PCD Day, June 2009

The PCD Day was held on Saturday, 27th June 2009 in a new venue in the village of Great Brickhill near Milton Keynes. It was well attended. We had a great day with over 50adults and 25+ children.

The children were entertained with a cookery workshop in the morning where they made their own pizza and cup cakes for lunch. They were also entertained by 'It's Him Entertainment' in the afternoon.

- **Fiona Copeland chaired the AGM of which the main points were: -**

What we have done since last year

- The video is in the process of being updated
- We have bought £7,000 of hearing equipment for the Royal Brompton Hospital.
- We have successfully bid for funds from Jeans for Genes and have been awarded £7,000 towards updating the video. Total cost for this is £17,000.
- Jeans for Genes have increased awareness of PCD and put it on their website and are sending information to schools
- There is an incidence of PCD of 1 in25 in Asian families (compared to 1 in 15,000 in the white families) therefore being more than cystic fibrosis. Fiona has therefore engaged with a hospital in Bradford.
- Gary and Myra attended GIG Rare Diseases Day at the House of Commons and met other groups and spoke to MPs.
- We supported research into physiotherapy
- The Medical Advisory Board Meeting was cancelled because of the tube strike
- Fiona resigned from the PPI meetings at the Brompton Hospital which now has Foundation Status
- Two newsletters have been sent out
- The British Thoracic Society Winter Meeting stall was manned by Royal Brompton Hospital staff
- Fiona went to University College Hospital to talk to medical students
- We signed up to everyclick.com which is a search engine that is free to use and raises money for PCD

Financial Review
Mick Wilkin, Treasurer, addressed the meeting. A copy of the latest accounts was distributed and accepted. He explained that: -

- We started and ended the year with£30k in the bank
- We raised £14k this year (£28k last year). Thanks to all fund raisers
- Interest on the capital is almost nil
- Costs have risen to £13,500 from£11,000 last year
- Leaflets were translated into Urdu and Arabic
- Independent Examiners were re-appointed

Financial Outlook

- The economic climate is difficult
- Fundraising and donations are down on2008, but please continue to do what you can
- Jeans for Genes funding is secured but we need to contribute £10k from our own funds
- We are a going concern for the fore seeable future

Way Forward

- The video will be updated and streamed on the website. It should be available in the New Year.
- We will support families
- We will support Jeans for Genes. Volunteers are needed to give presentations and raise awareness

- **Presentation from Claire Jackson, Postdoctoral Scientist Primary Ciliary Dyskinesia Diagnostic and Research Team of Southampton General Hospital**

Claire explained that there are three diagnostic centres in Leicester, London and Southampton. They are government funded and provide diagnostic services free of charge. The referrals are made to these centres by GPs. Around 250 patients per year are tested, and about 10% of these are PCD positive.
She explained that normal cilia act like a toothbrush and clean the lungs. There are motile and sensory cilia. They are in the airways, ear, brain, eye, respiratory tract, reproductive tract, inner ear, ventricles of the brain and the sperm tail structure.
There are about 70 PCD births per year and approximately 3,000 cases in the UK. The implications of under diagnosis are bronchiectasis, inappropriate ear nose and throat management (i.e. Grommets and surgery) and inappropriate hearing management. We want to broaden the profile of PCD and do more research across Europe.
Symptoms of PCD: -

- Respiratory distress in the newborn
- Reduced mucus clearance from the lungs and airways
- Recurring lung infections
- Sinusitis
- Otitis media
- Bronchiectasis
- Situs inversus in 50% of cases
- Subfertility/infertility in adulthood
- Danger of ectopic pregnancy

One in fifteen thousand of the general population have PCD, but this is about 1 in 2265 in Asian communities particularly around Bradford. If two carriers have children, then one in four of those children are likely to have PCD. In America there is funding for research into PCD. Five or six genes have been identified, but more research is needed.
The gold standard tests for PCD are: -

- Look at the clinical history
- Look at lung function - without good care, and with late diagnosis, lung function declines
- Nasal nitric oxide
- Nasal brushing biopsy
- Ciliary function by light microscopy
- Ciliary ultrastructure

There are several ciliary ultrastructure defects

- Dynein arm defect, which is the most common
- Central microtubule defects
- Compound cilia
- Radial spoke defects
- Extra microtubules

The cilia are looked at by light microscopy. The process is vibration controlled, the temperature is controlled to 37C, and there is high-speed video analysis, which is then slowed down so that the beat can be identified.
In normal cilia there is a strong forward stroke and then a recovery stroke, which moves the mucus along. However, in PCD the cilia move in a variety of ways and can sometimes be static. They may vibrate ineffectively as in atypical PCD. The cilia may by dyskinetic and have missing proteins. They may have dynein arm defects. They may have a transposition defect, which cause the cilia to rotate. Secondary dyskinesia occurs when cilia are damaged due to infection.
When cilia are tested using an air liquid interface, it takes about 8weeks to get a result. Light microscopy can give a result on the same day, but it is not enough for a diagnosis.
She talked about raising awareness of PCD and the need to target special care baby units and getting articles in the British Medical Journal, which GPs read. She explained that the route for testing started with the GP and referral to a consultant, and then to one of the three testing centres. However, Scotland and Wales are outside the testing zone of the three centres.

- **Presentation from Lizzie Flude, adult physiotherapist, and Nicola Collins, paediatric physiotherapist, both from the Royal Brompton Hospital**

It was explained that PCD is an inherited condition, but if appropriate and diligent care is taken, then the PCD patient can have a normal and active life.
Mucus traps dust and bacteria in the lungs, which is then swept out by normal cilia. However, in PCD mucus stays in the lung because the cilia do not work properly. In a normal person the cilia in the lung act like an escalator on the underground, but in PCD the cilia act like an escalator, which is not moving. Therefore physiotherapy is needed to get rid of this mucus.
A film was shown of a bronchoscopy which showed the airways of the lungs being blocked by mucus, underlining the need for regular physiotherapy. If PCD is managed well, then a normal and active life can follow. Prompt antibiotics are required, as well as regular sputum samples and ear nose and throat (ENT) reviews. It is important to break the cycle of sputum getting trapped which causes infection and inflammation. This in turn causes airway damage, which then leads to more sputum being trapped. Physiotherapy can help enormously to break this cycle, but lung damage can be irreversible. It is important to keep the chest clear which will reduce infection. Coughing is not enough, sputum needs help to move up. Plugging of the airways occurs if sputum is not cleared which can cause inflammation, pain, infection, breathlessness and wheeze. Physio helps by delaying lung damage, reducing wheeze, reducing the rattle of cough, and helps prevents infection and inflammation.
It was stressed that each patient is an individual, and that there are many different kinds of physiotherapy to suit each individual. There is: -

- Active Cycle of Breathing Technique (ACBT)

This method is flexible, and can be used with a lot of other techniques. It is easy to remember, and can be used in conjunction with postural drainage and blowing games etc.
The active cycle of breathing technique uses: -

- Breathing control
- Thoracic expansion
- Forced expiration/huffing

Breathing techniques for babies need to be passive, but blowing games can be used and huffing so that a mirror steams up.

- Postural Drainage (PD)

For this method the patient needs to be comfortable and lies on his/her side in a gravity assisted position. It can be done on a Physio table or pillows so that the patient lies in a tilted position which allows gravity to drain the lower lobe. If the patient lies further back then this drains the middle lobe. However this method should not be used if the patient has reflux problems. It can be used in conjunction with the ACBT technique.
The ACBT with PD techniques were demonstrated by Nicola. Thanks to Gregor for assisting with the demonstration.

- Autogenic Drainage

This method uses huffing but is much slower than ACBT. It is usually done in the seated position but can be done in the PD position as well. It is flexible and can be done at work.
Various devices can be used to increase the efficacy of physiotherapy, and can also help with compliance if there is reluctance to do it. Oscillatory and positive expiratory pressure(PEP) devices help mobilise and move secretions. They splint the airways, increasing the efficiency of mucus clearance. The flutter can really only be used in the sitting position because it needs to be held upright, but the PEP/acapella can be used in the postural drainage position or the seated position. PEPs can be used with ACBT. Acapellas are oscillatory devices which provide both resistance and vibration. It may be necessary to hold the cheeks to make sure that the vibration is in the lungs and not the cheeks. It varies from person to person as to what is best for that individual.
In PCD the sinuses become blocked and there are various devices that can be used to help clear the sinuses. These include a watering can type device, Sterimar and NeilMed sinus rinse.
Posture is very important, and research shows that impaired posture can impair lung function. Evidence also suggests that exercise is almost as effective as puffers. Trampolines are particularly effective. Activity is really important, and sport should be encouraged. It can be helpful to use a flutter before football.
Urinary/stress incontinence can result from so much coughing, even in youngsters, and as a result can discourage coughing. It is fairly easy to treat, and pelvic floor exercises can help
There are various aids to loosen mucus, thereby making it easier to cough it up. These are mucolytics and include hypertonic saline (i.e. salty water) which can be inhaled by nebuliser. This is not suitable for everyone, and the airways need to be checked for toleration. There is also Mucodyne and Dnase medication.
It was then explained what order is the most efficient for clearing the airways.

- Open airways (puffers)
- Use Mucolytics
- Airway clearance
- Steroid inhaler
- Inhaled antibiotics

The challenges for effective physiotherapy are adapting and tailoring techniques to age and circumstances e.g. transition experiences, adapting to work, relationships, pregnancy etc.
The following questions should be asked so that the patient knows they are doing what is right for them and what works for them: -

- Is my physiotherapy effective?
- Is my condition stable?
- Do my physiotherapy techniques fit with my life style?

Each patient should ask for a Physio review periodically.

- **The day closed with a question and answer session**

Questions were asked from the floor about swine flu. The advice given was to wash hands frequently, to use tissues and dispose of them promptly, and to avoid people with swine flu. Clare Hogg from the Royal Brompton Hospital is advising us on swine flu. If a PCD patient gets swine flu then the advice is to ring the consultant's secretary and tell them.
In answer to questions about sputum tests, it was explained that It takes five days to obtain sputum test results and that the respiratory nurse can be contacted to give an indication of the result. PCD patients sometimes need to huff for doctors to hear infection with a stethoscope.