Primary Ciliary Dyskinesia
The Facts

www.pcdsupport.org.uk
The condition is **genetically inherited** and may affect about **1 in 15,000 Europeans**. The incidences are higher in certain populations where consanguineous marriages are customary and those isolated for cultural geographical reasons. In the UK, tests are not available to determine who is a carrier and as yet no test for prenatal diagnosis is available. **Research** has identified a number of genes which are thought to cause about 38% of all PCD cases, but the total number of genes involved in PCD has not yet been determined. **If you are concerned about PCD genetics please ask to see a genetic counsellor.**

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**The PCD Family Support Group is a registered charity who:**

- Provide support to patients with PCD and their families.
- Bring PCD to the attention of the medical profession and the public
- Provide an up to date information service
- Raise money to promote research to aid diagnosis and treatment of children and adults with PCD
What is Primary Ciliary Dyskinesia?

Primary Ciliary Dyskinesia (PCD) is an inherited, relatively rare condition associated with an abnormality of cilia (see definition of cilia below). PCD may affect the lungs, nose, sinuses, ears and fertility.

The condition involves recurrent infections in the nose, ears, sinuses and lungs and if untreated can lead to a form of lung damage known as ‘bronchiectasis’.

Up to 50% of patients with PCD also have dextrocardia (heart on the right side) and situs inversus (internal organs being on the opposite side to normal).

The mainstay of treatment is chest physiotherapy and targeted antibiotics enabling individuals to lead normal lives. Any problems resulting from PCD vary from person to person.

Outlook

If appropriate and diligent care (e.g. undertaking regular physiotherapy and attending clinic appointments for reviews) is undertaken from an early age, children and adults with PCD can lead normal, active lives.

Cilia

Cilia are the microscopic hairs which beat in the airways to sweep secretions containing bacteria and particles out of the respiratory tract. In people with PCD the structure of the cilia may be abnormal, appear to be structurally normal but the cilia do not function properly. All abnormalities have the same end result, a predisposition to infections in the lungs, sinuses and ears, because secretions tend to sit in the airways rather than being cleared by the normal action of the cilia.

The tail of the sperm is almost identical to the cilia and because of reduced motility, some PCD males are sub-fertile. Female reproductive fertility may also be affected because the Fallopian tubes are lined by cilia. However, several female adults with PCD in the Family Support Group have produced children with and without techniques to assist conception.

Early Years

Newborn babies with PCD may often have some breathing difficulties, though often these improve naturally. It may be months or even years later that, if untreated, problems become apparent with recurrent nose, ears, sinus and chest infections. Chest physiotherapy to clear the airways helps prevent lung damage occurring. Without treatment it is inevitable that the lungs will eventually become damaged. At first the main symptoms noticed by parents may only be a loose cough, perpetually runny nose from birth and susceptibility to chest infections. In some there are hearing problems associated with glue ear.

Treatment

Chest Physiotherapy

Adults with PCD and parents of children with PCD should be taught by a respiratory physiotherapist how to do daily chest physiotherapy and their techniques should be reassessed regularly as the child grows up.

In very young children chest physiotherapy consists of positioning so that gravity helps to drain secretions, combined with different techniques to aid effective clearance of secretions from the lungs. As children grow they are taught how to do more of their physiotherapy independently.
People with PCD often need to do chest physiotherapy twice daily and more frequently during a cold or chest infections. If PCD patients don’t do their physiotherapy healthy areas of the lungs may become infected and permanent lung damage can occur in the form of bronchiectasis.

Children and adults with PCD should be encouraged to eat well and do plenty of exercise. Exercise is an important way of increasing the clearance of secretions from the lungs. As is singing and blowing wind instruments.

**Ear, Nose and Throat (ENT) Management**

People with PCD often experience ENT problems. This is because the cilia in the Eustachian tube may not be working and there can be a build-up of fluid in the middle ear, termed ‘glue’ ear which may cause a variable degree of hearing loss.

PCD patients should be seen by an ENT specialist with experience of PCD. From the time of diagnosis, all children should have regular hearing assessments and schools made aware of any hearing problems. The insertion of grommets is not advisable as their insertion is often complicated by a discharging ear.

It is recommended that people with PCD should get into the habit of blowing their nose at least twice a day (in the morning and before going to bed at night) to help keep the airways clear.

**Antibiotic and other drug therapies**

All chest infections for PCD patients must be treated as soon as they are suspected. A sputum test or cough swab for younger children, can be taken. The results of these tests will help patients to be given the right antibiotic (if needed) to fight the infection. Some PCD patients are on permanent prophylactic antibiotics. If the patient is unwell they may be admitted to hospital for a course of intravenous antibiotics.
Other forms of treatment can be nebulised treatments, inhalers and steroids.

**Diagnosis**

Early diagnosis is very important in order that appropriate daily chest physiotherapy and, where necessary, antibiotic treatment can be established. This helps to prevent lung damage occurring by keeping airways clear of infected mucus. A doctor who is aware of PCD can suspect the diagnosis, but confirmation requires a test from a specialised centre.

There are three centres that offer a diagnostic service, the Royal Brompton Hospital in London, the Leicester Royal Infirmary and Southampton Hospital. There are two simple screening test for adults and older children measuring the amount of nitric oxide gas present in the nose or a saccharin test. This will determine whether a nasal brushing sample is required. The nasal brushing is a simple test – a tiny brush is inserted in the nose. It is uncomfortable for a few seconds and will most likely make the patient’s eyes water. The sample then starts a complicated process which involves close scrutiny under a specialist microscope. The results will take a minimum of 6 weeks to be reported back to you.

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Further supplies of this leaflet, a Teenage PCD leaflet and an information DVD are also available.

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