Sonic Genetics

Prenatal single gene testing | Request form

FOR THE DOCTOR

This test should be requested by the doctor for a patient with a known familial mutation in a single gene.

Patient details

First name _____

Surname _____

Date of birth ____/____/____ Sex _____

Address _____

_____

_____

Phone (mobile) _____

Medicare No. ☐☐☐☐☐☐☐☐☐☐

PATIENT STATUS AT TIME OF SERVICE OR SPECIMEN COLLECTION
(Required by law for all patients) Was the patient a:

Private patient in a private hospital or approved day hospital?	☐ Yes	☐ No
Private patient in a recognised hospital?	☐ Yes	☐ No
Public patient in a recognised hospital?	☐ Yes	☐ No
Outpatient of a recognised hospital?	☐ Yes	☐ No

Test requested REQUIRED

Please include a copy of the original parental genetic testing report.

☑ **Prenatal**

Gene(s) requiring testing: _____

Optional: ☐ Microarray* ☐ Microarray* + FISH for 13/18/21/X/Y aneuploidy (**additional charge**)

*Opt in for the following:

☐ Incidental findings# – Genetic variants that are not associated with the clinical presentation, but may still be relevant to the current or future health of the fetus or their genetic relatives.

☐ Susceptibility variants with estimated penetrance >40%# – Genetic variants that confer susceptibility to an abnormal clinical phenotype, but which are known to display incomplete penetrance and variable expressivity.

☐ All established susceptibility variants#

#Please refer overleaf for full details.

Clinical information

Clinical testing (please include a copy of all relevant reports)

NIPT: _____

FTS: _____

Ultrasound findings: _____

Further information: _____

_____

Gestational age (weeks): _____ Sample type: ☐ CVS ☐ Amniotic fluid ☐ SD

Partner details

First name _____

Surname _____

Date of birth ____/____/____ Sex _____

Address _____

_____

Phone (mobile) _____

Requesting doctor

Name _____

Address _____

_____

_____

Phone _____ Provider No. _____

I confirm that this patient has been counselled about the purpose, scope and limitations of the test and has given consent.

DOCTOR SIGNATURE

X _____ Date

Copy reports to

Name _____

Address _____

_____

_____

FOR THE PATIENT – Patient and Financial Consent

I have read the Patient, Privacy and Financial Consent section on the reverse of this request form.

I confirm that I have been informed about the purpose, scope and limitations of the test. I understand that all or part of the test requested is not eligible for a Medicare rebate and that I will pay in full prior to testing being performed.

If I do not fulfil the Medicare criteria

ACCOUNT STATEMENT
I understand that as the test requested is not eligible for a Medicare rebate, I will pay in full prior to the sample being tested. Cancellation fees may apply.

If I do fulfil the Medicare criteria

MEDICARE ASSIGNMENT (Section 20A of the Health Insurance Act 1973):
I offer to assign my right to benefits to the Approved Pathology Practitioner who will render the requested pathology service(s) and any eligible pathologist determinable service(s) established as necessary by the practitioner.

PATIENT SIGNATURE

X _____ Date

Practitioner's Use Only (Reason for patient being unable to sign)

For pricing, please refer to our website – www.sonicgenetics.com.au/bpns
For any enquiries, please contact Sonic Genetics on 1800 010 447.

FOR THE COLLECTOR

I certify that I established the identity of the patient named on this request, collected and immediately labelled the accompanying specimen(s) with the patient's name, DOB and date/time of collection.

Collector initials	Date collected / /	Patient initials
Location code		PAY CAT
Collection type	Time collected :	

Your doctor has recommended that you use one of the subsidiaries affiliated with Sonic Healthcare Limited, an Approved Pathology Authority. You are free to choose 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 service. You should discuss this with your doctor.

Sonic Genetics

Prenatal single gene testing

Information for patients

Purpose

This test is used to determine whether a known familial genetic condition is present in a fetus. It looks at fetal material for specific genetic changes identified by testing in one or both parents (e.g. changes identified by the Beacon expanded carrier screen).

FISH or microarray can be carried out on the fetal material as an additional optional test, to determine if there is a chromosomal abnormality present in the fetus. FISH is a rapid screening test for the common chromosome aneuploidies involving chromosomes 13, 18, 21, X and Y. Microarray screens every chromosome to look for gains or losses of genetic material, called copy number variants (CNVs), some of which can cause severe conditions prenatally or in early childhood ('pathogenic' CNVs).

Information from these tests, together with other prenatal screening, can be used to guide pregnancy decisions, in consultation with an obstetrician or clinical geneticist.

Limitations

The gene test will specifically look for the genetic changes identified in one or both parents. It will not look for other genetic changes in the same gene and will not look for change in any other gene.

A microarray test may identify a CNV that is not definitely abnormal. We call these 'variants of uncertain significance', or VUS. If a VUS is found during pregnancy, testing of the parents will be carried out to determine if the VUS has been inherited from an unaffected parent. If it is, it will likely be reclassified as a harmless ('benign') familial CNV.

In a small proportion of tests, a microarray might identify an 'incidental' or 'secondary' CNV. These findings very likely cause a clinical condition, but not one presenting prenatally or in early childhood. An example of this is an abnormality including the BRCA1 gene. This change would increase the chance of certain types of cancer later in life. These results can be confronting to deal with in the prenatal setting. We have chosen to make these opt in. If the 'incidental findings' box is ticked, then these CNVs will be reported. If it is not ticked, these CNVs will not be reported, even if detected by the laboratory.

Some CNVs are known to result in an increased risk of certain conditions (e.g. developmental delay), but do not show features in all carriers. These are called susceptibility variants (SV). Penetrance of a SV refers to the proportion of people with the variant who will show some degree of clinical features. Some SVs are known to have 'high' penetrance, 40–80%. Some have low to moderate penetrance, 10–40%. Because the outcome of these variants cannot be predicted with any certainty, we have chosen to make these opt in. You can choose to receive information on high penetrance SVs only, or on all SVs. If neither box is checked, susceptibility variants will not be reported, even if detected by the laboratory.

It is important to note that these tests are not able to detect all possible disease-causing genetic mutations. This testing cannot guarantee the health of your baby.

Testing procedure

An invasive procedure by an obstetrician is necessary to obtain your sample. Your doctor will send your sample directly to your local Sonic Pathology Australia laboratory for processing.

Depending on what is requested, genetic material will be extracted from your sample at our Sullivan Nicolaides Pathology (SNP) laboratory and will be:

- Transported to Fulgent Genetics laboratory in the US for processing of the prenatal single gene test.
- A portion retained in our SNP laboratory for processing of the FISH and microarray test.

Privacy

Your sample will be transported to Fulgent Genetics in the US for analysis and interpretation. This means that your personal information will be subject to the privacy and data protection rules of Fulgent Genetics in the US, which may be different from those of Australia. Details in relation to the collection, use and storage of your information can be found on the Sonic Genetics website, www.sonicgenetics.com.au/privacy-policy.

Results

For the prenatal single gene test:

Your results will be sent to your doctor in approximately 4–6 weeks from when the sample is received in the laboratory.

Results of testing will confirm:

- the fetus has inherited mutations from one (X-linked) or both parents (autosomal recessive) and will likely be affected by the condition
- the fetus is only a carrier of the condition and will not be affected by the condition
- the fetus has not inherited mutations from either parent and will not be affected by the condition

For the FISH test:

Your results will be sent to your doctor in approximately 3 business days from when the sample is received in the laboratory. Results of testing will confirm if there is an aneuploidy involving chromosomes 13, 18, 21, X or Y.

For the microarray:

Your results will be sent to your doctor in approximately 15 business days from when the sample is received in the laboratory.

Microarray testing can return a range of results, as outlined (no clinically relevant CNV detected; pathogenic CNV detected; VUS detected; if 'opted in', incidental finding or susceptibility variant).

Your doctor can contact our genetic pathologists and senior medical scientists to discuss technical aspects of your result.

Patient, Privacy and Financial Consent (Please sign overleaf to confirm that you agree with these statements)

By consenting overleaf:

- You consent to the prenatal test(s) being performed and confirm that you have been informed about the purpose, scope and limitations of the test. Sources of information that you can access include your doctor, the Sonic Genetics website and brochures, a genetic counsellor and this request form. You have had the opportunity to ask questions and understand that you can request further information or genetic counselling.
- You understand that this test will only look for the genetic changes identified in you and/or your partner and additional FISH and microarray can be selected to look for a limited range of other genetic conditions in your baby, and that return of incidental findings and susceptibility variants is optional, based on your wishes. You also understand that this test cannot guarantee the health of your baby.
- You expressly agree and consent to the disclosure of your personal information and sample (including health information) to Fulgent Genetics, a CLIA-accredited US laboratory, for the purpose of analysing and interpreting your sample and data. You acknowledge that Australian Privacy Principle 8.1 will not apply to such disclosures and that Fulgent Genetics may not be bound by the laws that provide the same level of protection for personal information afforded by the Australian Privacy Principles (APPs). You agree that Sonic Genetics will not be responsible for any breach of privacy by Fulgent Genetics, and you will not be able to seek redress under the APPs.
- You understand that all or part of the test requested is not eligible for a Medicare rebate and that you will pay in full for the test before your sample is processed.

PRIVACY NOTE The information provided will be used to assess any Medicare benefit payable for the services rendered and to facilitate the proper administration of government health programs, and may be used to update enrolment records. Its collection is authorised by provisions of the Health Insurance Act 1973. The information may be disclosed to the Department of Health and Ageing or to a person in the medical practice associated with this claim, or as authorised/required by law.