

PCD Day June 2012

PCD Day was a great success, with lots of families travelling from various parts of the country to attend this enjoyable and informative event. The children were entertained by table football and arts and crafts activities.



Glenda Dalton held physio workshops, one for those under eight years old, and also a separate one for those over eight.

Meanwhile Fiona welcomed everyone and introduced the first speaker, Maya Waldman, an enthusiastic and inspirational singing teacher who works at the Royal Brompton and Harefield Hospitals.



Singing workshops were established there in 2007 and Maya has been involved for the last three and a half years. She demonstrated singing with postural and breathing exercises in a fun way, and promoted a positive association between the voice and the breath.

She demonstrated diaphragmatic breathing and singing techniques and explained the benefits for people with lung problems. This was a lively and enjoyable interactive session with everyone singing and taking part. A CD with singing and therapeutic exercises is available for £10 and through the hospital website (www.rbhcharity.org/gen/shop/)



Next was a lively and stimulating question and answer session. Beryl Adler, Consultant Paediatrician, Nicki Keech, Physiotherapist, Liz Meleady, Community Respiratory Nurse all from Luton and Dunstable General Hospital and Mustafa Munye, PCD Researcher at the Institute of Child Health, formed an impromptu panel.



They answered a wide range of questions from how to explain to family and friends what PCD is, to antibiotic use and possible resistance, pseudomonas, physiotherapy and appropriate devices. This was followed by a delicious lunch when everyone mingled and chatted to each other, exchanging experiences and tips about PCD.



Beryl Adler, Consultant Pediatrician, from Luton and Dunstable Hospital then gave a very interesting talk “Managing PCD in your local hospital”. She explained the history of the discovery of PCD. It was first observed in 1904 and in 1933 Kartagener’s syndrome was discovered. In the 1980s electron microscopy technology enabled PCD to be more readily diagnosed, with about one in 15,000 of the population being affected.

However, with more diagnosis now, PCD is expected to be found more prevalent than cystic fibrosis within the next five years. She explained that cilia are in the large airways, sinuses, middle ear and back of the nose. The impaired movement of cilia cause poor clearance of secretions and mucus, and disease of the upper and lower respiratory tract. Nodal cilia are implicated in the formation and position of the heart and other organs and can cause dextrocardia and organ reversal. There are sensory non motile cilia in the eyes, kidney, liver

and the drainage system in the brain which may explain some rare disease associations.

The symptoms of PCD are:-

- Unexplained newborn respiratory distress
- Recurrent unusual wet chest infections
- Dextrocardia
- Complex cardiology
- ENT problems, glue ear, hearing problems
- Persistent nose sinus
- Poor response to usual treatment
- Atypical asthma
- Unusually severe upper airway disease
- Rattly wet cough
- Bronchiectasis

Family history is very helpful. Several genes are implicated in PCD and it is an autosomal recessive disease with genes being implicated from both parents. The manifestation of PCD is different in each family but is the same at a molecular level.

Diagnosis needs to be made with the above symptoms by referral for nasal brush biopsy and infections need to be treated. Nasal nitric oxide is also measured and sometimes a saccharine test is also carried out. This diagnosis process can take as long as 16 weeks.

She then went on to explain how local care for PCD works, with respiratory clinics, dieticians, community teams etc. She also talked about transition to the adult clinic.

She explained the usefulness of hypertonic saline and how it breaks down secretions making them less sticky. She discussed intravenous antibiotics and pic lines and emphasised the importance of life long physiotherapy and exercise. Regular cough swabs and sputum tests should be taken to identify infections and suitable treatments. She recommended flu and pneumonia vaccinations.



Nicky Keech (physiotherapist) and Liz Meleady (community respiratory nurse) also addressed the meeting and explained their roles in PCD and how the support team works together to help patients.

Beryl Adler went on to say that she has 20 pediatric patients at Luton and Dunstable Hospital, with 17 being diagnosed within the last seven years. Five families have two to three children with PCD. The patients are mostly Bangladeshi and Pakistani. She explained that PCD is a learning treasure chest and emphasised the importance of finding out about the condition and its management.

Fiona then chaired the AGM, as she has done for the last seven years and the minutes were agreed. The achievements this year include:-

- The Cilia 2012 Conference which was held with 250 delegates talking about cilia from around the world.
- The BTS Conference where a stand was manned for PCD.
- The Luton and Dunstable PCD evening
- The Bradnet event for Rare Disease Day in Bradford for 40 patients
- Leicester Royal Infirmary Awareness Session where there is a high Asian population
- Talking to medical students at UCL
- Participating in the “Quality of Life Survey” for Southampton General Hospital
- LCI research at the Royal Brompton Hospital
- Working with others re cardiac surgery and the review at the Royal Brompton Hospital
- Obtaining PCD Pediatric funding.
- Online awareness: The website had 31,000 hits between 2011 and 2012. We also have a presence on Twitter and Facebook.
- Fund Raising: £1,000 was raised at the Stony Stratford AHA event. Debbie Richards selling Angel necklaces has also raised £1000. A zumbathon raised £700 and coffee mornings raised £390. The Richmond half marathon and the Great South Run also raised funds, as well as birthday parties. Graham Collins ran the London Marathon and raised £1,000.



- Purchased physiotherapy equipment for hospitals and individuals
- Alex Kupse and Justine Currie have both been nominated to carry the Olympic Torch

Plans for 2012/13

- Working with NHS, RBHT re changes to the hospital services. We don't want to see the Royal Brompton Hospital skills dissipated.
- Helping to implement the new PCD Management Service
- Helping to develop transition and the adult service
- Ciliopathy Alliance Patient Conference, Saturday, 15th June 2013. Four families can go free with an overnight stay included
- BTS Conference - Rare Disease Meeting December

Mick Wilkin, Treasurer, presented his Financial Review. He explained that the cash reserves are up to £17k from £12k last year. Fund raising was £13,404 which was similar to last year. This included a grant from Jeans for Genes of £1500 for PCD Day.

The accounts were approved and the Independent Examiners, Dove Naish, were reappointed. They do not charge us for what they do.

Mick explained that the financial outlook for 2012 in the current economic climate is still difficult and said that an increased fundraising effort was needed. He asked for help with researching potential trusts or big companies who may donate to us. The annual running costs of current plans are around 11k. We have restocked marketing materials this year. We are a going concern for the foreseeable future.



Fiona told us about Hannah Mitchison's genetic research. Lots of new genes are being discovered that are implicated in PCD. Blood samples are needed from patients with inner dynein arm defects to facilitate research. The Royal Brompton Hospital are looking for small children to help with research. We have done a quality of life survey, but we need to do another one. Fiona asked for people to participate promptly in this because it is needed soon. Volunteers are also needed for research with mucociliary clearance at Southampton. They will be paid £100 each, but it does involve a two day trip.

Fiona told us that Darren and Tracy Anderson have resigned from the committee and asked for new members. Fiona asked for volunteers to attend meetings in London, and for a fundraising co-ordinator. She also asked for volunteers who would give talks, and also for help in getting the website validated with a kite mark. She asked if anyone could help Myra with the secretary's role for the next six months because Myra has family problems at the moment. Justine Currie volunteered to help if needed.

Officers were re-elected:- Chairman - Fiona Copeland Secretary - Myra Tipping Treasurer - Mick Wilkin Adult Contact - Sylvie Prouse Child Contacts - Sarah Kirk and Joanne Wilkin Other re-elected members of the committee are:- Justine Currie, Glenda Dalton, Gary Tipping, Terry Irwin.

AOB Fiona asked everyone to fill in the feedback form. The day always provides further insights into PCD, whilst at the same time being enjoyable and interesting. The children had a great day too and took their art work home, including fascinating pictures of their perception of living with PCD.

