Pathology Request Form Maternal Serum Test

The Combined Screen for Down Syndrome and Trisomy 18

Victorian Clinical Genetics Services Limited (APA)

10th Floor, Royal Children's Hospital, Flemington Road, Parkville, Victoria 3052 Phone (03) 8341 6303

a NATA/RCPA accredited laboratory No. 3171

I.N.A/2196

Surname _____ First name _____

Hospital number _____ Date of birth _____/_____/_____

Medicare Number _____

Address _____

_____ Post Code _____

Telephone (0___) _____ _____ Current Weight: _____ kg

Ethnic Group: ☐ European ☐ Aboriginal ☐ Asian ☐ Afro-Caribbean ☐ Other

LMP_____/_____/_____ ☐ Certain ☐ Uncertain or EDD_____/_____/_____

Previous pregnancy with a chromosomal abnormality? ☐ YES ☐ NO

If YES, please specify:

Complications of this pregnancy:

☐ None ☐ Diabetes (IDDM only)
☐ Threatened Miscarriage
☐ Multiple Pregnancy
☐ IVF
☐ Other, specify:

Ultrasound booked for:

Date of scan: _____/_____/_____
At:_____
_____
_____
_____

Requesting Practitioner _____ Provider No. _____

Address _____

Telephone Number (0___)_____ Facsimile number (0___)_____

Signature _____ Date of Request _____/_____/_____

Copy of report to_____

PATIENT CLASSIFICATION Patient status at the time of the service or specimen collection

☐ Private patient in a private hospital or approved day hospital facility
☐ Private patient in a recognised hospital
☐ Hospital patient in a recognised hospital
☐ Outpatient of a recognised hospital

SAMPLES SHOULD BE TAKEN AT APPROX 10 WEEKS GESTATION (9-12 Wks Accepted)

7 mL clotted blood sample, plain tube - no anticoagulant (or 4 mL serum. if possible)

Store at 4°C (Do not freeze)

Date of collection____ /____ /____ @ ____ am/pm Collected by _____

Send the specimen to Genetic Health Services Victoria at the above address

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Part 1: The blood test.

The blood test, also known as the **maternal serum screening (MSS) test** measures the amount of two different proteins called PAPP-A and beta hCG which appear naturally in the mother's blood during pregnancy. A change in the level of these proteins may indicate that there is an increased risk that the baby has Down syndrome. Previously a similar test was available but only after 14 weeks of pregnancy. A result is not available from the blood test alone.

Part 2: The ultrasound.

The ultrasound can be done by a specially trained ultrasonographer between 11 weeks 3 days and 13 weeks and 6 days of pregnancy. It is usually performed through the abdominal wall but sometimes it is necessary to do an internal (vaginal) scan. The ultrasound has no known harmful effects on the mother or baby.

The ultrasound allows measurement of the amount of fluid in the skin at the back of the baby's neck. This measurement is called the **nuchal translucency.**

All babies at this stage of pregnancy have some fluid in this area, but on average a baby with Down syndrome or another chromosome abnormality has a larger amount of fluid. The nuchal translucency measurement can therefore be used to identify if a baby has an increased risk of Down syndrome.

The ultrasound also checks a number of other things, including:

- Confirming that the baby is alive
- Looking for twins
- Allowing an accurate estimation of the due date of the baby
- Determining if there are any obvious physical abnormalities of the baby.

When is the test done?

The nuchal translucency test can be done between 11 weeks 3 days and 13 weeks and 6 days of pregnancy (ideally at 12 weeks). **The blood test is most informative if collected during the tenth week of pregnancy**, however it can be collected between 9 weeks and 12 weeks 6 days of pregnancy.

How accurate is the test?

By **combining** the knowledge about a woman's age, the results of the blood test, nuchal translucency measurement, gestation and how much the woman weighs the test can identify 9 out of 10 pregnancies in which the baby has Down syndrome.

It is very important to realise that this test is only for Down syndrome and that a 'low risk' result does not ensure that the baby is free of other possible birth defects.

How do I get my result?

Once the ultrasonographer has completed the ultrasound the measurements are faxed to the MSS laboratory. The information from the blood test is combined with the ultrasound measurements and the final result sent back to the ultrasonographer. A copy of the result will also be forwarded to your referring doctor.

What if the screening test finds that my baby is at 'increased risk' of Down syndrome?

1 in 25 women will have an 'increased risk' result. This does not necessarily mean that there is a problem in your unborn baby. It means that further tests should be considered to see if there is a problem with your baby. Most women with an 'increased risk' result will go on to have a normal baby.

There are two diagnostic tests that may be offered to women identified with an 'increased risk' result to confirm whether or not the baby has Down syndrome or another chromosome abnormality. These tests are chorionic villus sampling (CVS) or amniocentesis. A CVS is performed at about 12 weeks whilst the amniocentesis is carried out at about 15 weeks of pregnancy. Both tests provide the same result and either one or the other is usually offered. Both have a small risk of miscarriage. Preliminary results from either of these tests may be available within 48 hours. Further information is available from your doctor about these tests.

What if the screening tests finds that my baby is at 'low risk' of Down syndrome?

24 out of 25 women tested will have a 'low risk' result. This means that the risk of having a baby with Down syndrome is very low. While women with this result are very unlikely to have a child with Down syndrome, a few will.

Does the test pick up any other birth defects?

Although the test is designed to detect babies at risk of Down syndrome, babies at risk of other chromosome abnormalities may also be identified. In addition the ultrasound may identify if a baby has an obvious physical birth defect. If there is any concern on the ultrasound, a further ultrasound may be suggested by the ultrasonographer in a few weeks time.

Do all pregnant woman have the screening test for Down syndrome in early pregnancy?

No. The decision whether or not to be tested is entirely up to you. Here are a few questions you may want to consider before you decide whether or not to have the test.

In what ways would Down syndrome affect my baby and my family?

What are the chances of my baby being affected?

Would I want to know ahead of time if there is a problem with the baby?

Would I consider termination if the baby were affected?

The test does not identify all pregnancies with Down syndrome, does this matter?

If I tested at 'increased risk' would I consider having a CVS or amniocentesis even though there is a small chance of a miscarriage?

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How much will it cost?

There is a Medicare rebate for the blood test, however this does not meet the full cost of the test. There are potential costs for the ultrasound scan. Individual practices need to be contacted regarding costs.

How can I get more information about this test?

You can discuss any of these issues with your doctor.

Some women many find thinking about these issues stressful. If you need help in dealing with worry or concern about this test, please speak to your doctor.

In addition, the Genetic Health Services Victoria offer an information and counselling service regarding this test:

- More detailed information about the test itself
- Assistance to understand the implications of the test
- Help to interpret the results.

For further information contact:

MATERNAL SERUM SCREENING
Genetic Health Services Victoria
Royal Children's Hospital
Flemington Road
Parkville, Victoria 3052
Telephone (03) 8341 6356 or 8341 6357

Another useful telephone number:
DOWN SYNDROME ASSOCIATION
Telephone (03) 9486 9600
Website www.vcgspathology.com.au

How the testing process works:

1. Your pregnancy is confirmed and your dates estimated from your last period.
2. An ultrasound is booked for about 12 weeks of pregnancy.
3. Your doctor gives you the form for your maternal serum screening blood test.
4. Blood is collected ideally during the tenth week of pregnancy. An appointment for blood collection can be made by phoning 03 8341 6303.
5. The ultrasound is performed, including the nuchal translucency measurement.
6. The 'combined' risk is calculated.
7. The result will be discussed with you by your doctor.
8. If you receive an 'increased risk' result, the options of further testing will be discussed with you.

VCGSPATHOLOGY
Victorian Clinical Genetics Services

Combined First Trimester Screening

Screening Test for Down syndrome in early pregnancy.

Every woman hopes that she will have a healthy baby. Most babies are born healthy, but three in a hundred babies are born with a problem that is serious and may require medical care. To help mothers in Victoria reduce the chance of having a baby with a disability we are offering a screening test during pregnancy which you may choose to have if you wish.

This test allows the combination of a blood test and an ultrasound test to be used at an early stage of pregnancy to see if your baby might be at risk of having Down syndrome.

This leaflet answers the most commonly asked questions about this test.

What is Down syndrome?

Down syndrome is a condition that results in intellectual disability of varying degrees and may cause physical problems such as heart defects, or difficulties with the sight or hearing. 1 in 350 pregnancies in the first trimester are affected by Down syndrome. All women are at risk of having a baby with Down syndrome, although this risk increases with the age of a woman. Your doctor can tell you the risk of having a baby with Down syndrome at your age.

Down syndrome is a chromosome abnormality. Chromosomes are the structures that contain the genetic information that we need to grow and develop normally. Each of our cells contains 23 pairs of chromosomes. Abnormalities in the number or structure of the chromosomes can result in medical problems. Down syndrome is usually caused by having 3 copies of chromosome number 21. There are other rarer chromosome abnormalities which may result in a child being born with a disability.

What does the screening test for Down syndrome involve?

The test has two parts.

1. A blood test
2. An ultrasound.

By combining the results of the 2 parts of the test it is possible to identify if a pregnant woman may be at increased risk of having a baby with Down syndrome.

By itself the test does not tell you whether the baby has Down syndrome or not. It identifies women who should be offered further testing to determine if their baby is affected.

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