INCIDENTAL FINDINGS

When a genomic (DNA) test finds something unexpected

Key points

- Genomic (DNA) testing is performed to work out whether a particular health condition is caused by a change in a person's genome
- Occasionally, a change in a person's DNA (genetic variant) is unexpectedly found that causes a different condition
- Incidental findings may affect you or a family member's current or future health
- Incidental findings may also be referred to as 'unexpected' or 'additional' findings

Why is genomic testing done?

Genomic (DNA) tests are performed to work out whether a particular health condition is caused by a change in a person's **genome**. Our genome is found in every cell in our body and is like an instruction manual for our bodies. The 'sentences' in this instruction manual are called **genes**, and each gives a different instruction to the body. Genes work together to tell our bodies how to grow, develop and function.

Changes in our genes are known as **variants**. Sometimes having a variant in a gene can affect our health, learning or physical development. Health conditions that are caused by variants in genes are called **genetic conditions**.

Genomic testing may find the variant(s) responsible for a health condition. However, in many cases a genetic cause cannot be found to explain an individual's condition or symptoms. This is called an uninformative, inconclusive or negative result. Occasionally, the testing laboratory will find a completely unexpected result, known as an *incidental finding*.

What is an incidental finding?

Genomic testing examines many thousands of genes. Occasionally, a variant is unexpectedly found in a gene that relates to a different condition than what the test was intended for. This is called an incidental finding. These are generally seen in less than 1% (1 in 100) of genomic tests done in Australia.

Incidental findings have always been a feature of medicine. For example, sometimes a chest X-ray that was done to investigate a cough, unexpectedly finds an enlarged heart. This incidental finding (the enlarged heart) will then be investigated further.

If an incidental finding on a genomic test is thought to be relevant to your current or future health, the laboratory that did the test will report it to your doctor. An example would be a variant in a gene that makes a person more likely to develop certain types of cancers. Your doctor will explain what the incidental finding is and what it may mean for you and/or other family members.

Occasionally, genomic testing may reveal unexpected family relationships. For example,a genomic test may reveal that a couple are closely related or that either the mother or father is not the biological parent of a child. If this is a concern to you or has happened, please discuss it with your doctor or genetic counsellor.

What happens now?

Your doctor or genetic counsellor will explain what the finding means for you, whether it may affect other family members and what is known about the condition. Often, if a person is identified as having an increased chance of developing a condition, they can be screened, tested and/or treated for the condition. Even if genomic testing shows that a person has an increased chance of developing a condition, it is not usually possible to predict exactly if and when this will happen, or the severity.

Content updated October 2021
OCT21/V1 NS12681
SHPN: (HETI) 240972

Incidental findings are unexpected and can cause worry or confusion. If this result has brought up concerns about the future, talking it over with family members, a close friend, your doctor, a counsellor or psychologist, or a genetic counsellor may help.

If you have any questions about the result or additional testing, you can ask the health professionals who gave you the genomic test result. Your doctor will continue to care for you or your family member and may refer you to other specialists for an appointment.

The genetic health professionals at a clinical genetics service can provide further information, advice and support, including **genetic counselling**. Details of your local clinical genetics service can be found at the **NSW Centre for Genetic Education**.

Certain incidental findings mean that the condition will develop. Other incidental findings simply indicate an increased chance of developing the condition.

Knowing about an increased chance means that you may wish to be screened and/or treated for the condition, or keep this in mind when planning a family. Others in the family may also benefit from knowing about the finding.

Will an incidental finding affect my personal insurances?

When companies are deciding whether to offer personal insurance such as life or disability insurance, they assess your health, including risk factors such as age and smoking history. Typically, information is also collected on health conditions seen in other family members.

Some genomic test results may affect your ability to obtain new personal insurance policies, and you may need to tell the insurance company the results of your genomic test. More information about the impact of genomic test results on insurance can be found at the **NSW Centre for Genetic Education**.

In Australia, the cost and availability of private health insurance is not affected by your personal or family health or genomic test results.

More information and support

- Human Genetics Society of Australasia (HGSA) Position Statement: **Genetic Testing and Personal Insurance Products in Australia**
- NSW Centre for Genetic Education: **Life insurance fact sheet**
- Australia's Financial Services Council (FSC): **Genetic tests and applying for life insurance – Key facts**
- Genetic Alliance Australia
- Rare Voices Australia

This fact sheet should not replace a consultation with a specialist healthcare professional.