



JOURNALIST FACTSHEET

GENE THERAPY

What is gene therapy?

Gene therapy is a way of treating or curing a disease by adding a copy of a healthy gene to do the job of a faulty one.

For gene therapy to be effective, the healthy gene has to be transported to the cells where it is needed and has to tell the body to turn it on when it gets there.

The carrier for transporting a healthy gene is called a vector or gene transfer agent (GTA). The body is told how to turn the gene on by using a promoter.

A gene therapy product is made up of the healthy gene, the gene transfer agent and the promoter.

Gene therapy in Cystic Fibrosis

Cystic Fibrosis (CF) is caused by a single faulty gene that controls the movement of salt and water through the cells in the body. In people with Cystic Fibrosis, this gene doesn't work or only partly works meaning that the internal organs such as the lungs, liver and digestive system become clogged with thick sticky mucus which attracts infection. In turn, the body tries to fight off infection, which causes inflammation and damage to the organs.

In 1989, scientists discovered the faulty CF gene. Since then, much time and money has been spent working to create gene therapy for people with Cystic Fibrosis. If scientists can find a way of adding a healthy copy of the gene to the lungs, this will allow normal movement of salt, thus breaking the cycle of infection, inflammation and damage.

90% of people with Cystic Fibrosis die of chronic lung damage. Effective gene therapy would help prevent the development of this lung damage by correcting the fault in the lungs.

Gene therapy to the lungs is difficult as they are designed to keep things out. In people with CF, the thick sticky mucus in the lungs creates another barrier to overcome.

Funding for Gene Therapy

In the early 2000s, the Cystic Fibrosis Trust brought together the UK's leading CF gene therapy teams into a single working group – the UK CF Gene Therapy Consortium. There are now scientists and clinicians in Edinburgh, London and Oxford dedicated to gene therapy for Cystic Fibrosis. It is the largest group studying CF gene therapy in the world.

The UK CF Gene Therapy Consortium has developed a product for CF gene therapy. Detailed trials to ensure it is safe are now being carried out.

The Cystic Fibrosis Trust has invested £30 million over 10 years to discover if lipid based (using a fat based carrier) gene therapy to the lungs of people with Cystic Fibrosis will have clinical benefit. This particular piece of funding came to an end in March 2012.

Clinical Trials

200 people with CF in Edinburgh and London were recruited to take part in a Run-in to a future clinical trial and were monitored for over a year to measure vital functions. Over 150 young people with CF completed the Run-in study.

In February 2009, a pilot trial for CF gene therapy began. 27 young adults with Cystic Fibrosis were given a single dose of the gene therapy product. This will provide important information on the correct level of dose and possible toxic effects.

Once the results of the pilot study have been analysed, a further round of safety tests is necessary. At the same time, up to 100 of the people recruited for the Run-in will be chosen to take part in the multi-dose clinical trial. Doctors and scientists are looking for the most suitable candidates from the 150 patients who completed the Run-in.

If all is well in the pilot trial and as long as the safety studies and the product are passed by the regulatory authority, these patients, all over 12, will be given either the gene therapy product or a placebo in a double blind multi-dose trial for a year.

This is the first time anywhere in the world that CF gene therapy has been studied in this way.

Next Stage March 2012:

Matthew Reed, Chief Executive of the CF Trust said:

"It is fantastic news that the EME programme has agreed to award the UK CF Gene Therapy Consortium funding for the gene therapy clinical trial. With the Herculean efforts of the families and friends in the CF community and the general public which has enabled the work to reach this stage, the groundbreaking research trial funded by the MRC-NIHR is now able to continue. "

Further good news for the CF community is that there is to be a second lab-based study, funded directly by the MRC and led by the University of Oxford. £1.2 million has been granted to investigate a more advanced version of the therapy using a modified virus to carry the replacement gene into the lungs, which could in future lead to a more efficient delivery mechanism.

Professor Eric Alton, Coordinator of the UK CF Gene Therapy Consortium said:

"We are delighted that the MRC-NIHR, after extensive peer review through its EME programme, has agreed to support the clinical trial. Starting in spring, about 130 CF

patients will receive monthly doses of either the CF gene or placebo over a one year period. The outcome should be known in spring 2014, once all patients have completed the study. Although no results will be available before then, we will provide further updates on the Consortium's website."

If successful, this research could be developed by a major pharmaceutical company through a large multi-centre trial, potentially including Europe and the USA. This would be the definitive study for safety and efficiency. The company could then develop gene therapy into a product for the market and normal clinical use. This is a best case scenario for CF gene therapy.

ENDS

For all media inquiries please contact:

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- The Cystic Fibrosis Trust is the UK's only national charity dealing with all aspects of Cystic Fibrosis (CF). It funds research to treat and cure CF and aims to ensure appropriate clinical care and support for people with Cystic Fibrosis.
- Cystic Fibrosis (CF) is one of the UK's most common life-threatening inherited diseases. Cystic Fibrosis is caused by a single defective gene. As a result, the internal organs, especially the lungs and digestive system, become clogged with thick sticky mucus resulting in chronic infections and inflammation in the lungs and difficulty digesting food.
- Each week five babies are born with Cystic Fibrosis and two young people die – 90% from lung damage. Only half of those living with Cystic Fibrosis are likely to live past their 41st Birthday, although improvements in treatments mean a baby born today is expected to live even longer.
- Further information can be found on our website www.cftrust.org.uk. Help and advice for those affected by Cystic Fibrosis is available through our Helpline on

0300 373 1000. For further information, media should contact Louise Banks on 0208 290 7912 or email: lbanks@cftrust.org.uk

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