

Two blood markers moderately accurate in screening for Down's syndrome

Clinical Question

How accurate are first trimester serum markers for the detection of Down's syndrome in the antenatal period?

Bottom Line

Tests involving two markers in combination with maternal age, specifically PAPP-A (pregnancy-associated plasma protein A), free β -hCG (beta- human chorionic gonadotropin) and maternal age were significantly better than those involving single markers with and without age. They detected seven out of 10 Down's affected pregnancies (3 in 10 were missed) and a fixed 5% of all tested women got a false positive result. The addition of further markers (triple tests) was not shown to be statistically superior; the studies included were small with limited power to detect a difference. The screening blood tests themselves had no adverse effects for the woman, over and above the risks of a routine blood test.

Caveat

Some women who have a 'high risk' screening test result, and are given amniocentesis or chorionic villus sampling have a risk of miscarrying a baby unaffected by Down's syndrome. Parents will need to weigh up this risk when deciding whether or not to have amniocentesis or chorionic villus sampling following a 'high risk' screening test result.

Context

Down's syndrome is the commonest congenital cause of mental disability and also leads to numerous metabolic and structural problems. Non-invasive screening based on biochemical analysis of maternal serum or urine, or fetal ultrasound measurements, allows estimates of the risk of a pregnancy being affected and provides information to guide decisions about definitive testing.

Cochrane Systematic Review

[Allred SK et al. First trimester serum tests for Down's syndrome screening. Cochrane Reviews, 2015, Issue 11. Art. No.: CD011975.DOI: 10.1002/14651858. CD011975.](#) This review contains 56 studies involving 204,759 participants (including 2,113 with Down's syndrome).