Great Ormond Street Hospital for Children
NHS Foundation Trust

# Musclemania

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## Transition edition

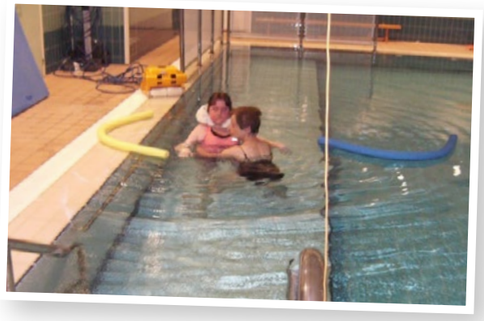
Find out what transition means and how it affects you and your child

# Velvet's story: living with chronic inflammatory demyelating polyneuropathy (CIDP)

Feature article


Velvet's computer photo montages

"Hello, I am a 16-year-old girl who was diagnosed with CIDP at the age of four. CIDP is a very rare condition, especially in children. This condition damages your nerves by making holes in the mylene sheath, and the messages that go round your body don't always reach where they're meant to. As a result, you fall over quite a bit and your muscles start to weaken. That is why it is vitally important that those with this condition must exercise to keep their muscles alive.



"In order to help stabilise my condition I was put on a high dose of steroids (prednisolone) when I was young. Over the years these have been reduced as my health improved, but if I got too weak the doctors would increase them a little to build my strength back up.

"Since the year 2011, I have been unable to walk and now use a wheelchair for health and safety reasons. The right side of my body is extremely weak, especially my right arm. That is why I am left-handed now and this side is a lot stronger as I tend to use it more. I have an unsafe swallow and since May last year have been fed by an NG tube. I have scoliosis and respiratory problems and now use BIPAP every night.

"Unfortunately over the last couple of years the steroids haven't been able to sustain me, so as a result my consultant has tried several other treatments. These have included two lots of IVIg [an intravenous infusion of immunoglobulin] and plasmapheresis [a procedure that removes plasma from the blood and replaces it with new plasma fluid], which I couldn't finish because the catheter cracked so they had to stop the treatment and take it out. I have also had azathioprine and I just recently finished a course of cyclophosamide, and now to follow that treatment I am taking mycophenolate mofetil. I'm not sure if this treatment will work or not, because it is in the same family as azathioprine, which caused me kidney problems.

"The biggest part of my life being happy is having my mum and dad always there to support me or to give me a cuddle when I'm upset."

"2012 was the worst year I have experienced with my condition so far because I was in hospital for over 130 days. I was in hospital the first time when I was rushed in by ambulance because I was having great difficulty with my breathing. The second time I was in hospital a few months later and one night I was woken up to be given some medication, then 10 minutes later I was sick and struggling to breathe and was admitted into the intensive care unit where I went down another two times but for different reasons.

"As a result I hardly went to school, but I did do my school work at home, and I finally went back to school January this year.

"As I am going to need a job when I leave school my dad is helping me to create my own business, which is using a computer to create imaginative photos. It's a great job as I am quite restricted but I can still use a computer to do this work.

"During my life I have had some really tough times, but I have still been able to enjoy my life as I have been to Disneyland Paris twice and a number of great shows in London, plus days out here and there. The biggest part of my life being happy is having my mum and dad always there to support me or to give me a cuddle when I'm upset.

"I keep positive because I am sure that one day something will come along to help me. Thanks for reading my story. Velvet."

# Transition explained

As your son or daughter gets older, you may hear people talking about 'transition'. In this issue of *Muscle Mania*, we explain what transition means and how it affects you and your child.

## What is transition?

Transition is a planned process through the teenage years to prepare young people for the move to adult healthcare and independence.

It aims to:
- Support teenagers to develop skills to become more independent in their healthcare.
- Support parents so they can help their teenager achieve independence to the best of their ability.
- Prepare you and your child for transfer from child-centred healthcare to adult healthcare services.

## Why do I need help with transition?

Research has shown that when young people and their carers first make the move or transfer from children's services to adult services, they may feel nervous about the change. However, when they are prepared for the move, they often find it easier to cope in the new situation.

## When should preparation for transition start?

Most young people will be ready to start between the ages of 12 and 14. We will discuss it with you and your teenager and introduce it gradually. We recognise that this time of transition will be different for everyone. We will work with you and your teenager to respond to their individual needs as much as we can.

If your teenager has a learning difficulty, it is still helpful for them to build on the skills they have and to become as independent as they can for the future.

## What are the aims of transition in the neuromuscular clinics?

- To provide support and information to teenagers and parents as they make the transition from children's to adult healthcare.
- To enhance a sense of control and independence.
- To support teenagers to develop the skills needed to move on to adult care.

## What is the difference between transition and transfer to adult care?

Transfer is an event at the end of the transition period when your teenager moves from paediatric to adult care. Ideally, there is no fixed age for transfer as this will depend on several factors including age, maturity and medical status. It is best to transfer a young person with a neuromuscular condition when they are going through a more stable phase of their condition.

## What will you notice in clinics?

If your child is between 12 and 16, we will be working on aspects of independence and becoming a young adult with a neuromuscular condition.

At each visit we will give young people an opportunity to use a skills plan, helping them to identify what they would particularly like help with.

For parents, we have written a booklet about transition so that you can support your teenager through the process.

### *Helpful resources for transition*

## Takin' Charge

The Takin' Charge project aims to work with several groups of people affected by Duchenne muscular dystrophy.

## Young Adults with Duchenne 14–19 years

This programme aims to help teenagers develop adult skills in addition to their GCSEs or other school subjects eg a Duchenne Duke of Edinburgh award. The course is delivered through active learning workshops and covers areas such as IT and social media, self-advocacy for medical care, workplace skills, skills for independent living including developing a social life, relationships and sexual health.

## Becoming a Man: life with Duchenne muscular dystrophy

The Muscular Dystrophy Campaign asked 40 young men aged 15 to 33 what it's like to live with Duchenne muscular dystrophy. The research aimed to share their experiences and help others find the support they need.

**School, college and next steps**

People had good and bad things to say about what school and college were like. Lots of people said they'd like it if things were easier: "It does seem to be that before you're 16 there is a plan – this happens, then that happens, and it's smooth. Then you hit a certain age and no one's thought about what happens next!"

If you have a statement of special educational needs at school you should have a meeting every year from the age of 14 until you leave school. This is called a transition plan and it helps you plan what you do next.

You can invite people to the meeting – your mum or dad, a teacher who you get on well with, your occupational therapist or social worker – anyone who might have good ideas about what you do next and be able to support you with making decisions. You are entitled to support with making choices as you get older and it is your school's job to organise this meeting.

**Having a social life**

Everyone wants a good social life: friends, mates, places to go, things to do. It's no surprise that the young men with Duchenne in this research wanted all these things too. Everyone wants to do things (which don't involve their parents) with their mates. It's an important part of getting older: "I wouldn't want to go out on my own. If you don't have friends, what's the point? It's just boring if you can't do what you want because you've got to be with your parents."

There were lots of things that people were into – music, computers, art, cars, TV, sport: "There's a form of wheelchair football we can play. A lot of us love football so a couple of parents set up a team. It's amazing, I love it."

Some people lived away from home at residential college or university. They said that their social lives were pretty good and that they had quite a lot of independence: "Everyone treats you as a normal person at uni. That's one of the main reasons I wanted to go – to get that contact with other people, to get out of the house, you know, like going out drinking."


**Driving**

Several young men said how much they wanted to have and drive a car. One 17-year-old had been through a driving assessment and got his provisional driving licence: "It's been my lifelong ambition to drive. In February I got my provisional driving licence and it felt awesome! I want to let anyone else who is in my position know that if driving is your thing it can be possible, so don't be put off."

For more personal stories from people living with muscular dystrophy, visit **www.muscular-dystrophy.org**

# Who's who

Meet the new members of the Neuromuscular team at Great Ormond Street Hospital (GOSH).

**Anna Sarkozy**
Consultant
Anna studied medicine at La Sapienza University in Rome, where she also completed her higher medical training in clinical genetics. Having worked as a specialty doctor in the Neuromuscular team in Newcastle for over six years, Anna has now joined the Neuromuscular team at GOSH as a consultant.

**Katie Groves**
Research Nurse
Katie is a Neuromuscular Research Nurse. She has been working for the Neuromuscular team for about two months and before this she worked as a General Research Nurse in the Somers Clinical Research Facility. She has worked in the trust for about five years on a number of different wards.

**Ruth Haslett**
Social Worker
Ruth qualified as a social worker in 2008 and has since worked in statutory and voluntary settings. She has a range of experience including working with disabled children, children in care and within a child protection team (both in hospital and community settings). Most recently, she was based in a charity assessing vulnerable children and then training and supervising volunteers to provide them with one-to-one, long-term mentoring support.

**Adeline Ngoh**
Registrar
Adeline hails from Singapore. She was trained in Barts and the London School of Medicine and Dentistry and graduated in 2005. She is currently training in Paediatric Neurology and has been working in the Paediatric Neurology department at GOSH since 2012. She will be working with us in the Neuromuscular department for one year.

**Nahla Alshaikh**
Clinical Research Fellow
Nahla is spending a year here as a clinical and research fellow to gain experience in paediatric neuromuscular diseases. She chose GOSH for its great reputation worldwide. She graduated from King Abdulaziz Medical School in Saudi Arabia in 2005 and earned her degree in Paediatric Neurology in 2011. Nahla is really enjoying London because of its beauty and cultural diversity.

**Pinki Munot**
Consultant
Pinki studied medicine at Mumbai University in India, where she also completed her postgraduate training in paediatrics. She moved to the UK in 2005 and completed her training in paediatric neurology in 2013 at GOSH. She has worked at the Trust since 2008. In her current role she will be working in both the Neuromuscular and General Neurology services.

# Patient registries

## UK Duchenne Registry

The UK Duchenne Registry, which is run by the charity Action Duchenne, was set up to find patients with Duchenne muscular dystrophy for new clinical trials. For more information, contact Angela Stringer on 0208 556 9955 or visit **www.actionduchenne.org**

## Facioscapulohumeral muscular dystrophy (FSHD)

There are a number of patient registries across the world that collect data on patients with FSHD to help advance the research and development of treatment, therapies and care for all those diagnosed with this condition. For more information and to register in the UK, visit **www.fshd-registry.org/uk**

## UK SMA (spinal muscular atrophy) Patient Registry

This registry is for patients in the UK and Ireland with SMA. The goal of the registry is to quickly identify suitable patients for clinical trials, inform patients about new treatments that might be relevant to them and give scientists important information about SMA. For more information and to register, visit **www.treat-nmd.org.uk/registry**

## The UK Myotonic Dystrophy Patient Registry

This national registry is for all patients in the UK who are affected by myotonic dystrophy type 1. The aim of the database is to help advance the research and development of treatment, therapies and care for all those diagnosed with myotonic dystrophy. For more information and to register, visit **www.dm-registry.org/uk**

## Congenital Muscular Dystrophy International Registry (CMDIR)

The CMDIR was created by the patient advocacy group Cure CMD to identify patients with congenital muscular dystrophy (CMD) on a global scale. This information is used to raise awareness, improve standards of care, accelerate clinical trials and work towards finding a treatment or cure. People with all forms of CMD can register. For more information and to register, visit **www.cmdir.org**

## Global FKRP (fukutin-related protein) registry

The Global FKRP Registry is an international database of genetic and clinical data about people affected by conditions caused by mutations in the FKRP gene.

These include limb girdle muscular dystrophy type 2I (LGMD2I), and the rarer conditions of congenital muscular dystrophy (MDC1C), muscle eye brain disease and Walker-Warburg syndrome. The information collected will help to identify patients suitable for clinical trials and allow them to participate more easily. For more information and to register, visit **www.fkrp-registry.org**

## Myotubular and Centronuclear Myopathy Patient Registry

This international database records information about patients with these conditions. The aim of the database is to help identify patients for relevant clinical trials, encourage further research into these conditions, provide researchers with specific patient information to support their research and offer up-to-date information to health professionals to help them deliver better standards of care for their patients. For more information and to register, visit **www.myotubulartrust.com**

**Leaflets for all registries are available in clinic.**

If you have any ideas for future issues of *Muscle Mania*, or would like to share your story, please get in touch...

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