

To confirm, your result has shown that the chance of your baby having Down syndrome is 1 in 78 which equates to around 1.3%. This means there is a 98.7% chance that your baby does not have Down syndrome. The Edward and Patau syndrome screening result was lower chance at 1 in 5,000.

The options we discussed are:

Have no further tests

Some women prefer to leave any further testing and just wait to see when the baby is born

Non-invasive prenatal testing (NIPT)

This is a more accurate method of screening and is performed by looking for placental DNA in a sample of your blood. Whilst this test is not 100%, it will give a more accurate estimation of the chance of your baby having Down, Edward or Patau syndromes than the test you have already had. However, sometimes it is not possible to get a result, and the test has to be repeated. Occasionally a result cannot be obtained at all. There is no risk of miscarriage with this option.

Diagnostic Testing

The only way we can let you know *for definite* whether your baby has Down syndrome or not is by an invasive test either Chorionic villus sample (CVS) or Amniocentesis. The CVS test is performed at St Mary's hospital in Manchester as soon as we can arrange it and has a miscarriage risk of 2-3%. The amnio is performed at Thomas Linacre Centre from 15 weeks and has a miscarriage risk of 0.5-1%.

The links below will take you to the national leaflets explaining the options we discussed in more detail

<https://www.gov.uk/government/publications/downs-syndrome-edwards-syndrome-and-pataus-syndrome-options-after-a-higher-chance-screening-result/your-choices-after-a-higher-chance-screening-result>

<https://www.gov.uk/government/publications/cvs-and-amniocentesis-diagnostic-tests-description-in-brief>

There is no rush to make a decision and please bear in mind that there is far more chance of your baby having 46 chromosomes than there is of it having the extra chromosome present in Down syndrome.

I would be more than happy to meet with you to discuss the results face to face if you wish to talk your options through in more detail. Please do not hesitate to contact me on the number/s below if you wish to arrange an appointment or have any questions, otherwise I will await your decision.