CLINPATH PATHOLOGY

The “How-To” Guide:

Accessing Reproductive Tests & Resources for your Patients

Presenter: Katelyn Henry

Today’s talking points:

1. Overview of RCS, ECS and NIPT and how to access
2. Genetic counselling for high risk results
3. Educational resources available
4. Genetic pathologist support and access

Our Reproductive Genetic Tests:

- 3-Gene Reproductive Carrier Screening (CF SMA FRAX)
- Expanded Carrier Screening (430+ genes)
- Non-invasive Prenatal Testing (NIPT) and Genome-wide NIPT (g-NIPT)

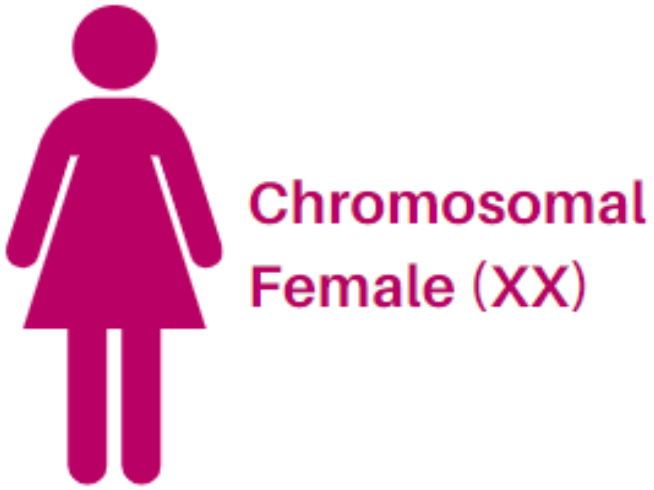
3 Gene Reproductive Carrier Screen

CF SMA FRAX

CLINPATH PATHOLOGY

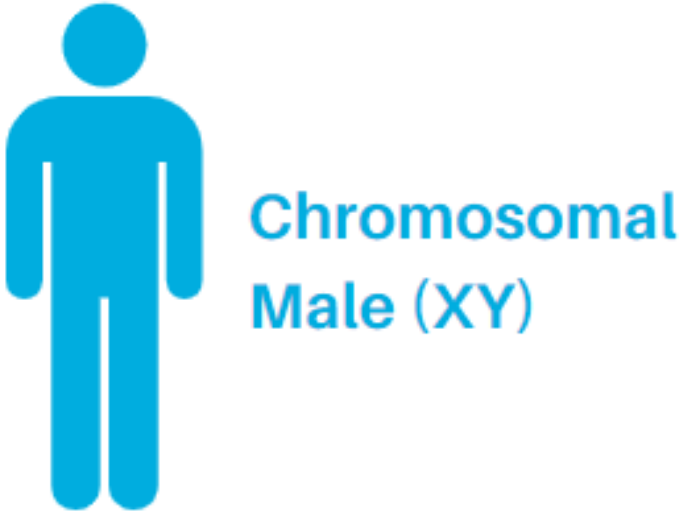
Medicare Rebate for 3 Gene Panel

MBS item 73451



Covers the initial screening of the female who is pregnant or planning pregnancy.

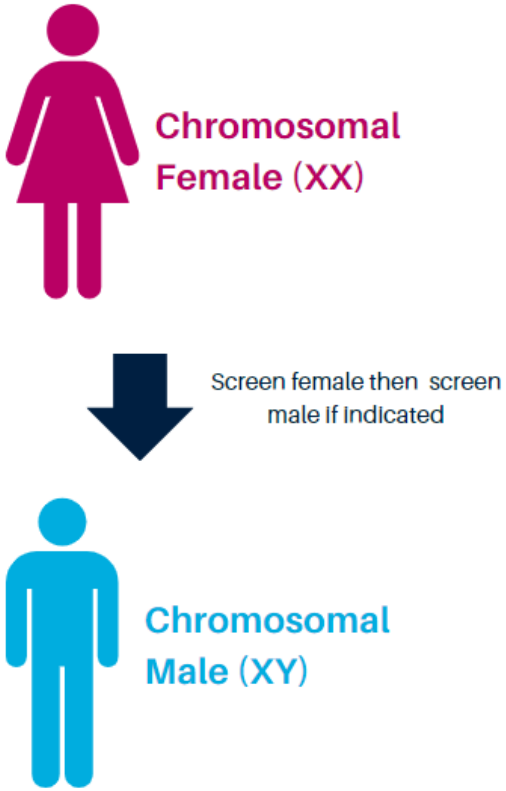
MBS item 73452



Covers the subsequent screening of the male reproductive partner for either CF or SMA, when detected in the female.

Pathways for RCS Testing

SUBSEQUENT



SIMULTANEOUS

Screened at the same time.

SUBSEQUENT TESTING

Chromosomal Female (XX)

Screen female then screen male if indicated

Chromosomal Male (XY)

Is she a carrier for...

CF and/or SMA?	FXS?	None of these?
Male partner to be tested to determine couples risk.	At increased risk of an affected child, counselling recommended	Low chance of having a child who is affected.

SUBSEQUENT TESTING

Chromosomal
Female (XX)

Screen female then screen male if indicated

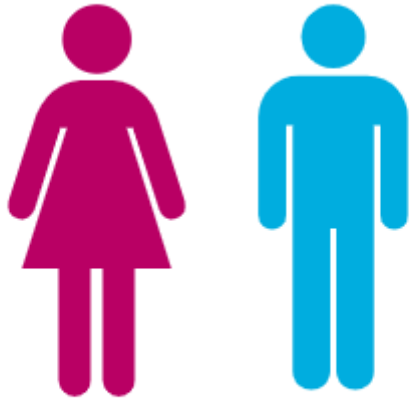
Chromosomal
Male (XY)

Medicare Rebated

TAT = 2-4 weeks

Depends on if male partner needs to be tested or not.

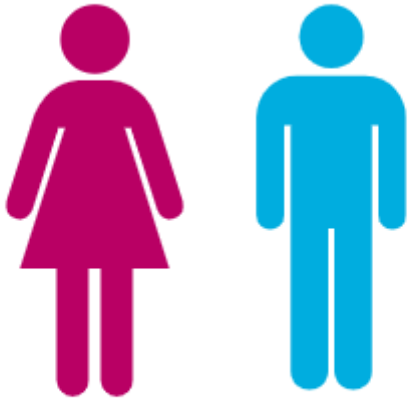
SIMULTANEOUS TESTING



Screened together

Dr will receive couple report

Pathways for RCS Testing



Screened together

Partially rebated

Female is rebated, male pays $385

TAT = 2 weeks

Recommended pathway if couple pregnant

Carrier Couple Identified!

Female is carrier of X-linked FXS **OR** both are carriers of mutations in CF or SMA

1. You will get a report that couple are carriers and at increased risk

2. If female is a FRAX carrier or the couple are carriers of mutations for CF or SMA then they are eligible for one session of genetic counselling through Clinpath / Sonic Genetics*

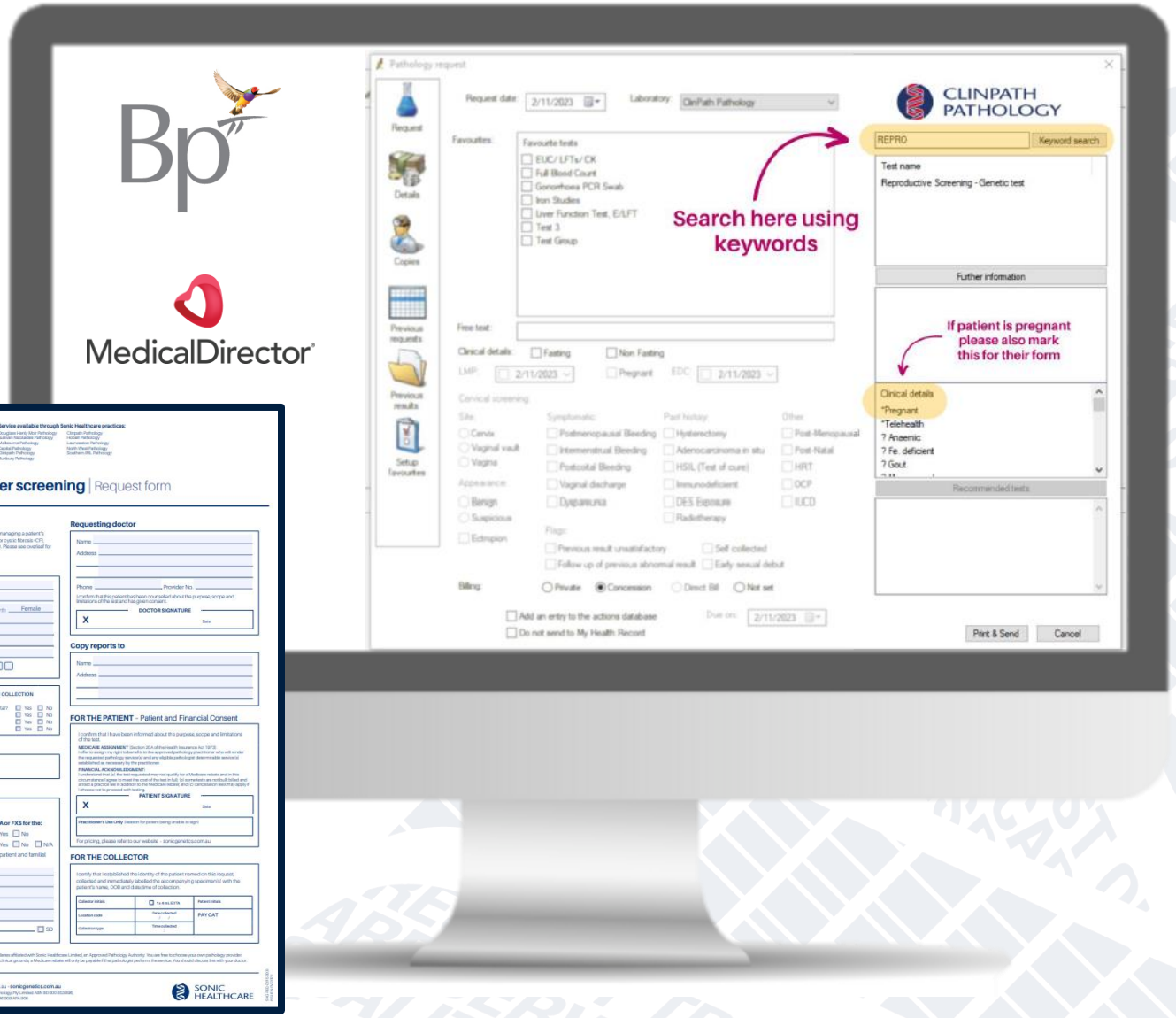
***at least one of the couple must be tested through Clinpath/Sonic.**

How do I order the 3 gene carrier screen?

Standard Clinpath form as:
“3 gene carrier screen” OR
“reproductive screening –genetic test”

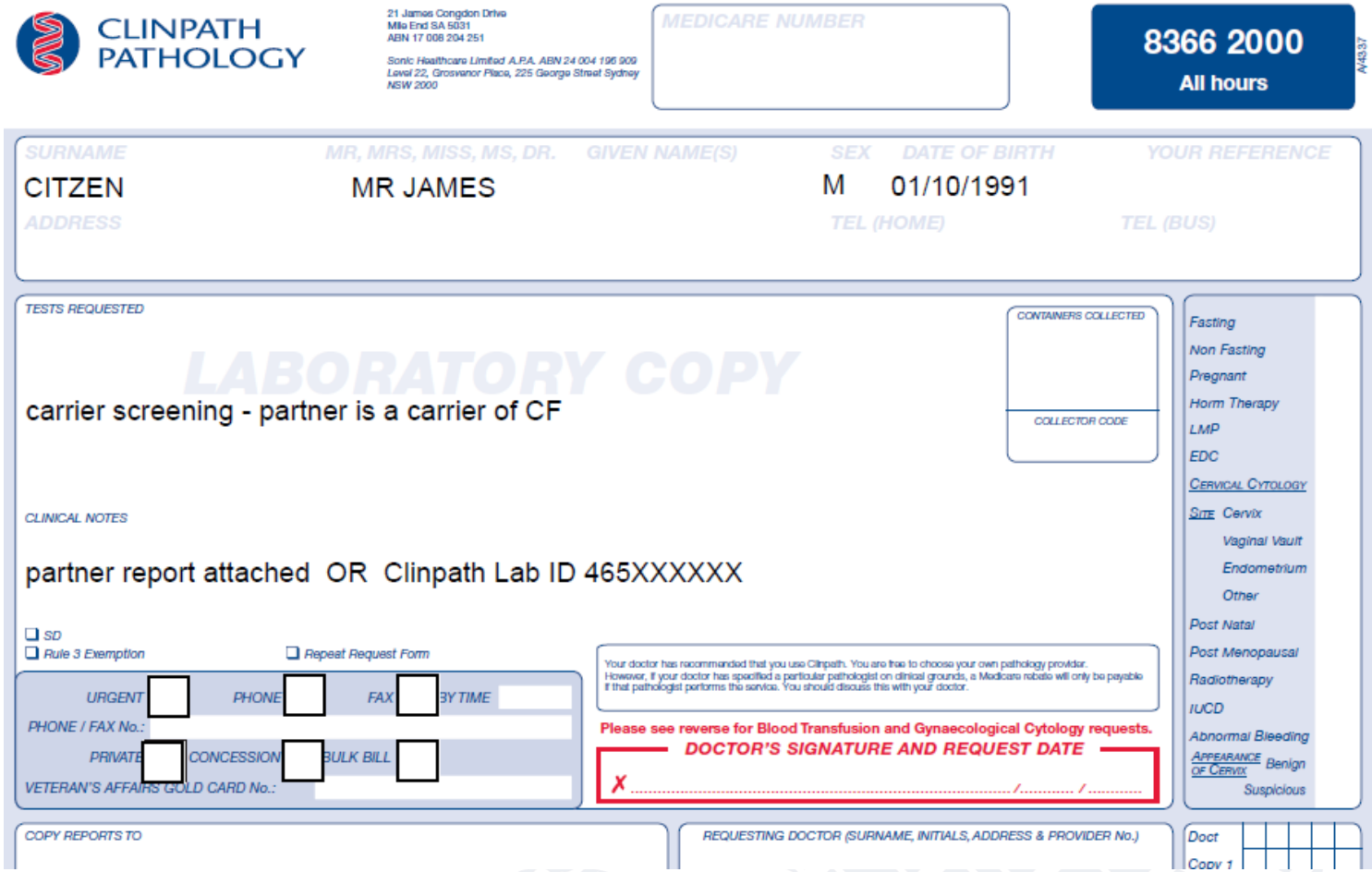
Through your e-collect:
“reproductive screening –genetic test”, search “REPRO” or set it up as a favourite

Dedicated RCS request form:
In BP and MD



How do I order partner testing?

Standard Clinpath form as:
“carrier testing, partner carrier of XXXX”



CLINPATH PATHOLOGY

21 James Congdon Drive
Mile End SA 5031
ABN 17 008 204 251

Sonic Healthcare Limited A.P.A. ABN 24 004 196 909
Level 22, Grosvenor Place, 225 George Street Sydney
NSW 2000

MEDICARE NUMBER

8366 2000
All hours

A/4337

SURNAME: CITZEN
MR, MRS, MISS, MS, DR.: MR JAMES
GIVEN NAME(S):
SEX: M
DATE OF BIRTH: 01/10/1991
YOUR REFERENCE:
ADDRESS:
TEL (HOME):
TEL (BUS):

TESTS REQUESTED

LABORATORY COPY

carrier screening - partner is a carrier of CF

CONTAINERS COLLECTED

COLLECTOR CODE

CLINICAL NOTES

partner report attached OR Clinpath Lab ID 465XXXXXX

Fasting
Non Fasting
Pregnant
Horm Therapy
LMP
EDC
Cervical Cytology
Site Cervix
Vaginal Vault
Endometrium
Other
Post Natal
Post Menopausal
Radiotherapy
IUCD
Abnormal Bleeding
Appearance of Cervix Benign
Suspicious

❑ SD
❑ Rule 3 Exemption
❑ Repeat Request Form

URGENT ☐ PHONE ☐ FAX ☐ BY TIME
PHONE / FAX No.:
PRIVATE ☐ CONCESSION ☐ BULK BILL ☐
VETERAN'S AFFAIRS GOLD CARD No.:

Your doctor has recommended that you use Clinpath. 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 this service. You should discuss this with your doctor.

Please see reverse for Blood Transfusion and Gynaecological Cytology requests.

DOCTOR'S SIGNATURE AND REQUEST DATE

✗ ...../...../.....

COPY REPORTS TO

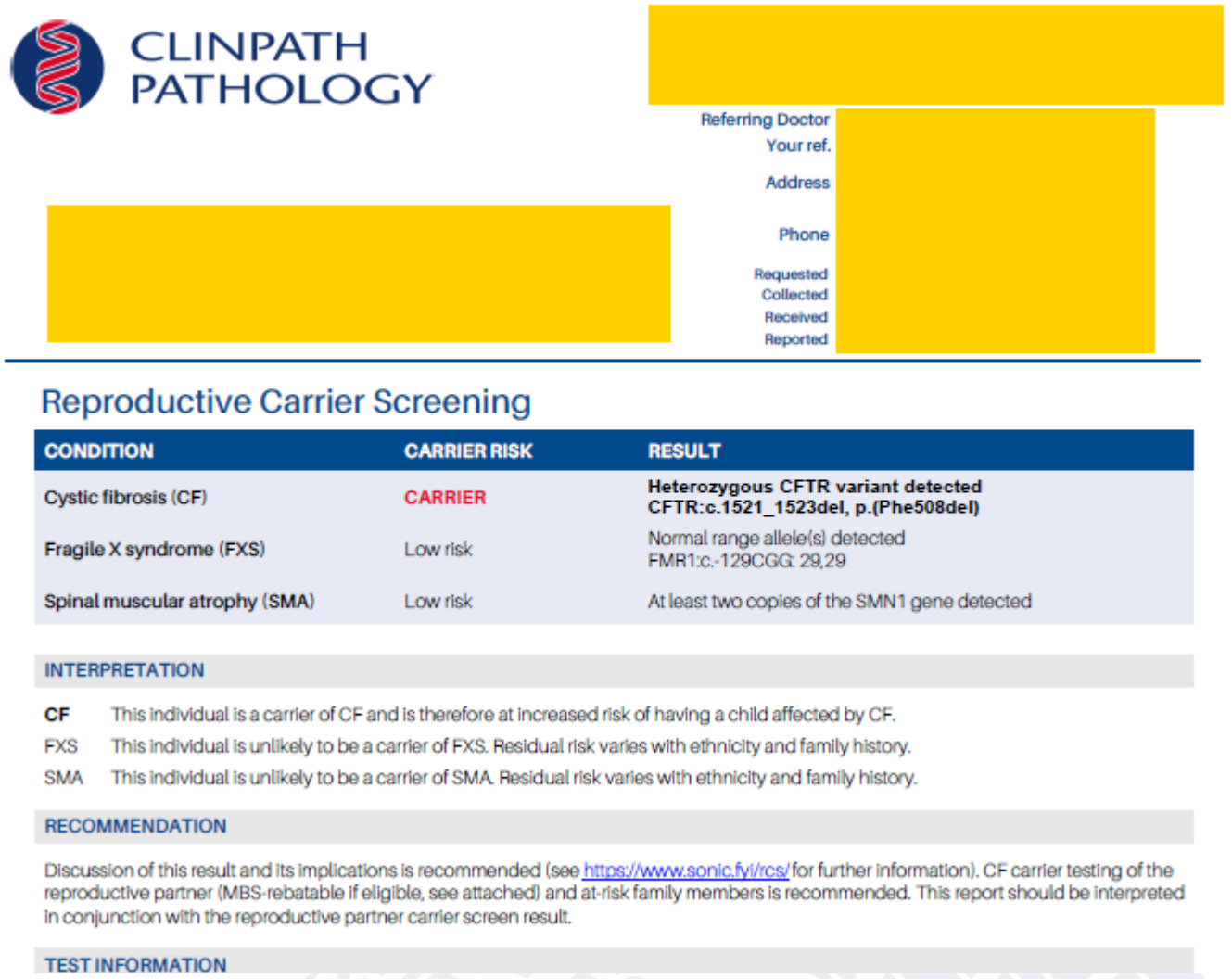
REQUESTING DOCTOR (SURNAME, INITIALS, ADDRESS & PROVIDER No.)

Doct Copy 1

How do I order partner testing?

Standard Clinpath form as:
“carrier testing, partner carrier of XXXX”

Partner testing form which is provided in females report



CLINPATH PATHOLOGY

[redacted]

Referring Doctor
Your ref.
Address
Phone
Requested
Collected
Received
Reported

[redacted]

Reproductive Carrier Screening

CONDITION	CARRIER RISK	RESULT
Cystic fibrosis (CF)	CARRIER	**Heterozygous CFTR variant detected CFTR:c.1521_1523del, p.(Phe508del)**
Fragile X syndrome (FXS)	Low risk	Normal range allele(s) detected FMR1:c.-129CGG: 29,29
Spinal muscular atrophy (SMA)	Low risk	At least two copies of the SMN1 gene detected

INTERPRETATION

CF This individual is a carrier of CF and is therefore at increased risk of having a child affected by CF.
FXS This individual is unlikely to be a carrier of FXS. Residual risk varies with ethnicity and family history.
SMA This individual is unlikely to be a carrier of SMA. Residual risk varies with ethnicity and family history.

RECOMMENDATION

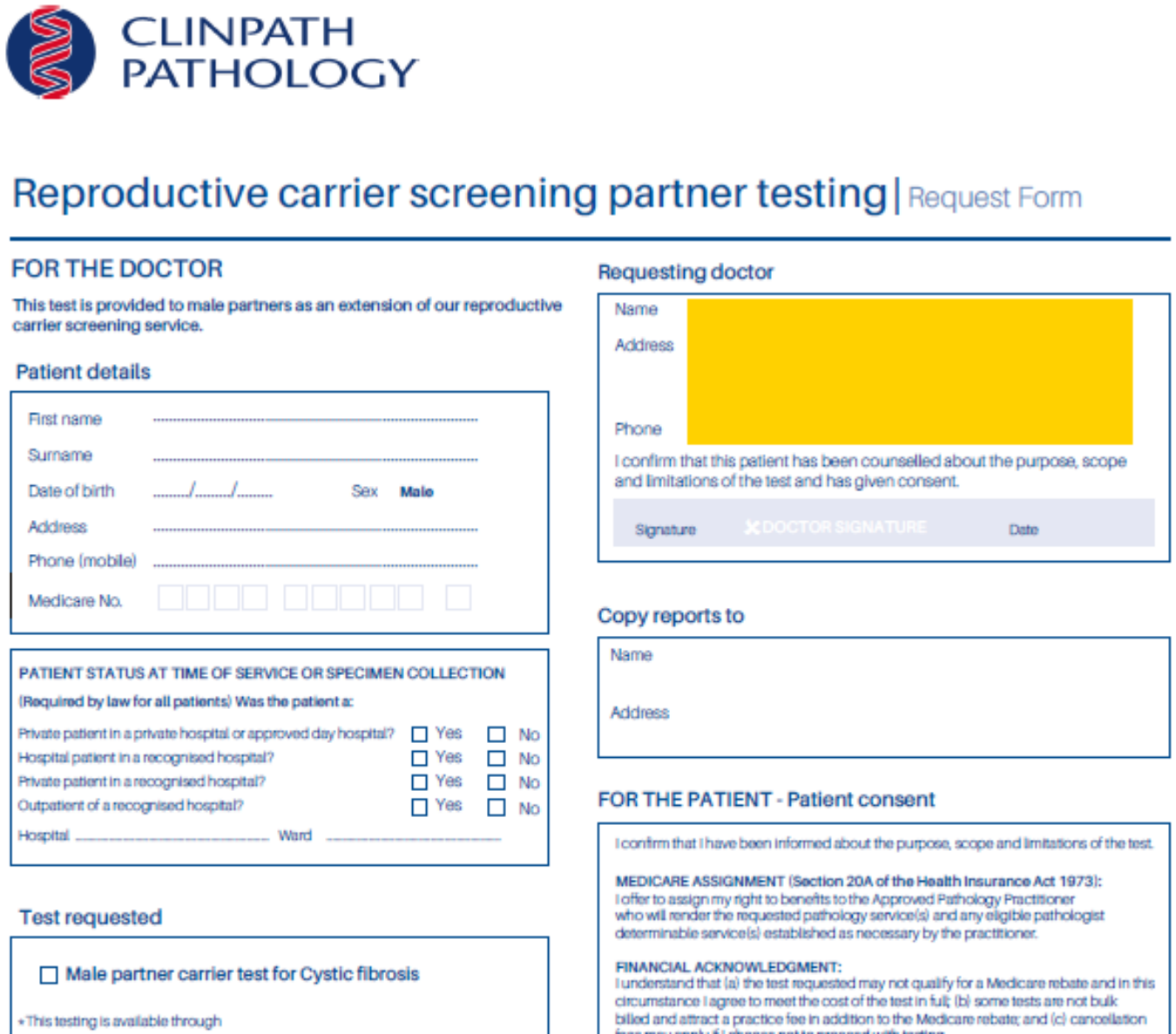
Discussion of this result and its implications is recommended (see https://www.sonic.fyi/rcs/ for further information). CF carrier testing of the reproductive partner (MBS-rebatable if eligible, see attached) and at-risk family members is recommended. This report should be interpreted in conjunction with the reproductive partner carrier screen result.

TEST INFORMATION

How do I order partner testing?

Standard Clinpath form as:
“carrier testing, partner carrier of XXXX”

Partner testing form which is provided in females report



CLINPATH PATHOLOGY

Reproductive carrier screening partner testing | Request Form

FOR THE DOCTOR

This test is provided to male partners as an extension of our reproductive carrier screening service.

Patient details

First name .....
Surname .....
Date of birth ...../...../..... Sex **Male**
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
Hospital patient in a recognised hospital? ☐ Yes ☐ No
Private patient in a recognised hospital? ☐ Yes ☐ No
Outpatient of a recognised hospital? ☐ Yes ☐ No

Hospital ..... Ward .....

Test requested

☐ Male partner carrier test for Cystic fibrosis

• This testing is available through

Requesting doctor

Name
Address
Phone

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

Signature ✕DOCTOR SIGNATURE Date

Copy reports to

Name
Address

FOR THE PATIENT - Patient consent

I confirm that I have been informed about the purpose, scope and limitations of the test.

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.

FINANCIAL ACKNOWLEDGMENT:
I understand that (a) the test requested may not qualify for a Medicare rebate and in this circumstance I agree to meet the cost of the test in full; (b) some tests are not bulk billed and attract a practice fee in addition to the Medicare rebate; and (c) cancellation

Expanded Reproductive Carrier Screen

430+ genes

Expanded Carrier Screening

Screens for 400+ genes to identify if a patient is a carrier of a genetic condition that could affect their baby.

This comprehensive screen can detect one in 44 couples at high risk of having an affected child. If a couple is found to be at risk of having an affected child they are then able to make an informed choice to accept that risk or consider a range of reproductive options such as IVF and prenatal testing to reduce the risk.

Expanded Carrier Screening

The disorders on the panel are AR and X-linked disorders which are **serious**, have **childhood-onset**, and for which there are **limited therapeutic options**.

Most patients will not have a family history.

For patients with a family history of a known mutation (such as FRAX or CF) this is not the appropriate test.

	Reproductive carrier screen (three-gene panel)	Expanded reproductive carrier screen (Beacon)
Specific request form required	No	Yes
Specimen required	Blood sample	Blood sample (or cheek swab, by arrangement)
Number of conditions screened (female)	3 common genetic conditions: Cystic fibrosis, spinal muscular atrophy, fragile X syndrome	>400 serious childhood-onset conditions with limited therapies
Number of conditions screened (male)	2 common genetic conditions: Cystic fibrosis, spinal muscular atrophy^	>350 severe childhood-onset conditions with limited therapies
Chance of a person being shown to be a carrier	~1 in 20 individuals (5%)	~3 in 4 individuals (75%)
Chance of a couple being shown to be at increased reproductive risk	~1 in 240 couples (0.4%)	~1 in 44 couples (2%)[1]
Medicare rebate	Yes#	No, ($595* per person)
Combined report for a couple available	No	Yes
Result turnaround time for individual test (or couple tested together)	2 weeks	~ 4 weeks
Testing laboratory	Accredited Sonic Healthcare laboratory (Australia)	Accredited Fulgent Genetics laboratory (US)
Counselling	Provided at no additional cost to couples tested by Sonic Genetics and found to be at high risk of having an affected child (details on how to refer eligible couples will accompany the results).	

	Reproductive carrier screen (three-gene panel)	Expanded reproductive carrier screen (Beacon)
Specific request form required	No	Yes
Specimen required	Blood sample	Blood sample (or cheek swab, by arrangement)
Number of conditions screened (male)	2 common genetic conditions (cystic fibrosis, spinal muscular atrophy)	>400 serious childhood-onset conditions with limited therapies
Number of conditions screened (female)	2 common genetic conditions (cystic fibrosis, spinal muscular atrophy)	>350 severe childhood-onset conditions with limited therapies
Chance of a person being shown to be a carrier	~1 in 20 individuals (5%)	~ 3 in 4 individuals (75%)
Chance of a couple being shown to be at risk	~1 in 240 couples (0.4%)	~1 in 44 couples (2%)
Medicare rebate	Yes#	No, ($595* per person)
Combined male and female test	No	Yes
Result of test (or couple tested together)	Within 2 weeks	~ 4 weeks
Testing laboratory	Accredited Sonic Healthcare laboratory (Australia)	Accredited Fulgent Genetics laboratory (US)

Due to the high chance of being a carrier, it is generally recommended to test both female and male at the same time.

There is no Medicare rebate and no free male partner testing.

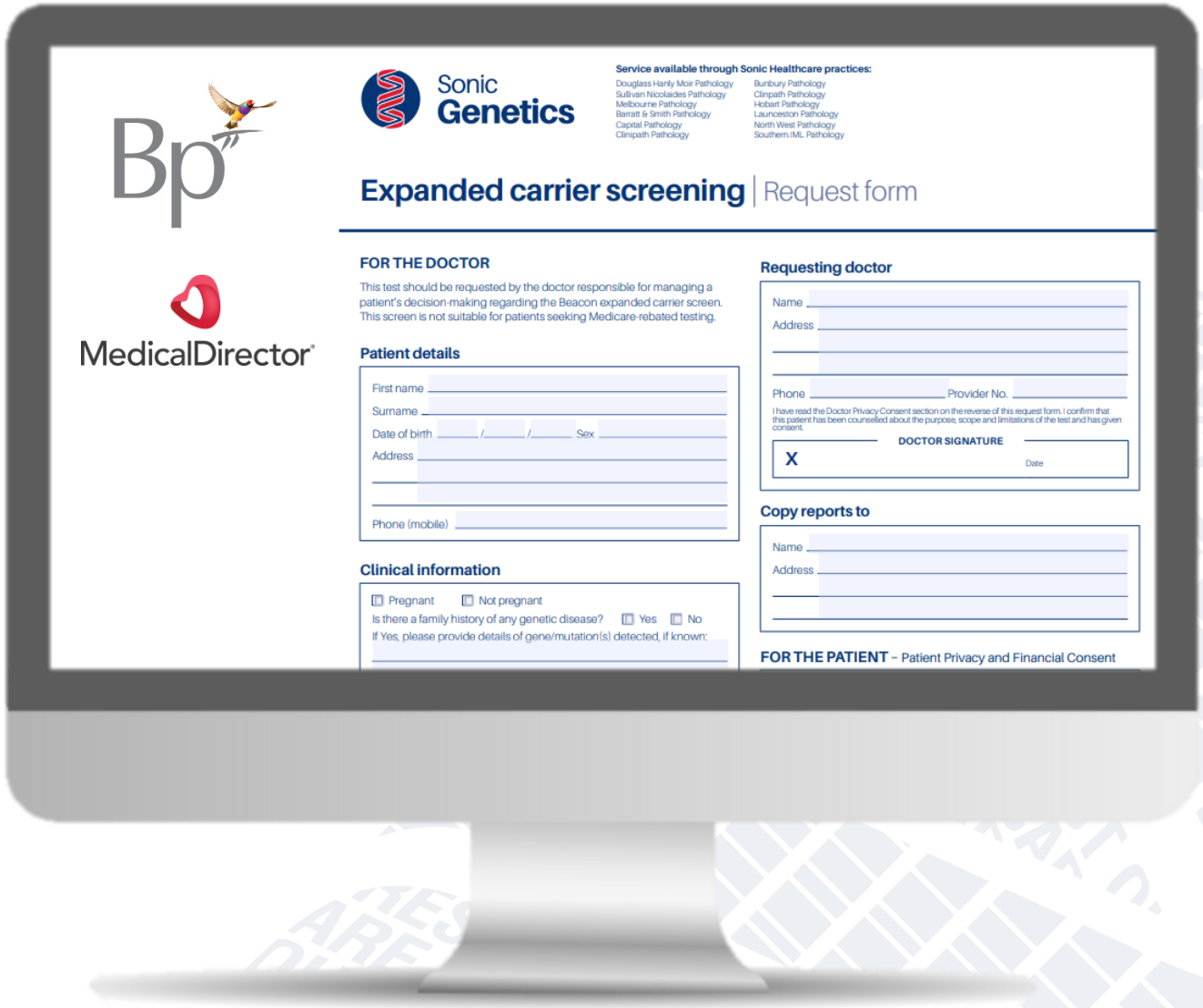
If couple pregnant, strongly advise testing simultaneously due to TAT.

How do I order expanded testing?

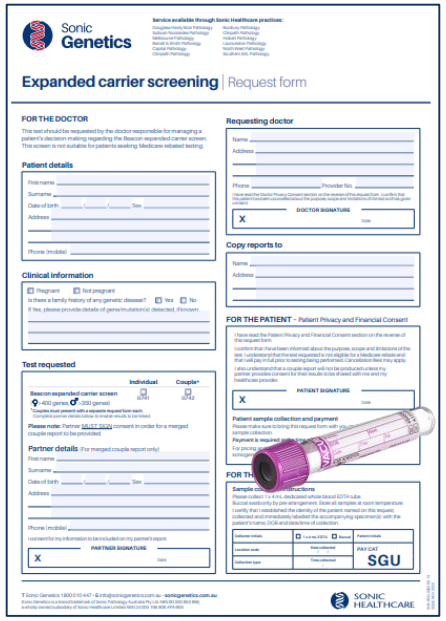
Standard Clinpath or request forms cannot be used for expanded testing

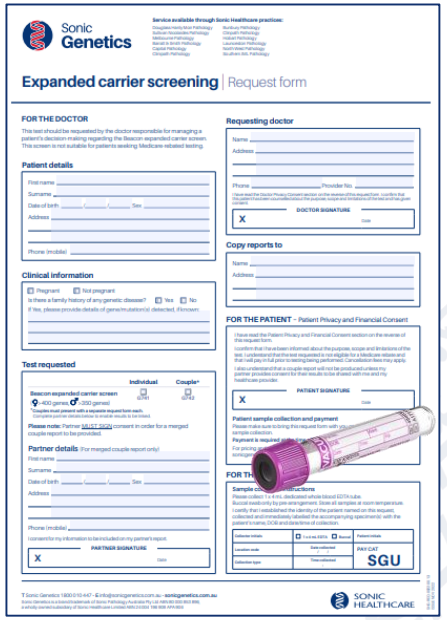
Expanded Carrier Screening form – in BP, Med Director or available at www.sonicgenetics.com.au

One form PER patient.



Ordering Expanded Carrier Screening





Each patient needs a form and they must sign each others forms for a couple test.

Genetic Counselling

One counselling session with our Genetic Counsellor for all high risk couples (X-linked carrier female, AR carrier couple).

Addresses informational and psychological matters relating to preconception carrier screening results

Performed by phone/video telehealth

Letter sent from counsellor to referring doctor to advise of discussion and key points

How do I arrange genetic counselling?

Not available for all genetic results

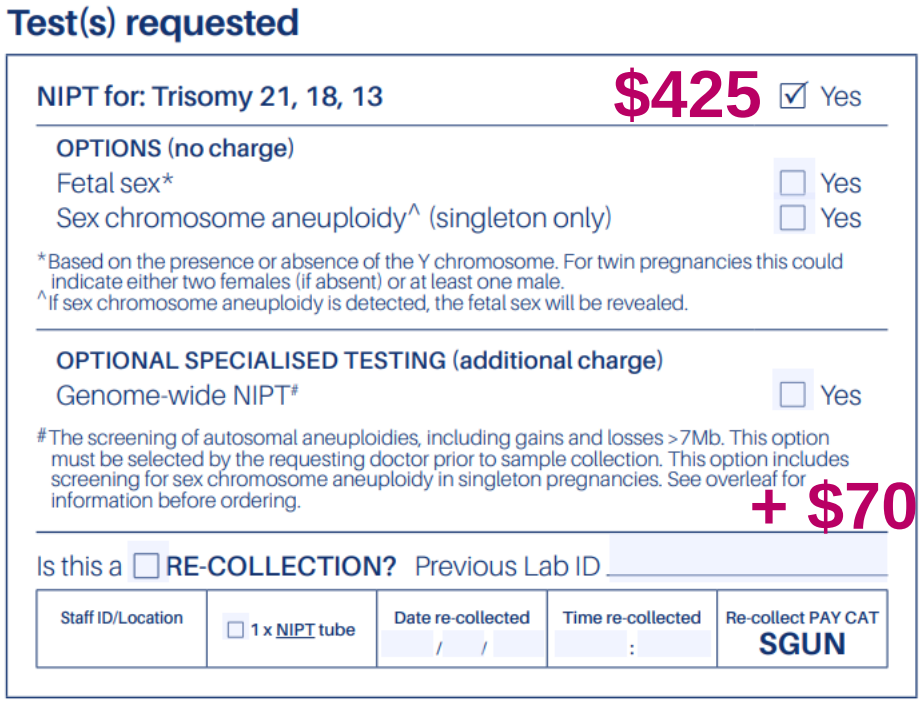
Available for reproductive carrier screening high risk couples where at least one patient is tested through Sonic

Telehealth and once off session for the couple

Non-Invasive Prenatal Testing

and Testing Options

Non-invasive Prenatal Testing (NIPT)



Test(s) requested

NIPT for: Trisomy 21, 18, 13 **$425** ☑ Yes

OPTIONS (no charge)

Fetal sex* ☐ Yes

Sex chromosome aneuploidy^ (singleton only) ☐ Yes

*Based on the presence or absence of the Y chromosome. For twin pregnancies this could indicate either two females (if absent) or at least one male.
^If sex chromosome aneuploidy is detected, the fetal sex will be revealed.

OPTIONAL SPECIALISED TESTING (additional charge)

Genome-wide NIPT# ☐ Yes

#The screening of autosomal aneuploidies, including gains and losses >7Mb. This option must be selected by the requesting doctor prior to sample collection. This option includes screening for sex chromosome aneuploidy in singleton pregnancies. See overleaf for information before ordering.

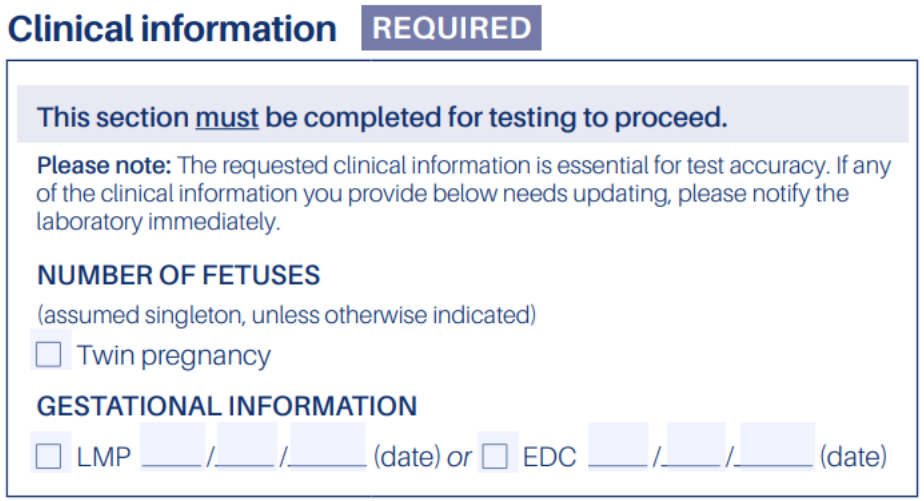
+ $70

Is this a ☐ RE-COLLECTION? Previous Lab ID

Staff ID/Location	☐ 1 x NIPT tube	Date re-collected / /	Time re-collected :	Re-collect PAY CAT SGUN

- **Standard testing is T21, T18, T13**
- **Fetal sex and SCA can be added at no extra cost to patient**
- **Genome-wide NIPT can be added at request of doctor for additional $70 bringing cost of NIPT to $495**

Non-invasive Prenatal Testing (NIPT)



Clinical information REQUIRED

This section must be completed for testing to proceed.

Please note: The requested clinical information is essential for test accuracy. If any of the clinical information you provide below needs updating, please notify the laboratory immediately.

NUMBER OF FETUSES

(assumed singleton, unless otherwise indicated)

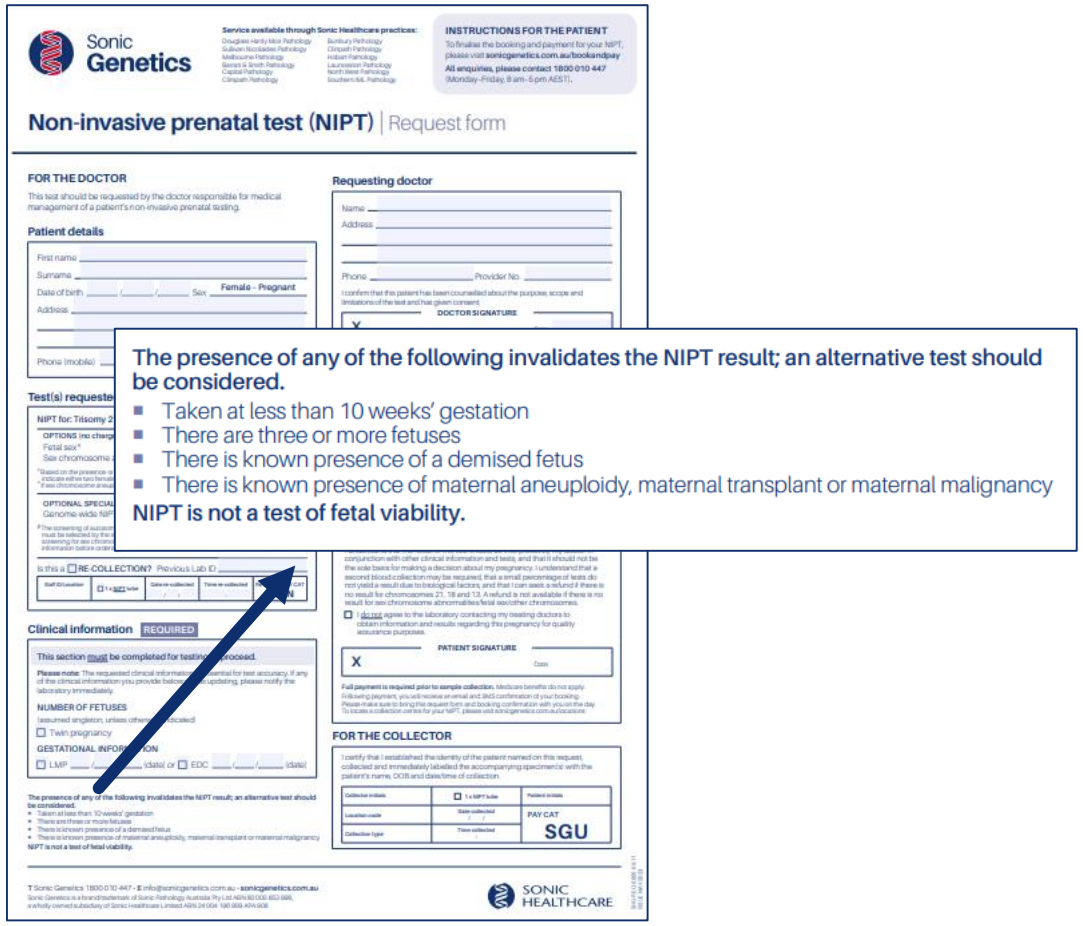
☐ Twin pregnancy

GESTATIONAL INFORMATION

☐ LMP ____/____/_____ (date) *or* ☐ EDC ____/____/_____ (date)

- **Ensure that twin pregnancy is marked if required.**
- **Ensure at least one of LMP or EDC is completed and if later scan shows different date then please call laboratory to amend.**

Non-invasive Prenatal Testing (NIPT)



Please ensure that you have noted this section of the form which explains some limitations of NIPT testing.

How do I order NIPT testing for my patient?

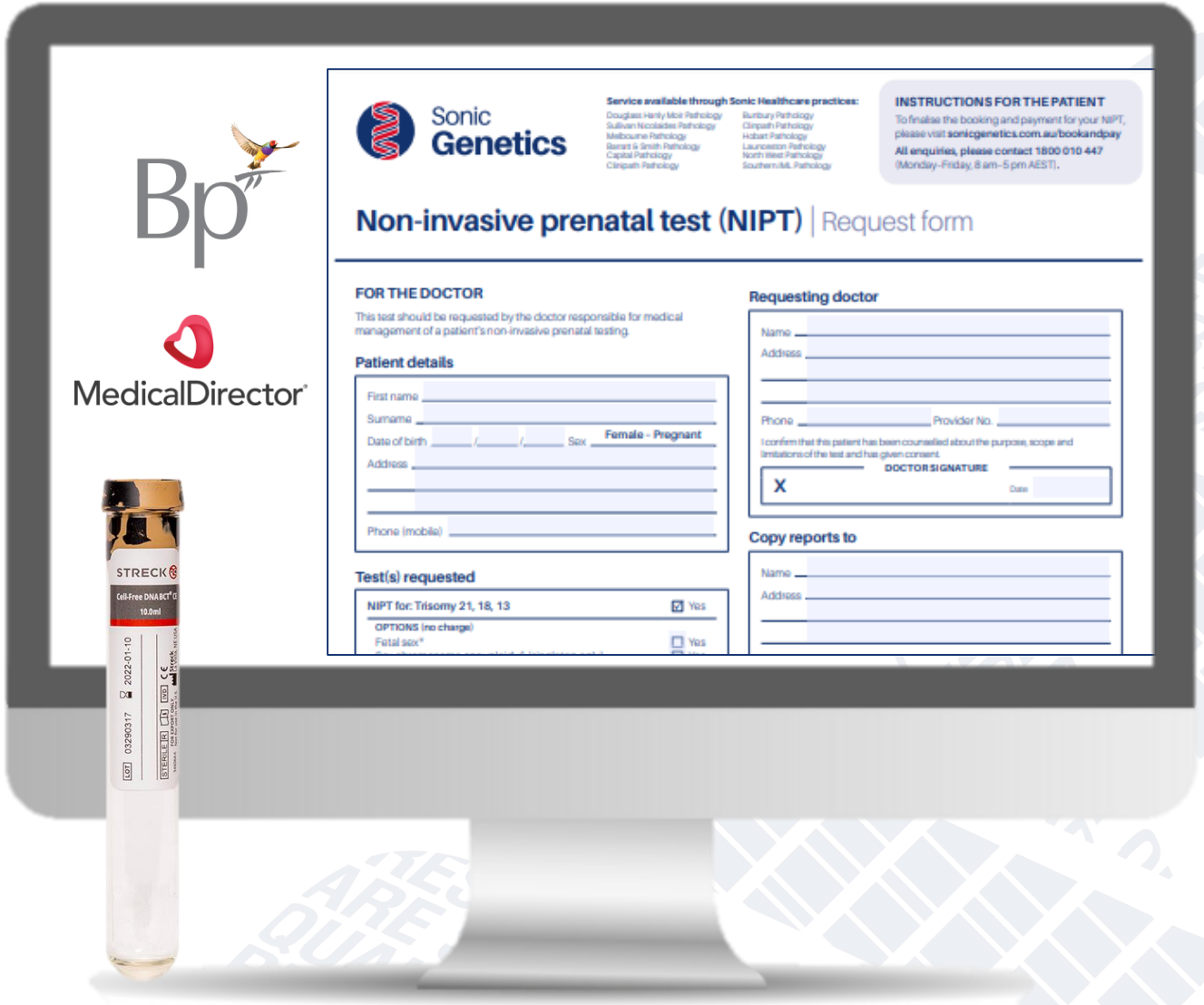
Order on a Clinpath NIPT form

Available in BP and Med Director or available in PDF form on our website.

Patient goes online to book & pay for test

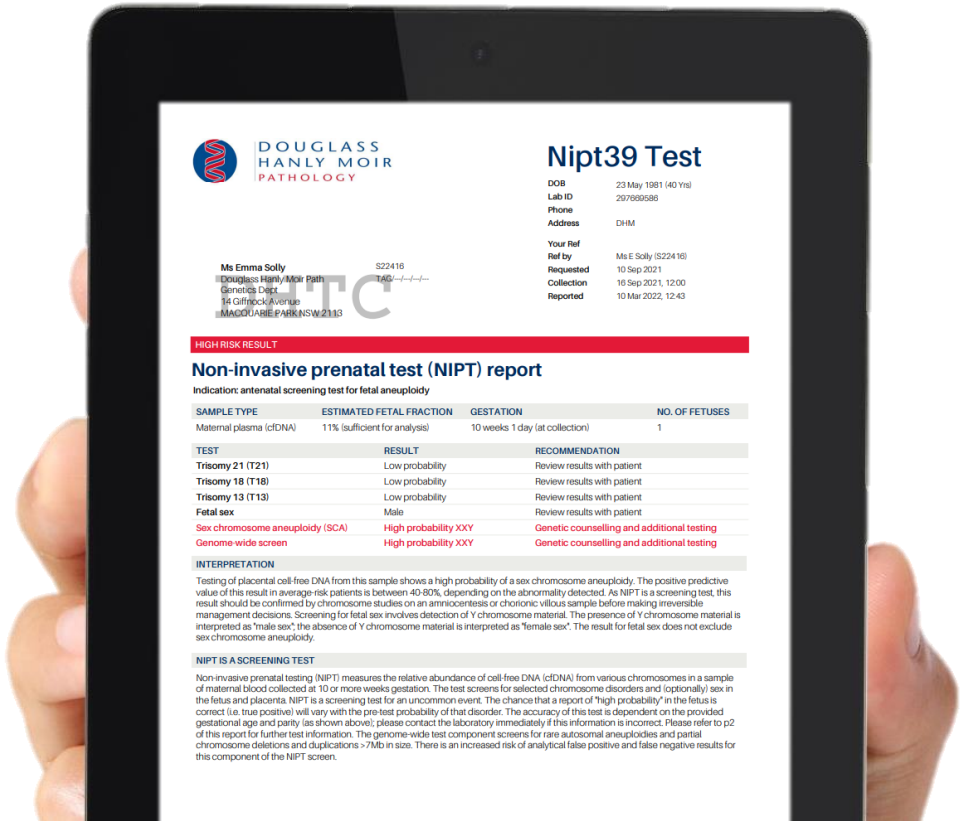
and attends preferred collection centre

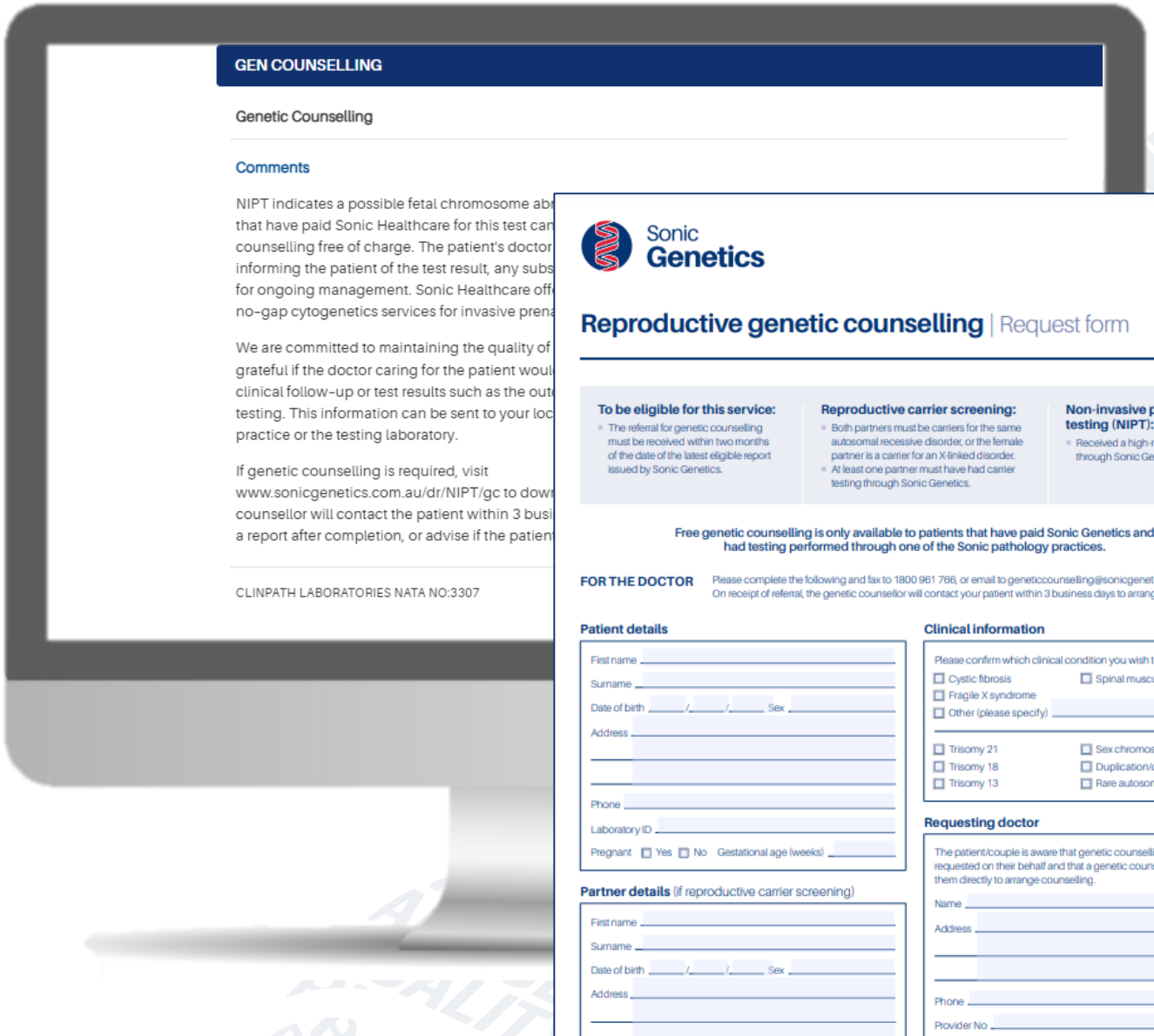
Results returned in 3-8 business days (typically 3-5)



Genetic Counselling for High-Risk Results

All high risk NIPT's are eligible for genetic counselling

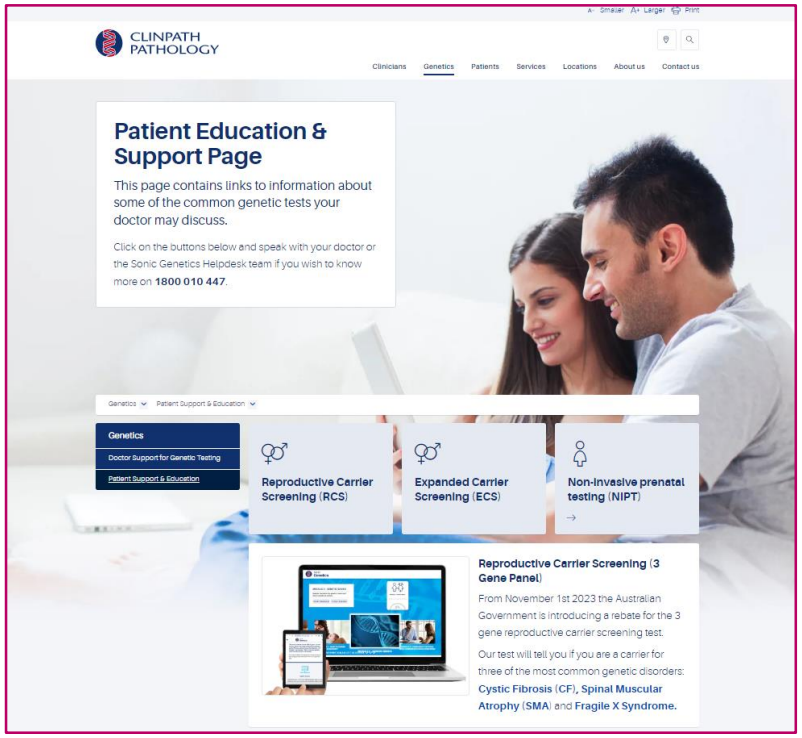




Resources for Patients

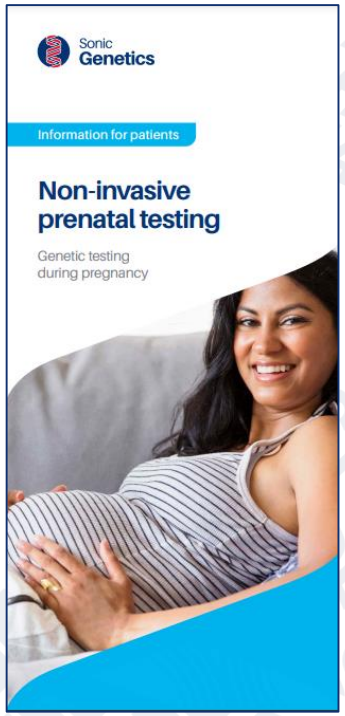
Pre-Test Resources

Clinpath Website



Patient Cards

Brochures



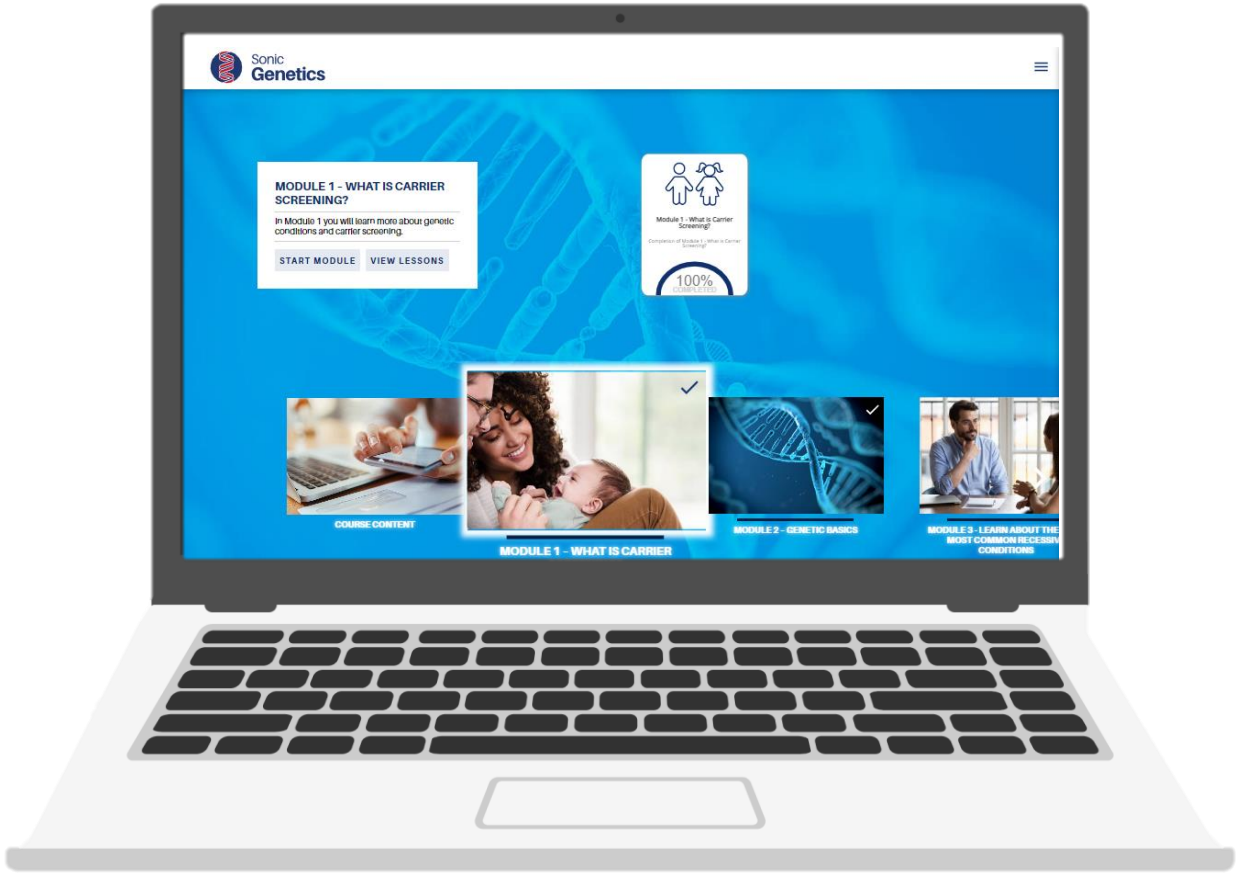
Carrier Screening Education

Patient Resources
Genetic Testing

Scan the QR Code for more information on
Reproductive Carrier Screening
or visit our website
clinpath.com.au/genetics

Sonic Genetics Helpdesk
P | 1800 010 447

Carrier Screening Education



Course content

The course seeks to inform the patient and guide them in identifying their own preferences, rather than directing them to a particular conclusion. At the end of the course, patients are able to print out their preferences to discuss with you.

The carrier screen patient course is delivered in six modules:

Module 1. What is carrier screening?

- Carrier screening basics
- Recessive genetic conditions
- Screening for recessive genetic conditions

Module 2. Genetics basics carriers

- Recessive condition statistics
- If I am a carrier for an autosomal recessive condition, what is the risk to our child?
- If a woman is a carrier for an X-linked condition, what is the risk to her child?

Module 3. Learn about the four most common recessive conditions

- Cystic fibrosis
- Spinal muscular atrophy
- Fragile X
- Thalassaemia

Module 4. Understanding your choices

- Carrier screening accuracy and results
- Being a carrier
- If I have a normal carrier screen, what is the chance that I am still a carrier?
- Before doing the test

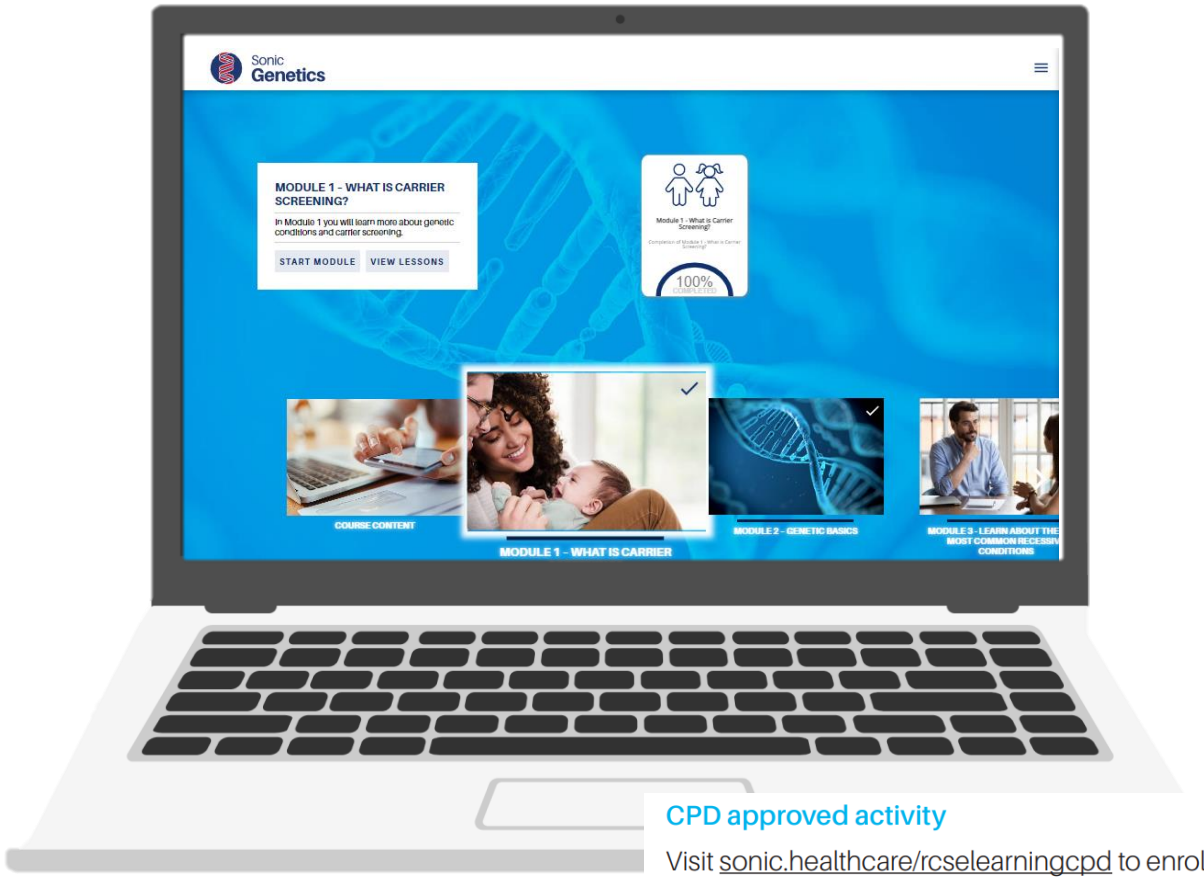
Module 5. Doing the test

- Getting a carrier screen
- Practical questions about carrier screening

Module 6. Making a decision

- What is my next step? (decision tool)
- Completing the course

Carrier Screening Education



This course can be used as a refresher for CPD hours.

CPD approved activity

Visit sonic.healthcare/rcselearningcpd to enrol.

RACGP CPD Approved Activity

Educational Activities	Measuring Outcomes	Reviewing Performance
2 hours	0 hours	0 hours

ACRRM CPD ACCREDITED ACTIVITY 2023-2025

Further Support

Your Local Support Team:

Katelyn Henry

Key Contacts

Katelyn Henry
Genetics Business Manager (SA)

M | 0408 819 430 E | khenry@clinpath.com.au

For more information including access to Dr. support page visit
www.clinpath.com.au/genetics/

Doctor Priority Phone line

8366 2111

Sonic Genetics Helpdesk

1800 010 447