What is a rare lung disease?

A rare disease is a disease that affects less than one person in every 2,000. This may sound like a very small number, but as there are 700 million people in Europe, one ‘rare’ disease could affect up to 350,000 Europeans - that’s almost the population of Malta.

Examples of rare lung diseases are cystic fibrosis and alpha-1 antitrypsin deficiency.

There are many rare lung diseases, so it is probable that millions of people suffer from these. This is a major problem, as many rare diseases are serious and long-lasting, and they are often life-threatening.

What is the difference between ‘rare’ and ‘orphan’ diseases?

An ‘orphan’ disease is the name given to a disease that is not widely researched and/or for which there is no specific treatment, thus making patients feel ‘orphan’ in the world of healthcare. Many rare diseases are orphans, although some orphan diseases are not rare (e.g. some parasitic diseases in poor countries).

On the next page of this factsheet, there is more information about some of the orphan diseases.

There is no list of all the rare and orphan lung diseases, although at www.orpha.net, a website devoted to these diseases, there is a large database. They can be diseases that affect only the lungs (such as idiopathic pulmonary fibrosis), or they may affect other parts of the body too (such as scleroderma).

A wide range of causes lie behind these diseases, but for many of them the cause is not known. A large number of the diseases are caused by faulty genes. This means they can be passed from parents to children, although they may occur randomly because of gene damage. Other diseases can be caused by the body’s immune system going wrong.

Sometimes, orphan diseases stop being orphans as scientists begin to study them more intensely. This has happened to idiopathic pulmonary hypertension in the past few years.
**Lymphangioleiomyomatosis** is often called LAM for short. It almost never affects anyone who is not a woman of child-bearing age. Abnormal cells invade the tissues of the lungs, including the airways, and can form cysts, destroying healthy tissue and making holes in the lungs.

**Symptoms:** LAM makes breathing more and more difficult, and may cause chest pain, because of a collapsed lung (pneumothorax). A computed tomography, or CT, scan is needed for diagnosis.

**Treatment:** There is no current specific treatment, and doctors must try to reduce symptoms, for instance by giving oxygen. Women with advanced stages of the disease may need a lung transplant.

**Scleroderma** is a disease that can affect many parts of the body, as well as the lungs. Whereas some people with the disease have only a localised form, that only affects their skin (the word scleroderma means ‘hard skin’), other people have a more serious form that is called systemic sclerosis and can affect the lungs - especially the alveoli and vessels - kidneys, heart and digestive tract. It is more common in women than in men, and is usually diagnosed between the ages of 30 and 50 years. The cause of the disease is not known.

**Symptoms:** Scleroderma is a complicated disease, but doctors may look for skin that becomes thick progressively, and will have to do tests before they are sure someone has the disease. Lung problems can range from shortness of breath due to some kind of pulmonary fibrosis or pulmonary arterial hypertension, which is high blood pressure in the vessels that carry blood from the heart to the lungs.

**Treatment:** There is no cure for scleroderma, but doctors try to treat the problems that it causes. In particular, there are drugs to treat pulmonary arterial hypertension. Some patients are also given drugs to make their immune system less active.
Idiopathic pulmonary fibrosis (often called IPF) is a disease that usually begins between the ages of 50 and 75 years of age. Tissue deep inside the lungs starts to become scarred and thickened. This means that it is more difficult for the body to get enough oxygen. Usually, the cause of the disease is not known.

Symptoms: The symptoms of IPF include difficulty in breathing, a cough and a crackling noise at auscultation (auscultation is when a doctor uses a stethoscope to listen to someone's chest) when the patient breathes in. IPF can be hard to diagnose, because many of the symptoms and other signs are similar to those in other diseases.

Treatment: Patients may be given oxygen to improve their quality of life, but there is no cure for IPF and, sadly, many patients die within 3 to 5 years of being diagnosed.

Idiopathic chronic eosinophilic pneumonia, often shortened to ICEP, is an accumulation of a type of white blood cells called eosinophils. The cause of this disease is not known. ICEP is twice as common in women as in men, and about half of the people who get it already have asthma and allergic rhinitis (an irritated nose and eyes, caused by allergy).

Symptoms: The symptoms of ICEP may not appear serious initially, so many patients are not diagnosed for some time after they become ill. Patients may have serious difficulty breathing and may wheeze or cough. They may feel weak, lose weight and sweat during the night. The doctor may have to carry out a range of tests, such as X-rays and blood tests, to be sure someone has the disease.

Treatment: Patients are usually given corticosteroids, usually for a long period (often a year or more). Many patients get the disease again, however, as they begin to come off the drugs. Some patients may develop asthma (or worse asthma if they already have it) after having ICEP.

Pulmonary alveolar proteinosis is also called PAP. In this disease, a grainy substance made mostly of fat and protein collects in the air sacs, or alveoli, of the lungs. The cause is usually unknown, although sometimes it can be the result of inhaling substances such as silica dust.

Symptoms: Like many other lung diseases, the symptoms can include a dry cough that will not go away, difficulty breathing, tiredness, weight loss and chest pain. To diagnose the disease the doctor needs a computed tomography (CT) scan which shows very peculiar opacities (a non-transparent area) suggesting the diagnosis.

Treatment: The commonest treatment is to wash the grainy substance from the lungs, using a large amount of water. This is called 'lavage'. Some patients may have to have this procedure regularly under general anaesthesia, while others may only need it once. Some patients are also given a substance called GM-CSF to treat the disease, but this is still experimental. People with severe forms of pulmonary alveolar proteinosis may need a lung transplant.
Will my doctor know if I have a rare lung disease?

One problem with rare diseases is that family doctors may not recognise them. It is impossible for medical schools to teach doctors about all the many thousands of rare diseases that exist. This means that when a patient's respiratory problems continue without a proper diagnosis, the family doctor should not hesitate to refer them to a specialist for further help. It is important that specialist respiratory doctors know enough to at least suspect that someone has a rare disease, so they can further be examined by an expert in that disease.

Is anything being done to improve care for people with rare lung diseases?

Governments in Europe and the USA have tried to encourage drug companies to look for treatments for orphan diseases, by offering financial and business incentives. This has led to some new treatments. Treatment for some rare diseases has also improved in other ways. Cystic fibrosis patients in many parts of the world now live longer thanks to better current treatment, and more research is being carried out into idiopathic pulmonary fibrosis. As well as this, drugs that are commonly used in other diseases, such as immunosuppressors and corticosteroids, have sometimes been found to be good at treating some rare diseases.

What organisations can offer support to people with rare lung diseases?

In recent years, partly thanks to the rise of the internet, there has been a large increase in the number of patient organisations devoted to individual rare lung diseases. Often, these organisations work only with patients in a single country, especially because of language barriers. However, one good place to start looking for an organisation that helps people with a specific disease is the European Organisation for Rare Diseases (Eurordis). Its website, at www.eurordis.org, contains the contact details of dozens of organisations that work on individual diseases or on rare diseases in general.

The size of the patient organisations varies, but in many cases they help to fund research, and they also provide support to patients, as well as pushing other organisations and governments to help patients.

Scientists and doctors have also taken an increased interest in rare lung diseases in recent years. Because not many doctors see many patients with rare diseases, doctors have joined forces to share information about rare diseases. Over time, this information may help to provide better diagnosis and treatment of these conditions.

Useful websites

The European Organisation for Rare Diseases - www.eurordis.org

Orphanet - www.orpha.net

It provides information on many orphan diseases including lung diseases.

Related information from the European Lung Foundation


The ELF is the public voice of the European Respiratory Society (ERS), a non-profit-making medical organisation with more than 8,000 members in more than 100 countries. The ELF is dedicated to lung health throughout Europe, and draws together the leading European medical experts to provide patient information and raise public awareness about respiratory disease.

This factsheet has been prepared with the help of J-F. Cordier, of, the Reference Center for Orphan Pulmonary Diseases, Lyon, France.