VCGS Leaders in Genetic Health

a NATA accredited laboratory No. 3171

PATHOLOGY REQUEST FORM
VCGS MATERNAL SERUM TEST

Victorian Clinical Genetics Ser vices,
(APA) Murdoch Childrens Research Institute
Flemington Road, Parkville VIC 3052 Australia
P 1300 934 355 F +61 3 8341 6389 W vcgs.org.au

MEDICARE CARD NUMBER

August 2017

Patient LAST NAME	GIVEN NAMES	SEX	DATE OF BIRTH	YOUR REFERENCE:
Patient ADDRESS			PHONE (home)	PHONE (business)

TESTS REQUESTED SELF DETERMINED: ☐

MATERNAL SERUM SCREENING TEST

OPTIONAL PRE-ECLAMPSIA SCREENING TEST IN THE FIRST TRIMESTER ONLY BLOOD SAMPLE AT 11 -13W6D ☐ **PLEASE TICK**

First Pregnancy YES / NO
Family hist. PE YES / NO

The requesting doctor should ensure both blood specimen and ultrasound reports are provided to the Maternal Serum Screening Laboratory for a complete Combined First Trimester Screening result.

CLINICAL NOTES

ETHNIC GROUP: ☐ EUROPEAN ☐ ABORIGINAL ☐ ASIAN ☐ AFRO-CARIBBEAN ☐ OTHER .....

LMP: _____/_____/_____ **MATERNAL WEIGHT** ……kgs

MATERNAL BLOOD PRESSURE SP……...../DP……..……mmHg

GESTATIONAL AGE TODAY ……..…….. ON _____/_____/_____

CLINICAL DUE DATE ……

DUE DATE BY ULTRA SOUND _____/_____/_____

PREGNANCY COMPLICATIONS: ☐ IVF EGG DONOR DoB ___/___/___

☐ DIABETES (IDDM ONLY) ☐ SMOKER ☐ THREATENED MISCARRIAGE

☐ TWINS: **PREVIOUS:** ☐ T21 ☐ NTD ☐ PREECLAMPSIA

MSS 1ST TRIMESTER
NAME OF IMAGING PRACTICE: ……

FOR FIRST TRIMESTER SCREENING AN ULTRASOUND REQUEST FORM IS REQUIRED FOR NUCHAL TRANSLUCENCY, 11-13W6D.

ULTRASOUND DATE _____/_____/_____ DATE: / /

DOCTOR'S SIGNATURE AND REQUEST DATE

x …… DATE: ___/___/___

COPY REPORTS TO:	REQUESTING DOCTOR (Provider Number, Initials and Address)

HOSPITAL STATUS OF PATIENT AT SPECIMEN COLLECTION OR DATE OF SERVICE

	YES	NO		YES	NO
Private patient in a private hospital or approved day hospital facility	☐	☐	Hospital patient in a recognised hospital	☐	☐
Private patient in a recognised hospital	☐	☐	Outpatient of a recognised hospital	☐	☐

LABORATORY COPY

5-10ML CLOTTED BLOOD SAMPLE
GEL OR PLAIN TUBE - NO ANTICOAGULANT
First trimester blood sample 9-13w6d
Second trimester blood sample 14-20w6d
First trimester plus Pre-Eclampsia blood sample 11 – 13w6d

PRIVACY DISCLOSURE

VCGS requires personal information contained in this request form for the purpose of test assessment and Program Audits.
VCGS may therefore request copies of ultrasound and cytogenetic reports from your doctor in order to complete its testing and audits.

I CERTIFY THAT THE PATHOLOGY ACCOMPANYING THE REQUEST WAS COLLECTED FROM THE PATIENT STATED ABOVE AS ESTABLISHED BY DIRECT INQUIRY AND/OR INSPECTION OF WRIST BAND.

SIGNED _____/_____/_____
PERSON COLLECTING SPECIMEN SPECIMENT DATE & TIME HRS

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MEDICARE CARD NUMBER

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Patient ADDRESS			PHONE (home)	PHONE (business)

TESTS REQUESTED

MATERNAL SERUM SCREENING TEST

OPTIONAL PRE-ECLAMPSIA SCREENING TEST IN THE FIRST TRIMESTER ONLY BLOOD SAMPLE AT 11 -13W6D ☐ **PLEASE TICK**

First Pregnancy YES / NO
Family hist. PE YES / NO

There is a Medicare rebate for this test, however there will be an out-of-pocket cost.

REQUESTING DOCTOR (Provider Number, Initials and Address)

FOR FURTHER INFORMATION CONTACT:
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Murdoch Childrens Research Institute
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Maternal serum Screening:

Your Pregnancy Your Choice

Every woman hopes she will have a healthy baby. Most babies are born healthy but there are about four in one hundred born with a problem which is serious and may require medical care. Victorian Clinical Genetics Service offers screening tests for some conditions detectable during pregnancy, which you may choose to have.

Maternal serum screening tests are the combined first trimester screening test and the second trimester maternal serum screening test. These tests are available to pregnant women to identify if they have an increased chance of having a baby with Down syndrome, trisomy 18/13, pre-eclampsia or neural tube defects (only second trimester screening). You may ask your doctor to request the combined first trimester screening test or the second trimester screening test, not both.

What is Down syndrome?

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

Down syndrome is usually caused by having 3 copies of chromosome number 21, instead of the usual 2.

What are trisomy 18 or 13?

Trisomy 18 or 13 are conditions, which result in some physical problems such as growth deficiency, heart defects, digestive system defects and intellectual disability. Most pregnancies with trisomy 18 or 13 are lost spontaneously. Babies born with trisomy 18 or 13 have a very poor prognosis; many do not live beyond the first few weeks of life.

Trisomy 18 or 13 are usually caused by having 3 copies of chromosome number 18 or 13, instead of the usual 2.

What are neural tube defects?

The most common neural tube defects are anencephaly and spina bifida. In anencephaly, the brain does not develop properly and the baby does not survive. Babies with spina bifida have an opening in the bones of the spine which can result in damage to the nerves controlling the lower part of the body. This can cause weakness and paralysis of the legs, and sometimes inability to control the bowel and bladder.
In around 1 in 750 pregnancies, the developing baby will have a neural tube defect.

MANDATORY PATIENT ADVISORY STATEMENT:

Your doctor has recommended that you use the nominated pathology provider. You are free to use your own pathology provider. However, if your doctor has specified a particular pathologist on clinical grounds, a Medicare rebate will only be payable if that pathologist performs the ser vice. You should discuss this with your doctor.

PRIVACY NOTE:

The information provided will be used to assess any Medicare benefit payable for the ser vices rendered and to facilitate the proper administration of government health programs, and may be used to update enrolment records. Its collection is authorised by the provisions of the Health Insurance Act 1793. The information may be disclosed to the Department of Health and Aging or to a person in the medical practice associated with this claim, or as authorised/ required by law.

What is Pre-elampsia?

Pre-eclamsia occurs in around 1 in 50 pregnancies. It is pregnancy induced high blood pressure and is one of the most common life-threatening conditions occurring in pregnancy. Increased blood pressure can be dangerous to both mother and baby. The only current treatment is early delivery, which will depend the stage of the pregnancy and therefore the maturity of the baby.

Screening during the first trimester can identify women at increased risk of early-onset pre-eclampsia. This could potentially improve the pregnancy outcome with early detection and appropriate treatment is commenced when required. There is extensive evidence that early-onset pre-eclampsia is associated with an increased risk of conditions such as fetal growth restriction which affects 1 in 20 pregnancies. These conditions are also influenced by a number of other maternal and placental factors.

Do all pregnant women have maternal serum screening?

No. The decision whether or not to be tested is entirely up to you.

How can I get more information about this test?

You can discuss any of these issues with your doctor. Some women may find thinking about these issues stressful. Please speak to your doctor about your concerns.

In addition, the Victorian Clinical Genetics Services offer information and genetic counselling:

Contact Details:

MATERNAL SERUM SCREENING
P. 1300 934 355
F. (03) 8341 6389
W. vcgs.org.au

Support Groups:

Down Syndrome Victoria
P. 1300 658 873
W. downsyndromevictoria.org.au

Spina Bifida Foundation Victoria
P. 03 9663 0075
W. sbfv.org.au

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Maternal serum Screening:

Combined First Trimester Screening

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 have Down syndrome or trisomy 18. This leaflet answers the most commonly asked questions about this test

What does the combined first trimester screening test 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 might have a baby with Down syndrome or trisomy 18.

By itself the test does not tell you whether the baby has Down syndrome or trisomy 18. It identifies women who are at increased chance and should be offered further testing to determine if their baby is affected.

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 free 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 chance that the baby has Down syndrome or trisomy 18. **The blood sample can be taken at any local pathology collection centre. A result is not available from the blood test alone.**

Part 2: The ultrasound

We recommend that the ultrasound is done by a specially trained ultrasonographer between 11 weeks 1 day 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 chance of Down syndrome or trisomy 18.
The ultrasound also checks a number of other things, including:

- The presence or absence of the nasal bone. If the nasal bone is absent the baby may have a higher chance of Down syndrome
- 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 blood test is most informative if collected between 10 and 11 weeks gestation, however it can be collected between 9 weeks and 13 weeks 6 days of pregnancy. The nuchal translucency test can be done between 11 weeks 1 day and 13 weeks and 6 days of pregnancy (ideally between 12 and 13 weeks gestation).

How accurate is the test?

By combining the information about a woman's age, the results of the blood test, the nuchal translucency measurement and nasal bone, the test can identify about 9 out of 10 pregnancies in which the baby has Down syndrome and 8 out of 10 pregnancies in which the baby has trisomy 18.

It is very important to realise that this test is only for Down syndrome and trisomy 18 and that a 'screen negative' 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 is faxed to the ultrasonographer and/or requesting doctor.
A copy of the result will also be forwarded to your referring doctor. You must contact your doctor for your test result. Results are usually available within 5 working days of the ultrasound report being received by the MSS laboratory.

What if the screening tests finds that my baby is 'screen negative' for Down syndrome or trisomy 18?

This means that the chance of having a baby with Down syndrome or trisomy 18 is very low. 24 out of 25 women tested will have this result. While women with this result are very unlikely to have a child with Down syndrome or trisomy 18, a few will.

What if the screening test finds that my baby is 'screen positive' for Down syndrome?

1 in 25 women will have a 'screen positive' 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 a 'screen positive' result will have a healthy baby.

What further tests can be done for those with a 'screen positive' result?

There are two diagnostic tests that may be offered to women identified with a 'screen positive' result to confirm whether or not the baby has Down syndrome, trisomy 18 or another chromosome condition. These tests are chorionic villus sampling (CVS) or amniocentesis. A CVS is performed at about 12 to 13 weeks whilst amniocentesis is performed from 15 weeks gestation, onwards. 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.
If further testing confirms that the developing baby has Down syndrome or trisomy 18, then you would have the option of deciding whether to continue your pregnancy.
Further information is available from your doctor about these tests.

Does the test pick up any other birth defects?

Although the test is designed to detect babies at increased chance of Down syndrome and trisomy 18, babies at increased chance of other chromosome conditions, may also be identified. In addition the ultrasound may identify if a baby has an obvious physical birth defect.

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.

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Maternal serum Screening:

Second Trimester

This test is available to see if your baby might have Down syndrome, trisomy 18 or neural tube defects. This leaflet answers the most commonly asked questions about this test.

What does the maternal serum screening test in the second trimester involve?

The maternal serum test in the second trimester is a **blood test** for pregnant women to find out if they have an increased chance of having a baby with Down syndrome, trisomy 18 or neural tube defects

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

The blood test, also known as the maternal serum screening test (MSST) measures the amount of four different proteins called alpha- feto protein, unconjugated estriol, free beta hCG and dimeric Inhibin A, 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 chance that the baby has Down syndrome, trisomy 18 or neural tube defects.

When is the test done?

A blood sample is taken from the woman's arm between 14 and 20 weeks of pregnancy. The blood test is most informative if collected between 15 and 17 weeks gestation. If collected during the 14th week, no neural tube defect assessment is given.

The blood sample can be taken at any local private pathology collection centre.

How accurate is the blood test?

The test will help to identify all pregnancies with anencephaly, four out of five of neural tube defects, and two out of three cases of Down syndrome. It is very important to realise that this test is only for Down syndrome, trisomy 18 and neural tube defects, and that a "screen negative" result does not ensure that the baby is free of other possible birth defects.

How long does it take to get the results?

Usually within one week.

How will the test results be given?

Test results will be available from your doctor. You must contact your doctor for your test result.

The results will come back as "screen negative" or "screen positive".

What does 'screen negative' mean?

This means that the chance of having a baby with Down syndrome, trisomy 18 or neural tube defect is very low. 19 out of 20 women tested will have this result. While women with this result are very unlikely to have a child with Down syndrome, trisomy 18 or neural tube defect, a few will.

What does 'screen positive' mean?

1 in 20 women will have a 'screen positive' result. This does not necessarily mean that there is a problem in your unborn baby. It means that further tests should be considered. Most women with this result will have a healthy baby.

What further tests can be done for women with a 'screen positive' result?

There are two diagnostic tests that may be offered to women identified with a 'screen positive' result to confirm whether the baby has Down syndrome, trisomy 18 or neural tube defect: Amniocentesis, to detect chromosome abnormalities, such as Down syndrome and trisomy 18 and Ultrasound to detect neural tube defects.
Amniocentesis is usually performed from 15 weeks gestation, whilst the ultrasound to detect neural tube defects is usually performed between 18 to 20 weeks gestation.
Amniocentesis carries a risk (less than 0.1%) of causing a miscarriage.
Ultrasound has not been shown to have any harmful effect on the mother or the developing baby.
Preliminary results from amniocentesis may be available within 48 hours. If further testing confirms that the developing baby has Down syndrome, trisomy 18 or a neural tube defect, then you would have the option of deciding whether to continue your pregnancy.

Further information about these tests is available from your doctor.

How much will it cost?

If the test is requested by a private doctor there will be out of pocket expenses.