ($p = 0.37$).

Discussion: In this study it becomes evident that AFS can markedly reduce the rate of false positive test results. In combination with a slight increasement of the sensitivity of this new method overrides the hitherto test performance by far. This is based on several modification to the classical firsttimester screening algorithm. The most meaningful change is to relinquish the mathernal age from risk assessment. If the results on hand can be veryfied again by larger, international multicenter studies, the new Advanced Firsttimester Screening would mean a great improvement in fetal aneuploidy screening.