Muscular Dystrophy Ireland Information Day
Rochestown Park Hotel, Cork
Saturday 10th October 2009

In conjunction with Dr. Aisling Ryan, Consultant Neurologist in Cork University Hospital, Muscular Dystrophy Ireland held an information day for adults affected by neuromuscular conditions on 10th October 2009. The following is a report on the day.

Dr. Aisling Ryan, Consultant Neurologist, Cork University Hospital

Dr. Ryan spoke about the pathway to diagnosis for adults with muscular dystrophy and management of the condition. There are currently 22 Neurologists in the public health service, which is one for every 200,000 people. Until recently the ration was 1:300,000 so it has improved, but is still the lowest number of neurologists per head in Europe. The ratio should be 1:100,000 so we have half the recommended number. There are three Neurologists in Cork University Hospital but there is only one Neurophysiologist outside Dublin, a half time Paediatric Neurologist covering the southern region and no neuromuscular nurse specialist. Dr. Ryan's own background in neuromuscular conditions includes a particular interest in skeletal muscle ion channel disorders, with work on neuromuscular channelopathies in Ulm, Germany and work in the Centre for Neuromuscular Disease in Queen Square London. She is also a member of the British Myology Society.

Dr. Aisling Ryan, Consultant Neurologist

The neuromuscular clinic in Cork University Hospital has a team comprised of a Neurologist, Neurology Registrars, and a Neurology Nurse specialist. It has a waiting time of around two months (at the time of this talk) in comparison to a wait of 1-2 years for a general neurology appointment. They see people with a diverse range of neuromuscular conditions, including the muscular dystrophies, congenital myopathies, inflammatory myopathies, metabolic

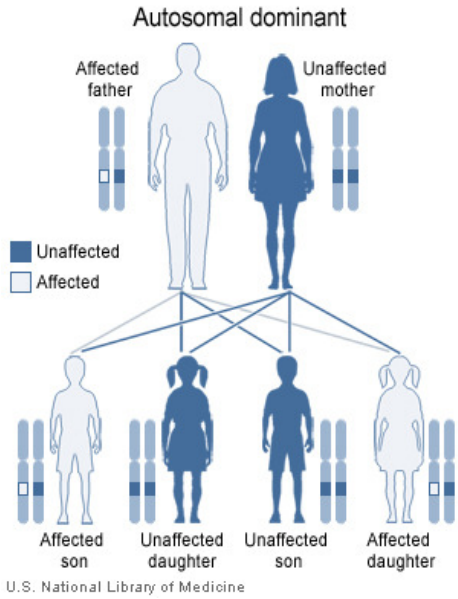
myopathy, myotonic dystrophy, periodic paralysis and hereditary neuropathy / CMT.

The muscular dystrophies are an inherited group of disorders, characterised by muscle weakness and wasting and “dystrophic” muscle biopsy changes. The onset can be prenatal, in childhood or in adulthood. They are clinically and genetically very variable. Some progress rapidly while others are more slow, and some have involvement of multiple systems (e.g. heart, brain). There are a number of key questions for each neuromuscular condition:

- When was it first described and by whom?
- What is the mode of inheritance?
- What is the general clinical pattern?
- Disease course or prognosis?
- How do I make the diagnosis?
- What are the treatment options?

<u>Inheritance</u>

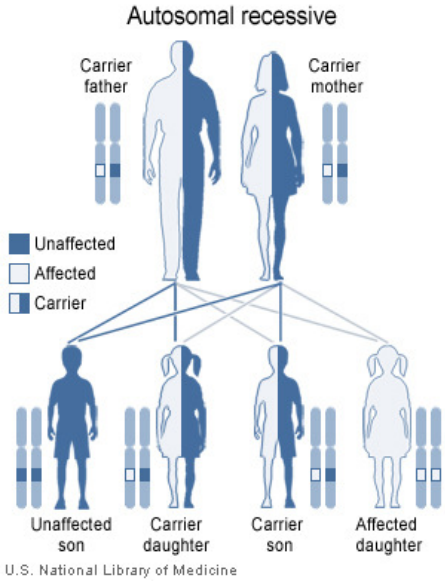
The muscular dystrophies can be inherited in different ways: autosomal dominant, autosomal recessive, x-linked or mitochondrial. The diagram below shows an autosomal dominant inheritance (one parent has the gene and they have the condition – in the example below it is the father who has it but females can also have it).



Examples of conditions inherited in this way are:

- Dystrophic: LGMD 1A, 1B, 1C, 1D, 1E
  Facioscapulohumeral MD
  AD Emery-Dreifuss MD
- Congenital: Central core disease
  Nemaline myopathy
- Myotonic: Thomsen myotonia
  Myotonic dystrophy
- Neuropathic: AD Charcot-Marie-Tooth

In Autosomal Recessive inheritance, each parent carries one copy of the gene but they don't have the condition as you need two copies to have it.



Examples of conditions inherited in this way are:

- Dystrophic: sarcoglycanopathies
  Calpainopathy (LGMD 2A)
  LGMD 2B – Miyoshi
  LGMD 2G, 2H, 2I
  Congenital muscular dystrophies
- Congenital: multi / minicore disease
  Selenopathy, nemaline myopathy, AR CCD
- Myotonic: Becker myotonia
  Schwartz-Jampel symdrome
- Neuropathic: Spinal muscular atrophies
  AR Charcot-Marie-Tooth

In x-linked inheritance, the mother carries the condition and every one of her sons has a 50% chance of having the condition while every one of her daughters has a 50% chance of being a carrier. Duchenne and Becker muscular dystrophies are two of the conditions inherited in this way.

<u>Clinical Patterns in Neuromuscular Conditions</u>

There can be different patterns of muscle weakness in different neuromuscular conditions, e.g. proximal weakness, distal weakness, muscle hypertrophy, facial weakness and contractures. In some conditions there can also be eye muscle weakness, scoliosis, heart involvement or respiratory problems.

<u>How to make the diagnosis?</u>

There are different laboratory tests available to diagnose neuromuscular conditions:

- Electrophysiology: EMG studies can show myopathic or neuropathic changes.

- Imaging: ultrasound, CT or MRI scans can be potentially useful in selecting the site for a biopsy, demonstrate selectivity or follow progression of the condition.
- Biochemistry, e.g. CK levels
- Muscle biopsy
- DNA studies

A muscle biopsy can be performed if a full clinical assessment deems it necessary, if limb girdle muscular dystrophy is suspected, if there have been no previous investigations, if there has been an inconclusive biopsy in the past or if the wrong muscle was biopsied in the past. Which muscle to select is determined by careful clinical assessment. There are two different techniques, a needle biopsy or open biopsy. The open biopsy is a little more invasive but gives more information – as a needle biopsy gives a smaller sample there is more chance that you might need to repeat it. After the sample is taken, it is carefully handled en route to the laboratory. The neuropathology lab at CUH receives biopsies from Cork, Limerick, Waterford etc and two Consultant Neuropathogists are based there. A Multidisciplinary Neuromuscular meeting has been held at CUH since 2006 to ensure a multidisciplinary approach to diagnosis. It is attended by Consultant Neurologists, Neuropathologists, Neurophysiologist, Nurse specialists and Neurology registrars so all muscle biopsies are reviewed and discussed and there is consensus on a diagnosis (where possible) or further investigations recommended.
Genetic tests may be done as a first diagnostic step if the clinical picture is suggestive but is more often done after muscle biopsy. This involves a blood test with informed consent, and samples are sent to the lab to be forwarded to the National Centre for Medical Genetics or to international laboratories performing the test as s diagnostic service. Conditions which may be tested for genetically include Duchenne and Becker MD, Facioscapulohumeral MD and myotonic dystrophy type 1 and 2. Genetic testing is useful to get an accurate diagnosis, on some occasions to get an indication of prognosis, for reliable genetic counselling, and to develop therapies or guide therapies.

There is a dilemma when the muscle biopsy might be inconclusive and the genetic test is negative. This can lead to further analysis of the biopsy and possible international collaboration, e.g. with the Centre for Neuromuscular Diseases in the UK.

<u>What are the Treatment Options?</u>
The multidisciplinary team incorporates a range of professionals:

- Neuromuscular physician
- Physiotherapy
- Respiratory management
- Speech therapy
- Occupational therapy / orthotics
- Nutrition / dietetics
- Orthopaedic expertise
- Genetic counselling

- Psychosocial support

Management of different symptoms is possible, e.g. heart complications, respiratory symptoms, scoliosis, weight loss / gain.
There are a number of options being researched as potential new therapies including gene therapies, stem cells and potential pharmacological therapies.

Conclusions

- Achieving a diagnosis is time consuming but it is increasingly important as new therapies are developed and it is also important for genetic counselling.
- The management of the physical aspects of disease are extremely important.
- Better services for patients and medical staff are of paramount importance.
- Developing a centre of excellence with international collaboration is important.

Liz O'Sullivan, Senior Physiotherapist, CUH

Liz O'Sullivan giving her talk

Access to the CUH Physiotherapy service is by consultant referral. There is an in-patient and an out-patient service, access to hydrotherapy and a service in primary care team venues, which are more accessible.
The physiotherapy assessment involves an individual measurement of:

- Muscle strength
- Joint range
- Functional performance
- Monitoring the chest

All treatment is specific to the individual, to deal with the muscle weakness and wasting as a result of their condition, and the knock-on effects of these.

Physiotherapy treatment can involve a range of things, including active exercises, core stability exercises, walking re-education, hydrotherapy,

passive stretches, assisted standing, mobilisation techniques, acupuncture for pain relief and electrotherapy.

The essential aims of treatment are:

- To maintain flexibility
- To maintain joint range
- Make the best use of available muscle power and delay contracture progression to minimise function
- To promote or prolong walking with appropriate aids and / or orthoses
- To give appropriate treatment and advice for acute injuries, e.g. strains or sprains
- To give postural advice
- Advice on moving and handling issues.

Keeping active is beneficial. The golden rule is to exercise until you feel slightly hot or sweaty but to pace yourself and not overdo it. Activities such as swimming, walking, cycling, yoga or pilates can be good, especially if friends or family can get involved as well, and these can be fitted into your daily schedule. It is important to warm up and cool down properly, Stretching the calf and back extensor muscles is good. Moderate resistance using small hand weights for example can be useful, but you should be careful to avoid heavy weights and excessive eccentric exercise (e.g. coming downstairs, downhill walking). Equipment such as a swiss exercise ball or a standing frame can be useful for some people.

Some of the audience at the Cork Information Day

Dr. Oisin O'Connell, Respiratory Specialist Registrar, CUH

Ventilation is the process of moving air in and out of the chest. People with muscular dystrophy can experience underventilation as a result of their condition. Symptoms of chronic underventilation include:

- Daytime fatigue
- Nightmares / night terrors

- Gradually increased nocturnal awakenings
- Morning headache
- Breathlessness
- Weakened or softened voice
- Weak cough
- Poor appetite, weight loss
- Loss of concentration / drowsiness
- Depression

If someone is experiencing these symptoms, they may need a respiratory assessment. This assessment can include:

- History – talk to the person
- Sleep questionnaire
- Physical examination
- Finger probe – pulse oximeter
- Arterial blood gas
- Breathing tests (spirometry)
- Chest x-ray
- Overnight sleep studies

Non-invasive and invasive ventilation

If necessary, it might be recommended that a person starts to use non-invasive ventilation. This is generally a BiPAP, which means Bi level positive airway pressure. The BiPAP can be individually set, is well tolerated, doesn't require any surgery and preserves baseline swallow, speech and cough. There are different types of masks that can be worn with the BiPAP. The nasal mask is the most common type. It does not impede swallow or speech and there are different sizes available. There is also a facial mask, which some people may prefer to use overnight or if they breathe through their mouth. It can also be used if there is a concurrent infection. People often only use their BiPAP at night, which results in improved sleep quality and improved daytime energy. Others need to use the BiPAP during the day as well as their condition progresses, and there are different things that can be used, such as a mouthpiece that people can take a breath through when they need it.

The benefits of non-invasive ventilation are

- Improved quality of life
- Improved and refreshed sleep quality
- More energy and less breathlessness
- Improved mood in general
- Louder voice
- Less chest infections
- Increased life expectancy for those with high carbon dioxide in the blood

Some people do experience problems or side effects with non-invasive ventilation however:

- Mask intolerance
- Airway dryness
- Nasal skin irritation and pressure sores
- Gastri distension

- Equipment familiarisation
- Family and support issues
- Contra-indicated if there is a risk of aspiration.

Some of these problems can be rectified by using a different mask or changing a setting on their BiPAP so if anyone is experiencing problems, they should contact their respiratory physician.
Some people might need invasive ventilation, or tracheostomy. This is a small surgical procedure, and is more powerful than non-invasive ventilation but does require suctioning. The advantage is that the face is unimpaired – there is no mask. It can incorporate a speaking valve as well.

Airway clearance
An ineffective cough can often go unnoticed, but this predisposes you to pneumonia. The diagnosis would be based on recurrent chest infections or specific breathing tests and treatments include physiotherapist teacing and mechanical cough assist devices.

Scoliosis
Scoliosis or curve of the spine, can be a problem in some people with muscular dystrophy, and it can impair ventilation. Surgery can correct this and the optimal time for surgery is based on spinal curve and breathing tests. Some people might require non-invasive surgery pre and short term post surgery.

Tips to Improve Respiratory Health
- Get the yearly flu vaccine (this year there are two vaccines – one is for H1N1 otherwise known as swine flu)
- Get the pneumococcal vaccine
- Do not smoke
- Avoid second hand smoke
- Avoid sleeping tablets and sedatives
- Good nutrition
- Monitor for scoliosis
- Present early to your physician with symptoms of poor sleep, daytime fatigue and drowsiness

Supports Available at Cork University Hospital
- Multidisciplinary team of physicians, allied health professionals and specialist nurses
- Specialist respiratory laboratory
- Overnight sleep studies available
- Speech and language therapists to assist ventilator associated speech
- Liaison with non-invasive ventilation reps and specialist respiratory physicians and technicians.

Additional Resources
- Muscular Dystrophy Association publication “Breathe Easy” – www.mda.org/publications/breathe/

- General resource "Medline Plus" – www.nlm.nih.gov/medlineplus/musculardystrophy.html
- Muscular Dystrophy Association publication "101 Hints to Help with Ease for Patients with Neuromuscular Disease" – www.mda.org/publications/101hints/

MDI Staff Members Margaret Goode (Family Support & Clinic Coordinator), Mary-Rose Howell (Youth / Respite Worker in the southern region) and Darryl Pearson (Family Support Worker in the southern region)

Ethne Mitten, Clinical Nurse Specialist in Neurology, CUH

A Clinical Nurse Specialist (CNS) is a nurse or midwife specialist in clinical practice who has undertaken formal recognised post registration education relevant to his or her area of specialist practice.

Role of the CNS

Nursing Assessment: Evaluation / Intervention

- Early detection of problems
- Prompt response to individual needs / expert assessment
- Evidence based practice
- Co-ordinate referrals to multidisciplinary team
- Assisting and reassuring patient
- Holistic approach
- Respite care

Patient Advocate

- Communication / negotiation / representation of patient values
- Encourage patient's own responsibility for well being
- Facilitate informed decision making
- Discharge planning

Education and Training

- Provide information, support, advice for staff, multidisciplinary team
- Liaise with Public Health Nurses and GPs
- Work with voluntary organisations

- Improve patient care

Work with Consultants

- Be available to advise
- Ensure referrals regarding patients' needs are made
- Provide support and leadership for colleagues
- Collaboration with multidisciplinary team / primary care

Research and Audit

- Demonstrate evidence based care
- Up to date knowledge base
- Contribute to nursing research
- Evaluation of nursing care
- Outcome measures

Management

- Coping strategies
- Retraining approach
- Compensatory approach
- Occupational therapy

Ethna Mitten's contact details are:
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Thank you to all speakers who gave their time on the day to give their excellent presentations.
If you need any further information, please contact Karen Pickering, Information Officer, Muscular Dystrophy Ireland, 71-72 North Brunswick Street, Dublin 7. Tel: 01 8721501. Email: karen@mdi.ie