PCD Day and AGM

Fiona Copeland introduced herself and welcomed everybody. She has been chairman for six years and has two boys with PCD. She is also the website manager and asked if anyone would like to take on this role.

The minutes of the last meeting on 29th June 2009 were agreed.

Achievements

- We participated in the Jeans for Genes campaign and generated lots of publicity for PCD
- We launched our new website and DVD
- Fiona spoke at the GIG conference (now the Genetic Alliance UK)
- We attended the BTS conference
- Fiona spoke to medical students at University College Hospital
- We helped to set up the Ciliopathy Alliance UK www.ciliopathyalliance.org.uk
- Fiona visited Luton and Dunstable Hospital an area where there is a high incidence of PCD
- Fiona is the Patient and Public Representative at the new gene therapy unit at the Royal Brompton Hospital
- We held the first PCD adult lunch in London which was a success and will be repeated

Financial Review

Mick Wilkin, Treasurer, introduced himself. He has been Treasurer for almost six years.

- Our cash reserves are down from £31k to £16k

- Fundraising donations amount to £7,729 whereas last year they were £13,641. This includes £5k from Jeans for Genes which is restricted to the website and DVD
- Interest received is £11
- Costs this year have risen to £22,773 from £13,558 last year
- £9,775 of the outlay is 50% of the payment for the multimedia resource
- £6,927 was paid out for medical equipment and grants. This includes audio equipment and acapellas for the Royal Brompton Hospital.
- £794 was paid in fundraising consultancy fees
- £1,193 was paid to fund the 2009 PCD Day and AGM
- The accounts were approved
- The independent examiners were re-appointed and it was agreed to continue with them as they do not charge us for this
- The economic climate is still difficult for everyone including charities
- We need to make an increased effort fundraise - can anyone help?
- We are looking at potential trust fund applications - can anyone help, particularly if experienced in this
- We are committed to a further £10k to pay for the multimedia resource and funds are running low
- We are a going concern for the foreseeable future, but we may have to refuse some funding requests for the moment

Plans for 2010/11

Fiona then addressed the meeting and outlined future plans:-

- To continue to support families
- To hold a PCD Awareness Day in Bradford in September 2010
- To continue to support the Jeans for Genes Campaign
- To attend the British Thoracic Society Conference
- To have a big fundraising push and raise awareness of PCD – currently Duathalon planned for September and Great South Run in October www.justgiving.com/rbht
- To develop relationships with the Ciliopathy Alliance
- To continue with the PCD Medical Board, ERS and the Genetic Alliance meetings
- To help the three diagnostic centres to obtain centralised funding for the ongoing care of paediatric patients
- To help improve the transition from paediatric to adult care
- To help to set the blue print for adult PCD care at the Brompton which can be used by other adult physicians throughout the UK

Fiona asked the audience to let us know if there was anything else that they would like us to do?

Election of Officers

- Chair - Fiona Copeland
- Secretary - Myra Tipping
- Treasurer - Mick Wilkin
- Adult Contact - Sylvie Prouse
- Child Contacts - Joanne Wilkin and Sarah Kirk
- Website Manager - Fiona Copeland
- Newsletter Editor - Myra Tipping
- Medical Advisory Board - Fiona Copeland
- Talks Co-ordinator
- Fundraising contact

The above officers were re-elected.

Fiona explained that we need a talks co-ordinator. Dr Jheeta volunteered to organise talks in the Midlands. We also need a fundraising contact as Gary is too busy at present to do this. Fiona asked if anyone had any specific skills that would be useful and explained that everyone is voluntary and asked for more volunteer committee members.

PCD Genetic Research Talk

Hannah Mitchison from the Institute of Children's Health at University College Hospital then gave a fascinating talk on PCD genetic research. She has worked in PCD research for ten years.

She explained that cilia are tiny microscopic hairs which line the cells in the nose and lungs and beat about 14 times per second. The function of the cilia is to move mucus and pathogens out of the lungs and they are a very important defence mechanism. There are motile cilia and flagella cilia. In PCD the respiratory system and the reproductive system are affected because of dysfunctional cilia. Laterality (ie when organs are reversed) occurs in 50% of PCD cases and is thought to be caused by defective fluid flow in the embryo. Normally this does not adversely affect the patient, but in about 6% of these cases there are complexities. Hydrocephalus can also be implicated in rare cases of PCD because cilia line the brain ventricles. Cilia also occur in the kidney and the eye and in rare cases can cause problems for PCD patients.

The trachea is the most densely ciliated part of the body. There are 200 to 300 cilia on top of each cell which beat in unison to sweep mucus and pathogens along. Each cell

consists of a nucleus, a cell body and a 'brush' of cilia at the cell surface. Normally, cilia beat and then have a recovery action and this is the process moves mucus from the lungs to be expelled. However, in PCD there are problems with this action. Cilia are made up of over 500 proteins and have a rigid tubular structure. Each respiratory cilia has a nine plus two arrangement of microtubules when seen in cross section with an electron microscope. There are two microtubules in the middle and 9 doublet microtubules around the edge. Attached to the outer doublet microtubules are the dynein arms. In most PCD families (over 80%) the outer dynein arm is absent and this causes the cilia to be immotile, since the motor protein that makes cilia beat is missing.

She explained that she is searching to identify the genes responsible for PCD and to understand the molecular abnormalities responsible for the disease. This will give understanding of how cilia are made and how they work. In PCD it is known that ten genes cause 17 - 38% of cases. Cystic Fibrosis patients in contrast have only one mutated gene. Six of the genes encode the structure of the cilia and cause the absence of part of the structure. In the last two years three genes have been discovered that preassemble the components needed for correct cilia structure within the cell, before they get taken up into the cilium by intraflagellar transport. In PCD it is known that more genes are involved and more genes need to be found. There is new technology coming called 'next-generation sequencing' that will make this easier since all the genes in the cell will be screened together to see if they have mutations that cause PCD.

Ciliopathy Alliance UK

There are other rare diseases caused by defects in the function or structure of cilia and a newly formed Ciliopathy Alliance UK is being launched with an Awareness Day on November 29th in London (www.ciliopathyalliance.org.uk). The idea of this alliance is to increase awareness, share knowledge and understanding, apply for research funding, encourage collaborative research and bank patient samples so that they can be part of genetic studies.

Researchers are looking for chromosomes that are similar in families to find gene defects. She explained that she is looking for families who would like to take part in this research by giving blood samples. The easiest way to donate a blood sample is at a hospital PCD clinic.

New Website and DVD Presentation

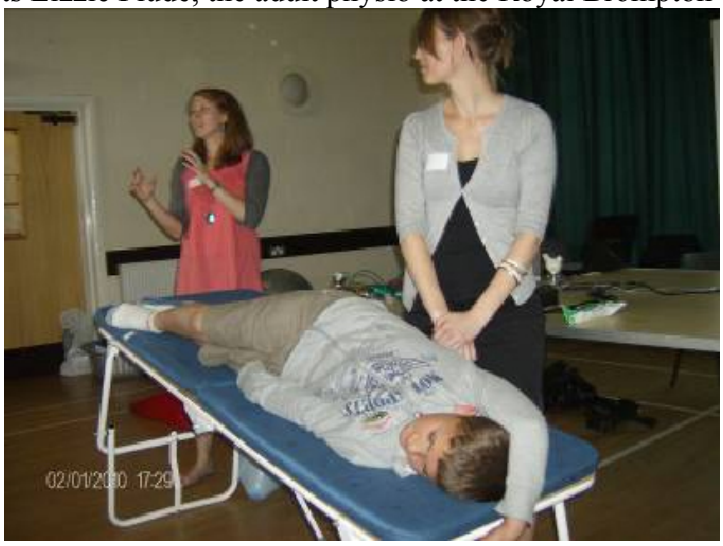
Fiona then presented the new website and DVD which are full of information and offer an opportunity to share best practices and experiences. She explained how the

website worked and how to access the case studies. She presented the physiotherapy page which recommends physio twice a day, with regular reviews, the importance of sport and becoming more responsible for your own treatment as you get older. She spoke about the medical team who care for PCD patients consisting of the GP, the consultants who may be local or may share care, and the respiratory nurse. She spoke about medication consisting of oral antibiotics, both prophylactic and for intervention, iv antibiotics, nebulisers (universal transformers can be obtained for foreign travel), inhalers and steroid treatment. She discussed holiday insurance which increases if there has been a hospital admittance or steroid treatment. She also encouraged and answered questions from the floor throughout explaining that the feedback from last year was to have more interactive sessions.

We then all had a delicious lunch. The atmosphere was very good and relaxed with everyone mingling and chatting and about PCD!

Physiotherapy Talk

After lunch the physiotherapists Lizzie Flude, the adult physio at the Royal Brompton Hospital, and Jo Keating, the paediatric physio at the same hospital gave us a really interesting talk and explained why we need to do physio and demonstrated various techniques and equipment.



They explained that we have two lungs, the right lung has three lobes and the left lung has two lobes. These are lined with cilia which move mucus and sweep in a straight and then recovery phase thus moving mucus out of the lungs. In PCD because of defective cilia movement this mucus gets trapped and a vicious cycle occurs. When sputum gets trapped it leads to infection, which then leads to inflammation and airway damage, which in turn leads to more mucus being trapped. Physio is an essential part of the tool kit of treatment. On the CT scan thick airway walls indicate inflammation. When lung damage (bronchiectasis) occurs the airways get bigger and have thick walls showing permanent damage which in turn trap more mucus. Bronchiectasis is not reversible but can be managed.

They showed us a film of a PCD patient having a bronchcoscopy which showed the sputum clogging up, blocking and inflaming the airways. It presented a very powerful argument for the importance of physio to get the sputum out of the lungs.

They explained that physio is essential to keep the chest clear and helps to reduce infection. Coughing alone is not enough because sputum needs help to move up the airways. Sputum can plug an airway and if it is not cleared and can lead to

inflammation, pain, infection, breathlessness and wheeze. It is very important to keep the airways clean and clear. Physio helps by getting rid of sputum, prevents inflammation and infection and prevents/delays lung disease. It improves how much air you can breathe in, the ability to exercise and reduces wheeze.

They then explained the following:-

The Active Cycle of Breathing Technique (ACBT)

- It is flexible, can be used with a lot of other techniques, is easy to remember, and can be used with postural drainage.
- Blowing games can also be used.
- Research has proved its efficacy.

Postural Drainage (PD)

- This involves gravity assistance and positioning is very important.
- It should not be done if there are problems with reflux.
- It can be done over a lap, with pillows or on a bed.
- It can be used with other techniques to make it more effective.
- PCD patients can really benefit from tipping. The tip should be at a 30 degree angle.
- Lying on one side drains the lower lobe and then changing the position still on the side but slightly towards the front drains the middle lobe.
- PCD tends to affect the middle and lower lobes rather than the top of the chest as in Cystic Fibrosis.
- The patient needs to be comfortable.
- Deep breathing and huffing is important.
- The forced expiratory technique is a huff.

The Active Cycle of Breathing Technique (ACBT) consists of:

- Normal breathing - breathe from tummy through the nose if possible
- Thoracic expansion ie deep breathing through the nose and out through the mouth. When the lungs are full, keep going for another couple of seconds. You might blow out through an acapella. A number of these should be done.
- Normal breathing ie breathing control.
- Huff (ie forced expiratory technique). Use tummy and chest muscles for this, but it should be fairly gentle. Do a couple of huffs and rest after each one.

Multiple cycles per physio session should be used. It should be done about four times lying on the left side and four times on the right side.

Percussion (chest clapping/shakes) can also be used. It helps to unstick mucus and can be used with postural drainage positioning, ACBT and blowing games. Cupped hands are used for percussion. The clap should be a hollow sound and not a slap. A towel or blanket can be used to make clapping more comfortable. The patient breathes deeply whilst being clapped. The patient then breathes out whilst a shake is

done. Four deep breaths are taken with clapping followed by four deep breaths with shakes and then normal breathing.

These physio techniques were demonstrated on a very willing volunteer boy with PCD.

Lizzie and Jo then showed us oscillatory and PEP devices which are also effective. They help mobilise secretions, can help with compliance and splint the airways open. The PEP and the acapella can be used in PD positions.

The flutter has a metal ball in it to create resistance and is used in sitting positions because it has to be used upright. The PEP creates resistance but not wobble and can have a manometer attached to it. It can splint the airways and is good if the patient is wheezy.

The acapella causes vibration which makes the chest "wobble" as you blow out. We were also shown a quake device which causes quite strong vibration and has a handle on it which has to be turned whilst breathing out.

Information on how physio needs to change through the years.

In the newborn physio techniques are more limited. Chest clapping and postural drainage can be used but not shakes. Physio should be done one hour after a feed with patting for 30 seconds and then a short break. Physio techniques should be taught to other family members so that they can help too. Exercises can also be done with babies.

With toddlers (2 - 4 years) blowing games like tissue football using straws are useful. Chest clapping and postural drainage can be introduced as well as shakes as the child is blowing out. Songs, games and rewards are important too. If the child is not very compliant then the physio can be broken down into smaller, more frequent sessions.

When the child is between 4 and 7 years old then the above techniques in addition to huffing and adjuncts are helpful.

During the transition to being an adult, increasing independence should be encouraged so that the patients can clear their chests themselves. This should be introduced gradually eg maybe independent sessions at weekends to begin with. Physio techniques need to be adapted to university and work, and to fit in with relationships.

Physio techniques also need to be reviewed in pregnancy. During pregnancy some medications also have to change as well as physio. It is important to establish routine and take exercise. Physio after birth also needs adapting.

No particular physio technique is better than another - it needs to suit the individual. Physio needs to be part of your routine.

Exercise is important as it mobilises secretions, improves cardiovascular fitness, improves body image, slows decline of lung function, reduces sensation of breathlessness and maintains bone mineral density. It enhances quality of life and increases muscle strength. Exercise for babies and young children is also important eg play row the boat etc. For older children gym balls and Wii fit are very good. Exercise testing is done at the Royal Brompton Hospital.

Mucolytics can help to make physio easier and are normally used before physio. Hypertonic saline can thin mucus and enhance motility. The airway surface liquid helps mucus to slide. Other mucolytics like DNase can also help by making the mucus thin but it is unsuitable for some patients. The order should be to open up the airways, use mucolytics, clear the airways, use a steroid inhaler and then use inhaled antibiotics.

It is also important to keep the nose and sinuses clear and there are various sinus rinse devices which can be helpful. Antibiotics can also be nebulised up the nose.

The most common question asked to physios is "how do I know if I am doing it properly?" If you are using correct techniques then the physio should be effective, with reduced infections and no unwanted symptoms. It should fit with your lifestyle and you should have regular physio reviews.

Gym Ball Exercise Demonstration

Glenda, the paediatric physio from Milton Keynes hospital and PCD committee member, then gave us an interesting talk and fun demonstration of gym ball exercises.

She explained that cardiovascular exercise is beneficial and that the gym ball can be used at any age and is adaptable for any level of fitness. It is also used by top athletes. It is easy to transport, is a reminder to exercise, is fun and anyone can use it. It has an unstable base and

therefore exercises and strengthens core muscles by training the deeper abdominal muscles. It trains specific muscle groups whilst balancing, and therefore tones the whole body and is excellent for stretching and flexibility. It improves posture and balance and is particularly good for PCD because it exercises the thoracic section of the body.

She explained that in chest conditions there is a tendency for pronated shoulders and reduced mobility of the thoracic spine and that the gym ball could be used for stretching of both the shoulder girdle and the thoracic spine. It can also be used for massage, chest physio and diaphragmatic breathing practise. She explained that a mirror should be used to check posture/positions and also an exercise mat to provide an anti slip surface and cushion the knees. Hand weights can be used for the upper body suitable to enable 12 to 16 repetitions.

She recommended starting without weights though and building up gradually. However weights are not recommended in pregnancy or with back/neck injuries. A smooth and steady movement should be used. When using the gym ball she explained you shouldn't hold your breath and should stay within your limits, building up gradually. Stop if you are short of breath, dizzy or have pain.

Make sure you have enough space to use it and seek your doctor's advice if you are concerned. There are different ball sizes for different heights so ensure that you use one that is suitable for your own height (size 45cm for under 4'11, 55cm for 4'11 to 5'4, 65cm for 5'5 to 5'11) Make sure you buy one that is anti burst which ensures that if there is a puncture, it will deflate slowly. They cost about £12 - £15.

She then demonstrated how to use the gym ball with lots of very willing volunteers. She explained that you should sit on the ball with good sitting posture and a natural curve in the spine and then demonstrated various exercises and games. Our volunteers certainly enjoyed it!