



**1. BIOLOGICAL MOTHER**

LAST NAME	GIVEN NAMES	SEX	DATE OF BIRTH	LABORATORY REF
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ADDRESS	POST CODE	MOBILE/PHONE (home)
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**2. TESTS REQUESTED**

Expanded carrier screening

Please email a copy (or image) of this completed form to [carrierscreening@vcgs.org.au](mailto:carrierscreening@vcgs.org.au) and we will coordinate sample collection.

**Sample type (both partners)**

2x Invitae saliva samples OR  2x 4mL EDTA blood

**3. CLINICAL INFORMATION**

It is recommended that samples be taken pre-pregnancy or before 10 weeks gestation

PREGNANT TESTING  
GESTATION (weeks): \_\_\_\_\_ EDD: \_\_\_\_\_

NON-PREGNANT TESTING

DONOR (egg, sperm, embryo)

**4. BIOLOGICAL FATHER**

FULL NAME	DATE OF BIRTH
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**5. IS THERE A FAMILY HISTORY OF A GENETIC CONDITION?**

Biological Mother  YES  NO specify: \_\_\_\_\_

Biological Father  YES  NO specify: \_\_\_\_\_

IF YES, PROVIDE DETAILS:

SPECIMEN COLLECTION	DOCTOR'S SIGNATURE AND REQUEST DATE
PHLEBOTOMIST SIGNATURE: _____ DATE: _____	SIGNATURE: _____ DATE: _____

COPY REPORTS TO:	REQUESTING DOCTOR (provider #, initials, address):
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**6. PATIENT CONSENT, SIGNATURES & EMAIL**

By signing this form, we request an **expanded carrier screen** be performed. We have read the patient consent included on the back of this form. The risks & limitations of this test have been adequately explained to us.

SIGNATURE (Mother): _____ DATE: _____	SIGNATURE (Father): _____ DATE: _____
EMAIL (Mother): _____	EMAIL (Father): _____

**Payment details**

Credit card details must be completed otherwise test results will not be available. If the payment fails, you will be contacted to verify and/or make payment by another method. If payment is not confirmed within 72 hours, the test will not be processed.

CARD TYPE  VISA  MASTERCARD

AMOUNT TO BE DEBITED **1. AUD\$750 - payment now to VCGS via credit card**  
**2. USD\$100 - you will receive an invoice from Invitae**

NAME ON CARD \_\_\_\_\_ SIGNATURE \_\_\_\_\_

CARD NUMBER                      EXPIRY DATE

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 expanded carrier screening is available at [vcgs.org.au/expandedcarrier](http://vcgs.org.au/expandedcarrier)

## Purpose of the test

- Expanded genetic carrier screening tests couples to see if they have an increased chance of having a child with one or more inherited genetic conditions (over 250 tested). These conditions may significantly impact quality of life and/or may reduce the lifespan of an affected child.
- These conditions are either 'autosomal recessive' (ie. if both parents are carriers there is a 1 in 4, or 25%, chance of having a child with the condition) or 'X-linked' (meaning that if the mother is a carrier there is up to a 50% chance of having a child with the condition).
- In general, carriers of genetic conditions do not experience health problems themselves. However, there are some conditions for which carriers may experience health effects. There are rare situations where this test can identify people with undiagnosed forms of genetic conditions.
- Most people who have expanded carrier screening will find out they are a carrier of at least one condition. In most cases, it is only if both parents are carriers of the same condition that there is a chance they may have a child with that condition.
- Your screening results may have implications for other family members.

## Test process

- For expanded carrier screening, you and your partner are tested at the same time. For couples using donor egg, sperm or embryos, the male and female 'biological' contributors to the pregnancy need to be screened. Contact your IVF clinic or VCGS for advice.
- After contacting VCGS to request testing, saliva test kits will be sent to you and your partner with return postage. Saliva samples should be collected and returned to VCGS at the same time to ensure they arrive together. Samples are then sent overseas to *Invitae (USA)* for testing ([invitae.com/en/](http://invitae.com/en/)).
- Payment is made in two parts: an upfront payment to VCGS (when ordering), followed by an invoice from *Invitae* once your samples are received overseas.
- Results will be available up to 5-6 weeks after both payments are made. Results will be issued to the healthcare provider who requested your testing. They will discuss results with you. VCGS genetic counsellors are also available to help you understand and discuss your results, at no cost (P +61 3 9936 6402).

## Limitations of the test

- Expanded carrier screening will not identify all individuals who are carriers of genetic conditions. Testing identifies most, but not all, disease-causing genetic changes, so rare or family specific changes may be missed. If you have a family history of one of the genetic conditions screened, please describe on the test request form so that we can provide accurate results.
- Expanded carrier screening does not screen for all possible inherited genetic conditions. Carrier screening does not eliminate the chance of having a child with a genetic condition.
- Your screening result is only relevant to you as a couple. Having children with a different partner will require another screening test.

## Privacy, confidentiality & use of information

- VCGS will only release your results 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.
- Your samples are being sent to *Invitae* in the United States for testing.
- The results of this test will not affect access to health insurance. Ability to obtain life insurance may be affected if an individual is found to have a genetic condition. This is unlikely with this test, as it looks for carrier status only.

## Financial responsibility

- You are responsible for fees incurred with VCGS & *Invitae* for services performed.
- Testing will not proceed until both payments have been made (VCGS + *Invitae*).