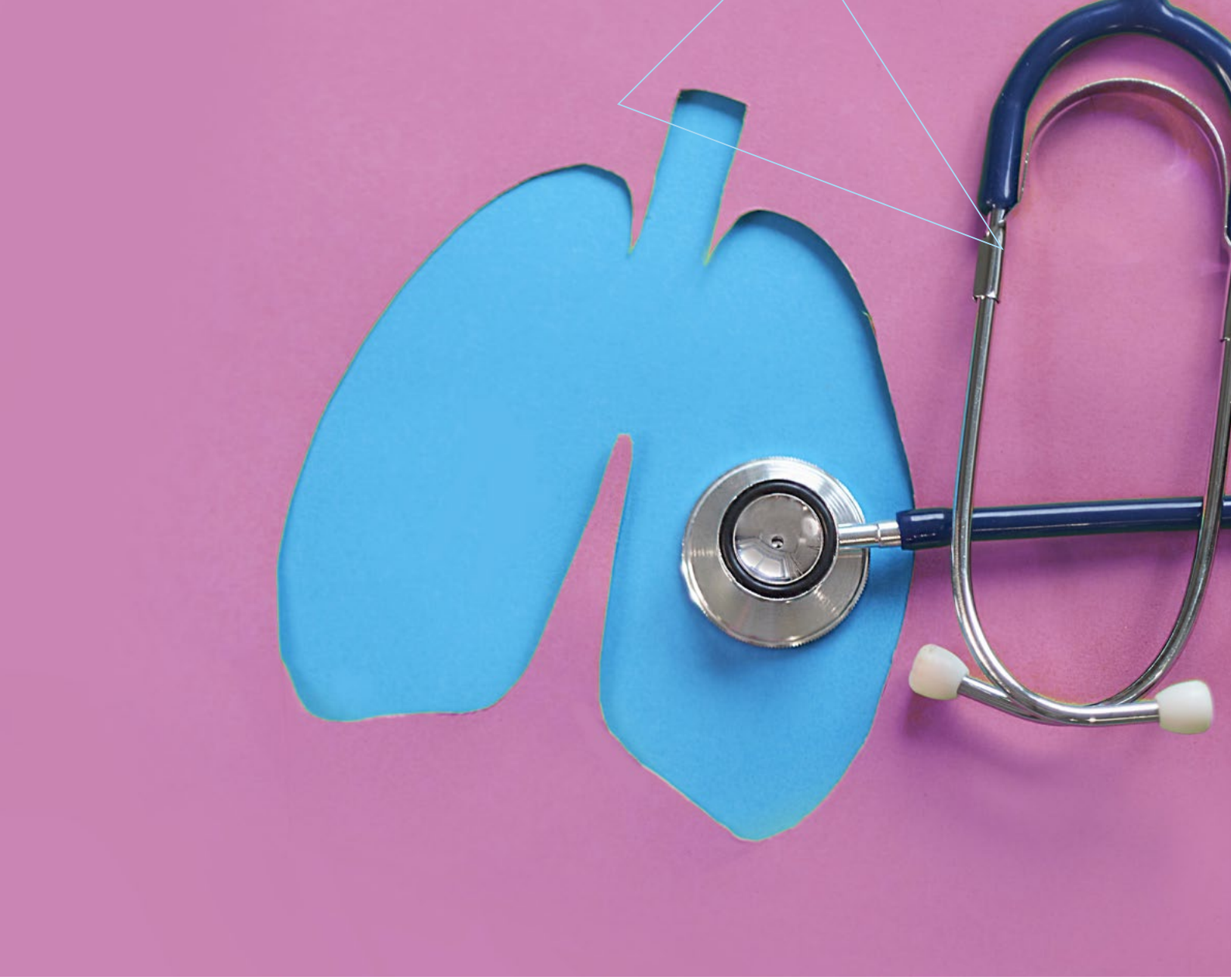


Therapy Guide

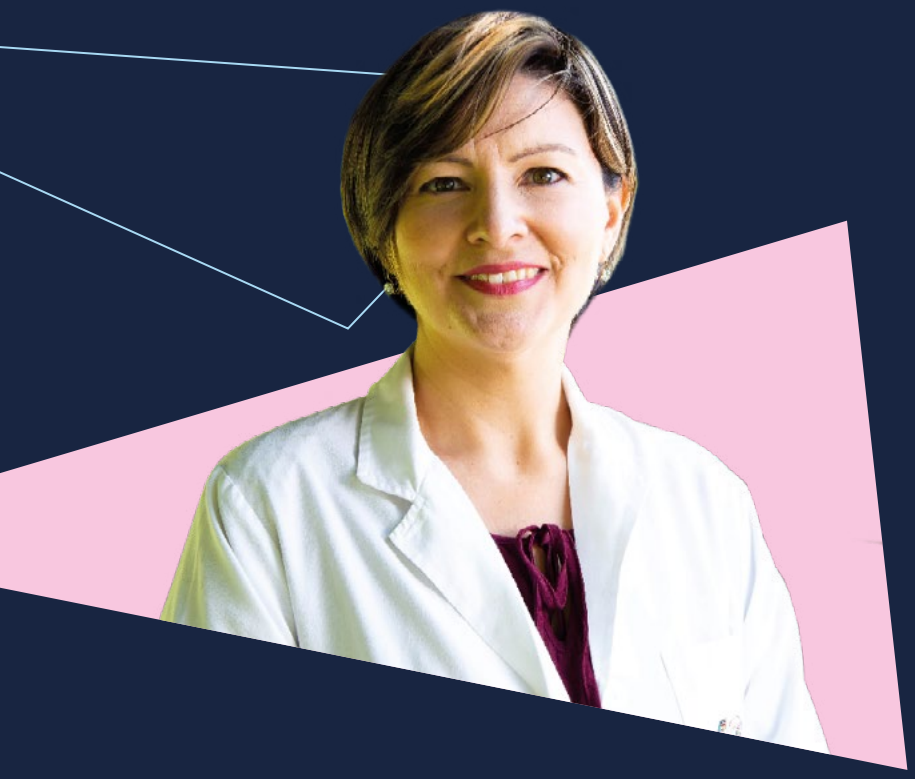
Cystic Fibrosis

GKA.

The GKA Difference

With a small patient population, many of whom are children, and a restricted number of specialists at a limited number of treatment centres, Cystic Fibrosis is a complex and challenging disease area for market researchers. However, GKA has overcome these challenges, leveraging its expertise and experience to sucessfully carry out studies on CF over the last 25 years.

The key to the GKA Difference is the company's strong relationships with key opinion leaders and specialists, including many at leading specialist centres such as Manchester Adult CF Centre and Great Ormond Street Cystic Fibrosis Centre. Its panel includes Respiratory Nurses and Respiratory Medicine Specialists, most of whom see CF patients on a regular basis. What's more, through its long-lasting relationships with CF patient organisations and dedicated finders, GKA can recruit validated CF patients across many different methodologies, including ethnographic studies, a key selling point for many clients.



The Lowdown

Cystic fibrosis (CF), also known as mucoviscidosis, is an inherited disease caused by a faulty gene. This gene controls the movement of salt and water in and out of sufferers' cells, causing the lungs and digestive system to become clogged with mucus, making it hard to breathe and digest food[4].

The name cystic fibrosis refers to the characteristic scarring (fibrosis) and cyst formation within the pancreas, first recognized in the 1930s by Dr Dorothy Andersen, a pathologist at the New York Babies Hospital[5]. Later, in 1985, the gene causing the disease – the cystic fibrosis transmembrane conductance regulator or CFTR – was identified[1].

Some facts about cystic fibrosis

2.5 million

people in the UK carry the CF gene[4]

9,000

people have CF in the UK[6]

Screening

is the main way CF is identified

40 years

More than half the CF poopulation live past 40 years[4]

Understanding Cystic Fibrosis

Cystic fibrosis is most common in white people of northern European descent. It is estimated that one in every 2,500 babies born in the UK is born with cystic fibrosis and there are more than 9,000 people living with the condition in this country. For someone to be born with cystic fibrosis, both parents must carry the faulty gene.

Babies are screened for cystic fibrosis at birth as part of the NHS newborn screening programme, where a small amount of blood is analysed for cystic fibrosis and four other inherited conditions, including sickle cell anaemia[2].

In the past, most children with cystic fibrosis would die of related complications before reaching adulthood, but the outlook has improved considerably in recent years due to advancements in treatment[2]. Today, more than half of the cystic fibrosis population in the UK live past 41 and with improved care and treatment a baby born today is expected to live even longer[4].

Causes and Symptoms

CF is the result of a faulty gene known as CFTR, which normally creates a protein that helps move salt and water out of cells. A defective gene results in the build-up of thick, sticky mucus in the body's tubes and passageways that damage the lungs, digestive system and other organs, causing swelling and, in the lungs, repeated infections[7].

Symptoms of CF usually start in early childhood and include a persistent cough, recurring chest and lung infections and poor weight gain. However, cystic fibrosis is a complex disease that affects many different organs and symptoms vary in severity from person to person[8].

Although not everyone is affected, many people with CF experience problems with lung function, most especially the build-up of mucus, which can damage the lung and result in lung infections. CF also affects the digestive system as thick, sticky mucus can block the ducts of the pancreas, reducing the amount of insulin produced and stopping digestive enzymes from reaching the intestines to aid digestion. This can cause malnutrition, leading to poor growth, physical weakness and delayed puberty, and some people can go on to develop CF-related diabetes when their blood sugar levels are no longer controlled. Poor nutrition can also contribute to the development of thin, brittle bones in older sufferers.

The condition can also cause fertility problems; in men, the tubes carrying sperm can become blocked and, in women, poor nutritional associated with the disease can cause irregular menstrual cycles and thicker vaginal mucus. CF can also cause the blockage of small ducts in the liver resulting in liver disease, which can require a liver transplant in severe cases[9].

Diagnosis

Most cases of cystic fibrosis in the UK are now identified through screening tests carried out early in life. However, some babies, children and even young adults are identified later following unexplained illness. There are three main ways of diagnosing cystic fibrosis:

Newborn testing – all babies are offered screening for CF at birth as part of the NHS newborn screening programme

Antenatal testing – a test on a pregnant woman to see if her unborn baby has CF. Such testing is usually only offered to mothers thought to be at high risk of having a child with the disease

Sweat testing – many parents first notice their child's symptoms when they kiss them, as their skin can taste salty and a harmless sweat test can confirm CF (**Genetic testing** is also available if the sweat test results are unclear[10]). In addition, a simple '**mouthwash**' test can determine whether a person is a carrier of the defective gene, an important precaution if a partner is known to carry the gene[11].

Treating Cystic Fibrosis

There is currently no cure for cystic fibrosis but many treatments are available to manage it, including medication, physiotherapy, exercise and nutrition[4].

Medication: Commonly taken medications include:

- Bronchodilator drugs open airways, relieving tightness and shortness of breath
- Antibiotics help to treat or control persistent lung infection
- Steroids reduce inflammation in the airways
- Mucolytics such as DNase break down mucus, makingit easier to clear from the lungs
- People with CF-related diabetes need to balance food intake with suitable diabetic treatment like tablets or insulin[12].

Physical Treatment: Traditionally, physiotherapy for CF focused on clearing mucus from the lungs and this still makes up a large part of daily treatment. However, the role of the physiotherapist has expanded to include daily exercise, inhalation therapy, posture awareness and, for some, the management of urinary incontinence.

Exercise is particularly important because it helps clear mucus from the lungs and improves physical bulk and strength. Children with cystic fibrosis should be encouraged to take part in as much physical activity as possible. Posture and chest mobility is also important; older children and adults may need to do stretching exercises to preserve full movement of the joints and muscles around their shoulders and chest. Younger children can do the same by taking part in games or activities that involve moving and stretching the trunk and arms[13].

Nutrition: Most people with cystic fibrosis take enzyme capsules with meals and snacks to help digest food. A suitable diet – high in calories and rich in fact and protein – is also very important as a good body weight helps to fight chest infections[14].For some rare types of cystic fibrosis, such as the G551D mutation, there are treatments which aim to compensate for a defective gene[2].

References
1 – http://cysticfibrosis.org.uk/about-cf/what-is-cystic-fibrosis/history-of-cf
2 – http://www.nhs.uk/Conditions/cystic-fibrosis/Pages/Introduction.aspx
3 – http://cysticfibrosis.org.uk/about-cf/what-is-cystic-fibrosis/what-causes-cystic-fibrosis
4 – http://cysticfibrosis.org.uk/about-cf/what-is-cystic-fibrosis
5 – https://www.princeton.edu/~achaney/tmve/wiki100k/docs/Cystic_fibrosis.html
6 – http://www.patient.co.uk/doctor/cystic-fibrosis-pro
7 – http://www.nhs.uk/Conditions/cystic-fibrosis/Pages/Causes.aspx
8 – http://www.nhs.uk/Conditions/cystic-fibrosis
9 – http://cysticfibrosis.org.uk/about-cf/what-is-cystic-fibrosis/symptoms-of-cf
10 – http://www.nhs.uk/Conditions/cystic-fibrosis/Pages/Diagnosis.aspx
11 – http://cysticfibrosis.org.uk/about-cf/what-is-cystic-fibrosis/how-is-cf-diagnosed
12 – http://www.cysticfibrosis.org.uk/about-cf/living-with-cystic-fibrosis/treatment/medication
13 – http://www.nhs.uk/Conditions/cystic-fibrosis/Pages/Treatment.aspx
14 – http://www.cysticfibrosis.org.uk/about-cf/living-with-cystic-fibrosis/nutrition

We are GKA.

If you have been asked to carry out a healthcare market research study surrounding Cystic Fibrosis or one of the related conditions, why not give us a call today?

+44 (0)1242 220420
quotes@gilliankenny.com
gilliankenny.com