Communication aid

Variants of uncertain significance (VUS)

This communication aid has been produced for clinicians to help support and guide conversations about variant classification and variants of uncertain significance with their patients.

What is a variant?

Variants are differences in DNA (gene changes) that make us unique

Most gene changes are harmless (**benign**), some cause genetic conditions (**pathogenic**).

Classifying variants

To find out if a variant causes a genetic condition, scientists gather evidence from lab tests, computer analysis, scientific research, population databases and a person's health and family history.

Using this evidence, the variant is classified based on how likely it is to cause a condition.

Only variants classified as "pathogenic" or "likely pathogenic" (disease-causing) are used for treatment decisions.

Correct classification is important, as getting it wrong can lead to incorrect treatment decisions for patients and their families.

Key terms

Variant: Changes in a gene or chromosome used to be referred to as 'mutations'. Now, they are more commonly called changes, alterations or variants.

Benign variant: A harmless gene change that doesn't cause any health problems.

Pathogenic variant: A gene change that can cause a health condition.

What is a variant of uncertain significance, or VUS?

A variant of uncertain significance (VUS) is a gene change that does not have enough evidence to know if it causes a condition.

A VUS result cannot be used by itself to make decisions about managing or treating a condition. Genomic testing for a VUS is not offered to relatives for management of their health.

A VUS may be reclassified at a later date as more information becomes available.

If a VUS is close to being considered likely pathogenic, it may be reviewed by a team of specialists. More tests or investigations may be ordered to gather more information.

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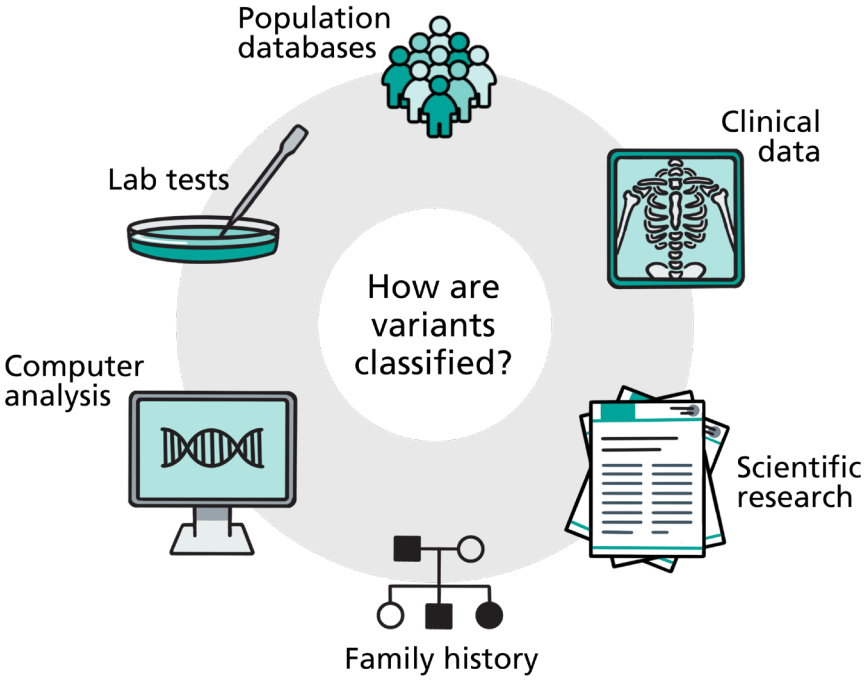

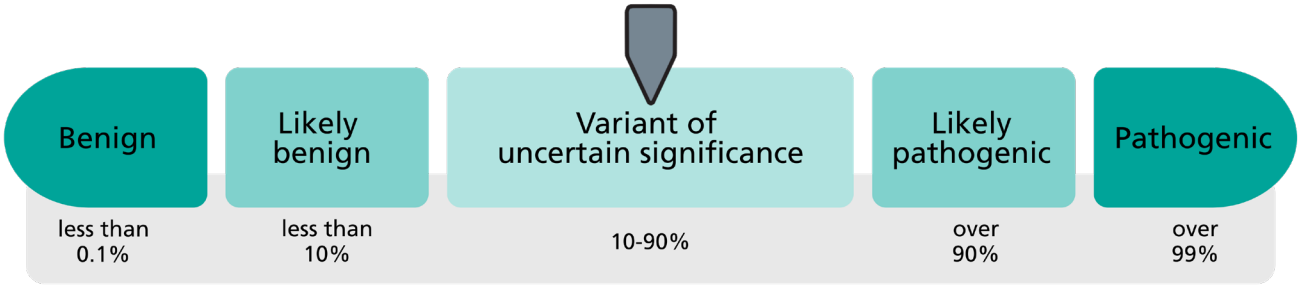

Likelihood a gene change is the cause of a condition

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