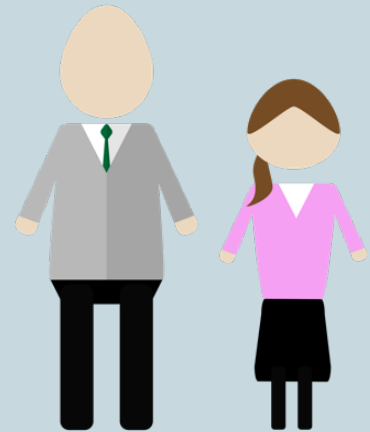


Case scenario 4 – Lola and Simon

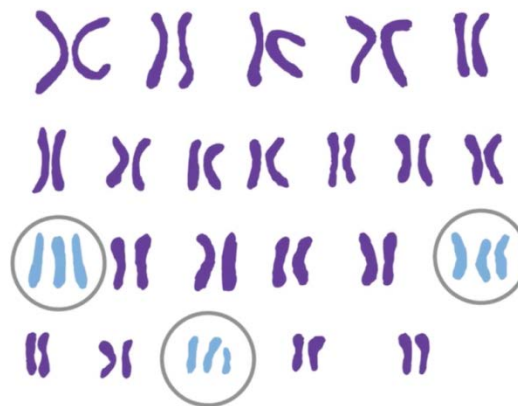
Part 1

- Lola and Simon attend a private clinic for an NIPT test at 16 weeks' gestation. Lola tells the clinic that she received a low chance Combined test result for trisomy 21, and she declined screening for trisomy 13 and 18.
- Lola reports that, after receiving news that her cousin has just given birth to a baby with Down's syndrome, she would like the NIPT test for reassurance.



Can the SAFE test screen for trisomy 21 only?

- a) Yes
- b) No

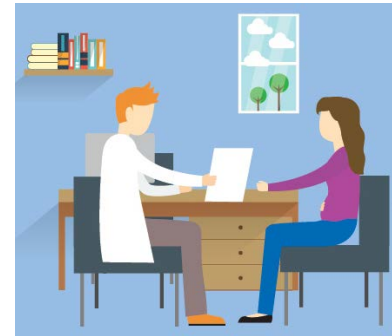


Answer

b) No

Additional note

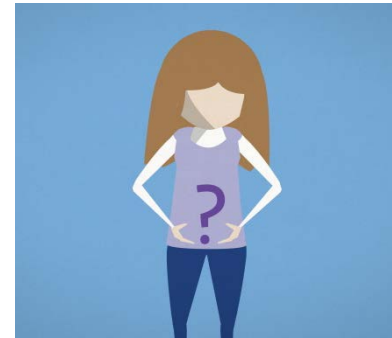
Currently, the software used for the SAFE test will report on all 3 trisomies. However, this may be something that is developed in future templates for the NHS Pathway



What are the disadvantages of NIPT?

Choose at least one

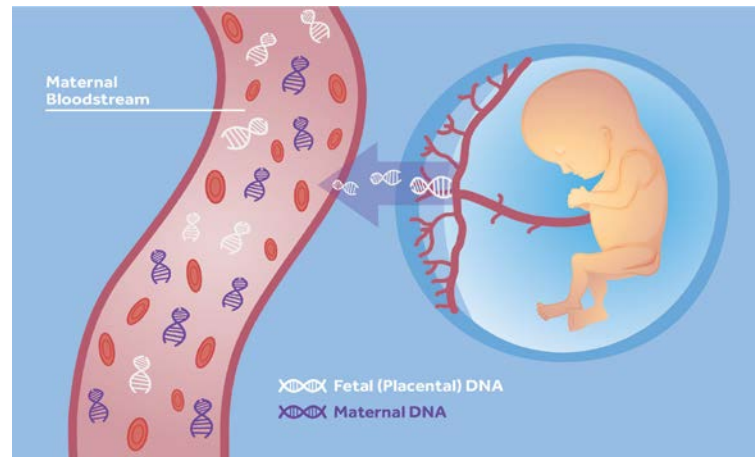
- a) It is not a diagnostic test
- b) NIPT cannot differentiate between DNA from the placenta or the fetus
- c) There is a small chance of test failure
- d) All of the above



Answer

d) All of the above

- *It is not a diagnostic test*
- *NIPT cannot differentiate between DNA from the placenta or the fetus*
- *Small chance of test failure*



What further information is required to complete the consent form?

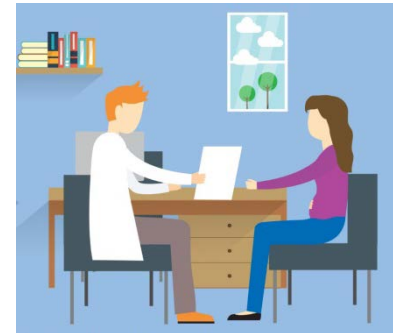
Choose one or more

- a) Combined or Quadruple screening results if available
- b) Husband's age
- c) Family history of genetic conditions
- d) Current cancer
- e) Any medications
- f) Previous blood transfusion
- g) Blood group
- h) All of the above



Answer

- a) Combined or Quadruple screening results if available
- c) Family history of genetic conditions
- d) Current cancer
- f) Previous blood transfusion



Additional note

Not all medical conditions or medications are requested on the consent form. Medications that are required are LMWH dosages. The Combined or Quad result can provide a patient specific result when used in a-priori replacing the age related risk.

Lola and Simon – Part 2

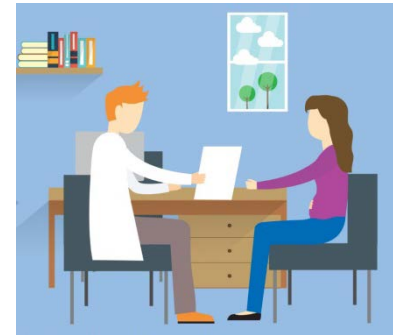
You receive Lola's NIPT report. It shows a 'greater than 75% chance' that the baby has trisomy 18.



Is the following statement True or False?

3 compulsory patient identifiers when completing a request form, blood tube and confirming a report are:

- Correct name
- Correct date of birth
- Correct hospital number



Answer

True



Additional note

If there is any incorrect information, it may cause a delay in reporting. If identifier information is incorrect on a report, liaise with the laboratory prior to giving result.

Why is the result given as a %?

- a) The report is not given as a percentage - it is a diagnostic test.
- b) It incorporates PAPP-A, beta hCG, ethnicity, weight and other maternal factors to provide a screening result.
- c) The data from the DNA fragments and the a-priori risk provide a patient specific result – it is not a diagnostic test.

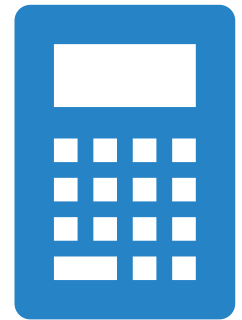


Answer

c) The data from the DNA fragments and the a-priori risk provide a patient specific result - it is not a diagnostic test.

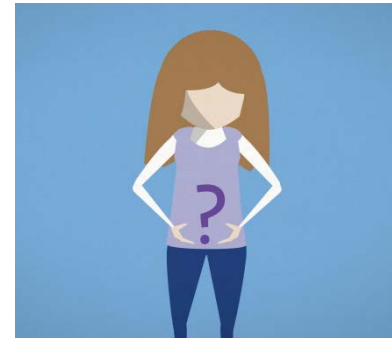
Additional note:

- *The percentage given may vary depending on the amount and the quality of cfDNA from the maternal circulation.*
- *The percentage is capped based on maternal and placental biological factors.*
- *The low chance score is capped at 0.67% (1 in 150).*



What is the recommended test to confirm a high chance result from NIPT?

- a) The combined test
- b) An anomaly scan
- c) A diagnostic test
- d) A repeat NIPT



Answer

C) A diagnostic test

Additional note

A woman can choose to decline a diagnostic test.

However, it should be highlighted that, due to both placental and maternal factors, the result is not diagnostic. A CVS or amniocentesis should be recommended to confirm a high chance result.

