Allele Profile

Cystic fibrosis, Allele C1

Basic Information

Genetic Disorder – Cystic fibrosis

Affected Gene – CFTR

Affected Protein – The affected gene codes for the protein cystic fibrosis transmembrane conductance regulator (CFTR)

Allele – C1

There are more than 1,200 versions, or alleles, of the CFTR gene. Some cause genetic disorders and some do not. Your assigned allele is one of a few hundred that cause cystic fibrosis.

Mutations & Alleles

The protein-coding portion of the CFTR gene is 4,440 nucleotides long, and it has 1,480 codons.

The DNA sequence of your allele is identical to a healthy allele for most of its length. The place where they differ is shown in detail:
Inheritance
Everyone inherits two alleles of the **CFTR** gene, and CFTR protein is normally made (expressed) from both. Once it is made, CFTR protein goes to the plasma membrane. Here, it moves chloride ions (from salt) from inside the cell to the outside.

The graph shows the average sweat chloride level from many people with each allele combination. A sweat test measures how well a person’s CFTR proteins are working. If CFTR proteins are working well, chloride levels are low. In people with cystic fibrosis, chloride levels are high.

Protein Function & Expression
1. The C1 allele is switched on in cells that line the lungs and digestive organs (same as healthy alleles).

2. Cells read the C1 allele and build CFTR protein—but they make much less protein than normal.

3. The small amount of CFTR protein that is made from the C1 allele is altered: it cannot move chloride ions.

4. People with two C1 alleles have cystic fibrosis. They usually have thick mucus in the airways of their lungs, and their digestive organs do not work properly.