What is genetic testing?

Genetic testing identifies changes in the DNA. Some changes have no effect on health and development, while others can cause genetic diseases with moderate to severe effect on our health.

What are genetic diseases?

Genetic diseases are caused by a variant (a version that is different from the standard) in a gene. Variants are also known as mutations. Depending on the gene involved, diseases are inherited in different patterns:

- Autosomal dominant diseases: The mutation is present in only one gene copy and is sufficient to cause the disease to be expressed in the person with the mutation. Examples: Huntington's disease, Neurofibromatosis
- Autosomal recessive diseases: The mutation is present in only one gene copy but it is not 'powerful' enough to cause the disease to be expressed in the individual with the mutation, who is known as a 'carrier'. Carriers are healthy, but two carriers of the same mutation could have a child who is affected if they both pass on their mutated gene copy. Examples: Cystic fibrosis, Beta-thalassemia
- X-linked diseases: The mutation is present on the X chromosome, one of the two chromosomes that define gender. Females have two X chromosomes, so when they inherit a mutation in one of their X chromosomes, they are carriers with or without exhibiting symptoms (see question 7). Males have only one X chromosome, so when they inherit a mutation in their X chromosome, they are affected. Examples: Duchenne Muscular Dystrophy, Fragile X

How are diseases inherited?

A healthy person has two pairs of 23 chromosomes in all the cells of their body (except for the gametes, egg or sperm, where they have just 23 chromosomes). Half of each pair is inherited from their mother and the other half from their father. Chromosomes have hundreds of small 'sections' on them called genes. Genes have the 'recipe' of our genetic material (DNA) and are responsible for executing it properly; they make up our physical characteristics and tell our bodies how to work and function properly. Sometimes, instead of the correct gene, a gene with a mutation (change) will be inherited instead. Depending on the mutation and where it occurs, mutations could have no effect, or they could cause genetic diseases by changing how the gene works, prevent it from working properly or from working altogether.

What is carrier screening?

Carrier screening is a test that can be taken by any individual to check if they are carriers of a genetic disease.

What is a carrier?

A carrier is an individual who has a mutation in one of their two gene copies. Carriers are not affected, and they don't show any signs or symptoms of a disease. Two carriers of the same mutation could have a child who is affected by the genetic disease if they both pass on the gene carrying the mutation.

What are monogenic diseases?

Monogenic diseases are caused by a mutation in a single gene. They are also known as single gene diseases.

Why do X-linked diseases affect males and females differently?

As males have one X and one Y chromosome, when their X chromosome has a mutation the disease always manifests. Females have two X chromosomes, and when one of their X chromosomes has a mutation they are carriers of an X-linked disease. They may exhibit some symptoms of the disease or no symptoms at all, due to 'X-inactivation' – a mechanism that always 'shuts off' one of the two X chromosomes in females so they don't have twice the number of genes as males, which would be toxic. As this process is random in the female cells, the X chromosome with the mutation can be silenced in varying degree in female carriers; thus explaining the symptom variability shown in X-linked diseases.