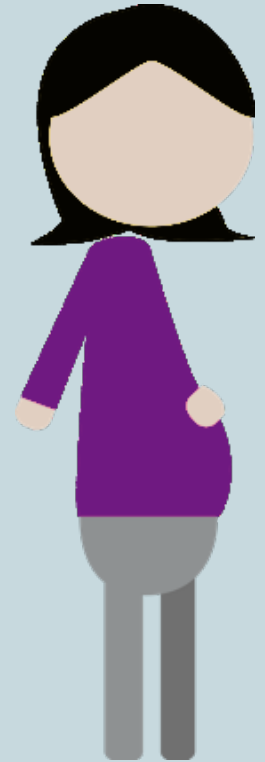


Case scenario 10 – Magdalena

Part 1

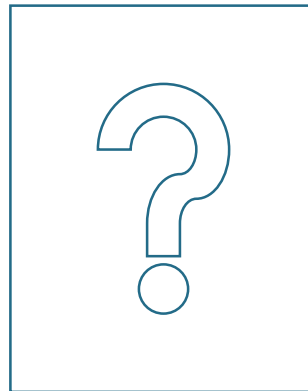
- Magdalena is 26 years old. She is 13 weeks pregnant and had a high chance combined test result of 1 in 140 for trisomy 13 with an NT measurement of 2.4mm.
- After a lengthy discussion about her options, she decided to have NIPT.
- The results reported a greater than 60% chance of the baby having Patau's syndrome. She decided to have an invasive test. A CVS was performed.



Would you have offered a CVS to this patient?

- a) No - she should be offered an amniocentesis only, due to the chance of placental factors affecting a result.

- b) Yes - the patient should be informed of risks and benefits of both CVS and amniocentesis.

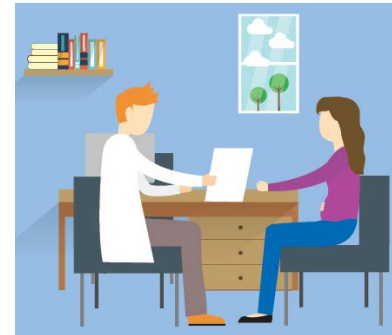


Answer

b) Yes - the patient should be informed of the risks and benefits of a CVS and amniocentesis.

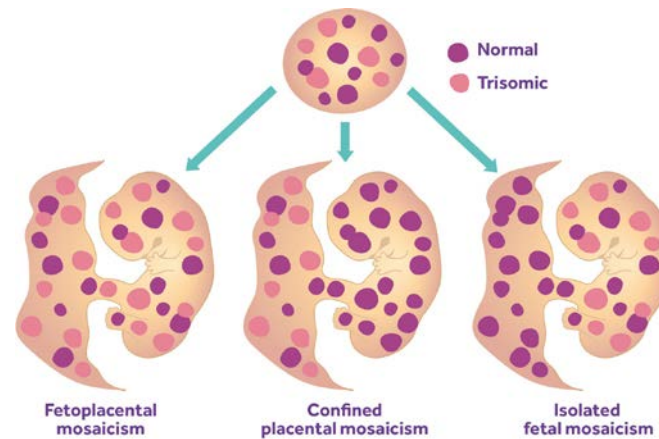
Additional note

A patient/couple should be informed of the options of both forms of diagnostic procedures. They should be informed of the benefits and limitations of each test, this should include timing of testing, possible results and risks of miscarriage. This discussion should also include the couples ethical, religious, social and individual beliefs.



Name the type of mosaicism that can cause a false-positive result with NIPT.

- a) Feto-placental mosaicism
- b) Placental mosaicism
- c) Fetal mosaicism

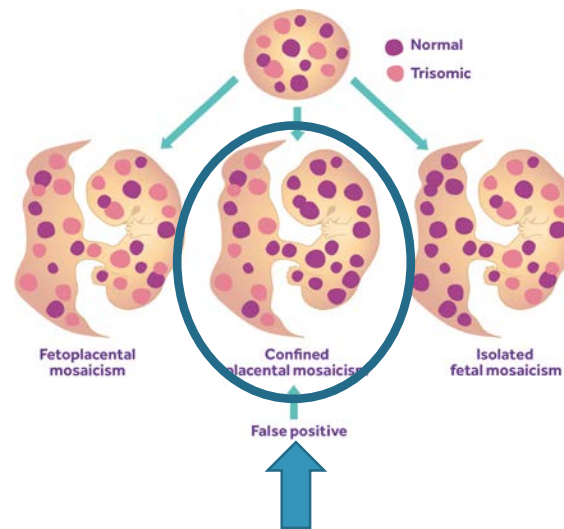


Answer

b) Placental mosaicism

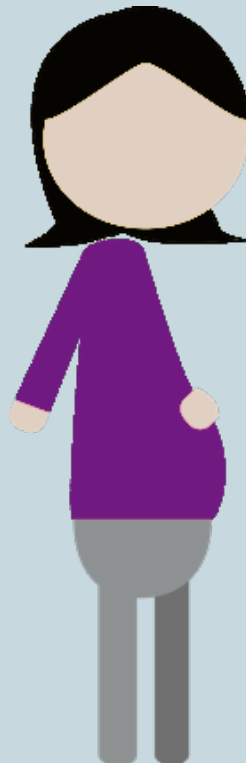
Additional note

There is a discrepancy of the cell line in the placenta and baby. The abnormal cell lines are seen in the placenta and not on the fetus. There is a small chance that a 'high chance report is caused by 'placental mosaicism', therefore a CVS may also report 'mosaicism'.



Magdalena – Part 2

The CVS reported a normal PCR – the baby does not have Patau's, Edwards' or Down's syndrome.



What is the definition of a false-positive result?

- a) When the pregnancy is affected with a condition, but the test indicates it is not
- b) The number of pregnancies correctly identified as being affected
- c) When the pregnancy is not affected with a condition, but the test indicates it is
- d) When the pregnancy is affected with a condition but the test fails

Answer

c) When the pregnancy is not affected with a condition, but the test indicated it is

		prediction outcome		total
		p	n	
actual value	p'	True Positive	False Negative	P'
	n'	False Positive	True Negative	N'
total		P	N	



Which of the following are maternal causes for a false-positive result?

- a) Vanishing twin
- b) Malignancy
- c) Maternal duplications
- d) Maternal balanced translocations
- e) Maternal complete or partial trisomy
- f) All of the above



Answer

- b) Malignancy
- d) Maternal balanced duplications
- e) Maternal complete or partial trisomy

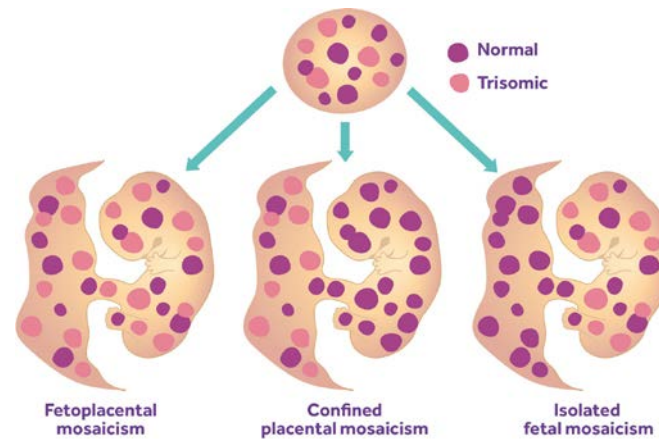
Additional note

Maternal duplication or mosaicism- a high proportion of cfDNA for T13, 18 or 21 circulating from the mother. The technology cannot separate fetal and maternal cfDNA.

Although very rare, cancer cells have major abnormalities and screening may detect this as a high chance result.

Name the type of mosaicism that can cause a false-negative result with NIPT.

- a) Feto-placental mosaicism
- b) Placental mosaicism
- c) Fetal mosaicism



Answer

c) Fetal Mosaicism

Additional note

A discrepancy of the cell lines in the placenta and baby. The baby is affected with an abnormal cell line (chromosome anomaly), but the placenta has a normal cell-line. Although this is very rare, isolated fetal mosaicism can lead to a false negative cfDNA result and diagnostic CVS test result.

