

Children's Services

Primary ciliary dyskinesia

What is primary ciliary dyskinesia?

Primary ciliary dyskinesia (PCD) is a rare genetically inherited condition where the microscopic hair-like structures within the body, known as cilia do not function normally. Cilia vibrate within the airways and other organs in the body such as the middle ear. Their function within the airways is to remove bacteria, dust and other particles. In PCD the cilia either do not function at all or their movement is disordered and therefore ineffective.

In the lungs the cilia beat in one direction moving unwanted dust or bacteria towards the throat and away from the lungs. In individuals with PCD the cilia do not beat effectively. Therefore they do not clear unwanted foreign particles from the lungs and nose. This places individuals with PCD at greater risk of recurrent respiratory infections.

Incidences

PCD is a relatively rare condition with about 3000 cases in the UK. The incidence is estimated to be 1 in 15000. There are about 70 new cases every year.

What are the symptoms?

- Symptoms vary with age.
- Newborns and infants often present with respiratory problems at birth such as a runny nose and nasal congestion. This will remain constant over time.
- Your child may also have a history of persistent moist sounding cough and constant coughs and colds.
- Rarer associations with PCD include congenital heart disease and gastro-oesophageal reflux.

How is it diagnosed?

There are two ways of making a diagnosis:

- A consistent history of symptoms suggestive of PCD is taken. A sample of cilia from the nose of your child is sent to a specialist centre to be analysed. This is the preferred method of diagnosis at Addenbrooke's.
- It is also possible to perform a saccharin test in older children. In this test a pellet of saccharin is placed inside the nose of the child and the time it takes for the child to taste the sweet pellet is measured.

How is it treated?

- Regular chest physiotherapy. This is to help clear the lungs of mucus which if left will congest the airways, reduce their functioning and increase the chance of developing recurrent chest infections. Simple techniques that can be performed at home will be taught to you by a physiotherapist.
- Antibiotics are important to prevent lung damage. Antibiotics are usually taken long term for life. Regular reviews with the medical staff are required. Sputum samples or cough swabs will be taken to detect the growth of bacteria within the lungs and appropriate antibiotics prescribed. Life expectancy is not felt to be altered if treated appropriately, although this life expectancy may be reduced if lung disease is allowed to progress untreated.

How is it caused?

It is an inherited condition. Research is underway to identify the faulty genes responsible for PCD. Genetic counselling may be appropriate as it is thought to be an autosomal recessive condition. This means the faulty gene must be inherited from both parents who are asymptomatic carriers of the faulty gene. There is a one in four chance of having a child with PCD with every pregnancy. There is a one in two chance of the child being a carrier for the condition with every pregnancy and a one in four chance of the child being neither affected nor a carrier for the gene.

What is the future?

At present there is no cure. Treatment will be necessary for life. There may also be issues surrounding fertility. Boys tend to be infertile and girls may experience problems with atopic pregnancies more than the normal population.

Further information

For further information, please contact the Paediatric Respiratory Nurse on 01223 348067.

Family support groups

The PCD family support group: www.p-c-d.org.uk



We are currently working towards a smoke free site. Smoking is only permitted in the designated smoking areas.

For advice and support in quitting, contact your GP or the free NHS stop smoking helpline on 0800 169 0 169

Help with this leaflet:



If you would like this information in another language, large print or audio format, please ask the department to contact Patient Information: 01223 216032 or patient.information@addenbrookes.nhs.uk

Polish

Jeżeli chciałbyś uzyskać te informacje w innym języku, w dużej czcionce lub w formacie audio, poproś pracownika oddziału o kontakt z biurem Informacji Pacjenta (Patient Information) pod numerem telefonu: 01223 216032 lub pod adresem patient.information@addenbrookes.nhs.uk

Portuguese

Se precisar desta informação noutra língua, em impressão de letras grandes ou formato áudio, por favor peça ao departamento que contacte a secção de Informação aos Doentes (Patient Information) pelo telefone 01223 216032 ou através do e-mail patient.information@addenbrookes.nhs.uk

Arabic

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Cantonese

如您需以另一語言版本、特大字體或錄音形式索取本資料，請要求部門聯絡病人諮詢服務：電話 01223 216032，電郵地址 patient.information@addenbrookes.nhs.uk

Turkish

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Urdu

اگر آپ کو یہ معلومات کسی دیگر زبان میں، بڑے الفاظ میں یا آڈیو طریقہ سے درکار ہوں تو برائے مہربانی اس شعبہ سے پیشینٹ انفارمیشن سے ذیل کے ذریعہ رابطہ کرنے کی درخواست کریں: 01223 216032 یا patient.information@addenbrookes.nhs.uk

Bengali

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