Sonic Genetics
Douglass Hanly Moir Pathology

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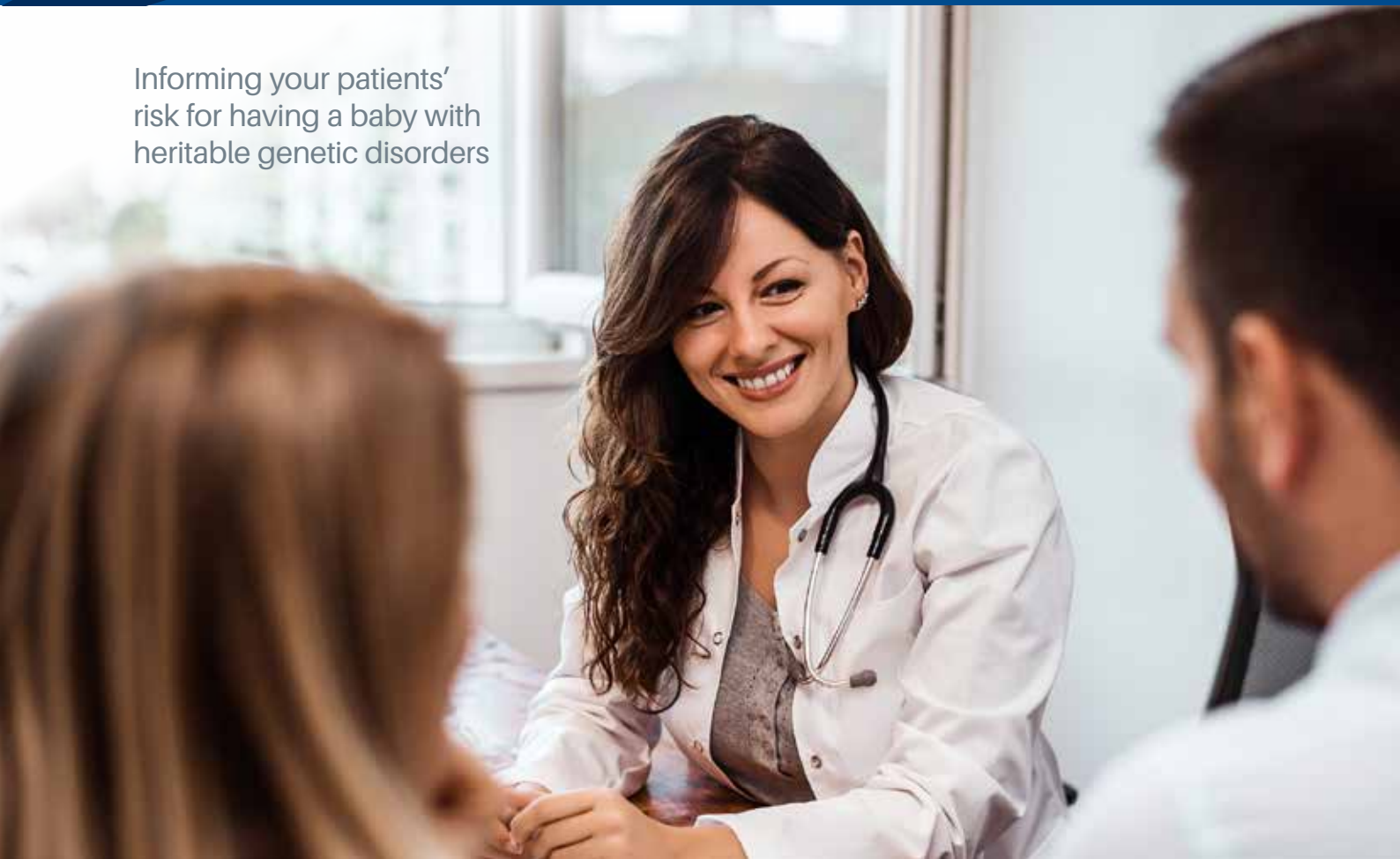
Expanded carrier screening

Information for Doctors

Informing your patients' risk for having a baby with heritable genetic disorders



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Introducing the most powerful reproductive carrier screen

Reproductive carrier screening can identify individuals or couples at high risk of having a child with a serious inherited condition. Best performed prior to conception, this test is becoming an essential part of prenatal care planning. It allows people the opportunity to explore their reproductive options, and helps ensure they can make properly informed decisions.

Expanded carrier screening

Sonic Genetics introduces the Beacon expanded carrier screen, developed in the US by Fulgent Genetics. This is a comprehensive and accurate carrier screen which provides you and your patients with valuable information for pregnancy and family planning.

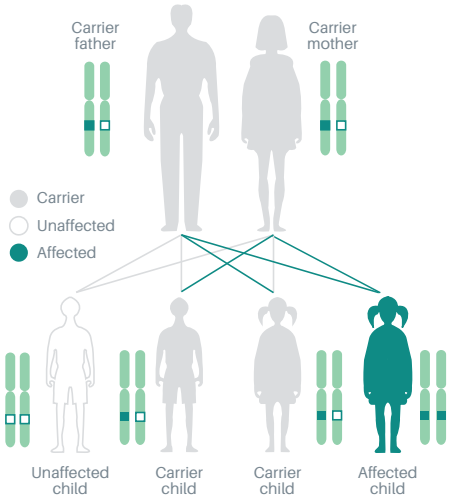
What is a carrier?

Every person has many thousands of genes. Most of those genes are present in pairs (autosomal genes), with each person inheriting one copy from each parent. Each person will have many autosomal genes in which one of the pair of genes has been inactivated by a genetic error (mutation), that is, the person is a carrier for the condition caused by that gene. This causes no medical problem because the second copy of the gene is working normally. However, there is a 50% chance that a child will inherit the abnormal gene from that person.

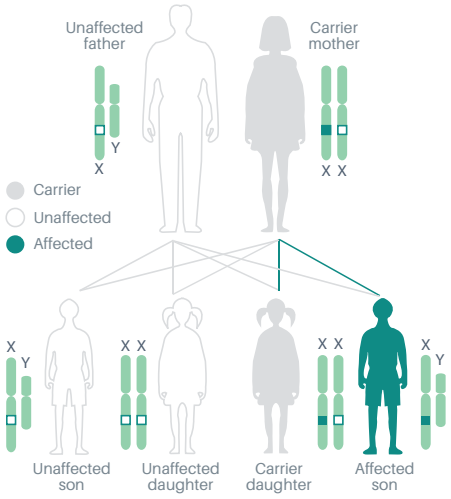
If a couple are both carriers for the same autosomal recessive disorder, then there is a 25% chance with each pregnancy that a child will inherit an abnormal gene from each parent and be affected by that condition. This applies to both boys and girls.

The story is different for genes located on the X chromosome. A man has only one X chromosome and would be affected with a serious childhood-onset disorder if he had a mutation in an X-linked gene. A woman has two X chromosomes and can be an unaffected carrier for an X-linked disorder. Her son is at 50% risk of inheriting the abnormal gene and being affected by an X-linked recessive disorder. Her daughter is also at 50% risk of inheriting the abnormal gene but would usually be protected from the adverse effects of this by the normal gene from her father. Overall, a woman who is a carrier of an X-linked disorder is at 25% risk of having an affected child.

Autosomal recessive inheritance



X-linked recessive inheritance



Why should I offer carrier screening to my patients?

There are hundreds of inherited genetic disorders which can affect children. Each of these conditions is individually rare, but together, they are more common than familiar chromosomal disorders, such as Down syndrome. In Australia, approximately one in 200 babies is born with a serious inherited disorder that will affect them during childhood. These disorders are inherited as autosomal recessive or X-linked recessive conditions, with one or both parents being carriers of the disorder.

Everyone is a carrier for at least one genetic disorder, but most people are not aware of their carrier status. A carrier will only know his or her status after the birth of a child with the disorder or by having a carrier screen.

For couples considering carrier screening, the test should be offered to women before conception, to their reproductive partners, and to gamete (egg or sperm) donors and recipients. Carrier screening will assist in preconception planning and prenatal diagnostic testing for those couples identified as carriers. Carrier screening is also possible during early pregnancy, however, the subsequent options for intervention are more limited and more stressful than preconception testing.

If a couple is found to be at 25% risk of having an affected child, a number of reproductive options can be considered prior to conception, for example, pre-implantation genetic diagnosis and IVF, the use of donor egg or donor sperm, prenatal diagnosis, or making an informed decision to accept the risk. The reproductive options that are available once the woman is pregnant are limited to prenatal diagnosis and making an informed decision to accept the risk.

Professional medical associations in the US, such as the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG), have published guidelines on expanded carrier screening and its importance in reproductive care.

Please note that carrier screening is not recommended for children. Being a carrier for a condition does not place a child at increased risk of developing a childhood-onset disorder. Carrier screening should be deferred until adulthood so that the person being tested can make their own decision about having the test. This also provides a better opportunity for testing of the reproductive partner.

What does the Beacon expanded carrier screen test for?

The Beacon expanded carrier screen is designed to identify carriers of autosomal and X-linked recessive disorders which are serious, have childhood onset, and for which there are limited therapeutic options. This test does not include mild disorders, autosomal dominant disorders, adult-onset disorders, or disorders for which there are effective interventions available in Australia, for example, haemochromatosis, Factor V Leiden, MTHFR variants, non-syndromic sensori-neural deafness, and familial breast cancer. A detailed list of disorders included in the Beacon expanded carrier screen is provided at the end of this brochure.

Carrier screening is designed for individuals and couples who are not aware of a family history of an autosomal or X-linked recessive condition. Carrier screening may not be the best test for a person who is likely to be a carrier for a specific condition. If there is a family history or other reason for high pre-test carrier risk, it may be more appropriate for a couple to be referred for genetic counselling and testing that is specific for that condition.

A carrier screen cannot identify *de novo* genetic disorders which occur for the first time during a pregnancy, for example, trisomy 21 and new autosomal dominant disorders, such as achondroplasia. Carrier screening does not provide information about the probability of a child having a congenital malformation due to the interaction of multiple genes or the influence of non-genetic factors, such as prenatal infections.

Why choose the Beacon expanded carrier screen?

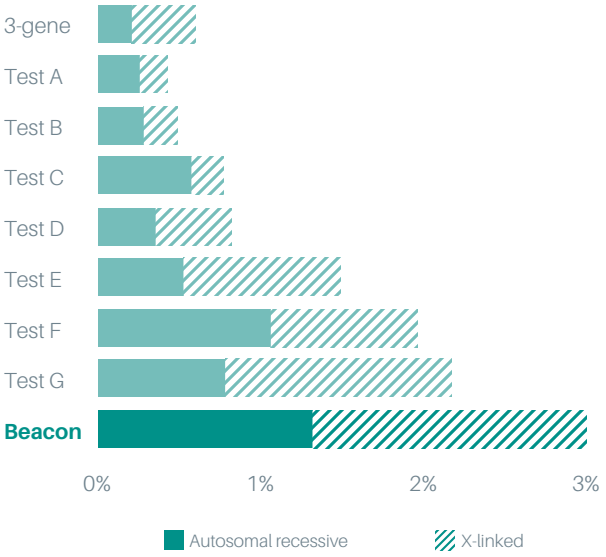
There are many hundreds of genes which can cause serious childhood-onset disorders, and many screening tests available through laboratories in Australia and overseas. It is tempting to think that the best measure of the value of a screening test is the number of genes analysed by the test. However, three additional factors must also be considered for each gene analysed: the frequency of mutations in the gene, the ability of a test to detect those mutations, and the mode of inheritance of the disorder caused by the gene.

A number of Australian laboratories (including Sonic Genetics) provide carrier screening for three common disorders: cystic fibrosis, spinal muscular atrophy and Fragile X syndrome. Approximately one in 160 couples (0.6%) will be identified as being at high risk of having an affected child by this three-gene panel. Some laboratories offer carrier screens of more than 100 genes which, despite examining many more genes, fail to identify more at-risk couples than the three-gene panel. This highlights the problem of relying on the number of genes analysed as the measure of a test's value.

Sonic Genetics has carefully evaluated the potential performance of nine popular carrier screens provided by laboratories in Australia and overseas. The number of genes tested by these laboratories varied from three to 327. We assessed the value of each test by calculating the proportion of Australian couples who would be identified as being at high risk of having a child with an autosomal recessive or X-linked recessive disorder (see Figure 1).

The best performance by far was obtained by the Beacon expanded carrier screen. Approximately one in 30 couples (3%) would be identified as being at 25% risk of having an affected child.

Figure 1: % couples identified as being at high risk



The Beacon test identifies five times more couples at risk of having an affected child, who can then make informed reproductive choices, than would be identified by the three-gene carrier screen and some large screens from other well-known providers.

The success of the Beacon test is due to the number of genes analysed (327), the frequency of mutations in these genes, the high detection rate for mutations (>98%), and the inclusion of 28 X-linked genes.

The Beacon expanded carrier screen

The Beacon expanded carrier screen consists of genes for 299 autosomal recessive disorders and 28 genes for X-linked recessive disorders. For female patients, we test the genes for both types of disorder (327 genes in all); for male patients, we test only for autosomal recessive disorders as it is assumed that the man does not have a severe childhood-onset X-linked recessive disorder.

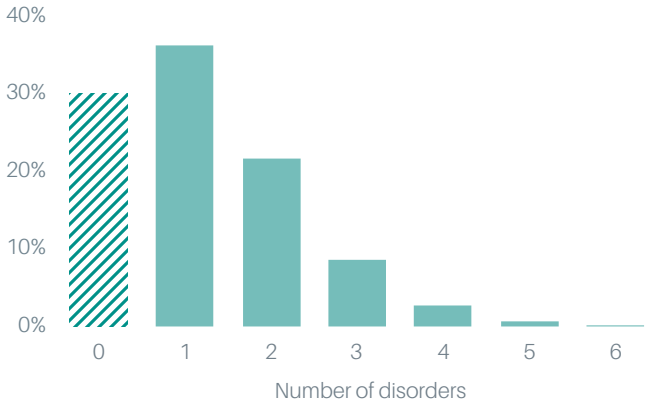
Overall, the Beacon expanded carrier screen will identify approximately 70% of Australian women as being a carrier of one or more disorders. The chance is slightly lower in men as it is assumed that they do not have a severe childhood-onset X-linked recessive disorder.

If a person is identified as a carrier of an autosomal recessive disorder, this is of little consequence unless the reproductive partner is also identified as being a carrier of the same condition. The probability of a person being identified as a carrier of one or more autosomal recessive disorders is shown in Figure 2. There is a one-in-three chance of a person not being identified as a carrier, the same of being shown to be a carrier of one disorder, and the same chance of being shown to be a carrier of two or more disorders. Approximately one In 77 couples (1.3%) will be identified in which both partners are carriers of the same autosomal recessive disorder.

If a woman is identified as a carrier of an X-linked recessive disorder, this is of immediate consequence in reproductive planning, as there is a 50% chance that her son would inherit the abnormal gene and be affected.

Approximately one in 60 women (1.7%) will be identified by the Beacon test as being a carrier of an X-linked recessive disorder. It is uncommon for a woman to be identified as a carrier of more than one X-linked recessive disorder.

Figure 2: Probability of being identified as an autosomal recessive carrier



Methodology

The Beacon expanded carrier screen is performed by the developers of the test, Fulgent Genetics, in their laboratory in California. In Australia, the Beacon test is available exclusively through Sonic Genetics.

The Beacon expanded carrier screen covers hundreds of genes, using a cost-effective and highly accurate technology. The Beacon test is not limited to common mutations in genes.

With a handful of exceptions, the Beacon test looks for different mutations in each gene, including sequence variants and deletions. The few exceptions are conditions in which a single type of mutation in one gene accounts for the great majority of diagnoses, for example, Haemophilia A and Fragile X syndrome. More than 45,000 mutations can be detected in the 327 genes using the methods described below.

The Beacon expanded carrier screen is the only carrier screen that offers sequencing and deletion/duplication analysis of more than 300 genes. Next generation sequencing (NGS) is used to analyse exons in multiple genes simultaneously. Fulgent Genetics has developed a sophisticated analysis, CNVexon™, which detects sequence changes and deletion/duplication via NGS; this methodology is only available through Sonic Genetics in Australia.

Sequence analyses can be compromised by pseudogenes (non-functioning DNA sequences which resemble genes) and by families of functional genes that contain highly similar sequences. Fulgent Genetics has developed highly sensitive tools that clearly separate genes from pseudogenes. When testing for Fragile X syndrome, the mutation (a CGG repeat expansion) is not reliably detected by NGS and a PCR method is used.

Detection rates and test limitations

The Fulgent laboratory uses a variety of laboratory and computer methods to detect mutations in gene sequence, splice junctions and deletions. The analytical detection rate for all genes is >98%. However, there remains a small chance that a person will have an undetected mutation in a gene and hence be an undetected carrier for a specific disorder. The Beacon report provides an estimate of this 'residual risk' of being an undetected carrier for an individual or couple.

Requesting the Beacon expanded carrier screen

If there is a family history of a specific condition, the risks in a pregnancy should be clarified specifically for that condition, possibly by referral for genetic counselling; carrier screening may not be the appropriate test for that specific condition. This does not preclude the individual or couple having carrier screening for other conditions.

Planning the test

We strongly recommend that carrier screening be performed prior to conception. This gives the doctor and individual, or couple, time to consider their options and make informed choices.

If a person is identified as a carrier of one or more autosomal recessive disorders, the reproductive partner should be screened to clarify the risk of an autosomal recessive disorder for the child. This is not necessary if a woman is identified as a carrier of an X-linked recessive disorder; the male partner need not be screened for X-linked recessive disorders, as the result would be of no consequence for his children.

Testing of a couple can be done sequentially, that is, test the woman first and then test her partner if indicated, or concurrently, by testing both partners at the same time. There are a number of reasons for proposing concurrent testing:

- The testing process will be quicker. Approximately 70% of women will be identified as carriers for at least one autosomal recessive disorder and testing of their male partners would then be recommended. This means that most couples will require testing of both partners.
- It is less expensive for the male partner to have the full Beacon screen rather than select individual genes for analysis. Of the women who are identified as carriers of autosomal recessive disorders, half will be carriers for more than one condition. Testing the partner to determine his carrier status for two or more conditions rapidly becomes more expensive than doing the Beacon expanded carrier screen.
- Sonic Genetics offers couples, who are tested at the same time, a 12.5%* discount for the Beacon test and provides a couple-specific report.

Pre-test considerations

An individual or couple having carrier screening must provide informed consent for the test and accept the following points:

- The purpose of carrier screening is to inform reproductive decisions and is not designed to diagnose current disease or the risk of future disease. There is an important exception to this general rule: female carriers of Fragile X syndrome are at increased risk of premature menopause and of late-onset tremor/ataxia, and should be referred for genetic counselling.
- It is impractical to provide detailed information for each of the 327 conditions screened by the Beacon test. The person being tested should be advised that the test screens for carriers of severe childhood-onset conditions. It does not screen for carriers of autosomal dominant disorders, adult-onset disorders, mild disorders, or all recessive disorders, and does not replace prenatal screening for chromosomal conditions.
- If a person is identified as carrier for an autosomal or X-linked recessive disorder, we recommend that they advise close genetic relatives. These relatives may also be carriers and may wish to use this information for their own reproductive planning. Please note that notification of relatives is recommended but not mandated.
- The genetic advice provided by carrier screening is only applicable to the couple who has been tested. If a patient has a different partner in the future, the reproductive risks need to be re-evaluated. Further testing may be required.

Pre-test genetic counselling is available throughout Australia on a fee-for-service basis. Please visit our website, www.sonicgenetics.com.au/counsellingservices.

Request forms

Please use our Expanded Carrier Screening Request Form which is available from your local Sonic Healthcare laboratory or from the Sonic Genetics website. This form prompts you to provide information necessary for us to deliver an accurate interpretation of the genetic analysis. It also provides billing and collection information for the patient. Please ensure that details of carrier screening for the reproductive partner are included if applicable.

Price

Please refer to the Sonic Genetics website, www.sonicgenetics.com.au/rcs/beacon, for current pricing. Please note that we do not screen the X-linked genes for unaffected men.

The price for a couple who have samples collected and are tested at the same time includes a 12.5%* discount.

Payment is required prior to sample processing. Please refer to the request form for payment details.

There is no Medicare rebate for carrier screening. If there is a family history of a known mutation, the Beacon expanded carrier screen may not be the most appropriate test and a rebate may be available for the appropriate genetic test (please check with the laboratory).

If a test is cancelled prior to processing (typically within 24 hours), a full refund may be possible. If a test is cancelled after sample processing has been initiated, but before a report is generated, there will be an administration charge per patient, with the balance being refunded.

*Correct at time of printing.

Sample collection and processing

Samples may be collected at any Sonic Healthcare 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.

We recommend that the patient or another adult check labelling of request forms and sample tubes, particularly if a couple is having samples collected at the same time. All samples are also tagged with molecular barcodes upon receipt in the Fulgent laboratory prior to analysis, enabling them to ensure the quality of sample processing. Sonic Genetics will extract DNA from the sample and then transport the sample to the Fulgent Genetics accredited laboratory in the US.

Turnaround time

Once the sample is received at the genetics laboratory at Douglass Hanly Moir Pathology (DHM), the results will typically be sent to your doctor within three to four weeks.

Post-test review

Reports

Each person has millions of variations in their DNA sequence when compared to a reference sequence. It can be challenging to determine if a given variant in a gene is serious enough to potentially cause a recessive disorder, or is a mild or benign variant of no clinical or reproductive consequence. Fulgent Genetics utilises their own experience of many thousands of Beacon tests, together with the pooled experience in international databases, to categorise each variant according to the internationally recognised ACMG system. The Beacon report will only include variants which are classified as 'pathogenic' or 'likely pathogenic' using this standard. Variants which are categorised as 'benign', 'likely benign', or 'of uncertain clinical significance' will not be reported.

The Beacon report details the mutations that have been identified in the genes analysed. The report will also identify the recessive disorder for which the patient is a carrier. If two people have been tested as a couple, the Beacon report will provide an integrated report which sets to one side carrier information that is not of direct relevance for reproductive planning for the couple. In this way, the couple report is focused on the material issues to be considered prior to a pregnancy.

Sonic Genetics has medical specialists in genetic pathology and clinical genetics, who are available to address your enquiries about these reports.

Genetic counselling

Sonic Genetics provides free post-test genetic counselling in specific situations:

- For a woman who has been identified by the Beacon expanded carrier screen as being a carrier of an X-linked recessive disorder
- For a couple who has been identified as being at 25% risk of having a child with an autosomal recessive disorder; at least one of the partners must have had the Beacon expanded carrier screen.

Genetic counselling is provided by telephone by independent genetic counsellors on the basis of a referral from the doctor responsible for the care of the patient or couple. The genetic counsellors are located in Australia and accredited by the Human Genetics Society of Australasia.

Referrals for free counselling need to be received by Sonic Genetics within two months of the latest Beacon report being issued.

Free counselling is limited to one episode per couple. A couple or individual is free to make their own subsequent arrangements with the genetic counsellor.

The free genetic counselling addresses informational and psychological issues arising from the Beacon report; the counsellor will not be responsible for the subsequent reproductive management.

Privacy

Patient information, samples and reports that are handled within Australia will be managed in accordance with the relevant legislation and professional stands.

Identifying details about each patient, together with pathology samples, will be sent overseas to the Fulgent Genetics laboratory in the US. Each patient will be required to acknowledge this on the dedicated Expanded Carrier Screening Request Form. The Fulgent laboratory is accredited by regulatory and professional bodies in the US and complies with US legal requirements for handling health-related information. All data analyses are performed on private server centres.

Why you should choose Sonic Genetics

We are Australia's largest private genetics referral laboratory. Our genetic pathologists and medical scientists work in NATA-accredited laboratories throughout Australia to provide quality testing and clinical support.

We are part of Sonic Healthcare, an acknowledged world leader, and Australia's largest pathology provider.

We are supported by Sonic's extensive network of state and regional laboratories, and benefit from the interdisciplinary collaboration between a large cohort of pathologists and scientists working across all specialities. Many are recognised nationally and internationally; they have established academic reputations and are actively involved in professional and regulatory oversight in Australia and overseas.

Conditions screened

This is a list of the autosomal and X-linked recessive disorders included in the Beacon expanded carrier screen. Please be aware that one gene may be responsible for more than one clinical condition, and one clinical condition may be caused by more than one gene.

This information is correct at the time of printing. Please contact us or refer to the Sonic Genetics website for an up-to-date listing.

- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency
- 3-ketothiolase deficiency
- 3-Methylcrotonyl-CoA carboxylase deficiency
- Abetalipoproteinemia
- Achondrogenesis, type IB
- Achromatopsia
- Acrodermatitis enteropathica
- Acyl-CoA dehydrogenase-9 (ACAD9) deficiency
- Adenosine deaminase deficiency
- Adrenoleukodystrophy, X-linked
- Aicardi-Goutieres syndrome
- Alkaptonuria
- Alpers-Huttenlocher syndrome
- Alpha thalassaemia
- Alpha thalassaemia, X-linked
- Alpha-Mannosidosis
- Alport syndrome, can be X-linked
- Alstrom syndrome
- Anauxetic dysplasia
- Andermann syndrome
- Arginase deficiency
- Argininosuccinate lyase deficiency
- Aromatase deficiency
- Arthrogryposis, mental retardation & seizures
- Arts syndrome, X-linked
- Asparagine synthetase deficiency
- Aspartylglucosaminuria
- Ataxia neuropathy spectrum
- Ataxia with isolated vitamin E deficiency
- Ataxia-telangiectasia
- Atelosteogenesis II
- Autoimmune polyendocrinopathy syndrome
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay
- Bardet-Biedl syndrome
- Bare lymphocyte syndrome, type II
- Bartter syndrome
- Bernard-Soulier syndrome
- Beta thalassaemia
- Bilateral frontoparietal polymicrogyria
- Biotinidase deficiency
- Björnstad syndrome
- Bloom syndrome
- Canavan disorder
- Carbamoylphosphate synthetase I deficiency
- Carnitine palmitoyltransferase
- Carnitine-acylcarnitine translocase deficiency

- Carpenter syndrome
- Cartilage-hair hypoplasia
- Cerebrotendinous xanthomatosis
- Charcot-Marie-Tooth disorder, can be X-linked
- Chediak-Higashi syndrome
- Chondrodysplasia punctata type 1, X-linked
- Choreoacanthocytosis
- Choroideremia, X-linked
- Chronic granulomatous disorder, can be X-linked
- Citrin deficiency
- Citrullinemia
- COACH syndrome
- Cockayne syndrome
- Cohen syndrome
- Combined malonic & methylmalonic aciduria
- Combined oxidative phosphorylation deficiency
- Combined pituitary hormone deficiency
- Congenital adrenal hyperplasia, X-linked
- Congenital amegakaryocytic thrombocytopenia
- Congenital disorder of glycosylation
- Congenital hyperinsulinism
- Congenital ichthyosis
- Congenital insensitivity to pain with anhidrosis
- Congenital myasthenic syndrome
- Congenital nephrotic syndrome
- Congenital secretory chloride diarrhoea
- Corneal endothelial dystrophy
- Corticosterone methyloxidase deficiency
- Costeff syndrome
- Creatine deficiency syndrome, X-linked
- Crigler-Najjar syndrome
- Cystic fibrosis
- Cystinosis
- D-bifunctional protein deficiency
- De Sanctis-Cacchione syndrome
- Dent disorder 2, X-linked
- Diastrophic dysplasia
- Dihydrolipoamide dehydrogenase deficiency
- Dihydropyrimidine dehydrogenase deficiency
- Duchenne muscular dystrophy, X-linked
- Dyskeratosis congenita type 5
- Dystrophic epidermolysis bullosa
- Ehlers-Danlos syndrome, dermatosparaxis type VIIC
- Ellis-van Creveld syndrome
- Emery-Dreifuss muscular dystrophy, X-linked
- Enhanced S-cone syndrome
- Ethylmalonic encephalopathy
- Fabry disorder, X-linked
- Factor XI deficiency
- Familial dysautonomia
- Familial hyperinsulinism, ABCC8-related
- Familial Mediterranean fever
- Fanconi anaemia
- Fetal akinesia deformation sequence
- Fragile X syndrome, X-linked
- Fukuyama congenital muscular dystrophy
- Fumarase deficiency
- Galactokinase deficiency
- Galactosaemia
- Gaucher disorder
- Gitelman syndrome
- Glutaric aciduria
- Glycine encephalopathy
- Glycogen storage disorder
- GM1-gangliosidosis
- GRACILE syndrome
- Guanidinoacetate methyltransferase deficiency
- Gyrate atrophy of choroid and retina
- Haemochromatosis (type 2A & 3)
- Haemophilia A & B, X-linked
- Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related
- Hereditary fructose intolerance
- Hermansky-Pudlak syndrome
- Holocarboxylase synthetase deficiency
- Homocystinuria
- Hunter syndrome, X-linked
- Hurler syndrome
- Hydrolethalus syndrome
- Hyper IgM syndrome, X-linked
- Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)
- Hypohidrotic ectodermal dysplasia, X-linked
- Hypophosphatasia
- Inclusion body myopathy type 2 (Nonaka myopathy)
- Infantile neuroaxonal dystrophy
- Isovaleric acidaemia
- Joubert syndrome
- Junctional epidermolysis bullosa
- Juvenile retinoschisis, X-linked
- Krabbe disorder
- L1 syndrome, X-linked
- Laryngo-onycho-cutaneous syndrome
- Leber congenital amaurosis
- Leigh syndrome
- Lethal congenital contracture syndrome 1
- Leukoencephalopathy with vanishing white matter
- Limb-girdle muscular dystrophy
- Lipoid congenital adrenal hyperplasia
- Lissencephaly, X-linked
- Liver failure, acute infantile
- Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
- Lowe syndrome, X-linked
- Lysinuric protein intolerance

- Lysosomal acid lipase deficiency
- Maple syrup urine disorder
- Maroteaux-Lamy syndrome
- Meckel syndrome
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- Megalencephalic leukoencephalopathy with subcortical cysts
- Menkes disorder, X-linked
- Metachromatic leukodystrophy
- Metaphyseal dysplasia without hypotrichosis
- Methylmalonic aciduria and homocystinuria, type cblC
- Methylmalonic aciduria, others
- Microphthalmia with or without coloboma
- Mitochondrial complex deficiency
- Mitochondrial myopathy and sideroblastic anaemia 1
- Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disorder
- Morquio syndrome A
- Mucolipidosis
- Mucopolysaccharidosis, can be X-linked
- Multiple epiphyseal dysplasia
- Multiple pterygium syndrome
- Multiple sulfatase deficiency
- Muscular dystrophy, LAMA2-related
- Muscular dystrophy-dystroglycanopathy
- Myocerebrohepatopathy syndrome
- Myotubular myopathy, X-linked
- N-acetylglutamate synthase deficiency
- Nemaline myopathy
- Nephrogenic diabetes insipidus
- Nephronophthisis
- Neuronal ceroid lipofuscinosis
- Niemann-Pick disorder
- Nijmegen breakage syndrome
- Non-syndromic hearing loss, can be X-linked
- Odontoonychodermal dysplasia
- Omenn syndrome
- Ornithine transcarbamylase deficiency, X-linked
- Osteopetrosis, TCIRG1-related
- Pendred syndrome
- Permanent neonatal diabetes mellitus
- Peroxisomal acyl-CoA oxidase deficiency
- Phenylalanine hydroxylase deficiency (phenylketonuria)
- Phosphoglycerate dehydrogenase deficiency
- Phosphoribosylpyrophosphate synthetase superactivity, X-linked
- Polycystic kidney disorder, PKHD1-related
- Pompe disorder
- Pontocerebellar hypoplasia
- Postnatal progressive microcephaly with seizures and brain atrophy
- Primary ciliary dyskinesia
- Primary congenital glaucoma
- Primary hyperoxaluria
- Progressive external ophthalmoplegia
- Progressive familial intrahepatic cholestasis
- Propionic acidaemia
- Pycnodysostosis
- Pyruvate carboxylase deficiency
- Pyruvate dehydrogenase deficiency, can be X-linked
- Renal tubular acidosis with deafness
- Retinitis pigmentosa
- Rhizomelic chondrodysplasia punctata
- Roberts syndrome
- Rosenberg-Chutorian syndrome, X-linked
- Sandhoff disorder
- Sanfilippo syndrome
- Schimke immunoosseous dysplasia
- Schopf-Schulz-Passarge syndrome
- Segawa syndrome
- Senior-Løken syndrome
- Severe combined immunodeficiency, can be X-linked
- Severe congenital neutropenia, can be X-linked
- Short-chain acyl-coA dehydrogenase (SCAD) deficiency
- Sialic acid storage disorder
- Sickle cell disorder
- Sjögren-Larsson syndrome
- Smith-Lemli-Opitz syndrome
- Spastic paraplegia
- Spinal muscular atrophy (SMA)
- Spondylocostal dysostosis
- Steel syndrome
- Stuve-Wiedemann syndrome
- Systemic primary carnitine deficiency
- Tay-Sachs disorder
- Tetrahydrobiopterin deficiency
- Thrombocytopenia, X-linked
- Trichohepatoenteric syndrome
- Trifunctional protein deficiency
- Tyrosinaemia
- Usher syndrome
- Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
- Weyers acrodental dysostosis
- Weyers acrofacial dysostosis
- Wilson disorder
- Wiskott-Aldrich syndrome, X-linked
- Wolcott-Rallison syndrome
- Xeroderma pigmentosum
- Zellweger syndrome

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For further information please refer to our website, www.sonicgenetics.com.au or call us on 1800 010 447

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Printed August 2018

DHMSG-ECS-Dr-V1-2018-D01224