

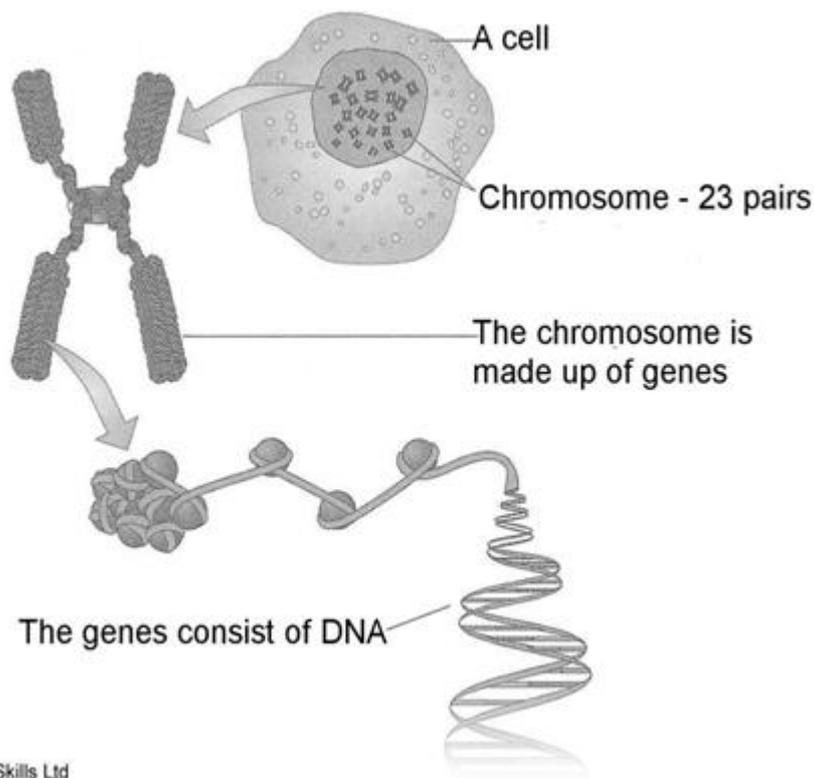
Genetics: A Guide to Some Common Used Terms Used in Genetics and Screening for CF

Introduction

Because Cystic Fibrosis is an inherited genetic disease, the following section seeks to provide an introduction or easy guide to some frequently used terms.

Genes are located on small thread-like structures called chromosomes. Usually we have 46 chromosomes in most cells. One set of 23 chromosomes we inherit from our mother and one set of 23 chromosomes we inherit from our father. So we have two sets of 23 chromosomes, or 23 pairs. Sometimes, there is a change (mutation) in one copy of a gene or chromosome which stops it from working properly. This change can cause a genetic condition because the gene is not communicating the correct instructions to the body. Some examples of genetic conditions include cystic fibrosis, alpha one and muscular dystrophy.

Genes, Chromosomes and DNA



Glossary of key terms related to genetics and screening for CF

Autosomal dominant genetic

Conditions:

These are conditions whereby a person needs only to inherit one changed copy (mutation) of the gene in order to be affected by the condition, or become affected by the condition later in life. The changed gene is dominant over the normal gene. For more information see the Dominant Inheritance leaflet.

Autosomal recessive genetic conditions:

These are conditions, including Cystic Fibrosis, whereby a person has to inherit two altered copies (mutation) of the gene (an altered copy from each parent) to be affected by the condition. A person who has only one copy of the changed gene will be an unaffected carrier of the gene and will not have the disease itself.

Autosomes:

We have 23 pairs of chromosomes. Pairs number 1 to 22 are called autosomes and look the same in men and women. Pair number 23 are different in men and women and are called the sex chromosomes.

Carrier of a mutated gene:

A person who is generally not affected with the condition (at that moment), but carries one fault copy of a gene. In the case of recessive conditions, the person will not be affected; in the case of dominant conditions, the person may become affected at a later stage.

Cell:

The human body is made up of millions of cells, which act like building blocks. Cells in different parts of the body look different and do different things. Every cell (except for eggs in women and sperm in men) contains two copies of each gene

Chromosomes:

Thread-like structures which can be seen under the microscope and contain the genes. The usually number of chromosomes in humans is 46. One set of 23 chromosomes we inherit from our mother and one set of 23 chromosomes we inherit from our father.

DNA:	A chemical substance which makes up the genes, and which contains the information needed for the body to work.
Family tree:	A diagram to show the people in your family who do and do not have the genetic condition, and how they are related to you and to each other.
Gene:	Information needed for the body to work, stored in a chemical form (DNA) on chromosomes.
Genetic condition:	A condition or disease caused by an abnormality in a gene or chromosome, including for example CF.
Genetic counselling:	Information and support for people who are concerned about a condition which may have a genetic basis. This is available from Crumlin children's hospital for parents with a newly diagnosed child with CF.
Genetic counsellor:	A specialist who gives information and support to people who are concerned about a condition which may have a genetic basis.
Genetic test:	A test which can help identify if there is a change in a particular gene or chromosome. It is usually a blood or tissue test.
Hereditary condition:	One that is inherited (passed down through families).
Metabolism:	The process your body uses to get or make energy from the food you eat. Food is made up of proteins, carbohydrates and fats. Chemicals in your digestive system break the food parts down into sugars and acids, your body's fuel. Your body can use this fuel right away, or it can store the energy in your body tissues, such as your liver, muscles and body fat.
Metabolic disorder:	Occurs when abnormal chemical reactions in your body disrupt the metabolic process. When this happens, you might have too much of some substances or too little of other ones that you need to stay healthy. A metabolic disorder can either be inherited or acquired and can affect major organs of the body.

Mutation:	A change in a gene that is sometimes also known as a gene alteration. Sometimes when a gene is changed, its information is altered so it does not work properly. This may cause a genetic condition.
Predictive testing:	A genetic test for a condition that may or will occur later in life. When the genetic test is for a condition that will almost certainly develop the condition in the future, the test is sometimes called a presymptomatic test.
Screening for CF:	The screening programme for CF measures the plasma level of immunoreactive trypsinogen (IRT). IRT is normally excreted by the pancreas but in individuals with CF this is 'regurgitated' back into the blood due to the thick mucus secretions blocking the pancreatic ducts. Levels of IRT may remain high in blood for about the first six months of life. If the blood IRT level is high the sample will be referred for CF mutational analysis. This DNA test screens for the presence of 38 mutations on the original bloodspot. If two mutations are detected then the baby probably has CF and this will be confirmed by a Sweat Test.
Sweat Test:	When cystic fibrosis (CF) is suspected, babies have a 'sweat test'. Babies with CF, and indeed all people with CF irrespective of age, have a large amount of salt in their sweat. Measuring the amount of salt in sweat will determine whether or not your baby has CF.