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Sonic Genetics

Information for Doctors

Expanded carrier screening

Reproductive carrier screening involves genetic testing of a couple to determine whether they are at relatively high risk of having a child with a serious recessive disorder. Screening tests may be limited to a few selected disorders, for example, cystic fibrosis (CF), spinal muscular atrophy (SMA), and fragile X syndrome (FXS). Carrier screening can also be expanded to cover hundreds of different disorders. Sonic Genetics, in partnership with Fulgent Genetics in the US, provides the Beacon expanded carrier screen for more than 400 disorders.

Advantages of the Beacon expanded carrier screen:

1. Covers serious childhood-onset conditions for which treatment is limited.
2. Identifies one in 20 couples as being at risk of having a child with such a disorder, the best performance of nine tests commercially available in Australia.*
3. Provides a succinct, yet comprehensive, report that details risks for the couple.
4. Prenatal testing is available through Sonic Genetics for all genes covered by the Beacon expanded carrier screen.
5. Sonic Genetics provides free genetic counselling support to all couples identified by the Beacon expanded carrier screen as being at risk of having a child with a serious genetic disorder.
6. Sonic Genetics has several experienced genetic pathologists in Australia who support the requesting doctors.
7. Sonic Genetics is part of Sonic Healthcare, which has provided exemplary medical testing in Australia for more than 25 years. Fulgent Genetics is an accredited laboratory in the US, with a long history of providing genetic testing to Australians.

1. What disorders can be detected by the Beacon expanded carrier screen?

The Beacon expanded carrier screen is limited to recessive disorders that are serious, occur in childhood, for which there are limited treatments available, and in which carriers are unlikely to develop features of the disorder.
Other expanded carrier screens may include conditions that are mild (e.g. Factor V Leiden), readily treated (e.g. haemochromatosis), adult-onset (e.g. familial breast cancer), or that may subsequently occur in the parent (e.g. hypercholesterolaemia).

2. How useful is the Beacon expanded carrier screen in identifying at-risk couples?

The Beacon expanded carrier screen is predicted to have the best performance of expanded carrier screens available in Australia. It identifies one in 20 (5%) couples as being at 25% risk of having an affected child. Approximately one in 160 couples (0.6%) are at risk of having a child with CF, SMA or FXS ('three-gene' screen in Figure 1). Using this as a baseline, we examined the performance of nine different expanded carrier screens available in Australia. Each tested more than 100 different genes; we excluded genes that do not cause serious childhood-onset disease. By analysing the number of genes, carrier frequency, detection rate and inheritance, we could estimate the proportion of couples who would be identified as being at risk of having an affected child. The Beacon expanded carrier screen was clearly the best.

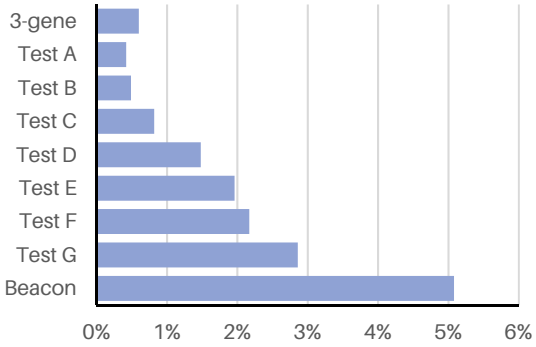

Figure 1. Percentage of couples identified as being high risk

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3. How are the Beacon expanded carrier screen results presented?

The report summarises the risks for the couple (if tested together), identifying the disorders for which they are at high risk of having an affected child. The Beacon expanded carrier screen also provides a calculator for estimating the residual risk for each condition for which the couples do not appear to be at risk, that is, the false-negative rate of the test.

4. What follow-up testing is provided to couples?

Sonic Genetics provides prenatal testing through Fulgent Genetics for any couple identified by the Beacon expanded carrier screen as being at high risk of having an affected child. For a rare condition, there may be few accredited laboratories that provide such a prenatal test, and some providers of carrier screening do not offer prenatal testing for the conditions they cover. Sonic Genetics will provide such testing for any at-risk couple identified through the Beacon expanded carrier screen.

The cost of the prenatal test for rare mutations detected by the Beacon screen is $595.* Microarray can be included with the prenatal testing at no additional charge. This will screen the pregnancy for additional chromosomal abnormalities that may be present. FISH testing can also be included in the prenatal screen for an additional $100.*

5. How is patient confidentiality maintained?

Sonic Genetics and our partner, Fulgent Genetics in the US, do not release or sell carrier screening results to third parties. The results of carrier screening are potentially useful to drug companies who gather information about the frequency of genetic variants in populations. The policy of some providers regarding the sale of this information to third parties is not clear.

6. What support does Sonic Genetics provide to couples being tested?

Sonic Genetics provides detailed literature for patients, which can be used in pre-test counselling. We also provide free genetic counselling with qualified Australian genetic counsellors for couples identified as being at high risk of having an affected child. Some providers offer no support for couples being tested, while others offer genetic counselling by counsellors located overseas.

7. What support is provided for doctors ordering the test?

Sonic Genetics has several genetic pathologists in Australia who provide information and advice for doctors ordering the Beacon expanded carrier screen. Most providers of expanded carrier screening have no local expertise to assist doctors ordering the test.

8. Does the testing laboratory have a sustainable program of testing?

Sonic Genetics is part of Sonic Healthcare, a profitable Australian company with a track record of providing exemplary medical testing for more than 25 years. Fulgent Genetics is a younger company, and is profitable and growing.

Some carrier screening companies are relatively new to the market and attract business by providing very low prices. While this may be appealing, it comes at the corporate cost of high debt and negative cash flow. The financial viability of such companies in the immediate-to-long-term is uncertain.

9. How much does the Beacon expanded carrier screen cost?

We provide the Beacon expanded carrier screen at a price of $595* per person.

If you would like further information about this test, please refer to sonicgenetics.com.au/rcs/beacon or contact us on info@sonicgenetics.com.au or 1800 010 447.

*Correct at time of printing

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