

First trimester screening for T18 / T13

From **April 2015**, the Fetal Anomaly Screening Programme recommends all eligible pregnant women in England are offered first trimester combined screening for Edwards' syndrome (Trisomy 18 / T18) and Patau's syndrome (Trisomy 13 / T13) in addition to the existing Down's syndrome (Trisomy 21 / T21) screening programme.

What is the screening policy?

The policy is to offer screening to assess the risk of the baby being born with T21 or T18 / T13. The test of choice for both singleton and twin pregnancies is first trimester combined screening. Women can choose to have:

- no screening
- screening for T21 and T18 / T13
- screening for T21 only
- screening for T18 / T13 only

What are T18 and T13?

Most babies with T18 or T13 will die before they are born, will be stillborn or will die shortly after birth. Some babies may survive to adulthood but this is rare.

In T18, there is an extra copy of chromosome 18 in each cell. T18 affects about 3 of every 10,000 births. In T13, there is an extra copy of chromosome 13 in each cell. T13 affects about 2 of every 10,000 births.

All babies born with T18 or T13 will have a wide range of problems, which are usually extremely serious - these may include major brain abnormalities.

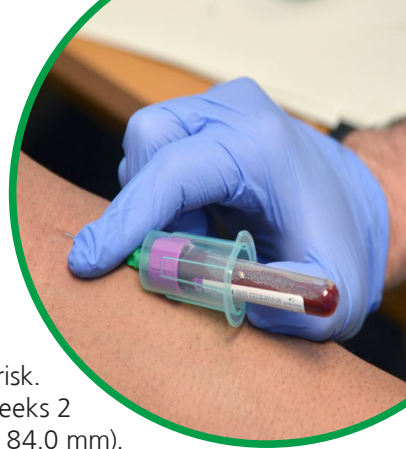


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Are additional samples or scans required?

No – the combined screening test currently undertaken for T21 is used by the laboratories to calculate risk for T18 / T13.

The combined test uses maternal age, nuchal translucency measurement and two biochemical tests together with the gestational age calculated from the crown rump length (CRL) measurement to calculate the risk. The optimal time for the combined test is between 11 weeks 2 days to 14 weeks 1 day of gestation (CRL of 45.0 mm to 84.0 mm).



What are the national standards?

Screening Strategy	Standardised detection rate	Standardised screen positive rate
T21	85%	2.5%
T18 / T13	80%	6.2%
T21 / T18 / T13	80%	1.5%

What is the risk cut-off?

A risk cut-off determines women who are in the 'higher risk' group and considered 'screen positive'. The national cut-off is 1 in 150 at term for both first and second trimester screening tests.

A woman with a risk of 1 in 150 or greater (1 in 2 to 1 in 150) of having a pregnancy affected by T18, T13 in the first trimester or T21 only in the second trimester is considered to be in the 'higher risk' group. Women in this group are offered a diagnostic test such as chorionic villus sampling or amniocentesis to directly investigate the fetal chromosomes.

What happens if the results are lower risk?

All women should be notified of their screening test result within two weeks of testing. The results should be documented in the health record.

What happens if the results are higher risk?

Maternity services should inform women of the result within three working days of receiving the result from the laboratory. A face to face discussion should be offered with a health professional who has the experience and knowledge to explain the results and discuss further options which include no further testing or diagnostic testing.

Women must be informed that diagnostic testing will give a result for T21 and T18 / T13 irrespective of the initial screening choices.

For more information visit

www.gov.uk/topic/population-screening-programmes/fetal-anomaly