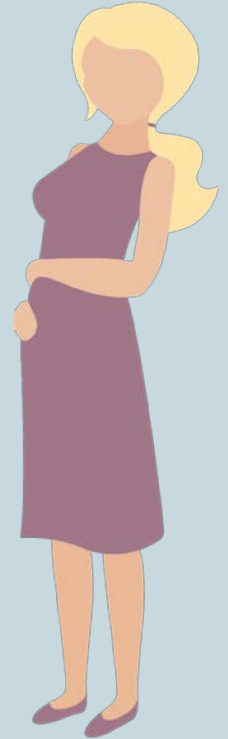


Case scenario 5 – Rachel

- Rachel is 27yrs old; At her 1st trimester scan she was informed that the NT measurement was 3.8mm. Rachel had her Combined screening bloods taken following the scan, and was advised the results will take approximately 3 working days.
- She was seen by a screening co-ordinator and was advised to attend the tertiary Fetal Medicine Unit (FMU) for further support and information. She was provided information on diagnostic testing only.
- Prior to the FMU appointment, Sarah finds information on NIPT. She contacts the local hospital and asks if she is eligible for this test.



Can NIPT be used with an NT >3.5mm?

- a) Yes
- b) No



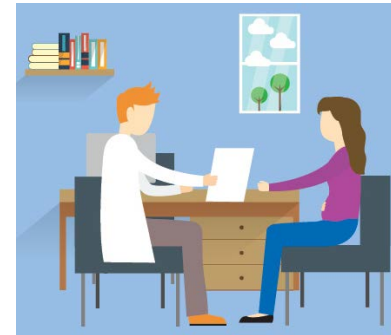
Answer

a) Yes - with limitations

Additional note

If a patient declines invasive testing and would like NIPT, the healthcare professional is responsible for ensuring the patient is aware of limitations of the test and documenting the discussion.

Patients should be informed that fetal structural defects are associated with a higher risk of non-aneuploid fetal chromosomal abnormalities.



Is the following statement True or False?

It is important to find out the social, ethical and religious beliefs and values of the individual when discussing screening.



Answer

a) True

Additional note

Values and beliefs of the patient should be explored to insure individualised choice and decision making.

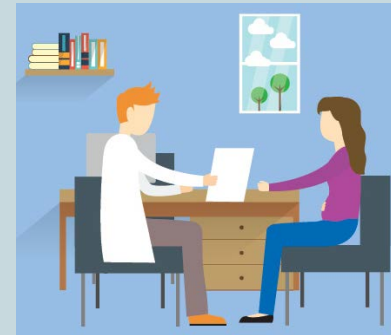


Rachel – Part 2

Rachel attends the clinic for a consultation. You are completing the consent form.

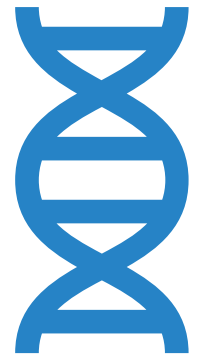
She informs you that she had a Kidney transplant 5 years ago. She is not aware of any chromosome or genetic history.

Rachel also reports a previous miscarriage at 14 weeks, 8 months ago. She informs you that a karyotype was performed and reported a baby with Triploidy.



Can the SAFE Test screen for Triploidy?

- a) Yes
- b) No



Answer

b) No

Additional note

The test is analysing an increase in levels of trisomy 13, 18 or 21, Triploidy will give a balanced increase in all chromosomes, and therefore this technology will not detect this and will report a low chance result.



Will a previous history of trisomy 21, 18 or 13 affect the circulating cfDNA in a new pregnancy?

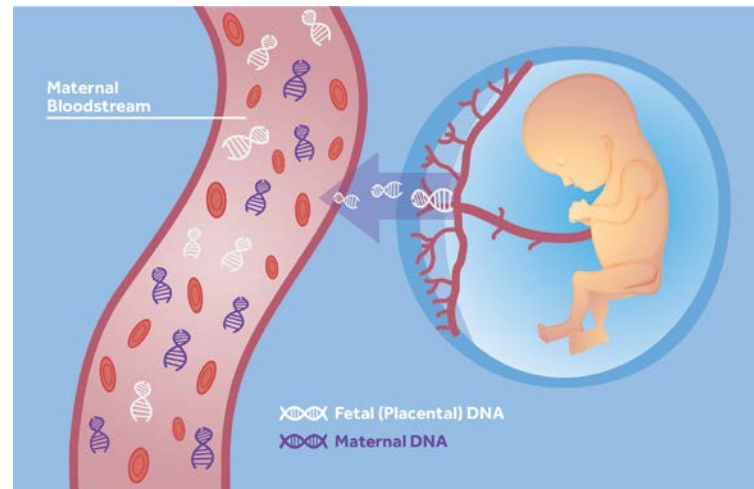
a) Yes

b) No



Answer

b) No



How long does it take for feto-placental DNA to leave the maternal circulation?

- a) It does not leave the circulation - it will only reduce
- b) 8 weeks
- c) 2 weeks
- d) A few hours

Answer

d) A few hours

Additional note

Previous pregnancy's will not affect the circulating DNA, as feto-placental secretes from the placenta. Therefore, once the placenta is expelled, no further circulating feto-placental DNA fragments exist.



Can Rachel have NIPT?

- a) Yes
- b) No



Answer

b) No

Additional Note

Transplants are a contraindication. There may be a small chance that the donor DNA is also circulating, this may very rarely affect the NIPT result (most likely when analysing the 'Y' chromosome for sex determination in some tests).

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

Unsuitable if the mother has:

- ✗ Cancer
- ✗ Trisomy
- ✗ Undergone Stem Cell Therapy or Immunotherapy
- ✗ Received an organ transplant