Holistic & multidisciplinary care, management and research for children with Neuromuscular Disorders at The Royal Children’s Hospital

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Overview

- MDA Neuromuscular Clinic
  - Purpose, aim, goal
  - Clinic Team
  - Evaluation
  - Contacts

- Neuromuscular Research Program
  - CINRG – network, mission, values
  - Clinical trials/studies @ RCH
  - Research team
MDA Neuromuscular Clinic

- Commenced in February 2008
- Multidisciplinary clinic which brings together relevant medical specialties, allied health therapists and other health professionals.
- “One-stop-shop” specifically designed for children with neuromuscular disorders and their families.
MDA Neuromuscular Clinic Team

- Neurology
- Respiratory
- Cardiology
- Orthopaedics
- Genetic Counseling
- Physiotherapy
- Occupational Therapy
- Orthotics
- Social Work
- Nurse Coordinator
- Mental Health
- Teacher Consultant
- MDA Staff Support
- Diagnostic testing as required
Why come to clinic?

- Each child’s overall physical function, pulmonary function, muscle strength and bone health are monitored
- A regular review of medical management
- Any side effects of treatment are assessed and monitored
- To receive up-to-date information and support
- Ongoing documentation of each child’s condition & progress, that can be communicated with others involved in the child’s care
- Planning for current and future issues e.g. surgery, equipment, preparation for schooling, transition to adult services
Evaluation & Quality control

- To date just over 470 patients have been seen in clinic
- Constant evaluation & implementation of new/different techniques suggested by staff
- Clinical audits for the MDA Neuromuscular Clinic

Feedback
Results show that families rate our clinic as:

- “excellent” in regards to information provided prior to clinic, coordination and organisation of their child’s appointments and the clinic team meeting their requirements.

- Family’s free comments include: “Fantastic organisation and friendliness!”; “… You all make what can be a daunting visit very relaxed. Thanks!”; “You do a great job including everyone”;
Muuum!!!! I’m bored!!

- Generous donations of an XBOX 360, WII and games from the MDA assist in providing entertainment in the waiting room and eliminated negative feelings for children regarding their experience in the clinic.
Neuromuscular Research @ RCH

29 August 2004 - launch of the affiliation of the Neurology department at RCH with CINRG
The Cooperative International Neuromuscular Research Group, (CINRG), was formed in 1999 as the clinical research arm of the Duchenne Muscular Dystrophy Research Center (DMDRC) and the Research Center for Genetic Medicine at the Children’s National Medical Center (CNMC) in Washington USA.

CINRG’s network expands across 23 clinical trial sites in 10 countries.
CINRG Network

Washington, DC
Richmond, VA
Pittsburgh, PA
Rochester, MN
Minneapolis, MN
St Louis, MO
Sacramento, CA
Dallas, TX
Houston, TX
Memphis, TN
Melbourne, Australia
Sydney, Australia
Gothenburg, Sweden
Buenos Aires, Argentina
Calgary, CA
Pavia, Italy
Jerusalem, Israel
Chennai, India
Puerto Rico
Leuven, Belgium
Toronto, CA
Edmonton, CA
CINRG’S MISSION

- To be a pre-eminent neuromuscular research group striving to advance knowledge of Duchenne Muscular Dystrophy and other neuromuscular disorders through the implementation of cutting edge clinical trials throughout various countries and cultures over the world.
CINRG’S VISION

- To find a cure and advance the standard of care for all patients and families with Duchenne Muscular Dystrophy over diverse geographies and cultures. No child with Duchenne Muscular Dystrophy will be left untreated or without access to health care professionals, education or compassionate care.
CINRG CLINICAL TRIALS

- Prednisone (high-dose weekly vs daily) in Duchenne muscular dystrophy study

- A double blind randomized placebo-controlled study of daily Pentoxifylline as a rescue treatment in Duchenne Muscular Dystrophy

- The UCD (University of California, Davis campus) Longitudinal study of the relationship between impairment, activity limitation, participation and quality of life in persons with confirmed Duchenne muscular dystrophy
The UCD Longitudinal study

- Five year study
- Collects information from boys and men with DMD and their families
- Information to be collected will include studies of patient’s physical abilities, medical problems and use of healthcare services.
- A second goal is to look for an association between modifying genes (which contain instructions for how the body works) and response to treatment of DMD
How is the data collected?

- Questionnaires
- Muscle strength testing
- Lung function testing
- Saliva sample
How many participants?

- Total international target 395
- RCH currently have 20 boys aged between 3 – 23 yrs old
A clinical study to compare two dosing regimens of GSK2402968 for effectiveness and safety in Duchenne muscular dystrophy.
What is the research about

- The muscles of Duchenne muscular dystrophy (DMD) patients are unable to make a protein called dystrophin, due to an error in their DNA. Without the dystrophin protein, the muscles of DMD patients become gradually weaker.

- The purpose of this study is to see whether the experimental medicine, GSK2402968, is safe to use as medication for DMD patients and to see if it might be effective in preventing or slowing loss of muscle strength in this condition.
What is GSK2402968?

- Investigational, sub cutaneous administered drug
- GSK2402968, is an "antisense oligonucleotide" (AON for short) which is a new class of medicine for DMD patients. It is designed to change how the dystrophin protein is made in the muscles.
- Commonly known as ‘exon skipping’
Study Purpose

- Understand whether GSK2402968 will increase the production of dystrophin in muscle cells, preserving muscle strength in boys with DMD. I.e. can it improve walking, activity, muscle function & strength
- Whether the drug is safe to be given over a long period of time
Study design

- International
- Multicenter
- Randomized
- Double-blind
- Placebo-controlled
- Evaluates two different regimes of the drug administration – weekly and intermittent
How is the data collected?

- 49 weeks of treatment – twice weekly/weekly clinic visits (includes injections)
- Questionnaires
- 6 minute walk test
- Muscle strength, functional, timed, memory testing
- Various diagnostic safety testing
Participants

- 160 international participants – 35 sites over 4 continents
- Specific inclusion/exclusion criteria
- RCH currently have 2 enrolled
....more research

- A randomized controlled trial of the efficacy of Vitamin C treatment for children with CMT type 1A
- A prospective multicentre study of the natural history of Charcot-Marie-Tooth disease (CMT) – The Australasian CMT Registry
- Collaboration with the Australian Paediatric Surveillance Unit in January 2007 saw the commencement of a prospective study of the incidence of inherited neuromuscular disorders of childhood.
Research Team

- Primary & Sub Investigators:
  A/Prof. Monique Ryan, A/Prof. Andrew Kornberg

- Study Coordinator: Daniella Villano

- Physiotherapists: Kate Carroll, Katy DeValle, Rachel Kennedy
How can I find out information about clinical trials?

- www.rch.org.au
- www.mda.org.au
- daniella.villano@rch.org.au
Contact

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THANK YOU!!
Questions????