

Cystic Fibrosis is a simple genetic puzzle, but finding a cure remains difficult

On the face of it, CF is a perfect candidate for gene therapy. It's literally a textbook genetic disease – if you were taught about inherited disease at school in the last 25 years, you'll likely have studied CF as the example of a simple, one-gene, recessive disease that happens to be relatively common in the Western world.

Mutations in the CFTR gene lead to faulty versions of an important protein being made, but only people who have two mutated copies, one from each parent, suffer from CF. People who have only one mutated copy are carriers with no symptoms, approximately one in every 22 people in the UK.

Scientists still do not know exactly how CFTR is responsible for causing the symptoms of CF. The leading theory is that, in people with CF, ions and water do not move across the epithelium, the thin layer of tissue that lines all parts of the body. This makes it harder for tiny hair-like structures called cilia to wave around or 'beat', which would normally move mucus and the airborne bacteria it traps out of the lungs and airways.

Yet, theoretically, CF is one of the easiest genetic conditions to fix. Healthy copies of the CFTR gene could be delivered to cells in the lung in much the same way that people with CF already inhale treatments through nebulisers. Once they got into the lung cells, the healthy copies of the gene could make functioning CFTR proteins.

"It was very alluring at the beginning to say I have a gene, I have a delivery device, surely I can get it in," says Eric Alton, professor of gene therapy and respiratory medicine at Imperial College London. People expected that once you'd identified the broken gene and what it did, you could do gene therapy by adding the normal gene. "Neither of those [discoveries] have been forthcoming rapidly," he reflects.

The 25 years since CFTR's discovery seems an awfully long time to wait – and still be waiting. The disease has "contributed much more to science than science has contributed to the disease", Jack Riordan, one of the gene's discoverers, told the journal *Nature* in 2009.

Read the full, original story: [Surviving through science: life with cystic fibrosis](#)