

Monogenic Conditions



vs.



Complex Conditions

Some health conditions are caused by single genetic variants with a big impact. These are called monogenic conditions. Other health conditions are more complex and are influenced by many genetic variants across the genome, plus lifestyle, environment, and more. This handout describes some of the differences between these types of conditions.

Genetic

Genetics plays a primary role

The cause



Combination

Genetics, lifestyle, and environment all play important roles

One gene

A single genetic variant has a very large impact and can cause a condition or trait entirely on its own

Underlying genetics



Many genes

The additive effect of hundreds to thousands of genetic variants across many different genes, each with a small impact

Predictable

Follow a predictable, simple inheritance pattern, such as passing directly from a parent to child

Inheritance Pattern



Unpredictable

Often run in families but do not follow a single, predictable pattern

Rare

Each condition is rare, but together they affect many people, especially in certain ancestry groups

How common



Common

Most common health conditions and many human traits fall in this category

Clear answer

Genetic testing often gives a clear "yes/no" answer about the presence of a specific genetic variant

Genetic testing



Probability

Risk is estimated using a polygenic risk score (PRS), which combines the effects of many genetic variants

Sickle cell anemia, cystic fibrosis, familial hypercholesterolemia, BRCA1/2-related cancers

Example conditions



Type 2 diabetes, depression, most heart diseases, most cancers