

Module 5: cfDNA Screening Test Results

Module 5 will cover:

- Low chance results
- High chance results
- What causes a high chance result
- Care pathway for a high chance result

What does a low chance result mean?

A cfDNA result is a risk result - it cannot give a yes/no answer

LOW CHANCE IS NOT ZERO CHANCE

- The SAFE test result is reported as a ratio and percentage.
- A low-chance result means that it is very unlikely that the pregnancy is affected with a trisomy.
- This result is not a diagnosis test, it is a screening test.
- With a low-chance result, routine antenatal care is recommended, unless otherwise specified by the hospital or clinic.
- It is extremely rare but very occasionally there can be a false negative result (FNR).
- This occurs when the mother is informed the fetus is low chance, but later in the pregnancy it is discovered that the fetus has the condition.

What does a high chance result mean?

A high chance result is given as a risk ratio and percentage. It means that there is a >1:150 chance that the baby is affected with a trisomy.

The SAFE test uses a cap for the higher end of risk/chance. This means that the highest risks provided are >95% for trisomy 21, >75% for trisomy 18 and >60% for trisomy 13.

TEST RESULTS:			
TRISOMY	*BACKGROUND RISK (before the SAFE test)	SAFE TEST RISK SCORE	CLINICAL SUMMARY
Trisomy 21	1 in 38 ^{PT}	Greater than 95%	HIGH CHANCE — INVASIVE TEST RECOMMENDED
Trisomy 18	1 in 537 ^{PT}	Less than 1 in 1,000,000 (<0.0001%)	LOW CHANCE
Trisomy 13	1 in 537 ^{PT}	Less than 1 in 1,000,000 (<0.0001%)	LOW CHANCE

the SAFE test is indicated for screening NOT diagnosis — (results should be reviewed and discussed with the healthcare provider)

The result will be available on the MyNIPT[®] portal to download. It is clearly highlighted in red to alert the healthcare professional. On the screening report it states:

HIGH CHANCE - INVASIVE TEST RECOMMENDED

What can cause a high chance result?

There are numerous biological reasons that can cause a high chance result:

- An affected pregnancy (true positive)
- An unaffected pregnancy (false positive)
- A 'vanishing twin'
- Confined Placental Mosaicism (CPM)
- Maternal factors (cancer, immunotherapy, organ transplant, maternal mosaicism)

These will be discussed in detail in module 7.



Recommended care pathway for a high chance result

Confirm patient details are correct

Inform of result

Offer appointment for a face to face discussion

Advise:

- Frame the chance result according to percentages that the mother or her family would understand
- Provide written information and contacts for national support groups

Options:

- 1) Routine antenatal care
- 2) CVS or Amniocentesis