Communication aid

Penetrance and expressivity

This communication aid has been produced for clinicians to help support and guide conversations about penetrance and expressivity with their patients.

In genomics, the terms 'penetrance' and 'expressivity' explain how a **genetic change** might affect people differently.

Penetrance: the proportion of people with a genetic change that show signs or symptoms of a condition.

- If a condition has **complete penetrance**, this means everyone with the genetic change will develop the condition.
- If it has **reduced (incomplete) penetrance**, this means some people with the genetic change develop the condition and some do not.

Expressivity: how a condition affects different people.

- Many genetic conditions show different symptoms or severity in different people. This is called **variable expressivity**.

Key terms

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

In conditions with reduced penetrance and variable expressivity:

- Not everyone with the genetic change will be affected.
- Among those who are affected, symptoms can look different from one person to the next.

Remember, conditions may show variable penetrance or expressivity even between people in the same family with the same genetic change.

Want to learn more?

Scan to read or download a guide from Unique on reduced penetrance and variable expressivity.

View the full collection of communication aids on GeNotes

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Complete penetrance

Everyone who carries the genetic change is affected.

Complete penetrance and variable expressivity

Everyone who carries the genetic change is affected, but they may show different signs or symptoms.

Reduced penetrance

Some people who carry the genetic change are affected and some are not.

Reduced penetrance and variable expressivity

Some people who carry the genetic change are affected, some are not. People who are affected may show different signs or symptoms.

Please scan to leave feedback about this resource

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