Facts about rare, genetic and undiagnosed conditions

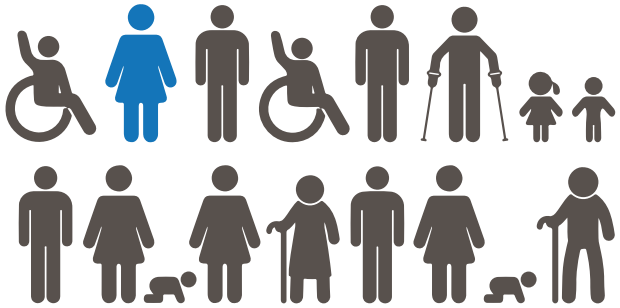
Rare conditions

Individually, rare conditions are rare but together, there are around 3.5 million people living with a rare condition in the UK.

3.5 million
people in the UK

436,000 in Scotland,
180,000 people in Wales
110,000 people in Northern Ireland

1 in 17
people are affected by a rare condition at some point in their lifetime



A rare condition affects fewer than

1 in 2000

There are over

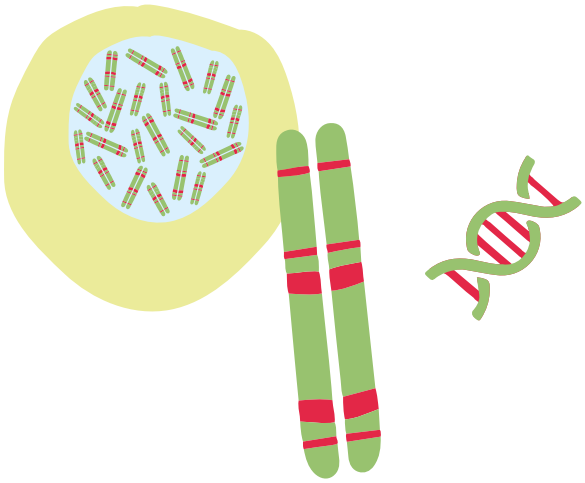
7000
rare conditions, with new conditions regularly identified through scientific progress

7 in 10
rare conditions affect children. And more than

3 in 10
children with a rare condition die before their fifth birthday

GENETIC ALLIANCE UK

Genetic Conditions



A genetic condition is caused by a change in an individual's genome.

The most common genetic conditions such as sickle cell disease affect around 17,500 people in the UK and cystic fibrosis affects 10,800 people in the UK. The rarest genetic conditions may affect just one family in the UK, and a handful of people across the globe.

8 out of 10

rare conditions are caused by a change to someone's genetic code

Undiagnosed Conditions

An undiagnosed genetic condition is known as a 'syndrome without a name' or SWAN

Each year around

6000

children in the UK are born with a genetic condition so rare that it does not yet have a name.

This might be because the right test has not been developed to diagnose it, or the genetic cause of the condition has not yet been discovered.

People living with genetic and rare conditions and their families face a lifetime of complex care. They need vital support from the NHS, social care and education services to live their lives to the full.

See our 2024 Rare Disease Day Report: '**Stats behind the stories**' for more.