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NORTH LONDON Adult Haemophilia Network

Haemophilia **link**

Newsletter for patients, children and their families with Inherited Bleeding Disorders

Welcome to the first edition of the 'Haemophilia Link' – an annual newsletter for patients, children and their families with Inherited Bleeding Disorders who are registered at Haemophilia Centres in the North London Adult Haemophilia Network.

The Network is made up of the Royal Free London Hospital Comprehensive Care Centre (CCC), the Royal London Hospital CCC and Hammersmith Hospital Haemophilia Centre. The Network was launched in April 2011 to share and develop staff expertise and experience to improve the quality and efficiency of services with the overall aim of improving patient experience.

The Network haemophilia teams are working together on a number of different projects covering all aspects of patient care. Nursing and Allied Healthcare Professionals such as Physiotherapists and Psychologists meet monthly and are developing plans for enhancing community care and links with District General Hospitals as well as discussing how to improve access to services such as physiotherapy and counselling. They are also working with Great Ormond Street CCC to look at how to further support teenagers as they move their care to adult centres.

Other areas that the Network centres are developing together include clinical guidelines for patient and service management, audit and research, Network Multi-disciplinary team meetings (known as MDTs) and work on developing patient information, haemophilia centre websites and patient education and social events.

World Haemophilia Day - 17th April 2013

For world haemophilia day 2013, the teams at the Royal Free and the Royal London Hospital Haemophilia Centres hosted a number of events.

The Royal Free team held an information stand to raise awareness of inherited bleeding disorders. The team made cakes and raised over £55 for the Haemophilia Society which goes towards services such as the Haemophilia Society Helpline and a children's activity weekend. The Royal London team ran an awareness campaign on educating visitors and clinical haematology patients visiting the Haematology Day Unit on the diagnosis and treatment of Haemophilia care.

Interview with **Nilanthi Karawitage**

(Senior Biomedical Scientist at Hammersmith Hospital)

"I find the work interesting and challenging"

Tell me what a typical day is like working in the laboratory?

I start at 07.30 and check outstanding work and plan for the day ahead. I need to ensure work is performed to the highest level and to teach and train staff.

No two days are the same. I really enjoy the wide range of tests performed in the special coagulation section. I enjoy setting up and evaluating new methods and new machines.

What made you want to work in a Haemophilia Laboratory?

I did a biomedical science degree. Over the years I became more fascinated with coagulation. Coagulation is a complicated biochemical pathway. I find the work interesting and challenging, as well as enjoying laboratory work. We have weekly coagulation meetings to discuss results with the coagulation consultants and the team.

I also get pleasure from teaching and training staff as well as evaluating new methodologies and machines.

What is the best thing about working in a laboratory?

I enjoy working with people who share the same fascination for coagulation as I do. We regularly meet with the Haemophilia Team and I get immense satisfaction combining the clinical details with laboratory results.

What keeps you busy outside of work?

Yoga, meditation, gardening; I enjoy socialising and entertaining friends. I do charity work at my local Buddhist temple and enjoy Buddhist retreats.

I have a mild/moderate bleeding disorder. Why do I need annual follow-up with my Haemophilia centre?

Whilst many patients have milder bleeding disorders and do not have problems day to day, we still need to have some contact on at least an annual basis.

This may be as a face to face appointment or in some centres in the network, could be done as a telephone clinic (please contact your centre to discuss this option further).

Reasons to keep in regular contact with your Haemophilia team

1. Update on the background of your bleeding disorder, genetics and any new treatment options
2. Update the team on any new medications you may be taking or new medical issues you may have
3. Reminder of how to contact the team (during normal working hours and out of hours)
4. Update the team with your contact information
5. Reminder of medications to avoid
6. You may need testing of your blood levels again or follow up of other investigations
7. An opportunity to ensure all members of your family have been tested as appropriate

Margaret's story

"What I have learnt is to always make the effort to get out and do things and spend time with your friends."

Margaret talks about living with Type 3 von Willebrand's disease. This is the most severe and rare form of the condition. Margaret is 72 and lives with her husband Frank.

Tell us about your early memories and experiences of hospitals?

It was a long while before they knew what was wrong with me and I wasn't diagnosed until I was 20. Before that the main problems I had were bleeding into the ankles.

Right from when I was born my mother knew that something was wrong. When I was 3 I ran into the kitchen and caught my mouth on the corner of a cabinet and the bleeding wouldn't stop. If I grazed my knee it would bleed for ages. I was sent to various hospitals and nobody could tell her what was wrong. I think they told her it was like haemophilia – but girls didn't get haemophilia.

I spent a lot of time in hospital; I've got memories of lots of doctors who were all discussing me and were very interested. I remember being very young and bleeding and having my arms tied to the sides of a cot because I was having a transfusion. I then had some teeth taken out and we didn't want them taken out because my mother knew I would bleed. I was in for 2 weeks after that because the bleeding wouldn't stop and there was no treatment.

What about school days?

I remember being pushed to school in a pushchair when I couldn't walk because the ankles were so bad. My ankles were damaged by more bleeds into the joints over time. The movement became less and they became fixed. I wore elastic supports. I missed a lot of school and did not pass my eleven-plus. I never did PE or games and always had to sit out. My father taught me to swim and I continued swimming for many years. There was a good technical school though and I went to the needlework school there as I'd always been good at needlework.

How did you finally get diagnosed with von Willebrands disease?

We were always going to different hospitals and they all said it was haemophilia. My dentist got so fed up with the bleeding problems that he referred me to the Eastman Dental Hospital. They took blood tests and did a bleeding time with the blotting paper. I knew that it wasn't normal and I told them "you will sit there for ever" waiting for the bleeding to stop – they gave up after 45 minutes. Within 24 hours of the appointment at the Eastman I got a telegram from Dr Dormandy asking me to go to Great Ormond Street Hospital to see her. Within hours it was diagnosed. We had never heard of von Willebrands disease until we were told. I was a bit overawed with it all

Once we knew what it was, I knew what to do when I had a bleed. I would ring up and then go to the caravan by the old Royal Free. What sticks in my mind is the bags of plasma that had to be put in a trough of warm water. And it worked – no tablets had been able to take the pain away but this worked.

What about having children?

I'd always been told "no children" by doctors. But when I was 25 we had our daughter. We had discussed this for quite a while with Dr Dormandy and she said it was fine to go ahead. It was all very well worked out and planned so that everyone knew to phone straight away when I went into labour. I didn't have any bleeding problems during my pregnancy. During labour and afterwards they treated me regularly with cryoprecipitate for about two weeks. We didn't have the Tranexamic Acid then so I didn't get that as well. All along we had been told that there was a good chance that our daughter would be affected by the von-Willebrands and they tested her within the first two days. They told us that she had a milder form than me. She was a bit of a tomboy so there were times when she was growing up when she would bleed like when she fell off the garden wall. But her bleeding would stop of its own accord.

How has von Willebrands disease affected you in later adult years?

I was on the pill for about twelve years and I was told I should come off because this was a time when there were a lot of concerns about being on the pill for too long. This meant I went back to having bad periods and when I was 39 I begged the gynaecologist to do a hysterectomy. They were very reluctant but I was clear that this was what I wanted. Once it was done, I felt I could really start living.

My problems have really come as I've got older and my joints have got worse. I had a knee replacement recently and can walk now without discomfort.

With your lifetime of experience, is there anything you wish you had known and understood when you were younger?

I wish they could have diagnosed it earlier. To go all those years and not to know what was wrong, when you know something was wrong, was hard.

We had our wedding anniversary last year. Frank is a very tolerant man and has always been very good with all my problems. We go out and go to the bowls club and have holidays away together. I always plan well for holidays – make sure I have the Haemate P and the right insurance. Its important to be totally honest with the insurance company so you know everything is covered if you get a problem. It makes the premiums high but you shop around.

What I have learnt is to always make the effort to get out and do things and spend time with your friends. You need to be involved and with people. We had people round yesterday and I'm tired today but it was brilliant and I'm glad I did it.

Gene Therapy For Haemophilia B

Since the isolation and characterisation of the genes responsible for the production of factor VIII (8) and factor IX (9) some 30 years ago, a longstanding goal has been the development of successful gene therapy for Haemophilia.

Research led by Professor Amit Nathwani, Director of the Haemophilia Centre at the Royal Free shows encouraging results of patients treated with Haemophilia B (factor IX (9) deficiency) with gene therapy. The findings were published in the New England Journal of Medicine.

If a mutated gene causes a necessary protein, in this case factor IX, to be faulty or missing, gene therapy aims to introduce a normal copy of the gene to restore the function of the protein. A gene that is inserted directly into a cell usually does not function. Instead, a carrier called a vector is genetically engineered to deliver the gene.

How is the gene transferred?

The vector or "transfer agent" is the means by which functioning factor IX gene is transferred into the cells of the person with Haemophilia so that factor IX can be made.

Why did we choose the vector to be used in this trial?

Many different viruses that affect mammals have been modified into vectors, to carry genetic material, and each has strengths and weaknesses. The class of virus called Adeno-Associated Virus (AAV) was chosen as the vector since it does not cause disease in humans. As the factor IX gene is contained in a version of the virus that homes to the liver (AAV8) it was possible to administer the vector by infusion into a limb vein.

All the patients who volunteered had severe haemophilia B with a factor IX of less than 1%. A single treatment was given as an intravenous infusion over one hour. None of the participants had any side effects from the infusion itself. Two patients who had the highest dose each had a short period where their liver enzymes were increased, suggesting inflammation of

the liver. The liver inflammation got better following a short course of steroids. Currently the factor IX baseline level in the first 6 patients is between 1% and 6%. The follow up involving blood tests and physical examination will continue at least annually for 15 years.

Having taken the decision to participate following a demanding consent process with a lot of information to grasp, the participants have all reported that they were glad they had gone ahead. Even modest increases in factor IX can dramatically improve quality of life and reduce bleeding episodes. The patient experience is therefore changed from that of living with severe Haemophilia to that of moderate or mild Haemophilia where bleeding generally only occurs following trauma or surgery.

In summary, this gene therapy trial is the first to achieve long-term production of a clotting factor protein at levels that make a significant difference to the condition. All participants have been able to reduce or eliminate the need for regular factor IX infusion. The research team also developing a vector for factor VIII, in the hope of starting a clinical trial this year.

For further information please contact Professor Amit Nathwani at the Royal Free Haemophilia Centre on 020 7830 2068

Why is a Physiotherapy assessment an important part of your Haemophilia review?

Physiotherapists are professionals who are concerned with skeletal function and maximising its potential. In Haemophilia, bleeding episodes into joints and muscles can affect the health of that joint, as well as alter how the body moves. It is important to have at least a yearly review with the physiotherapist to see if there are any small changes happening that you may not be noticing.

The physiotherapy assessment looks at your 'musculoskeletal health' –the movement, strength and function of your arms, legs and body as a whole. This will include a review of your elbows, knees and ankles, and sometimes your shoulders and hips. We look at how you walk, discuss with you any problems you have with activities at home and also if you have any pain.

The results of your assessment are recorded as a 'joint score'. We review this each time you come to see us to ensure that your joints are remaining well.

The assessment may highlight problems that require input from the physiotherapist in the form of 'rehabilitation'. Rehabilitation can help with muscle weakness, pain management, improving mobility and balance and provide better support for joints.

Patient Evening

We will be holding a patient evening on the evening of Wednesday 12th March 2014 at the Society of Medicine, Wimpole Street W1. This event, for those aged 18 and over, will include presentations on the history of haemophilia, new treatments and gene therapy and includes information stalls supported by our staff on areas such as ageing, rare bleeding disorders, physiotherapy and women's bleeding disorders. Refreshments will be available.

Please see posters and leaflets in your local centre for information on how to book a place on the evening.

Contact Details:

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Haemtrack

Haemtrack is an electronic therapy support service for people with bleeding disorders who are receiving home treatment. It is a convenient electronic web-based system that can be used to record product usage and replaces paper treatment sheets. It is free of charge for patients registered at Haemophilia Centres. Haemtrack has been a useful tool in monitoring patient's bleeds, treatment regimes and tracking treatment batches.

The North London Adult Haemophilia Network have been using Haemtrack since it was launched in 2011. More than 150 patients are using Haemtrack across the 3 sites.

Here is some of the feedback from Haemtrack users:

"I have found Haemtrack quick and easy to use; it takes about 2 minutes to enter a treatment".

"The Haemtrack app has made it easier for me to record my treatment".

"Haemtrack is practical, time saving and easy to access from these aspects; I can rate Haemtrack 100/100".

To register with Haemtrack online at https://apps.mdsas.nhs.uk/haemtrack/.

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