Media backgrounder

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About genomics

Genomics has huge potential to improve healthcare. Looking at a patient's genome may help doctors diagnose illness more quickly and easily, as well as working out what treatment/management might best help.

Genomics is a rapidly advancing field worldwide, which examines an individual's complete set of genetic information (the genome) to identify changes (variants) that may impact on health. Genomics offers enormous potential to improve diagnosis and provide more personalised treatment/management of medical conditions.

The genome is your complete set of DNA, including all of your genes. Each gene has a specific function. When a gene contains a variation or change, it may not work properly and this may affect your health.

A genetic test looks at one or a small number of genes at a time. A genomic test can capture information from a large number of genes at the same time to see whether changes are present.

How are gene changes ('variants') identified?

Computer programs are used to analyse the large amounts of raw genomic data generated by sequencing of a person's genome. The computer scans this data for differences or 'variants' between the genetic sequence of the person being tested and a reference sequence.

This list of variants is then examined to see if any can explain the medical condition someone might have and/or the most appropriate treatments.

The first step is to establish whether a variant or change in a gene will stop it working properly. To do this, a medical scientist examines large databases or scientific papers to identify what is known about the relevant variants.

Then a team of experts, including medical laboratory scientists, bioinformaticians (statistical specialists who deal with biological data), doctors and geneticists, match the list of variants which may impair gene function against a person's health and family medical history. This is how they determine if a particular variant is likely to be the underlying cause of a genetic disease. Or, in other cases, whether a variant might affect how a person might respond to particular treatments.

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