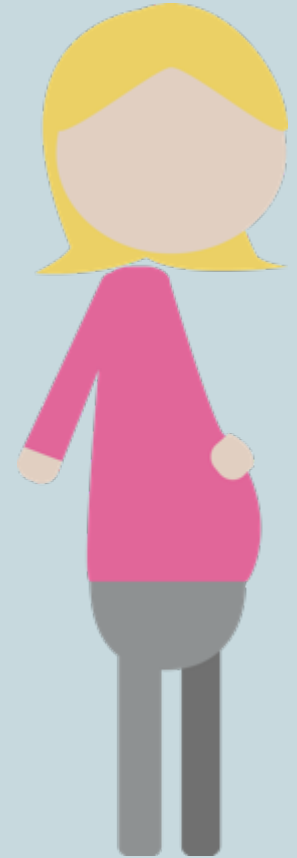


Case scenario 1 – Josie

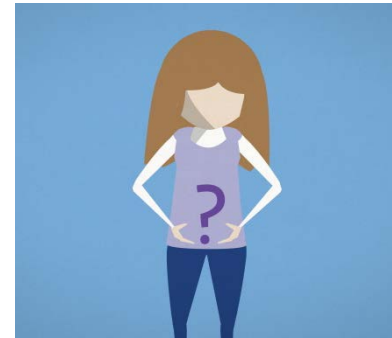
Part 1

- Josie is seen by the Fetal Medicine team following a high chance result from the Combined test, performed at 13 weeks and 6 days gestation. Her Trisomy 21 risk ratio is 1 in 28.
 - PAPP-A 0.3MoM
 - free β hCG 1.7MoM
 - NT 2.2mm
- After a full discussion, Josie and partner decide to undertake NIPT.



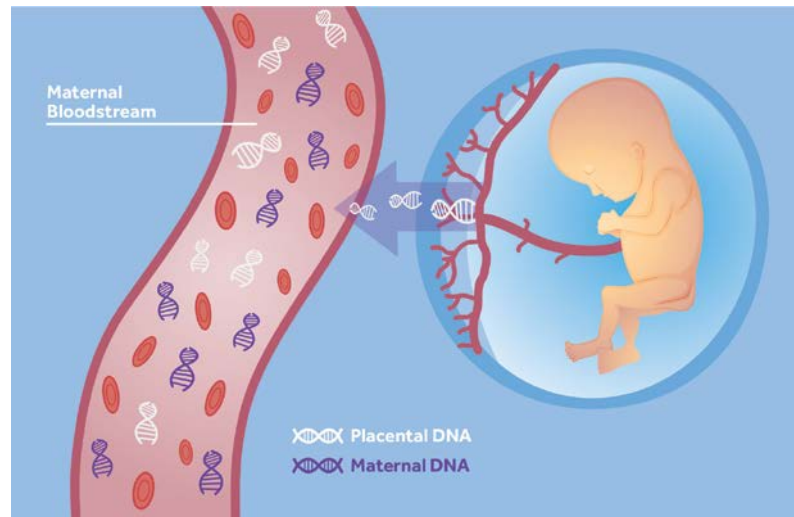
What is NIPT?

- a) It is a maternal blood test that analyses fetal chromosomes to predict if the baby is affected with a chromosome condition
- b) It is a maternal blood test that analyses the baby's DNA to provide a result to predict if the baby is affected with a chromosome condition
- c) It is a maternal blood test that looks at circulating cell free DNA to analyse chromosome levels and predict the likelihood of the baby having a chromosome condition



Answer

C) It is a maternal blood test that looks at circulating cell free DNA to analyse chromosome levels and predict the likelihood of the baby having a chromosomal condition.



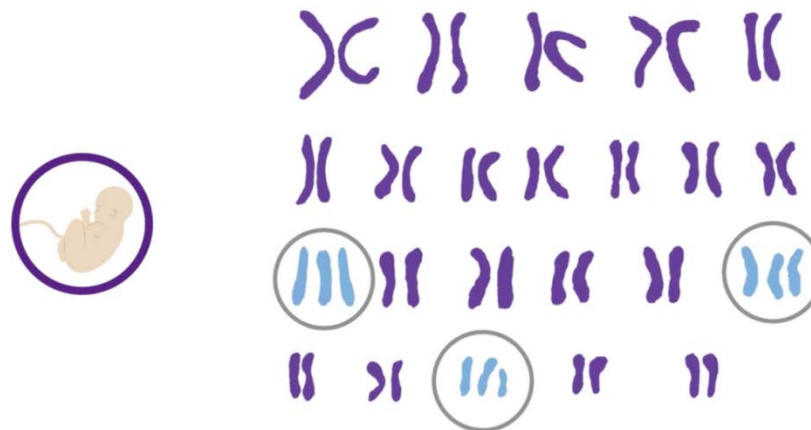
What does the SAFE test screen for?

- a) Down's Syndrome only
- b) Down's, Edwards' and Patau's Syndrome
- c) Down's, Edwards', Patau's and Turner's Syndrome



Answer

b) Down's, Edwards' and Patau's Syndrome



Additional note

Other NIPT tests may offer screening for sex aneuploidies, microdeletions or microduplications. These are NOT recommended for routine care and can lead to a high false positive rate with increased requirement for diagnostic testing.

Are there any contraindications for NIPT for Josie?

- a) Yes - based on the Combined test results she is not eligible
- b) No - based on the Combined test results she is eligible
- c) Unknown - The result from the combined test is important for the a-priori risk, however details of medical history are required to assess contraindications

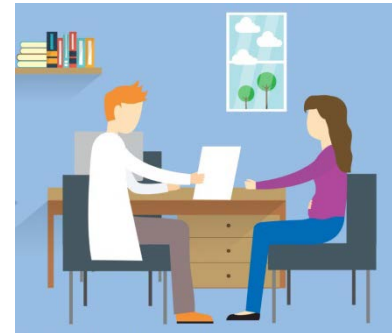


Answer

c) Unknown - The result from the combined test is important for the a-priori risk, however details of medical history are required to assess contraindications

Additional note

Based on Josie's results, there are no contraindications to screening. However, medical history, previous pregnancies and Josie's own personal values should all be considered when discussing screening/diagnostic tests.



What information should you provide to patients on NIPT testing? Choose 1 or More

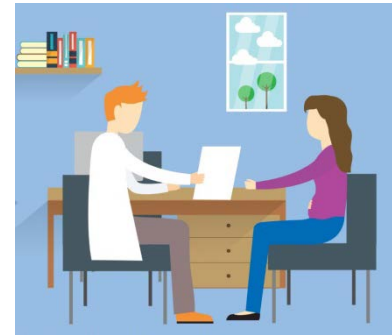
- a) What conditions the test is screening for
- b) How the blood sample is taken
- c) Sensitivity and specificity
- d) How and when the result will be given
- e) All of the above



Answer

e) All of the above

- What conditions the test is screening for
- How the blood sample is taken
- Sensitivity and specificity
- How and when the result will be given



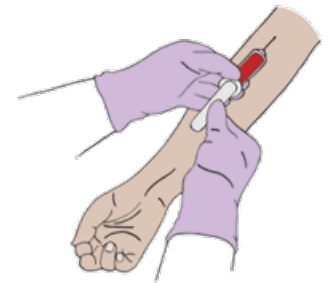
Josie – Part 2

- There were no contraindications for Josie and a sample of blood was taken with consent.
- The sample reaches the Laboratory the following day. By the 4th working day the laboratory was unable to issue a result. The sample was processed for a second time. Josie is 15+6 when she receives a second failed result.



How many times can one SAFE Test sample of blood be processed?

- a) Once
- b) Twice
- c) Three times
- d) Four times

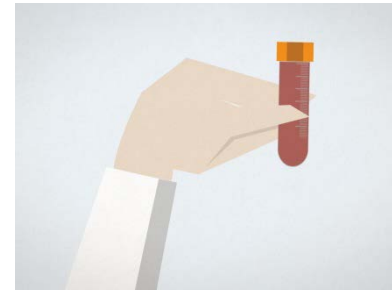


Answer

b) Twice

Additional note

2.5mls of plasma is required for one sample. Streck tubes hold approximately 5mls of plasma (10mls of peripheral blood)



If the SAFE test fails, what test(s) would you offer the patient? Choose one or more

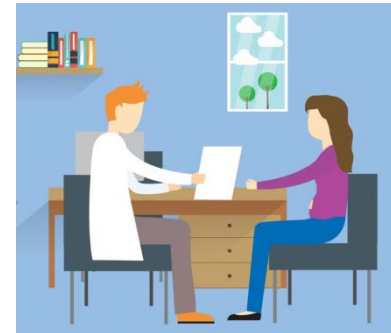
- a) No further testing - the test has failed, but the patient is informed there is a chance of failure with NIPT. We cannot offer any further screening.
- b) Offer Amniocentesis only - The test is more likely to fail therefore a patient cannot be offered a re-draw.
- c) Offer a redraw only - As the patient is now 15+6 there is likely to be more fetal fraction at this gestation.
- d) It depends on the reason for failing - The individual case should be discussed with the laboratory for a recommendation.
- e) Offer a choice - Inform the patient of the options: repeat NIPT/Invasive testing/no further testing.



Answer

d) It depends on the reason for failing - The individual case should be discussed with the laboratory for a recommendation.

e) Offer a choice - Inform the patient of the options: repeat NIPT/Invasive testing/no further testing.



Additional note

You should check with the laboratory the indication for a failed result, if the result is due to high levels of fetal fraction, the test is likely to fail again. Patients should be advised of the benefits and limitations of all options. Consider gestation and patients opinions on continuing or terminating the pregnancy. For example, a patient not wishing to risk miscarriage with a diagnostic test may want to repeat NIPT even if later in the pregnancy for preparation.

Why can the SAFE test fail?

Give two or more possible reasons

- a) NIPT does not fail
- b) Low fragment count
- c) Low or High fetal fraction
- d) The CRL from scan information has not been provided- critical information to provide a report
- e) Incorrect sample storage
- f) The pregnancy is affected with a trisomy



Answer

b) Low fragment count

c) Low or High fetal fraction

e) Incorrect sample storage e.g. Broken bottles, clotted samples, refrigerated samples

f) Pregnancy affected with a trisomy

