

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

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MDA Neuromuscular Nurse Coordinator

Overview

- Neuromuscular Research Program
 - CINRG – network, mission, values
 - Clinical trials/studies @ RCH
 - Research team
- MDA Neuromuscular Clinic
 - Purpose, aim, goal
 - Clinic Team
 - Evaluation
 - Contacts



Neuromuscular Research @ RCH

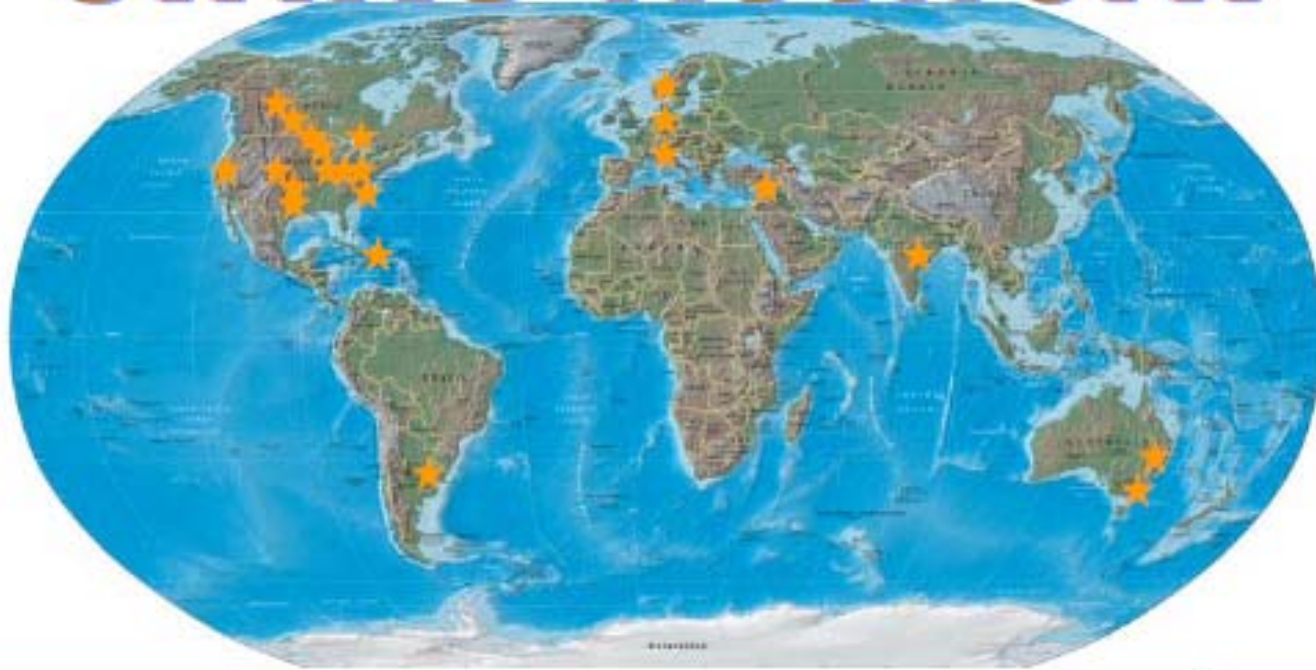
29 August 2004 - launch of the affiliation of the Neurology department at RCH with CINRG



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 16 boys aged between 3 – 12 yrs old (in process of recruiting older participants before end 2008)

A phase 2b efficacy and safety study of PTC124 in subjects with nonsense-mutation-mediated Duchenne and Becker muscular dystrophy.



What is PTC124?

- Investigational, orally administered drug
- Targets a particular genetic alteration known as a nonsense mutation
- Nonsense mutations are errors in the genetic code that cause a genetic disorder by prematurely stopping the production of a critical protein, dystrophin in DMD/BMD.
- PTC124 has the potential to overcome the premature stop signal & produce dystrophin protein to increase muscle strength

Study design

- International
- Multicenter
- Randomized
- Double-blind
- Placebo-controlled
- Evaluates two different dose levels of the drug

Study Purpose

- Understand whether PTC124 can improve walking, activity, muscle function & strength
- Whether drug is safe to be given over a long period of time



How is the data collected?

- 48 weeks of treatment – 2 day clinic visits every 6 weeks
- 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 6 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:
Prof. A Kornberg, Dr. Monique Ryan, Dr. Victoria Rodriguez-Casero
- 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



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
- 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 200 patients have been seen in clinic
- Constant evaluation & implementation of new/different techniques suggested by staff
- A clinical audit for the MDA Neuromuscular Clinic Feedback form was approved by the RCH Human Research Ethics Committee in June 2008

Preliminary 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”;

Muum!!!! I'm bored!!



- Generous donations of an XBOX 360 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.

Contact

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THANK YOU!!



Questions????

