First trimester screening clinic

Women's Health & Maternity Unit

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Yeovil Hospital Healthcare

This booklet gives you information about screening tests for Down's Syndrome (Trisomy 21), Edwards' Syndrome (Trisomy 18) and Patau's Syndrome (Trisomy 13). These are offered between 11-14 weeks of your pregnancy.

This information aims to help you to make an informed choice regarding your screening options.

Please read this booklet prior to attending the First Trimester Screening Clinic and bring your hand held pregnancy notes with you to the appointment.

What is Trisomy?

Inside all the cells of our bodies are tiny structures called chromosomes. Humans have a total of 46 chromosomes – 23 pairs, including the sex chromosomes.

Chromosomes are the structures that hold all of our genes. Genes are simply the instructions that tell our body how to grow and develop, and determine things like the colour of our eyes and hair.

In a chromosomal condition such as a Trisomy a person has three copies of a chromosome in each cell of their body, instead of the usual two. In Down's Syndrome a person has three copies of chromosome number 21, in Edwards' Syndrome a person has three copies of chromosome number 18 and in Patau's Syndrome a person has three copies of chromosome number 13.

Trisomy 21, 18 and 13 are not diseases or a hereditary condition (they are not passed down through generations of a family); it is caused by chance at conception. As yet we do not know what causes the extra chromosomes to be present, they can come from the mother or father, but we do know it occurs more commonly when mothers are over the age of 35 years.

What are Down's, Edwards' and Patau's Syndromes?

Down's Syndrome

Babies and children with Down's Syndrome can be affected with learning disabilities along with medical conditions such as

thyroid problems, cardiac issues, hearing and vision difficulties, among others, which may require medical input. The life expectancy of people with Down's Syndrome is 50-60 years with some people living into their 70s.

Edwards' Syndrome

Babies with Edwards' Syndrome can be affected with multiple anomalies, including structural heart defects, breathing and feeding difficulties, growth restrictions, cleft lip and palate, Neurological problems - severe learning disabilities, seizures, jitters and gastrointestinal problems among others.

Patau's Syndrome

Babies with Patau's Syndrome can also be affected with multiple anomalies which may include, congenital heart defects in 80 per cent of babies, midline facial defects, including cleft lip and palate, small eyes which are closer to the centre of the face and nasal malformation; along with neural tube defects, urogenital, gastrointestinal problems and also abnormalities of the hands and feet.

50 per cent of Edwards' and Patau's Syndrome babies won't survive to term (37-40 weeks of pregnancy) and there is an even slimmer chance of the babies surviving birth. If an Edwards' or Patau's Syndrome baby survives birth, their life expectancy is usually a few days/weeks. One in 10-12 will survive to see their first birthday; there is a ten per cent chance of survival after one year.

How common are Trisomies?

Some People think that only older women can have a baby with a Trisomy, but this is not true. Anyone can have a baby with Down's, Edwards' or Patau's Syndrome; however the risk does increase with maternal age.

Down's Syndrome occurs around 1 in every 1000.
Edwards' Syndrome occurs 3 in every 10,000 live births.
Patau's Syndrome occurs 2 in every 10,000 live births.

Will the screening test tell if my baby definitely has a Trisomy?

No it will not. We begin by offering all women a screening test for Trisomy 21, 18 and 13 that carries no risk of miscarriage. Screening tests do not give us a definite answer; instead they help to tell us which babies have an increased chance of having a Trisomy by calculating a risk score.

What does the screening test involve?

There are two parts to the screening test; the first is a blood test. This measures the amount of some substances which are released by the pregnancy naturally into the mother's blood. These substances have passed from the baby to mother and measuring them can help to determine your risk of having a baby with Trisomy 21, 18 and 13.

The second part of the test is an ultrasound scan; in addition to dating your pregnancy the sonographer will measure the thin film of fluid under the skin at the back of the baby's neck. This is known as the nuchal translucency (NT). Babies with a Trisomy can have an increased amount of fluid in this area.

The results are combined with your age, weight and stage of pregnancy to work out the risk of your baby having a trisomy. This is known as the combined test and your result is given as a ratio figure, for example 1:100.

Please note: Having a thickened nuchal translucency (NT) can be associated with other fetal anomalies, such as heart defects. If you have chosen not to have the screening for trisomy 21, 18 or 13 and are booked for a dating scan only, the sonographer will inform you if the NT appears larger than expected and may suggest they measure the thickness of the NT as a measurement above 3.5mm can be linked to other problems for your baby.

What is a high risk result?

If your risk is less than 1:150 then it will be classed as a high risk result and you will be offered further testing. If it is more than 1:150 then it will be classed as a low risk result and no further tests will be required.

For example if your risk is 1:50 it is a high risk result and you will be offered further tests. If your risk is 1:1000 it is a low risk result and you will not be offered any further testing.

Most screening test results (around 97 per cent) will fall into the low risk category. If you fall into the low risk category it does not mean your baby won't be affected by a Trisomy, just that the chance of it happening is low. Some babies with a Trisomy (about five per cent) are not identified through this screening process.

How long will it take to get my results?

Your results are usually available within three to five days of having the blood sample taken. High risk results are telephoned to you directly so you will need to make sure we have your correct telephone numbers at the time of the screening test. Low risk results are sent to you via post to be put into your hand held notes ready to show your midwife or consultant at your next appointment.

What if I have a high risk result?

If the result of the screening test shows that your chance of having a baby affected by a Trisomy is high (ie. less than 1:150) you will be offered the opportunity to come and talk to the Antenatal Screening Midwife and/or a doctor about your results.

You will be offered a diagnostics test (CVS or an amniocentesis) which the antenatal screening midwife or doctor will explain to you in detail.

You can decide not to have a diagnostic test but this leaves you with the uncertainty of not knowing if your baby will be born with a Trisomy.

You can decide to have the diagnostic test; this will give you a definite answer but carries a risk of miscarriage in one per cent of cases.

What are diagnostic tests?

The only way to be certain whether a baby has a Trisomy or not, is by performing a diagnostic test – an amniocentesis or a Chorionic Villus Sampling (CVS).

A CVS is performed from 11 weeks and involves taking a sample form the placenta; an amniocentesis involves taking a sample of fluid that surrounds the baby and is usually done after 15 weeks of pregnancy.

Both provide a sample that contains tissue that has the same genetic make up as the baby and will allow the baby's chromosomes to be looked at in detail.

However, there is a risk to undergoing these invasive tests and they can cause a miscarriage. For every 100 women who have and amniocentesis one will miscarry and for every 100 women who have a CVS one or two will miscarry.

If you are offered one of these diagnostic tests you will be given more detailed information and literature at the time. If you feel you want to see this literature before making your decision about screening please call 01935 384 856.

What else will happen at the screening clinic?

If you have not already had them taken during your first midwife appointment, the assistant practitioner will also take your routine pregnancy bloods.

These include finding out your blood group and full blood count (iron levels) as well as offering you infection screening for HIV, Hepatitis B and Syphilis. You will also be offered screening for other inherited blood disorders. More information about all of these tests can be found on page six of your pregnancy notes.

You will also be weighed whilst at the clinic, as your weight on the day of the test is needed to accurately calculate your results for Trisomy screening.

You can get more information about screening and Down's Syndrome from these organisations:

Antenatal Results and Choices ARC
Website: arc-uk.org
Helpline: 0207 631 0285

AnSWeR
Website: antenataltesting.info

DipEx
Website: dipex.org/antenatalscreening

Contact a Family
Website: cafamily.org.uk
Helpline: 0808 808 3555

Down's Syndrome Association
Website: www.downs-syndrome.org.uk
Phone: 020 8682 4001

More information about screening for Trisomy's

If you want to talk with someone before making your decision you can contact the antenatal screening coordinator, a midwife who specialises in screening tests in pregnancy, on 01935 384 261 or 07990 663 302.

If you would like this leaflet in another format or in a different language, please ask a member of staff.

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