The value of non-invasive prenatal testing (NIPT).

A supplement for a Genetic Counselor's flipbook

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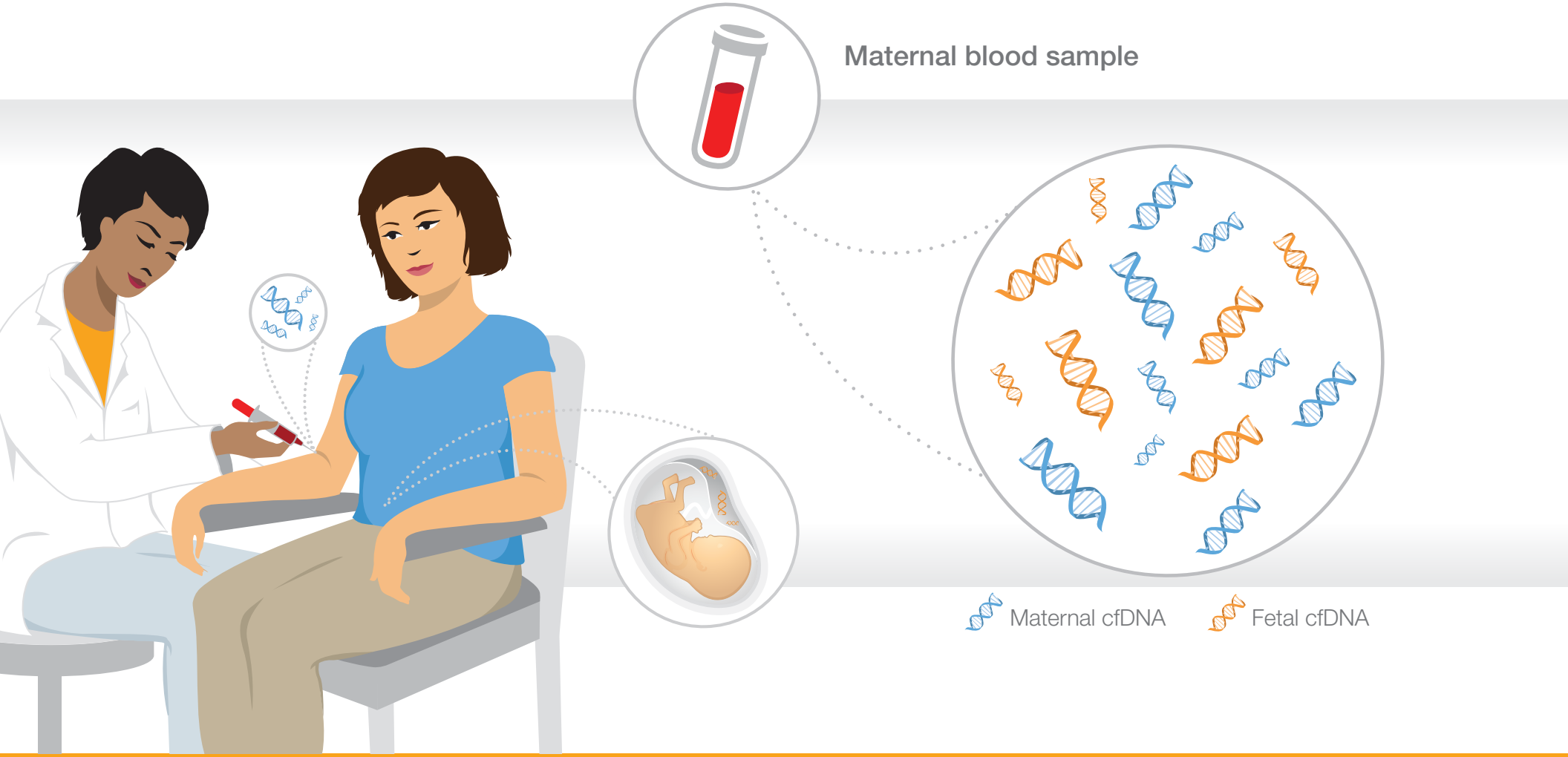
your questions – answered

NIPTs use cell-free DNA.



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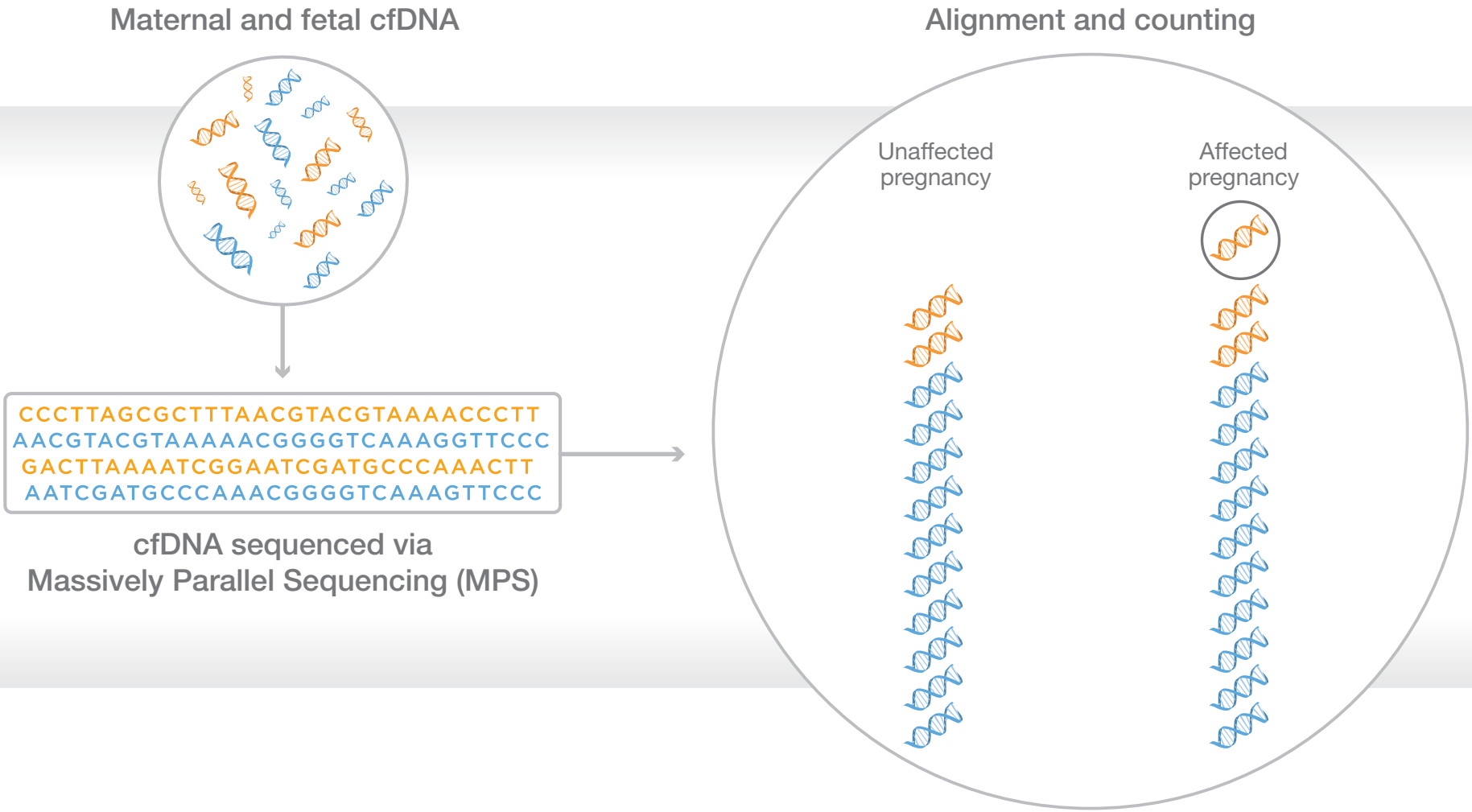
NIPTs use cell-free DNA.

- The maternal blood sample contains mixture of both maternal and fetal cfDNA.
- Both maternal and fetal fragments are counted and analyzed in NIPT.
- Aneuploidies are detected by comparing the amount of chromosomal material against a set of reference chromosomes.

MPS — a deeper approach to sequencing.



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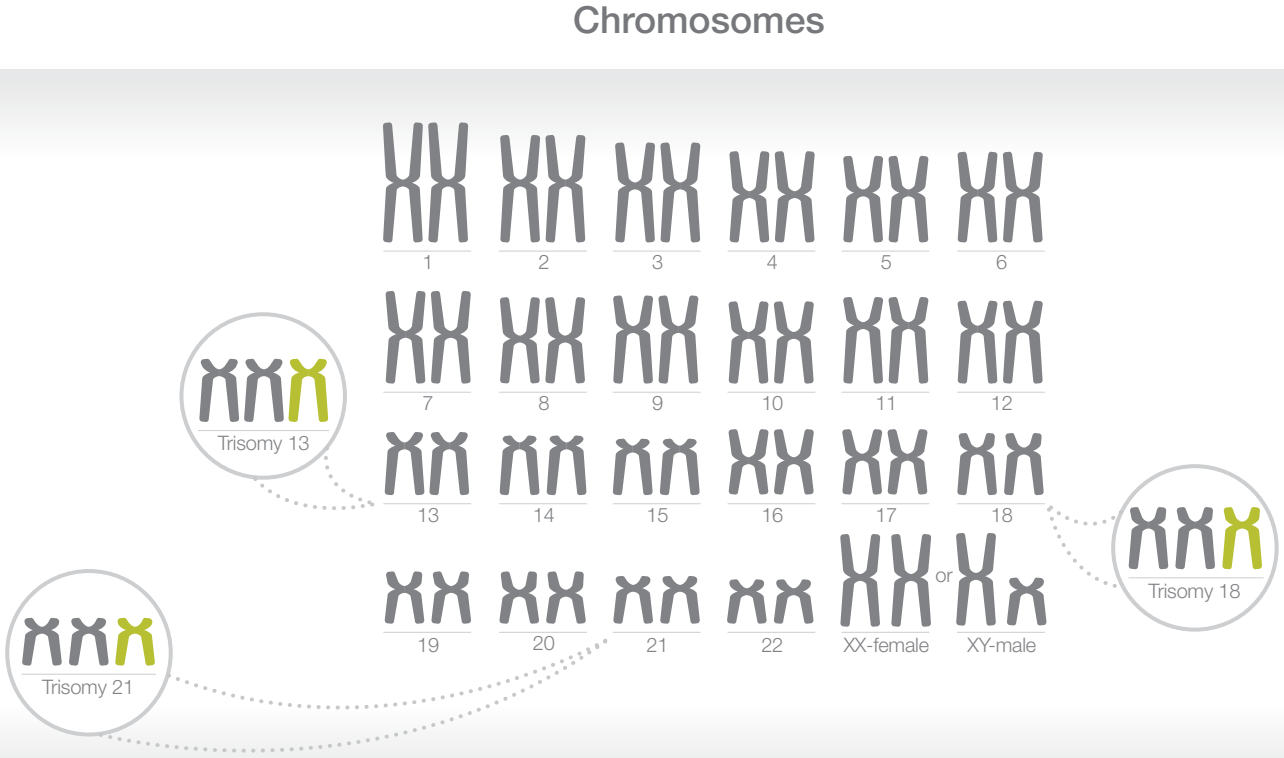
MPS — a deeper approach to sequencing.

- Maternal blood sample contains a mixture of both maternal and fetal cfDNA.
- MPS, or Massively Parallel Sequencing, analyzes cfDNA fragments from a maternal blood sample.
- Sequences are aligned to genome to identify the chromosome origin of the fragment.
- Aneuploidies are detected by comparing the amount of chromosomal material for a specific chromosome in a patient sample against a set of reference chromosomes.

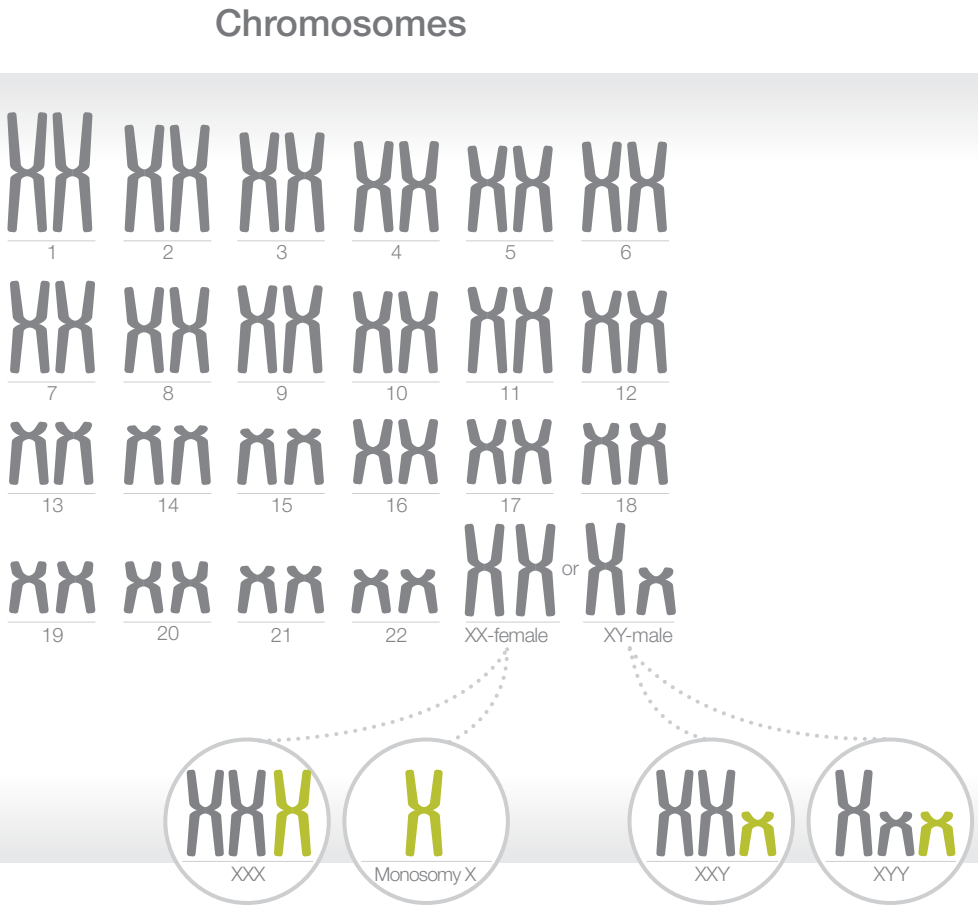
Detecting fetal chromosome abnormalities.



Detecting fetal chromosome abnormalities.

- The verifi® prenatal test detects trisomies 21, 18, and 13 in both singleton and twin pregnancies.

Detecting sex chromosome abnormalities.



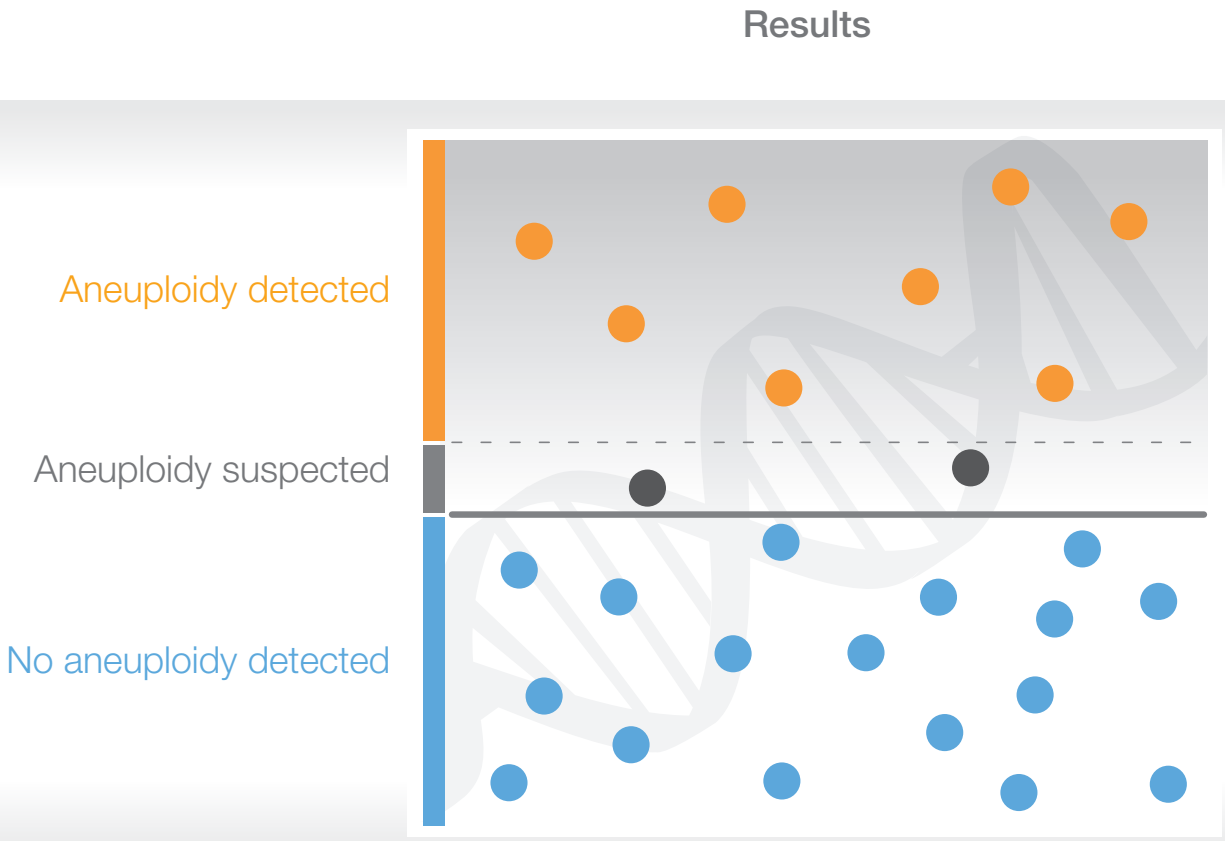
Detecting sex chromosome abnormalities.

- In singleton pregnancies, sex chromosome aneuploidies can also be detected. This test is optional based on patient and physician preference.

  – Sex chromosome aneuploidies detected include 45,X, 47,XXX, 47,XXY, 47,XYY.

- In twin pregnancies, the presence or absence of Y is detected. This test is optional based on patient and physician preference.

How the verifi® test is reported.



Test results.

There are three possible results for the autosomes:

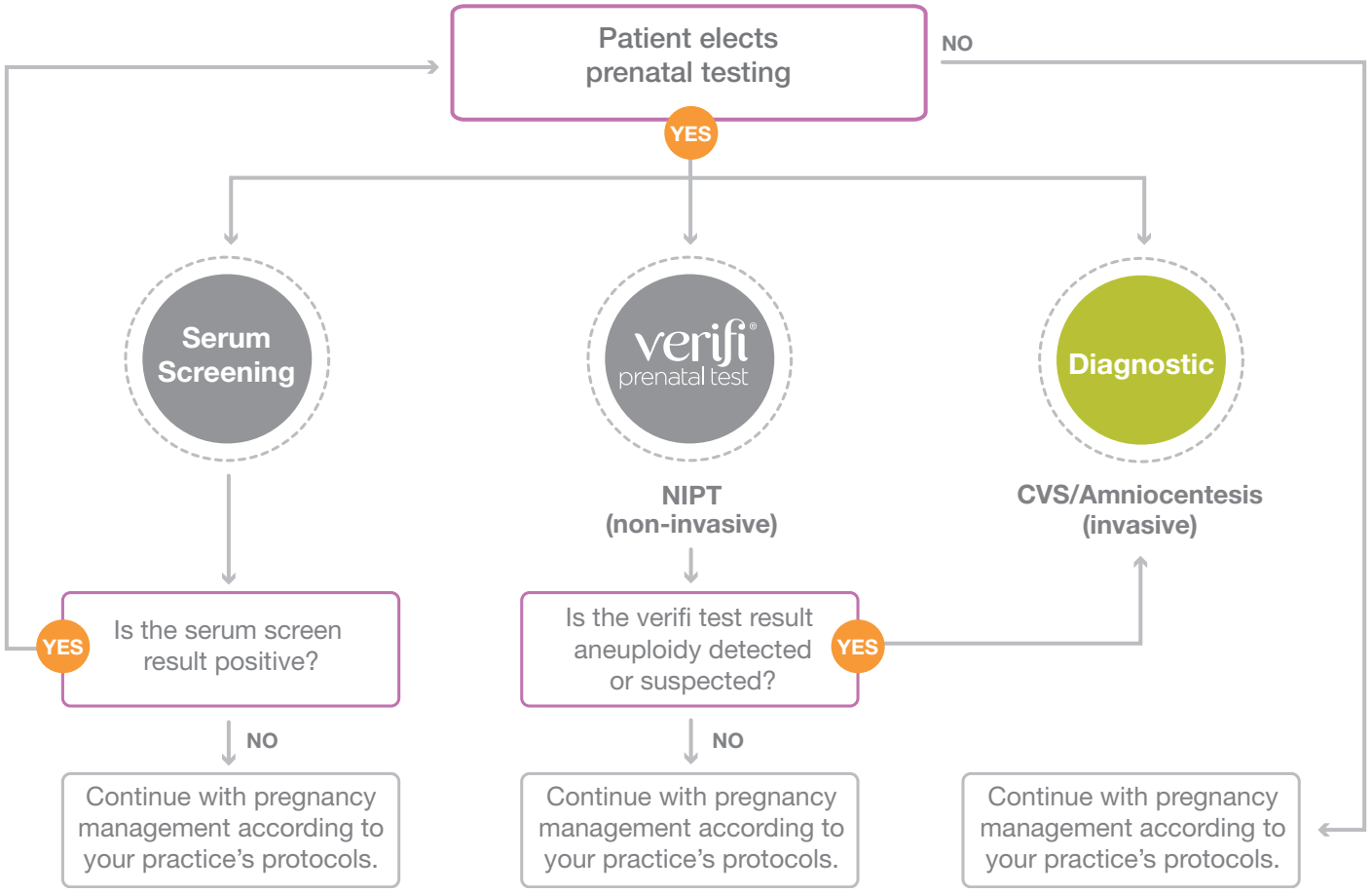
- Aneuploidy Detected, Aneuploidy Suspected (borderline value), Aneuploidy Not Detected.

There are two possible results for the sex chromosome analysis in singletons:

- Aneuploidy Detected and Aneuploidy Not Detected.
  - If aneuploidy is not detected, fetal sex will be reported.
  - For twin pregnancies, the presence or absence of Y will be reported.

Patients who receive an Aneuploidy Detected or Suspected result should be offered a follow-up discussion of results and options of invasive testing (CVS or amniocentesis) for confirmation.

Typical prenatal testing sequence.



Typical prenatal testing sequence.

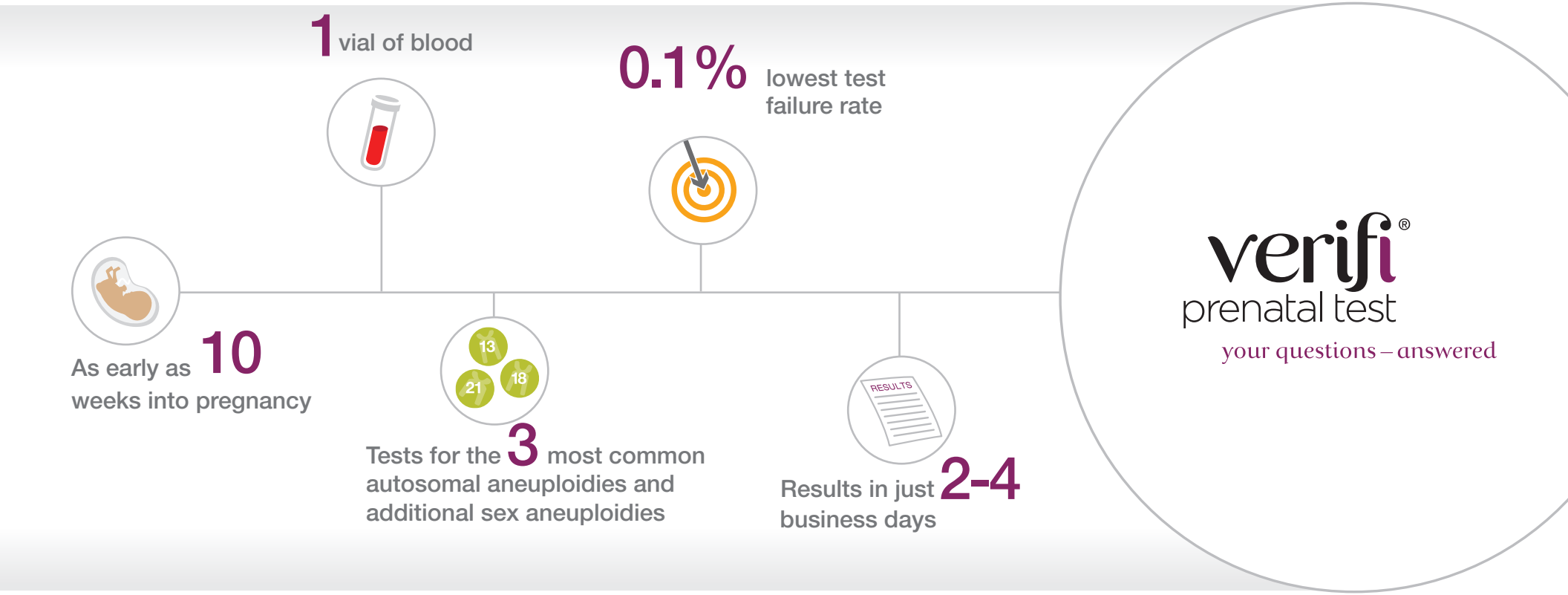
Pre-test Counseling Considerations

- Aneuploidy testing should be an informed choice.
- The verifi prenatal test screens for aneuploidies of chromosomes 21, 18 and 13. Sex chromosome testing is optional.
- Any test offering should be based on clinical context.
- CVS and amniocentesis remain options for all pregnant women.

Post-test Counseling Considerations

- 3 possible results for each chromosome tested.
  - **No Aneuploidy Detected** – This result is reassuring but does not ensure an unaffected pregnancy.
  - **Aneuploidy Suspected/Borderline Value** – This result is suggestive of aneuploidy. There is a small possibility that the results might not reflect the chromosomes of the fetus but may reflect changes to the placenta or the patient.
  - **Aneuploidy Detected** – This result is consistent with aneuploidy. There is a small possibility that the results might not reflect the chromosomes of the fetus but may reflect changes to the placenta or the patient.
  - Genetic counseling and CVS/amniocentesis are recommended for Suspected and Detected results.
- Open neural tube defect screening is not a part of NIPT testing and should be offered separately.
- All results should be reviewed in the context of the clinical, medical, and family history.

NIPT profile: the verifi® test by the numbers.



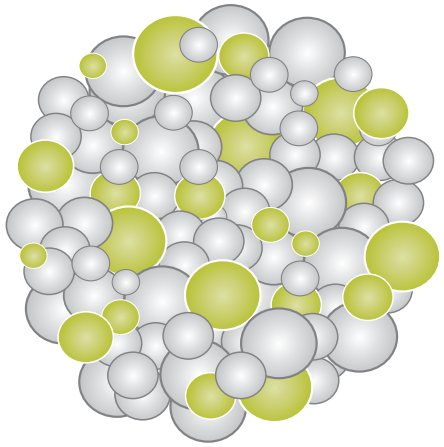
What you can't put a number on –
the reassurance of knowing

How is the verifi® prenatal test different?

- The verifi® test is an easy, in-office blood test – only one tube of blood is required.
- It can be performed as early as 10 weeks in pregnancy.
- It tests for trisomies 21, 18 and 13 and has an option for sex chromosome aneuploidies (Monosomy X, XXX, XXY, XYY).
- Test reports are usually available in 2 – 4 business days after sample receipt.
- It has the lowest failure rate among NIPTs (0.1%).

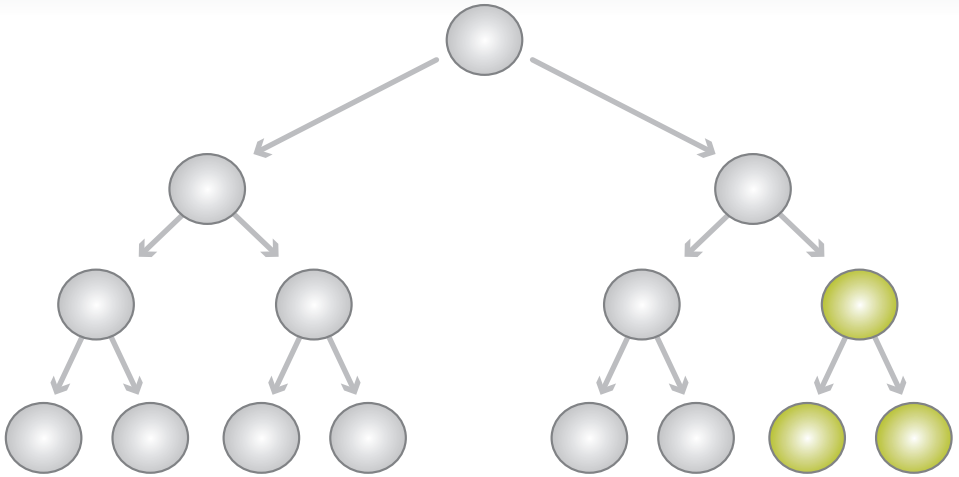
Because of this, the verifi® test is able to provide reliable answers about the most common chromosome abnormalities.

What is mosaicism?



Mosaic tissue

Cell division



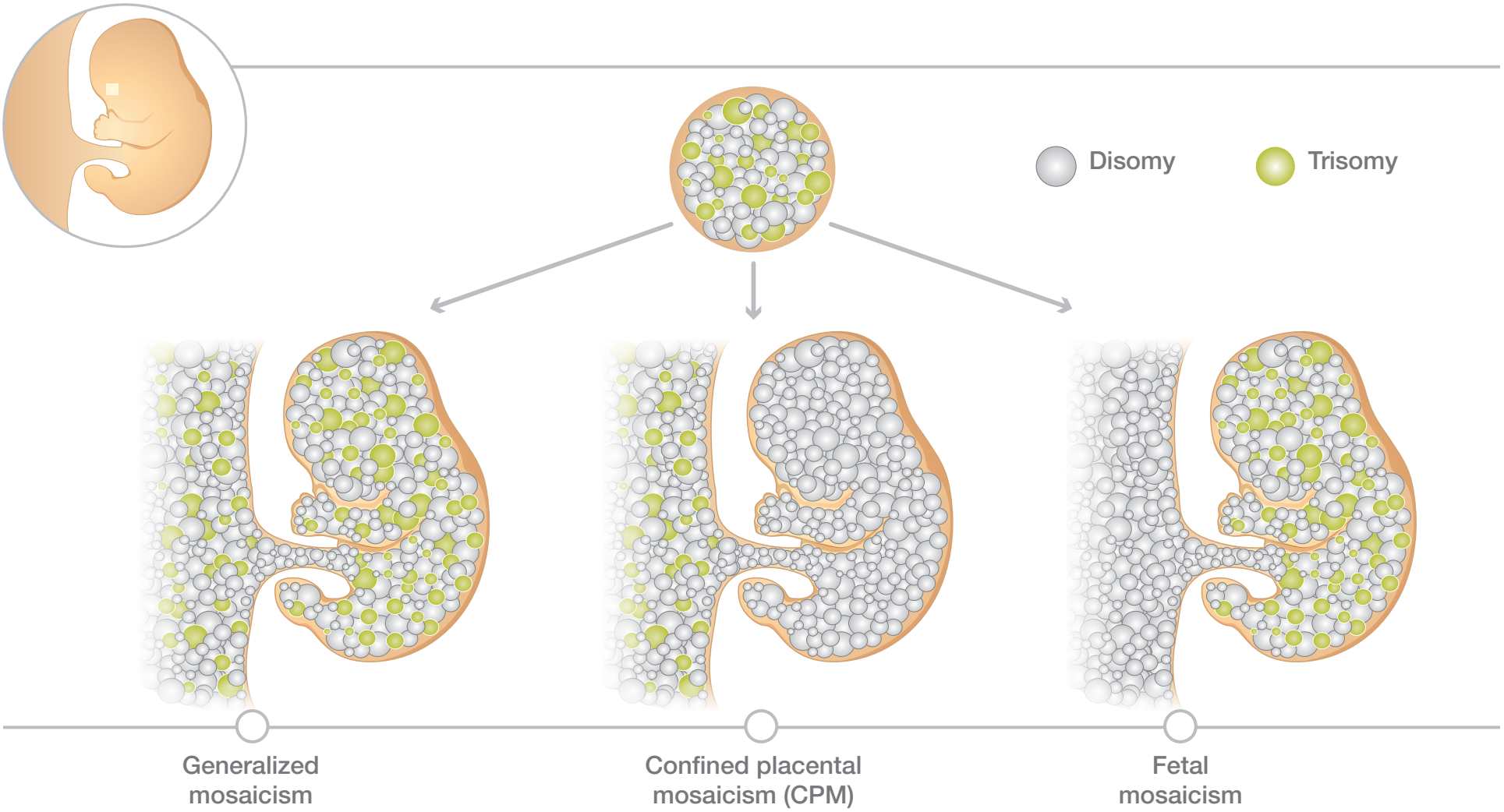
Normal cells

Cells with genetic change

What is mosaicism?

- Mosaicism is a biological phenomenon which results from the presence of cells with differing genetic compositions in the same individual. It usually occurs during the process of mitotic cell division.
- Mosaicism can be an underlying biological cause of discordant NIPT results.

Types of mosaicism.

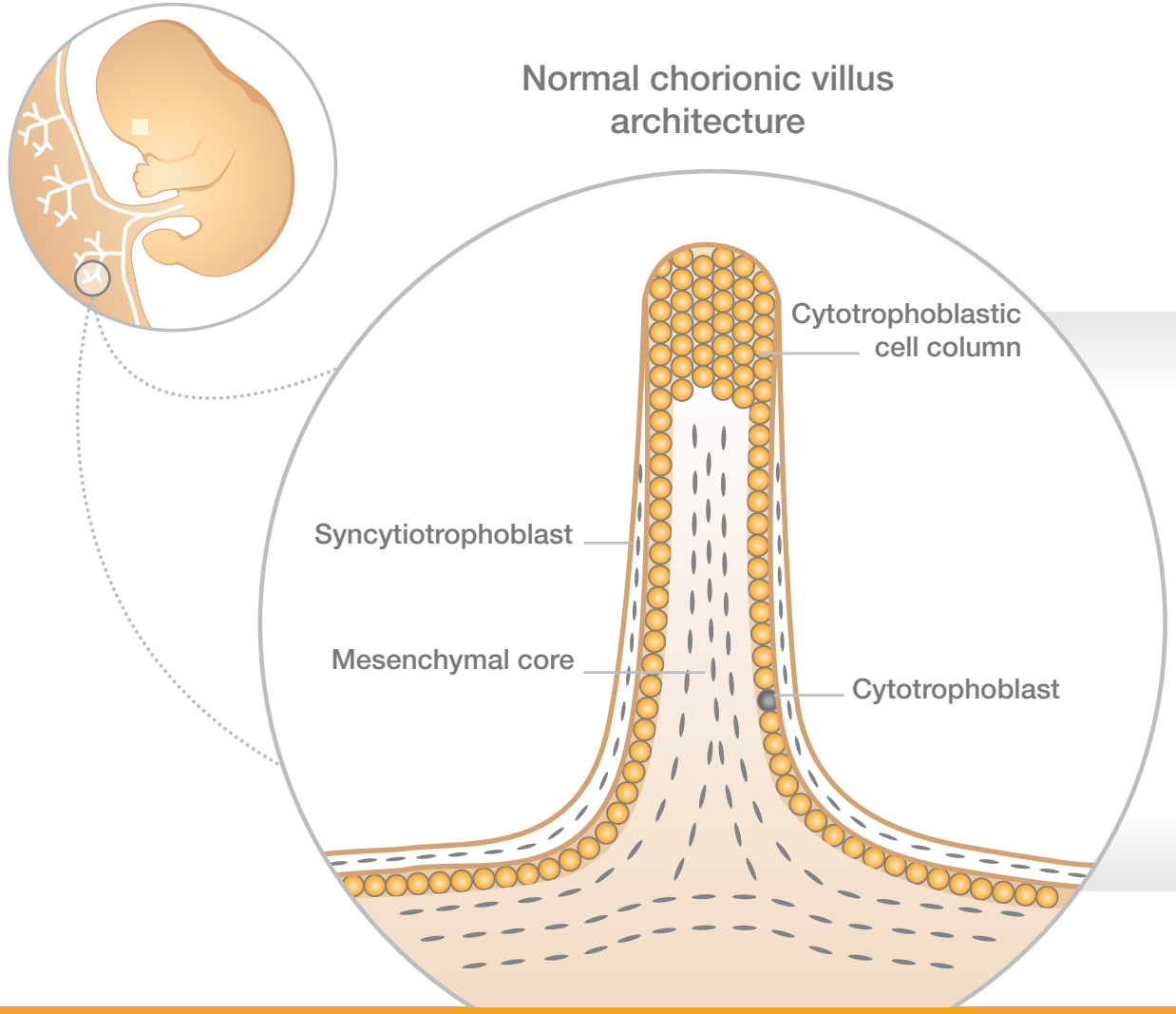


Types of mosaicism.

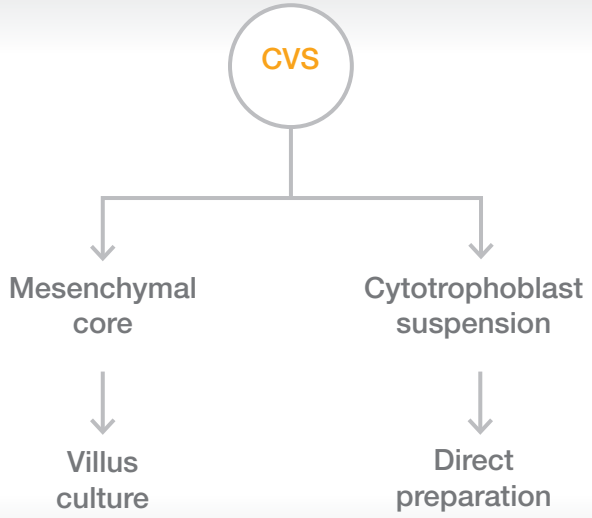
There are three types of mosaicism:

- **Generalized mosaicism** – Presence of two or more karyotypically different cell lines in both the placenta and the fetus. In cases of generalized mosaicism, there is a possibility of a false positive or a false negative NIPT result depending on the origin of the cfDNA.
- **Confined placental mosaicism** – Presence of two or more karyotypically different cell lines that are confined to the placenta and not present in the fetus. In cases of confined placental mosaicism, there is a possibility of a false positive NIPT result.
- **Fetal mosaicism** – Presence of two or more karyotypically different cell lines that are present in the fetus but not present in the placenta. In cases of fetal mosaicism, there is a possibility of a false negative NIPT result.

Confined Placental Mosaicism.



Laboratory techniques used for Chorionic Villus Sampling (CVS)



Confined Placental Mosaicism (CPM).

- Confined placental mosaicism (CPM) is defined as the presence of two or more karyotypically different cell lines that are confined to the placenta and not present in the fetus.
- The cfDNA that is analyzed in NIPT arises from the cytotrophoblasts; thus an NIPT result is most similar to a CVS direct preparation result.
- **Type 1 CPM** – Mosaicism is confined to trophoblastic cells.
- **Type 2 CPM** – Mosaicism is confined to the cells from chorionic villus stroma.
- **Type 3 CPM** – Mosaicism is present in both: trophoblastic and stroma cells.
- Type 1 and Type 3 CPM could result in false positive NIPT results.

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