VCGS Leaders in Genetic Health

VCGS Maternal Serum Screening

MEDICARE NUMBER

Victorian Clinical Genetics Services
Murdoch Children's Research Institute
The Royal Children's Hospital
Flemington Road, Parkville VIC 3052
P +61 1300 118 247 F +61 3 8341 6366
W vcgs.org.au

1. PATIENT DETAILS

LAST NAME | GIVEN NAMES | SEX | DATE OF BIRTH | YOUR REFERENCE

ADDRESS | POST CODE | PHONE (home) | MOBILE

2. TESTS REQUESTED

Combined first trimester screening (CFTS) or Second trimester maternal serum screening (2TMSS)

☐ CFTS (sample @**9-13W6D**)

☐ CFTS + PE (pre-eclampsia) (sample @ **11-13W6D**)
- family history of PE ☐ YES ☐ NO
- first pregnancy ☐ YES ☐ NO

☐ 2TMSS (sample @**14-20W6D**)

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

3. CLINICAL INFORMATION

LMP: _____/_____/_____

CLINICAL DUE DATE: _____/_____/_____

GESTATIONAL AGE: _____ ON _____/_____/_____

U/SOUND DUE DATE: _____/_____/_____

MATERNAL WEIGHT: _____ kg

MATERNAL BP: SP _____/DP_____ mmHg

PREVIOUS: ☐ T21 ☐ NTD ☐ Pre-eclampsia

4. CLINICAL NOTES

☐ IVF ☐ own egg pick-up date ___/___/___ ☐ donor egg DOB ___/___/___ pick-up date ___/___/___

☐ Diabetes (IDDM only) ☐ Smoker ☐ Threatened miscarriage ☐ Twins

Ethnic group: ☐ European ☐ Aboriginal ☐ Asian ☐ Afro-Caribbean ☐ Other: _____

5. ULTRASOUND

For first trimester screening a request form is required for nuchal translucency, 11-13W6D.

Name of imaging practice:

Date of ultrasound: _____/_____/_____

DOCTOR'S SIGNATURE AND REQUEST DATE

SIGNATURE: DATE:

COPY REPORTS TO:

REQUESTING DOCTOR (provider #, initials, address):

HOSPITAL STATUS OF PATIENT AT SPECIMEN COLLECTION OR DATE OF SERVICES

	YES	NO
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	☐	☐

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 & program audits. VCGS may therefore request copies of ultrasound and cytogenetic reports from your doctor in order to complete testing & audits.

SPECIMEN COLLECTION

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.

PHLEBOTOMIST SIGNATURE:

COLLECTION DATE:_____/_____/_____ COLLECTION TIME (hrs):_____

VCGS Leaders in Genetic Health

Pathology Request Form
Maternal Serum Screening Test
Victorian Clinical Genetics Services
Flemington Road, Parkville VIC 3052
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MEDICARE NUMBER

PATIENT DETAILS

LAST NAME | GIVEN NAMES | SEX | DATE OF BIRTH | YOUR REFERENCE

ADDRESS | POST CODE | PHONE (home) | MOBILE

TESTS REQUESTED
Maternal Serum Screening Test (MSST)

☐ CFTS (sample @**9-13W6D**)

☐ 2TMSS (sample @**14-20W6D**)

☐ CFTS + PE (pre-eclampsia) (sample @ **11-13W6D**)

REQUESTING DOCTOR (provider #, initials, address):

There is a partial Medicare rebate for this test. There WILL be some out of pocket cost.

We're Australian & not-for-profit – all proceeds support medical research.

Patient Information

Genetic counselling with a VCGS genetic counsellor is available to anyone considering this screening test (1300 118 247). VCGS genetic counsellors can also discuss other prenatal screening and testing options with you. More information about this test is available at vcgs.org.au/mss

Purpose of the test

VCGS offers screening for some conditions that can be identified during pregnancy. Maternal serum screening has two options: screening during the first trimester or screening during the second trimester.

- **Combined first trimester screening (CFTS)**: provides an estimate of the chance that a pregnancy has a chromosome condition such as Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) or Patau syndrome (trisomy 13). An optional extra with CFTS is screening for early-onset pre-eclampsia (EO-PE). Early detection and treatment can improve pregnancy outcomes.
- **Second trimester screening (2ndTMSS)**: provides an estimate of the chance that a pregnancy has Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) or a neural tube defect, such as spina bifida.

Test process

CFTS combines a blood test with an ultrasound.

1. A blood test between 9 weeks + 0 days and 13 weeks + 6 days (if having PE as well, blood must be collected between 11 weeks and 13 weeks + 6 days). This measures two proteins called PAPP-A and free beta hCG. These are found naturally in the blood during pregnancy. A change in the level of these proteins may indicate a greater chance of a chromosome condition.
2. A nuchal translucency ultrasound (NT scan). This ultrasound is performed by a specialist ultrasonographer between 11 weeks + 1 day and 13 weeks + 6 days. The ultrasound measures the thickness of a fluid filled space at the back of baby's neck and also the presence or absence of the nasal bone. An enlarged space and/or absence of the nasal bone may indicate a greater chance of a chromosome condition.

The results of the blood test and the NT ultrasound are combined with the patient's age and weight to give a combined estimate of the chance the pregnancy may have a particular condition.

2ndTMSS is a blood test only.

Blood is collected between 14 and 20 weeks. Although if the sample is collected between 14 and 15 weeks, no estimate will be given for neural tube defects.

A list of blood collection sites can be found on our website: vcgs.org.au

Limitations of the test

- Maternal serum screening is not a diagnostic test. Therefore, not all pregnancies with a chromosome condition will be identified. Screening only looks for a small number of particular chromosome conditions: no screening test can identify all possible conditions that may affect a pregnancy.
- A screen negative result means it is very unlikely the pregnancy has a chromosome condition. The majority of women will receive a screen negative result. As with all screening tests, there is always a small chance the result is incorrect (false negatives).
- A small number of women will receive a screen positive result. This does not mean the pregnancy definitely has a chromosome condition - only that there is a higher chance. If you receive this result, you will be offered diagnostic testing to confirm the result. In most cases, the screen positive result turns out to be a 'false positive' and the pregnancy does not have the chromosome condition. If you receive a screen positive result, there are two options for further testing. You can have *percept* non-invasive prenatal test (NIPT), which is a more accurate screening test or you can have a diagnostic test. A diagnostic test requires an invasive procedure such as a CVS or amniocentesis.

Privacy, confidentiality & use of information

- Your test results will be kept confidential. Results will only be released to your healthcare provider, other healthcare providers involved in your medical care, or to another healthcare provider as directed by you, or otherwise as required or authorised by applicable law.
- Collecting information on your pregnancy after testing is part of our laboratory's standard practice for quality purposes and test evaluation. VCGS may contact your healthcare provider to obtain this information.

Retention & use of samples

- In line with best practices and clinical laboratory standards, leftover de-identified specimens (unless prohibited by law), de-identified genetic material, as well as other information learned from your testing, may be used by VCGS for purposes of quality control, laboratory operations, laboratory test development, laboratory improvement, and generation of new scientific knowledge. All such uses will be in compliance with applicable law.

Financial responsibility

- You are responsible for fees incurred with VCGS for services performed.

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Maternal Serum Screening: Your Pregnancy Your Choice

Prenatal screening is offered to all pregnant women because of the small chance that a pregnancy may have a chromosome condition or other physical condition. While most women will have healthy babies, in about 1 in 25 pregnancies (4%) a baby will be born with a condition that may require medical attention. These conditions vary a lot, from mild to severe. Prenatal screening tests are used to identify those pregnancies that are more likely to have one of these conditions. However, no screening test can identify all possible conditions. Prenatal screening is optional.

Maternal serum screening options

Two possible screening options are the *combined first trimester screen* (CFTS) or the *second trimester serum screen* (2ndTMSS). These tests give you information about your chance of having a baby with Down syndrome, Edwards syndrome, Patau syndrome or a neural tube defect (second trimester screen only). CFTS can also determine your chance of developing early-onset pre-eclampsia (high blood pressure). You can choose CFTS or 2ndTMSS- you don't need both.

Down syndrome (trisomy 21)

Down syndrome is a chromosome condition caused by an extra copy of chromosome 21 (3 instead of 2). People with Down syndrome have characteristic physical, medical and intellectual features. Most have some level of intellectual disability and may also have physical problems affecting their heart, vision or hearing. All women have a chance of having a baby with Down syndrome, although the chance increases with maternal age.

Edwards syndrome (trisomy 18) & Patau syndrome (trisomy 13)

Edwards syndrome is caused by an extra copy of chromosome 18 and Patau syndrome is caused by an extra chromosome 13. Most pregnancies with these chromosome conditions will miscarry naturally. Babies born with trisomy 18 or 13 usually have severe physical and intellectual disability and survival beyond 1 year is rare.

Neural tube defects (NTDs)

Neural tube defects occur very early in development when the spinal cord and brain fail to develop properly. The most common NTDs are spina bifida (affecting the spine) and anencephaly (affecting the brain). Babies with anencephaly rarely survive long after birth however babies with spina bifida can show varying levels of disability. Surgery may or may not be an option.

Pre-eclampsia (PE)

Pre-eclampsia is high blood pressure caused by pregnancy. It is one of the most common and life-threatening conditions occurring in pregnancy. 1 in 20 pregnant women are affected by PE.

Screening during the first trimester (as part of CFTS) can identify women at increased risk of early-onset pre-eclampsia. Women at risk can be treated with low-dose aspirin and in some cases, may need to deliver early (depending on gestation).

Do all pregnant women have screening?

No. Prenatal screening tests are optional and many women choose not to have screening.

Some women choose to have screening so they can have more information about their pregnancy and to be reassured that everything is OK. For most women, they will receive this reassurance. For some, they will receive results they weren't expecting. Talk to your doctor or midwife if you have any questions.

Where can I get more information about prenatal screening?

You can always talk to your doctor or midwife if you have any questions about prenatal screening. In addition, you can find more information about screening on our website. Alternatively, you can contact one of our prenatal screening genetic counsellors on (03) 9936 6402.

Contact details:

P 1300 118 247

F (03) 8341 6366

W vcgs.org.au/mss **E** vcgs@vcgs.org.au

Genetic Support Network Victoria

P (03) 8341 6315

W gsnv.org.au

E info@gsnv.org.au

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.

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Maternal Serum Screening:

Combined first trimester screening

Combined first trimester screening (CFTS) combines a blood test with an ultrasound to provide an estimate of the chance that a pregnancy has Down syndrome, Edwards syndrome or Patau syndrome.

What does combined first trimester screening involve?

This screening test has two parts: a blood test and an ultrasound. This test will *estimate the chance* that a pregnancy has a particular condition by combining the results of the blood test and the ultrasound.

This *screening test is not diagnostic*- it does not give a 'yes or no' answer. It only identifies a pregnancy as having a high or low chance of a particular condition, such as Down syndrome. If you receive a high chance result, you can opt for further diagnostic testing to confirm the result.

Part 1: the blood test

The blood test measures the amount of two proteins called PAPP-A and free beta hCG. These are found naturally in the blood during pregnancy. A change in the level of these proteins may indicate an increased chance of a particular condition. A result cannot be calculated from the blood test alone. A lits of blood collection sites can be found on the VCGS website (vcgs.org.au)

Part 2: the ultrasound

The nuchal translucency (NT) ultrasound is performed by a specially trained ultrasonographer between 11 weeks 1 day and 13 weeks 6 days of pregnancy. This measures the thickness of a fluid filled space at the back of the baby's neck. An enlarged space (NT measurement) may mean a higher chance of a chromosome condition.

When is screening done?

The blood sample is best collected between 10 and 11 weeks gestation, however, a sample between 9 weeks and 13 weeks + 6 days is acceptable. The NT ultrasound can be performed between 11 weeks + 1 day and 13 weeks + 6 days- ideally between 12 and 13 weeks.

How accurate is this screening test?

CFTS is not a diagnostic test, but it can accurately identify about 9 out 10 pregnancies that have Down syndrome. It is less accurate at identifying trisomy 18 or trisomy 13. *It's important to note that screening does not look for ALL possible conditions*.

How do I get my result?

Once the NT ultrasound results have been sent to the MSS lab, it will be combined with your blood test results. In approximately 5 working days, your result will be sent to your referring doctor and ultrasound clinic. You can contact your doctor to discuss your results.

What does a 'screen negative' result mean?

A screen negative (or low chance) result means it is very unlikely the pregnancy has a chromosome condition. The majority of women (96%) will receive a screen negative result. As with all screening tests, there is always a chance the result is wrong.

What does a 'screen positive' result mean?

A small number of women will receive a screen positive (or high chance) result. This does not mean the pregnancy definitely has a chromosome condition- only that there is an increased chance. If you receive this result, you will be offered diagnostic testing to confirm the result. In most cases, the screen positive result turns out to be a false alarm and the pregnancy will not have a chromosome condition.

Further testing after a 'screen positive' result

If you receive a screen positive result, there are two options for further testing. You can have a non-invasive prenatal test (NIPT) which is a more accurate screening test or you can have a diagnostic test. While NIPT is a more accurate test than CFTS, there is an out of pocket cost and if you receive a high chance result, the only way to confirm is with diagnostic testing.

Diagnostic testing requires a sample of fetal tissue to be collected by an invasive procedure- a chorionic villus sample (CVS) or an amniocentesis (depending on your gestation). Both procedures have a small risk of miscarriage. Diagnostic testing will give you a 'yes or no' confirmation as to whether the pregnancy has a particular condition. You can speak to your doctor about your options.

How much does screening cost?

There is a partial Medicare rebate for the blood test- so there is some out of pocket cost. You will receive an invoice from VCGS which can be taken to Medicare. There is also a cost for the NT ultrasound which varies depending on the clinic you attend.

Second trimester screening

If you miss first trimester screening, second trimester screening is an option. This test is a blood test only. It estimates the chance that a pregnancy has Down syndrome, Edwards syndrome or a neural tube condition.

Timing: Your blood sample is best collected between 15 and 17 weeks gestation.

Accuracy: 2nd TMSS is not a diagnostic test, but it can accurately identify 2 out of 3 pregnancies with Down syndrome.

Cost: Second trimester screening is usually free for public patients in a public hospital. If the test is requested by a private doctor, there will be an out of pocket cost & you will receive an invoice from VCGS.