SCN2A INFORMATION SHEET #7

Genetic Testing

What is genetic testing and why is it done? [1-5]

- **Genetic testing** is a special kind of medical test that looks at a person's **genes** (see Information Sheet #3 'Genetics 101' for more information about genes and genetics).
- Genes carry the body's genetic code – the information that tells the body how to grow, develop and function. Each gene tells the body to produce a specific protein.
- A change in one or more genes can cause a **genetic condition.**
- **Genetic testing** is done to find out whether an individual's health condition could be caused by a change in their genetic code.
- Knowing the genetic cause of a condition may have many benefits for the individual and their family, such as:
  - Avoiding further diagnostic testing
  - Helping families understand the condition, access appropriate treatment and plan for the future
  - Helping with planning future pregnancies
  - Providing information about whether other family members may develop the same condition.

In short...

- **Genetic testing** is carried out to determine whether an individual's health condition could be caused by a change in their **genetic code.**
- This type of testing may be carried out in individuals with epilepsy, developmental delay, intellectual disability, and/or autism spectrum disorder (ASD).
- A type of genetic test called **genomic testing** can detect changes in the SCN2A gene.

What sort of genetic tests are available in Australia? [1,6,7]

CHROMOSOME MICROARRAY

- Used to detect small segments of missing or extra genetic code
- Usually the first test carried out in individuals with developmental delay, intellectual disability and/or autism spectrum disorder

GENE PANELS

- Examine more than one gene
- May be used when an individual has specific symptoms that my be caused by one of many different genes
- For example, epilepsy

SINGLE GENE TESTS

- Look at a single gene
- Usually carried out when a particular condition is suspected, or if other family members have a particular health condition
- The test may look for a specific genetic change or any change in a single gene
- For example, Fragile X Syndrome

GENOMIC TESTING

- Two types available in Australia:
  - Exome sequencing
  - Whole genome sequencing
- Both read or 'sequence' every gene

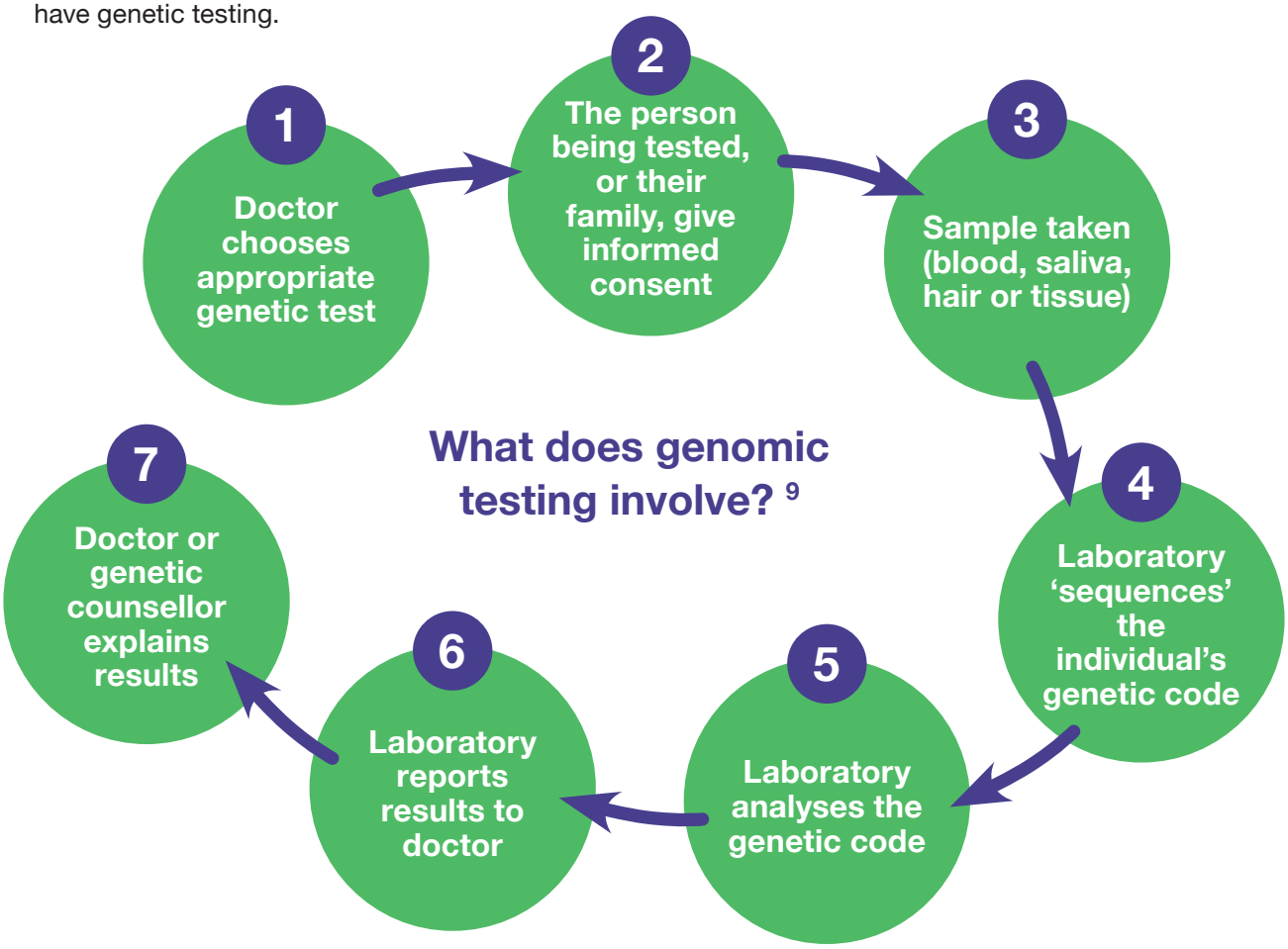
How to access genetic testing in Australia

- The first step for individuals wishing to have genetic testing is to discuss the testing with a doctor – either a general practitioner (GP) or a specialist doctor.
- GPs can order several types of genetic tests, including chromosome microarray and Fragile X testing
- Other specialist doctors can order genetic testing in certain clinical situations, for example paediatricians.
- Doctors who specialise in caring for patients with or at risk of a genetic condition are called **clinical geneticists**. A referral is needed from a GP or other doctor to see a clinical geneticist.
- Genetic testing can be expensive. Many tests are covered by Medicare or the hospital ordering the test. Sometimes the family will have to pay for some or all of the cost of the test.

Genetic testing for individuals with developmental delay, intellectual disability and/or autism spectrum disorder[1,5,8]

- Changes in many different genes have been found to cause developmental delay, intellectual disability and/or autism spectrum disorder (ASD). There is not always a genetic cause, but genetic testing may identify one.
- Chromosome microarray and Fragile X testing are usually the first genetic tests carried out. If these are negative, more detailed testing such as exome sequencing may be ordered.
- Studies suggest that genomic testing can find a genetic cause in more than a quarter (25%) of individuals with developmental delay, intellectual disability, speech delay and/or ASD.
- Experts now recommend that individuals with developmental delay or intellectual disability, undergo genetic testing. People with learning difficulties and ASD and / or epilepsy are also recommended to have genetic testing.

What does genomic testing involve? [9]



What are the possible outcomes of genetic testing? [10]

- Genetic testing does not always provide an answer.
- Several outcomes are possible:
    - **A genetic cause is identified for the condition.**
      The name of a specific gene and the change in that gene will be given.
    - **A genetic cause is not identified.**
      This does not mean the condition is not genetic. It just means the test did not find a change in any genes that are currently known to cause the condition. This is also known as an uninformative result.
    - **Unclear/uncertain result.**
      Sometimes a possible genetic cause is found but there is not enough evidence available to be certain. These results are known as variants of uncertain significance.
    - **Incidental finding.**
      Rarely, a change is identified in a gene that is relevant to another health condition.

Depending on the type of genetic test, the results may have wider implications for the individual being tested, and their immediate and extended family. It is important to understand these implications before having a genetic or genomic test. More information is available at the websites below.

More information

- MedlinePlus
    - What is a gene?
    - What is genetic testing?
    - What are whole exome sequencing and whole genome sequencing?
- Centre for Genetics Education
    - About genomic testing
    - Genetic and genomic testing
    - Genetic and genomic test results
- Melbourne Genomics: A genomic test step-by-step
- NSW Health: Genomic testing information (fact sheet and video)

References

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