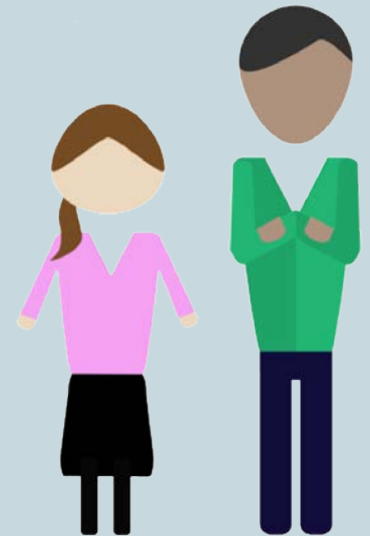


Case Scenario 3 – Sarah and Phil

Sarah and Phil have had repeated IVF cycles for over 8 years. Sarah is now 11+2 weeks pregnant, and has attended a clinic for a scan and Combined bloods. Sarah informs the midwife that she had an NIPT test that had reported a low chance result for Trisomy 21, 18 and 13.

The midwife advises the couple to have the Combined test as it will give additional information including PAPP-A levels; important for monitoring fetal growth. The couple decide to have the Combined test.

3 days later Sarah receives a phone call to inform her that the Combined test indicates a high chance of having a baby with Down's Syndrome.



Would you agree with the advice provided by the midwife?

- a) No - the Combined test is offered for trisomy screening only. PAPP-A levels cannot be reported independently.

- b) Yes - for the PAPP-A. Women should be informed that trisomies will not be screened.



Answer

- a) No - the Combined test is offered for trisomy screening only.

Additional note

PAPP-A is not an indication for performing the combined test, it is a coincidental finding that should be monitored if low. Currently the RCOG do not recommend routine screening of PAPP-A independently.



What information should have been offered to ensure Sarah made an informed choice?

- a) The NHS pathway recommends that all women must be offered screening regardless of prior testing. But a clear discussion of the risks and benefits should be included. It must be made clear the screening is for trisomies only. Women can decline; however an NT scan should be recommended.

- b) The NHS pathway recommends that, any woman who has accessed NIPT prior to entering the pathway, be advised that they are no longer eligible for further screening.



Answer

a) The NHS pathway recommends that all women must be offered screening regardless of prior testing. But a clear discussion of the risks and benefits should be included. It must be made clear the screening is for trisomies not other coincidental findings. Women can decline, however an NT scan should be recommended.



Additional note

A woman who has undertaken private NIPT should still be offered standard NHS Combined/Quadruple testing. She should be informed that the standard NHS tests are significantly less specific and sensitive (not as accurate) when compared to NIPT and that there is a significant risk that she may be incorrectly assigned a high-chance category by such testing. Under these circumstances, she may well decline further NHS screening as is her choice and right.

How early can NIPT (The SAFE Test) be performed?

- a) At any gestation
- b) From 10 weeks of pregnancy
- c) From 12 weeks of pregnancy
- d) From 4-5 weeks of pregnancy



Answer

b) The SAFE Test can be performed from 10 weeks of pregnancy



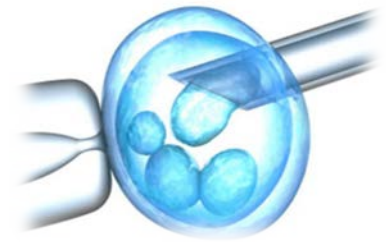
Additional note

Placental cell free DNA is present in the maternal circulation from approximately 4-5 weeks of pregnancy. Adequate quantities for clinical testing appear at about 10 weeks gestation.

Is IVF a contraindication to this test?

a) Yes

b) No



Answer

b) No

Additional note

Not all NIPT technologies can offer screening for IVF pregnancies, however this is not a contraindication for the SAFE test.

- ✓ From 10 weeks gestation
- ✓ Singleton or twin pregnancies
- ✓ IVF, donor egg or surrogate pregnancies

What information do you require from a patient with an IVF pregnancy?

Please choose one option

- a) The date the eggs are fertilised
- b) The date of embryo transfer
- c) The age of the mother or donor age at egg harvest
- d) All of the above



Answer

c) Age of mother or donor at egg harvest

Additional note

The age at egg harvest is used for the a-priori risk, as this is the most accurate for the individual's pregnancy.



Why was the result from the Combined test different to NIPT screening?

Choose one or more options

- a) They use different techniques for screening
- b) The Combined test is screening and NIPT is a diagnostic test
- c) The Combined test is more accurate as an initial screening test, because it is analysing more risk factors, whereas the NIPT is only looking at cell free DNA, and does not use risk factors
- d) The laboratory has made a mistake. The Combined test result and NIPT result should match



Answer

a) They use different techniques for screening

Additional note

The Combined test looks at hormones, maternal age and the NT to provide a risk, whereas the NIPT evaluates circulating cell free DNA to analyse chromosome levels and predict the likelihood of the baby having a chromosomal condition. The combined test has a higher false positive rate in comparison to NIPT.

