Patient Information

Following your Down’s and/or Edward’s/Patau’s Syndrome Screening Test Result

Your result has shown you to be of increased chance of having a baby with Down’s and/or Edward’s/Patau’s syndrome.

For most people this will not have been anticipated and you may not have thought about what you want to do now. In this section there is a list of common questions that people ask and some information which may help you to decide what you would like to do.

What does my screening test actually mean?

The screening test has not told you anything definite about your baby. Everybody who has a screening test will be given a result expressed in the form of their chance of having a baby with Down’s and/or Edward’s/Patau’s syndrome. The reason we offer the test is to identify a group of women to whom we offer a further ‘diagnostic’ test (either chorionic villus sampling or amniocentesis). These ‘diagnostic’ tests can tell you if your baby has Down’s, Edward’s or Patau’s syndrome but they also carry a chance of miscarriage. Everyone who has an increased chance screening test result is offered one of these tests.

Only a very small percentage of babies in the increased chance group will have a syndrome. For example if you were told your chance is 1:100 this represents only a 1% chance of having a baby with this syndrome. If your chance is 1:10 there is a 10% chance. It is also the case that there will still be some babies with this syndrome in the lower chance group who have not been identified by the screening process.

What do most women do in my situation?

Every woman’s circumstances and life experience is different. The important thing is to be able to have all the information and support you need to make the right decision for you.

It may not be possible for you to know at this stage what you would do if you found out that your baby did have Down’s, Edward’s or Patau’s syndrome. Many couples find that until they are faced with that reality it is impossible to really know how they would feel.

If you are certain what you would do, then deciding whether or not to have a diagnostic test may be easier. If you know for sure that you would continue a pregnancy knowing your baby has this syndrome you need to weigh up the benefits of knowing in advance with the risk of miscarriage. If you are sure you would wish to end the pregnancy, then you need to balance the chance of having a miscarriage with the possibility of having a baby with a syndrome that you would not have chosen to have.

It may be helpful to talk things through with your own midwife. Alternatively a midwife who specialises in screening is available for you to meet with at the Fetal and Maternal Assessment Unit. ARC (Antenatal Results and Choices) offers a telephone helpline to support people with their decision making. The Down’s syndrome Association and Support Organisation for Trisomy (SOFT) organisation can provide information and support and their contact details are at the back of this leaflet.
There is also an option of a private blood test called ‘Cell Free DNA test’ or Non-Invasive Prenatal Test (NIPT) which is an advanced screening test for Down’s, Edwards’ & Patau’s syndrome, with better detection rates than the current NHS screening test. Although you would have to pay for this test, we can perform it at the Centre for Women’s Health and send the sample to a laboratory in the UK. If you wish you can look online at available options or ask the midwives on FMAU if you wish to find out more.

What exactly do the further diagnostic tests involve?

A separate leaflet is available on these tests. If you have not received it please contact the Fetal and Maternal Assessment Unit on 01392 406540.

How soon can I get the result of a CVS or Amniocentesis?

The result, which will tell you about Down’s, Edwards’ and Patau’s syndrome should be available 3-4 working days following the test. If additional problems are seen on scan an additional test may be performed which will take about 2 weeks.

How accurate is the result?

The result is extremely accurate for these syndromes.

What will happen if the test shows my baby has one of these syndromes?

You will be offered an appointment as soon as possible to come and discuss the result with an obstetrician at the hospital. You can make the decision about what to do next in your own time. At the hospital we are able to give you booklets produced by ARC which may help you with your decision making. Again many people find their helpline service useful.

What are my options if I wanted to end the pregnancy?

The ARC booklet gives you some detailed information about the medical and surgical options available which we will discuss further with you.

If you would like any further information or wish to make an appointment to discuss your result please telephone the Fetal and Maternal Assessment Unit on 01392 406540.

Useful organisations and websites

- Antenatal results and choices
  www.arc-uk.org
  Tel: 0845 0772290

- Down’s syndrome Association
  www.downs-syndrome.org.uk
  Tel: 0845 230 0372

- Support Organisation for Trisomy 13/18 (SOFT)
  www.soft.org.uk
  Tel: 0121 351 3122

Reference Number: RDE 19 057 001 (Version date: September 2019)