

Case scenario 8 – Emma

Part 1

Emma is a 43 year old single parent with 2 young children.

Emma attends a clinic for a private NIPT test. She is 10 weeks and 2 days pregnant.

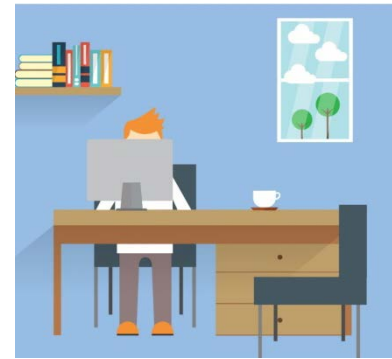
Emma tells you that this was originally a twin pregnancy and that the other baby was lost at 8 weeks gestation.



Can NIPT be performed on this pregnancy?

a) Yes

b) No



Answer

a) Yes

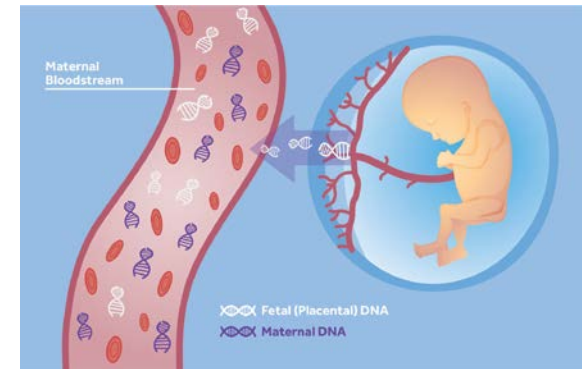
Additional note

A woman with a vanishing twin pregnancy should be advised that cfDNA testing performance is similar to that of a DCDA twin pregnancy.



What is the sensitivity of NIPT in this scenario?

- a) >85%
- b) >90%
- c) >95%
- d) >99%



Answer

c) >95%



Case scenario 8 – Emma

Part 2

Emma is informed that her baby has a greater than 95% chance of having Down's syndrome.



What are the possible explanations for this high chance result?

Select 2 correct answers based on the information provided in this scenario.

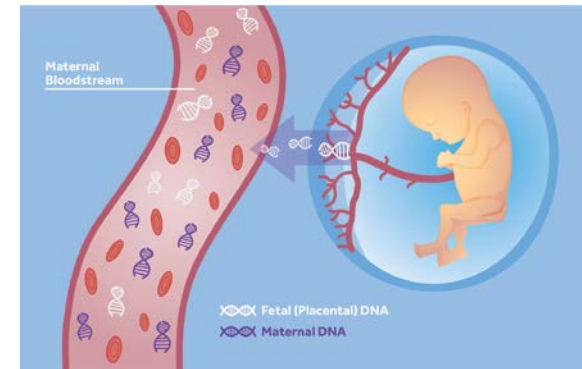
- a) She is under 12 weeks gestation
- b) It is caused by the vanishing twin
- c) She is over 35 years old
- d) The baby has Down's syndrome



Answer

b) It is caused by the vanishing twin

d) The baby has Down's syndrome



Is the following True or False?

It is suggested it takes up to 8 weeks for cfDNA from a vanished twin to completely leave the maternal circulation.



Answer

a) True

Additional note

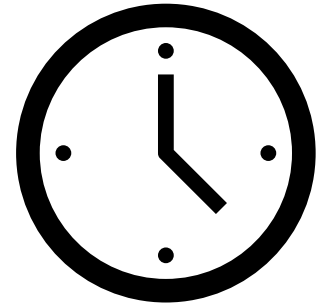
There is no data to indicate how long cfDNA from the demised twin remains in the circulation, but it has been suggested that 8 weeks is a safe period, and is less likely to affect a result.



Does a patient have to wait 8 weeks from when the vanishing twin is identified to having the SAFE test?

a) Yes

b) No



Answer

b) No

Additional note

Patients should be informed that vanishing twins may predispose to a high chance result. Women should be advised to have a diagnostic test to confirm an affected pregnancy (twin pregnancies have 5% chance of false positive result).



What further tests would you offer this patient following a high chance NIPT result?

- a) No further testing
- b) Consultant review and scan
- c) Diagnostic testing
- d) All of the above



Answer

- d) All of the above
- No further testing
 - Consultant review and scan
 - Diagnostic test



Case scenario 8 – Emma

Part 3

Emma informs you that, based on the NIPT result, she wants a termination.



Can Emma have a termination for trisomy 21 based on the NIPT result?

- a) Yes - NIPT is >99% sensitive, it is unlikely to be a false positive result. Patients should be informed that the result shows the baby has Down's syndrome. High chance NIPT does not require confirmation.
- b) No - NIPT requires confirmation from a diagnostic test, however a patient has the right to terminate a pregnancy for other personal reasons up to 24 weeks gestation. Patients should be informed of the small chance of a false-positive result.



Answer

b) No - NIPT requires confirmation from a diagnostic test, however a patient has the right to terminate a pregnancy for other personal reasons up to 24 weeks gestation.

Patients should be informed of the small chance of a false positive result.