

Genetic Disorders



What are genetic disorders?

'Our bodies are formed from DNA . A genetic disorder is a disease that is caused by a change, or mutation, in an individual's DNA sequence. These mutations can be due to an error in DNA replication or due to environmental factors, such as cigarette smoke and exposure to radiation, which cause changes in the DNA sequence. The human genome is a complex set of instructions, like a recipe book, directing our growth and development. However, unlike a printed book, the human genome can change. Our DNA provides the code for making proteins, the molecules that perform most of the functions in our body. However, when a section of our DNA is changed in some way, the protein it codes for is also affected and may no longer be able to carry out its normal function. Depending on where these mutations occur, they can have little or no effect, or may profoundly alter the biology of cells in our body, resulting in a genetic disorder.'

Taken from www.yourgenome.org

How might I know if my child has a genetic disorder?

Some genetic disorders are identified before or shortly after birth. They can be identified as a result of testing where there is a family history, whether a mother's age means she is more likely to carry a baby with genetic differences or where there has serious health issues. Some disorders are identified as the child grows and triggers that may lead to a diagnosis could include developmental delays / missed milestones, emerging health issues and physical differences or where there is a new diagnosis in a family. Whilst identifying a genetic disorder can help with understanding, possible medical treatment and a better understanding of a child's needs, for some a diagnosis does not provide further information. Some families find it helpful as it enables them to find tailored support for their child and them.

How can I find out if my child has a genetic disorder?

Most children where a genetic disorder may be part of a diagnosis will be offered genetic testing by the doctors working with them. Not all parents take up this offer. If you have concerns about your child, and there is not ongoing medical support, then you should share your worries with your GP.

Where can I find out more?

The NHS has some introductory information: <https://www.nhs.uk/conditions/genetic-and-genomic-testing/>

Once a diagnosis is confirmed there are specialist groups that support specific diagnosis as well as organisations that support all genetic disorders.

Where to find out more on website:

www.pinpoint-cambs.org.uk and look under Useful Information