

Helping patients understand their reproductive health

We introduce the Beacon expanded reproductive carrier screen for more than 300 severe childhood-onset genetic disorders

Reproductive carrier screening is fast becoming an essential factor in pregnancy planning with RANZCOG now recommending that information about reproductive carrier screening for common disorders be offered to every woman either prior to conception or in early pregnancy.

Reproductive carrier screening tests a woman and her partner for the presence of gene mutations which, if present in both, can affect their children through autosomal inheritance. We now offer the Beacon expanded reproductive carrier screen for 320+ severe disorders that although rare individually, collectively are more common than the familiar chromosomal disorders such as Down syndrome for which prenatal screening is widely available and accepted. Although uncommon, these conditions may have serious implications for a family when they occur. Informing a woman of her risk before she becomes pregnant can help her and her partner understand the implications of having a shared genetic mutation.

Beacon expanded reproductive carrier screen

After assessing the technology available at the present time, we have selected the Beacon expanded reproductive carrier screen, which is the most accurate reproductive carrier screen available in Australia. This test is developed in the USA by Fulgent Genetics.

About 70% of Australians are identified as carriers for one or more disorders using this investigation. Most carriers do not have a family history of relatives affected by the disorder and are unaware that they are carriers. About 3% of Australian couples will be found to have a 25% chance of having a child with one of these conditions.

Autosomal and X-linked recessive disorders

The Beacon expanded reproductive carrier screen identifies carriers of serious autosomal and X-linked recessive disorders that have childhood onset and for which there are limited therapeutic options. It does not include mild disorders, autosomal dominant disorders, adult-onset disorders, or disorders for which there are effective interventions available in Australia.

There are hundreds of inherited autosomal and X-linked recessive disorders that present in infancy and early childhood. When both parents are carriers of mutations in the same gene located on one of the autosomes (chromosomes 1-22) the chance of a child inheriting the abnormal gene from each parent and developing an autosomal recessive disorder is 25%. In the case of a woman with a recessive mutation on an X chromosome, each of her sons is at 50% chance of inheriting an

abnormal gene and being affected, and half of her daughters will be carriers.

Requesting the Beacon expanded reproductive carrier screening

This test can be requested by any medical practitioner. If you would like more information or advice, our specialist genetic pathologists can be reached on 1800 010 447.

Please use our [dedicated request form](#) for the Beacon expanded reproductive carrier screen which sets out the information needed by the specialist genetic pathologist in order to provide an accurate interpretation of the genetic analysis. This form also provides billing and collection information for the patient. Ensure that details of carrier screening for the reproductive partner are included on the request form if applicable.

Please note on the referral form whether or not the patient has a family history of a single-gene disorder. If there is a family history, please provide as much information as possible about the family pedigree and any previous testing, with results if available.

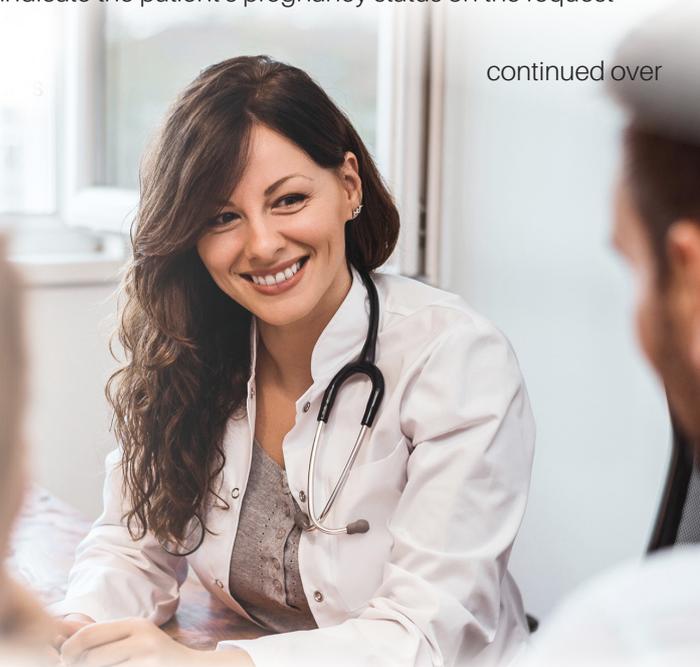
If the partner is being tested at the same time, they will require a separate request form. Please ensure that details for both are noted on their respective request forms.

The best time to establish carrier status is before conception; however, screening can still be performed in early pregnancy. Please indicate the patient's pregnancy status on the request form.

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Sonic Genetics

We are part of Sonic Genetics, Australia's largest private genetics referral laboratory, and the national genetics laboratory of Sonic Healthcare. Our genetic pathologists work alongside their colleagues in Sydney.





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Collection

Any Sullivan Nicolaides Pathology collection centre.

We require 1 x 4 mL blood in an EDTA tube. Buccal swabs can be used by prior arrangement, however, this method of collection is not recommended because it does not provide DNA of equal quality and quantity to that of a blood sample.

Cost

For up-to-date information about prices, please refer to the Sonic Genetics website www.sonicgenetics.com.au. Please note that if a couple chooses to be tested together, there is a reduced cost.

There is no Medicare rebate for carrier screening. If there is a family history of a known mutation, such as Fragile X or cystic fibrosis, the Beacon expanded carrier screen is not the appropriate test, so please check with the laboratory for more information on requesting these tests.

Turnaround time

Once the sample is received at our laboratory, the results will typically be available within three to four weeks.

Genetic counselling

Couples tested by us and found to be at high risk of having an affected child will be offered genetic counselling free of charge. A referral is required. Further information for counselling will be provided with the test report.

We have a range of resources to help guide you and your patients

General information is available on the Sonic Genetics website: <https://www.sonicgenetics.com.au/rcs/beacon/>

[Request form](#)

[Information booklet for doctors](#)

[Technical bulletin for doctors](#)

[Information booklet for patients](#)

Cervical Screening Test

Important to check expiry dates on ThinPrep® vials

Surgeries are reminded that ThinPrep® vials used for collection of CSTs have an expiry date and must be stored between 15-30°C.



Collection media used outside manufacturer specifications may be unreliable

To allow for processing and mandatory specimen retention times, do not use vials within 6 weeks of expiry.

If a sample is received in an expired vial, we may need to ask you to repeat the collection to ensure the integrity of the sample.

Please check your stock regularly and order new stock on the SNP Stores order form.

Pathologist Profile

Dr Anna Eastman BMed FRCPA

We are delighted to introduce Dr Anna Eastman who has recently joined the practice at Toowoomba as a consultant histopathologist. She reports across all areas of histopathology with a special interest in gynaecological pathology and dermatopathology.

Dr Eastman undertook her medical internship and residency at the Toowoomba Hospital after graduating in Medicine from the University of Newcastle. She began training in anatomical pathology in Newcastle but moved back to Toowoomba to be closer to her family. She worked as a clinician at Toowoomba Hospital, and then at Sullivan Nicolaides Pathology's Toowoomba laboratory, before completing her anatomical pathology training program in Brisbane.

Dr Eastman was awarded Fellowship of the Royal College of Pathologists of Australasia in 2018. She is Australia's first known pathologist of Aboriginal and Australian South Sea Islander descent.

She values working in a large and busy regional practice and the opportunities it provides for exposure to a broad range of histopathology cases. She also appreciates being part of a small team and the close working relationships with local clinicians that are possible in a rural centre such as Toowoomba.

Phone: (07) 4699 2000

Email: Anna_Eastman@snp.com.au



Changes to services for Easter 2019

Warfarin Care enrolments

To ensure the safe and complete enrolment of patients into our Warfarin Care program, enrolments will be closed between the following dates:

Community patients: closing 5 pm Thursday 4 April 2019 and re-opening 9 am Monday 29 April 2019.

Hospital patients: closing 5 pm Thursday 11 April 2019 and re-opening 9 am Monday 29 April 2019.

Collection centres

Please see www.snp.com.au for information regarding collection centre closures.

Cardiology services

The Cardiology department is closing 5 pm Thursday 18 April 2019 and re-opening Tuesday 23 April 2019 for all services. Monitoring services are unavailable during this time.