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Editorial

Serum Tests for Down Syndrome Screening

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ABSTRACT

Down's syndrome is an incurable genetic that causes severe physical and mental health disorders and disabilities. There is considerable variance, however, on how Down's affects individuals. Whilst some have minor difficulties and are able to lead relatively normal lives, some people are seriously affected. There is no way of predicting how badly a baby might be affected.
KEYWORDS: Down Syndrome, Trisomy 21, Chromosome Abnormality

INTRODUCTION

The most reliable tests for Down's include testing fluid for the irregular chromosomes associated with Down's from around the baby (amniocentesis) or placenta tissue (chorionic villus sampling (CVS)). Both these tests include sticking needles into the abdomen of the mother and the risk of miscarriage is known to increase. Therefore, the tests are not appropriate for all pregnant women. Rather, tests are used for screening to measure markers in the mother's blood, urine or on the baby's ultrasound scans. They will miss instances of Down's and even offer a 'high risk' test result to a number of women whose babies are not affected by Down's. These screening tests are not ideal. Therefore, to confirm a diagnosis of Down's, pregnancies classified as 'high risk' using these screening tests require additional testing using amniocentesis or CVS.

Down's syndrome happens once an individual has 3, instead of 2 copies of body twenty one; or the particular space of body 21 involved in inflicting subnormality. it's the most common innate reason for mental incapacity and conjointly results in various metabolic and structural issues. It will be life-threatening, or cause sizeable unhealthiness, though some people have solely gentle issues and might lead comparatively traditional lives. Having a baby with Down's syndrome is probably going to possess a major impact on family life.

Based on biochemical analysis of maternal serum or urine or fetal ultrasound measurements, non-invasive screening makes it possible to quantify the likelihood of pregnancy being compromised and provides information to guide decisions regarding conclusive testing. No exam, however, can predict the severity of issues that a person with Down's syndrome may have.

The purpose of this analysis was to estimate and compare the accuracy of the first trimester serum markers, both as individual markers and as combinations of markers, for the detection of Down's syndrome in the prenatal period.

Accuracy is defined by the proportion of pre-birth screened fetuses with Down's syndrome and the proportion of women with a low-risk screening test result who subsequently had a baby without Down's syndrome (specificity).

Tests involving two markers, specifically PAPP-A, free βhCG and maternal age, in conjunction with maternal age, are substantially better than those involving single markers with and without age. For a fixed 5 percent FPR, they detect seven out of 10 Down's affected pregnancies. It was not shown to be statistically superior to the inclusion of more markers (triple tests); the studies included are small with limited power to detect a difference.

There are no adverse effects on the woman from the screening blood tests themselves, beyond the dangers of a regular blood test. Nevertheless, certain women who have a 'high risk' screening test outcome and undergo amniocentesis or chorionic villus sampling (CVS) are at risk of miscarriage of a child not affected by Down's. When determining whether or not to have an amniocentesis or CVS following a 'high-risk' screening test result, parents would need to consider this risk.

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