Great Ormond Street Hospital for Children
NHS Trust

Musclemania

Autumn 2011 Issue 6

Clinical trials

Turn over to find out more.

Welcome
to our newsletter

This is a newsletter for parents and young people who come to the neuromuscular clinic. Its aim is to help keep you informed about the service at Great Ormond Street Hospital, to give you useful information and to respond with what you want to see in the newsletter.

The title of the magazine was chosen by one of the young people coming to the clinic.

Martin Chainani,
Editor
Neuromuscular Care Advisor

Next issue: Winter 2012

Front cover: Patient Amelia on her trike.

Who's who?
Newcomers to the Neuromuscular service

Dr Ros Quinlivan
Transition Consultant
Ros has been a Consultant in Neuromuscular Diseases for 16 years, prior to that her post graduate training had been at Guy's Hospital. She moved back to London to join the Neuromuscular teams here at Great Ormond Street Hospital and The MRC Centre for Neuromuscular Diseases, National Hospital, Queen Square. Previously she was the clinical lead for the Wolfson Neuromuscular Centre in Oswestry and Birmingham Children's Hospital. She still lives in Shropshire and commutes to London on a weekly basis. In her spare time she likes to walk in the Shropshire Hills and play golf (although claims she is not very good at it!).

Katie Finnegan
Senior Physiotherapist
Katie trained at Brighton University and has been here at Great Ormond Street Hospital for about three years. She previously worked at special needs schools and a variety of acute paediatric settings. She likes windsurfing, cycling, ballroom dancing and adventure holidays. I felt exhausted just talking to her!

Dr Ann Mathew
Clinical Fellow in Neurology
Ann is with us for the next 12 months and did her medical training in Bangalore, India. She did her Paediatrics training mostly in Scotland and has been doing her Neurology training here. Ann wants to go back to India after her training and provide a service for children with neuromuscular conditions. She loves cooking and sings in a church choir.

Martina Andrews-Rouckova
Physiotherapy Assistant
Martina trained as a qualified Physiotherapist in the Czech Republic and moved to the UK in 2007. As her registration is not valid here, Martina first worked in a variety of caring jobs including as a Healthcare Assistant here at Great Ormond Street Hospital. She hopes to get her registration in the UK in the future so that she can work as a qualified Physiotherapist. Martina is a sporty person and swam competitively at National level in the Czech Republic. She also loves in-line skating and ice skating but finds the ice rinks here too small compared to those back home!

If you have any ideas for next issues, any comments or if you would like to share your story please get in touch...

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What's new?

New Neuromuscular Care Advisors

Following a lot of hard work by the Muscular Dystrophy Campaign, there are many new Neuromuscular Care Advisors around the country employed within the NHS. I have also benefited from this as I am now employed directly by Great Ormond Street Hospital. Particularly relevant to us are the Care Advisors in the East and South West of England. These are areas where traditionally a lot of patients have come to us because of a shortage of Neuromuscular services in those areas.

However there are still some areas where there is campaigning ongoing. For more information or if you want to get involved, you can contact the Muscular Dystrophy Campaign on 0207 803 4800 or check their website at **www.muscular-dystrophy.org**

Information

I am busy at the moment looking at the information that is provided in clinic and to schools. So be prepared I may be coming at you with a questionnaire in the near future! At the moment there is an information cabinet in the clinic area which I regularly keep stocked up with information and we have a section on the GOSH website: **www.gosh.nhs.uk/gosh/clinicalservices/neuromuscular_services/homepage**

The Muscular Dystrophy Campaign and Contact A Family provide fantastic information and you can get in touch with Contact A Family on freephone 0808 808 3555 or visit their website at **www.cafamily.org.uk**

Neurobehavioural phenotypes of Duchene

Recently we have been focussing on the behavioural problems that some boys with Duchenne can have because we have seen more and more how much these can affect family life and schooling. This is backed up by research in this country and in Italy. We are doing an audit over a 15 month period by giving out questionnaires to parents and teachers of boys to find out how often the problems occur and to understand them more. Our goal is to be able to make recommendations for future assessment and management of behavioural difficulties and make referrals to other services.

Read more on Duchenne muscular dystrophy on the back page.

Clinical trials

Feature article

GSK2402968

This drug is being studied to see how effective it is in producing dystrophin (the lack of this protein causes the muscle weakness) in boys with Duchenne muscular dystrophy with certain deletions of the XP21 gene, the genetic fault that is the cause of the disease. This is done by skipping exon 51 of the gene. GSK2402968 or a placebo are being given to five boys at our centre for a period of 48 weeks. It is part of a worldwide study involving 180 boys in 18 countries. Recruitment is now closed.

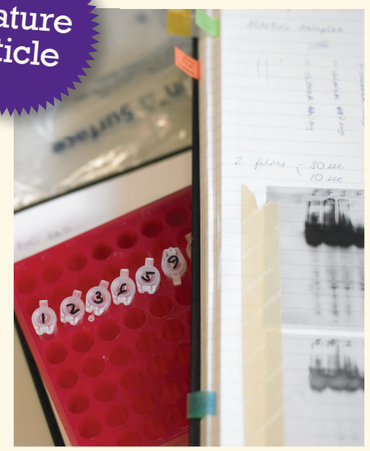
You can find out more information at **www.prosensa.eu/press-release/gsk-and-prosensa-announce-start-phase-iii-study-investigational-dmd-medication**

Olesoxime (Trophos)

This drug is being assessed to check the safety and effectiveness of it over a two year period in patients with Spinal Muscular Atrophy types 2 and 3, aged from three to 25 who are not walking. This trial has started in several European countries, (25 hospitals in France, Germany, Italy, Poland, Belgium and Netherland and United Kingdom). In the UK the study will be here at GOSH, and also in Newcastle and Birmingham. In England there will be 19 patients involved (10 in London). Recruitment has now closed. You can find out more information at **http://www.treat-nmd.eu/sma/overview/**

AVI-4658

The study of this drug that was recently finished, was also looking at a drug that aims to produce dystrophin in boys with Duchenne muscular dystrophy with certain deletions of the dystrophin gene, again starting with skipping Exon 51, because this is the one that has the potential to benefit the greater amount of boys. There were some promising results and you can find out more information about this and the other trials at the following website **www.mdex.org.uk**



Find out more

There are lots of other clinical trials going on in other places for many other muscle conditions and you can find out about these on the Muscular Dystrophy Campaign website at the following address **http://www.muscular-dystrophy.org/research/clinical_trials**

First targeted treatment success for Duchenne muscular dystrophy

A team led by scientists at the UCL Institute of Child Health (ICH), funded by the Medical Research Council (MRC) and AVI BioPharma, have made an important breakthrough in the development of a treatment for Duchenne muscular dystrophy (DMD).

Together with the MDEX Consortium, chaired by the ICH's ProfessorFrancesco Muntoni, the group showed that a gene-based drug treatment was effective in restoring the dystrophin protein that is missing in sufferers of DMD, in seven out of 19 trial participants.

DMD is a devastating and life-limiting condition that affects one in 3,500 male births in the general population, with around 100 cases diagnosed in the UK each year.

Three of the participants in the two highest dose cohorts showed dystrophin levels that exceeded 18 per cent of those found in normal muscle cells. There was significant statistical increase across the cohorts.

Thirteen per cent of boys with DMD could be treated with this gene specific, exon-skipping therapy, the largest group by a single antisense. Overall scientists say this approach could work for at least 70 per cent of DMD sufferers. DMD causes progressive muscle weakness due to the breakdown and loss of muscle cells. Patients lack a single important protein in their muscle fibres called dystrophin.

In this clinical trial of 19 patients, study participants aged five to 15 at Great Ormond Street Hospital and the Royal Victoria Infirmary, Newcastle, were given weekly doses of the drug, AVI-4658. The drug had already been tested for safety and efficacy by the MDEX Consortium and AVI Biopharma in an earlier phase of the study (Kinali et al, Lancet Neurol 2009).

Brothers Jack, 11, and Tom Bosanquet, 8, were enrolled on the trial. Both have DMD with a deletion from exons 45-50. Their mum, Claire, said:

"The diagnosis of DMD came as a complete shock, neither me nor my husband Ian had heard of the condition before. Receiving the diagnosis was like falling into a black hole, you don't know how you will cope and you really feel like your whole world will fall apart.

"Jack and Tom were placed on a DMD genetic registry, co-ordinated by Action Duchenne, which is how we were approached about the clinical trial at Great Ormond Street Hospital. Enrolling was a no brainer for us, we felt from the outset that by taking part we were getting some control over something which for so long had been completely out of our hands. We felt, at last, we could do something positive about something negative.

"Coming to the hospital was amazing; we knew we were at one of the best children's hospitals in the world with access to some of the most experienced health care professionals.

"The boys were on the trial for 12 weeks between 2009 and 2010. Our whole family noticed a marked difference in their quality of life and mobility over that period.We feel that it helped prolong Jack's mobility and that Tom has been considerably less fatigued."

Muscle project news

Experiences of Transition

Since moving here to Great Ormond Street, we have been working very closely with the Adult Neuromuscular team at the National Hospital for Neurology and Neurosurgery in Queens Square, next door to us.

Ruth Barratt, our Nurse Specialist, and I have been meeting with Dr Quinlivan and the Nurse Specialists at Queens Square to improve our Transition service.

Transition Questionaire

Our first step has been to design a questionnaire to ask those who have already moved over to adult services how their experience was.

Focus Group

Our second step will be to get together a group of young people from Great Ormond Street Hospital and Queens Square to ask them what they would like from our services during the transition period.

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