NIHR | BioResource

CFT. Cystic Fibrosis

NIHR BioResource – Rare Diseases study project

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Prof. Andres Floto (left) and Dr Alex Horsley, CFT project Leads

Summary

Cystic fibrosis (CF) is one of the UK's most common life-threatening inherited diseases. CF is caused by by faulty variants in both copies of the Cystic Fibrosis Transmembrane conductance Regulator (CFTR) gene, each inherited from one parent.

The gene and the protein it makes, help control the movement of salt and water in and out of cells. When the gene is faulty, it can cause thicker mucus. 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.

Cystic fibrosis is a genetic condition with one person in 25 carrying the faulty CFTR, usually without knowing. If two carriers have a baby, the child has a one-in-four chance of having cystic fibrosis. Around 10,600 people in the UK have cystic fibrosis; that's 1 in every 2,500 babies born.

Different mutations of the CFTR gene affect the CFTR protein in different ways and more than 2,000 CFTR variants have been identified, which can have different impacts on an individual's disease severity. In addition, these variants will impact on which of the currently available treatments for CF they are eligible for or will benefit from.

This is an extraordinary time in the history of CF with the development of breakthrough highly effective CFTR modulator therapies, which target the basic protein defect by enhancing the function of the CFTR. Most recently, a triple combination therapy developed by Vertex Pharmaceuticals, Kaftrio, has been made available to eligible people with CF in the UK.

However, while a high proportion of the CF population in the UK will now be eligible for this new treatment, there is still a need to do more research to make sure that all people with CF can live a long and full life. For example, some people have different responses to these therapies, some will also have ongoing healthcare needs due to CF-related conditions such as inflammation, infection, diabetes and other co-morbidities due to the increased longevity expected to be seen with these highly effective modulators. And, importantly, the need to develop effective therapies for these individuals, who may feel "left behind" by the recent Kaftrio success, is also increasingly pressing.

The BioResource will provide a unique and ground-breaking opportunity to join up existing clinical data registries, providing information on the characteristics of CF, with genetic data. This will drive the development of more targeted CF therapies and large-scale clinical trials for the benefit of all CF patients.

Recruitment Criteria

Inclusion

Cystic fibrosis diagnosis.