



ST VINCENT'S
HOSPITAL
MELBOURNE

What can an adult neurologist do for you?

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UNDER THE STEWARDSHIP OF MARY AIKENHEAD MINISTRIES



THE UNIVERSITY OF
MELBOURNE

Disclosures

- **Stroke Fellowship 2009 joint funding Boehringer-Ingelheim and Stroke Unit Monash Medical Centre**
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- **Education grant - Novartis**
- **Advisory boards/working parties – Biogen, Roche, Teva, Medtronic, Abbott**
- **Medical advisory board Hearts4Heart (NFP)**

Overview

- **Transition to adult neurology**
- **What does a neurologist do?**
- **Neurologist role in NMD**
- **Access to treatments & trials**
- **Questions & Discussion**





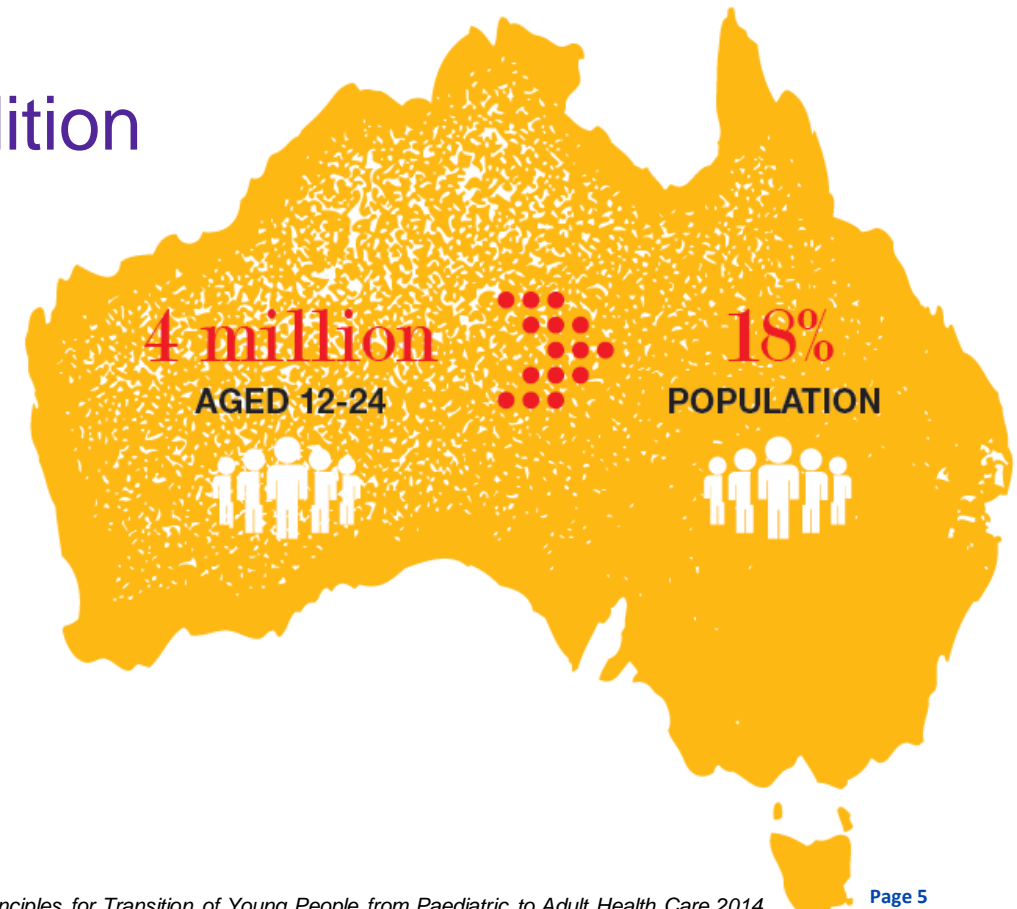
TRANSITION TO ADULT NEUROLOGY

Where are we now?

Diseases *of* childhood → Diseases *beginning in* childhood

- 12% Young Adults have chronic condition
- 90% with childhood conditions reach adulthood

→ *transition to adult health system*



From meerkats to mayhem.....



***“It’s not as well
co-ordinated!”***

~ young man with DMD ~

The Adult Neurology Clinics

- Often not multidisciplinary
- Rarely nurse/co-ordinator
- Different specialty days (historical)
- Appointment time (in the box)
- DNA not chased

MBS funding

132 (New visit) - \$231.35

133 (review) - \$115.85



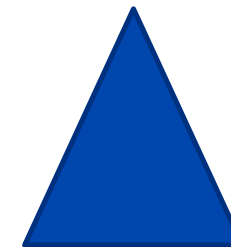
Transition to adulthood



Independence
&
Autonomy

↑ Health
needs

↑ Physical
dependence



Why does transition matter?





WHAT DOES A NEUROLOGIST DO?

Becoming a Neurologist

Medical school

↓ (6-7 years)

Internship

↓ (1 year)

Basic training

↓ (3 years)

Advanced training

↓ (3 years)

Neurologist

Total: ~ 13-14 years

Paeds training

3-4 months

Young Adult training

3-4 hours

- Fractional/VMO
- Staff Specialist

Then sub-specialisation

- Neuromuscular
- Stroke
- Headache
- Epilepsy
- Movement disorders
- Neuroimmunology
- Neurocritical care
- Neurogenetics
- Memory & cognition

Academia



Research



Clinical

Education



THE ROLE OF A NEUROLOGIST IN NEUROMUSCULAR CONDITIONS

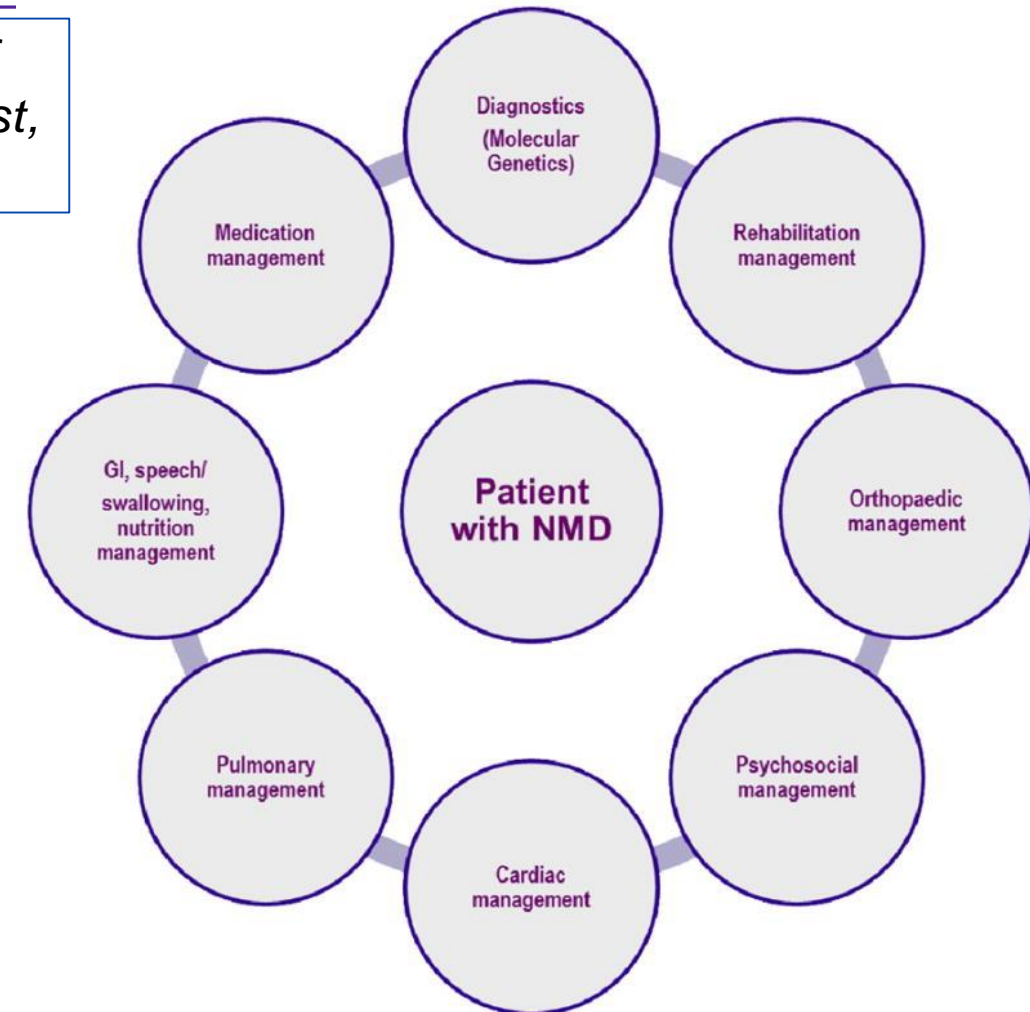
Interdisciplinary care co-ordination

“multidisciplinary approach.....should be coordinated by one of the physicians, generally the neurologist or pediatric neurologist, who is aware of the disease course and potential issues”.

Neurologist responsibilities

- Health maintenance
- Maximizing health and functional capacities
- Monitoring of disease progression
- Complications prevention
- Preventive care
- Evidence based management
- Promoting access
- Advocacy
- Advice to other disciplines

- Anaesthetics
- Medications
- Vaccinations



Diagnosis & Genetics

- **Correct diagnosis**
- **Genetic characterisation**
 - Prognostication
 - Individualised treatment
 - Clinical trials
- **Access to testing**
 - Cost
 - Type of testing



Physical assessments


- Haven't been routine in adults
- Now recommended
- Physiotherapy shown to slow progression
 - Stretching
 - Positioning
 - Mobility
 - Chest physio

Facioscapulohumeral
Dystrophy Composite
Outcome Measure
(FSHD-COM)

Revised Upper
Limb Module for
SMA
(RULM FOR SMA)



Timed Function Tests



10-meter walk/run 4-stair climb Time to rise from supine

North Star Ambulatory Assessment


Assessment measures — from most to least difficult

Hop right leg	Climb box step right
Hop left leg	Climb box step left
Stand on heels	Stand on one leg right
Rise from floor	Stand on one leg left
Run	Get to sitting
Jump	Rise from chair
Lift head	Walk
Descend box step right	Stand
Descend box step left	

How measures are scored:

2 Can perform 1 Can perform with difficulty 0 Unable to perform

MFM self-training DVD
Motor Function Measure
For neuromuscular diseases



Hammersmith Functional
Motor Scale – Expanded (HFMSE)
Manual of Procedures

Patient reported outcome measures

Table 1

Selected Problems Impacting Health-Related Quality of Life in NMD (n = 1169)

How much difficulty do you have performing the following functions?	Moderate or Severe	Slight	Not a problem
Difficulty with muscle weakness.	72.4%	11.6%	6.0%
Difficulty getting exercise.	66.1%	17.5%	16.3%
Difficulty with fatigue.	70.8%	21.7%	7.6%
Difficulty controlling weight.	36.0%	27.9%	36.2%
Difficulty with sleeping.	36.2%	28.6%	35.1%
Difficulty with muscle contractures.	33.6%	29.9%	36.6%

How much does your health limit you in the following activities?	A lot	A little	Not at all
Vigorous activities such as running?	93%	3%	4%
Walking more than a mile?	84%	9%	7%
Walking several blocks?	73%	17%	10%
Walking one block?	54%	27%	18%

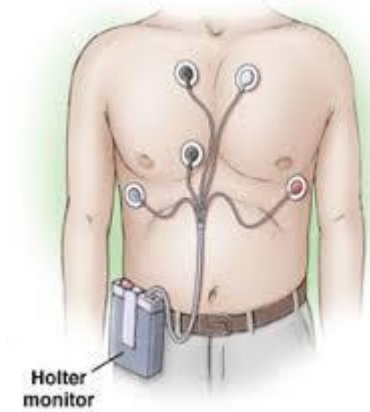


Heart Care

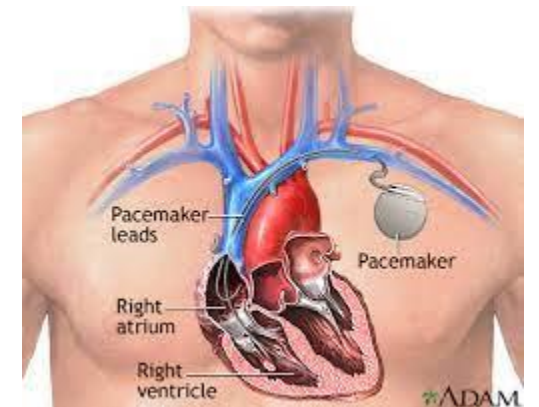
- ECG/Holter
- Echocardiogram
- Cardiology physician
- Medications
- Cardiac MRI
- Cardiac devices



ST VINCENT'S
HEART CENTRE

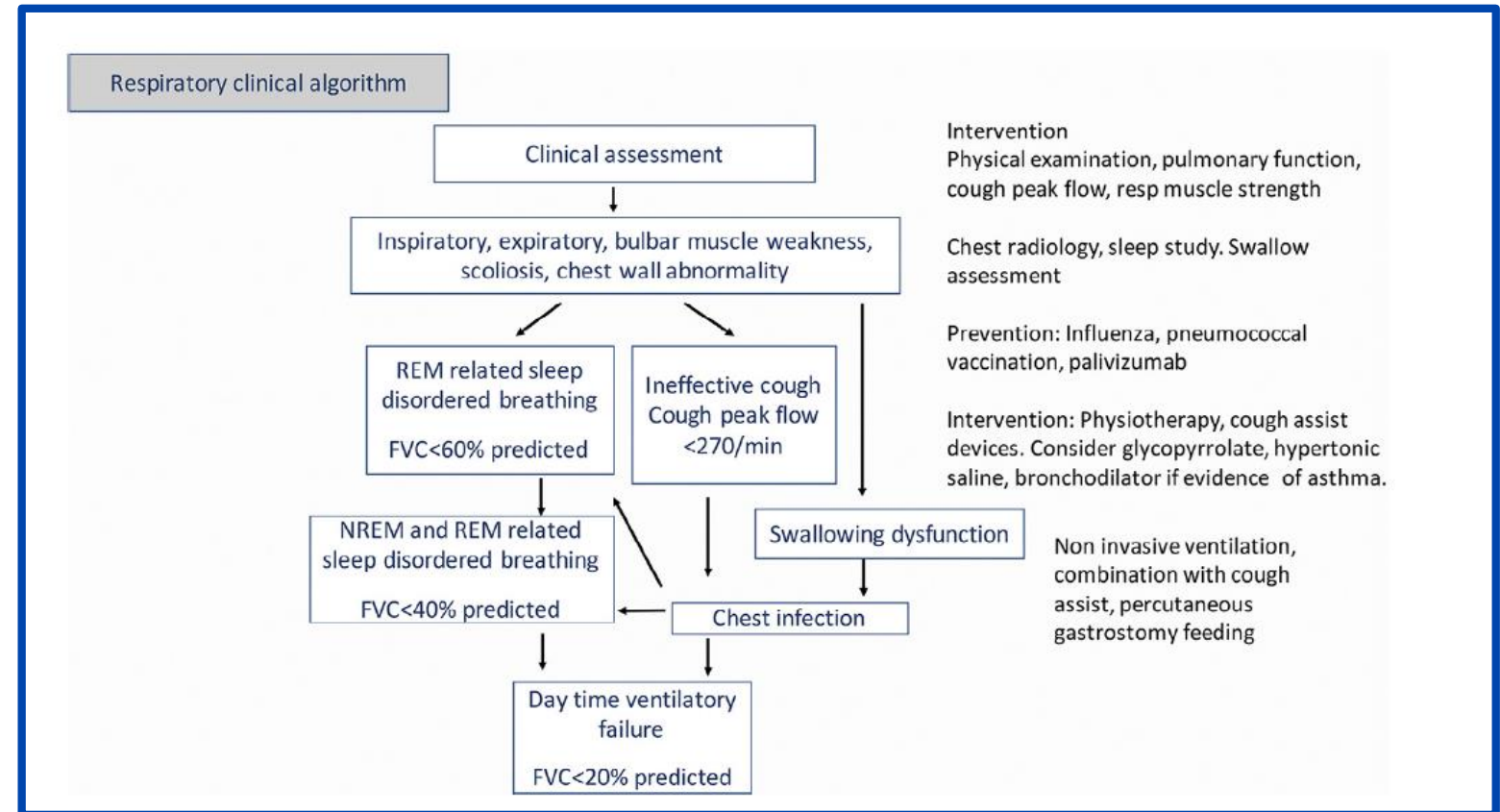


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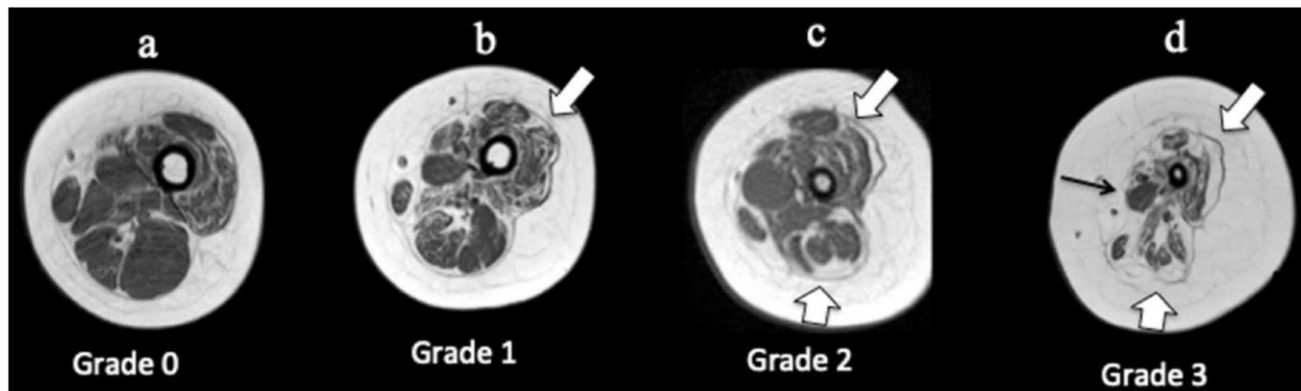
Lung Care

- Lung function tests
- Sleep study
- Respiratory physician
- Proactive airway clearance
- Physiotherapy/ cough assist
- Ventilation support



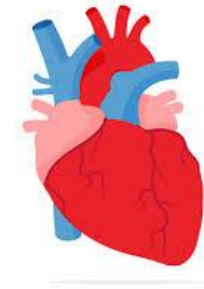
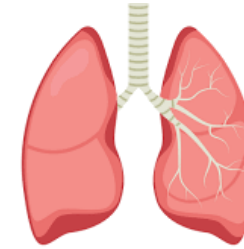
Bone & Muscle health

- modified bone–muscle cross-talk \Rightarrow \uparrow bone fragility
- Exposure to steroids
- Increased risk osteopenia and fractures
- Yearly DEXA & vitamin D
- Metabolic Bone Clinic & Consideration of medication









Other medical conditions

- Preventative health
- Immunisations
- Conditions of aging (!)
- Emergency plans
- Surgical management



Advance Care Planning

- **Appoint a Medical Treatment Decision Maker**
- **Chat and Communicate**
- **Put it on Paper**

<p>Patient Story</p>  <p>Dorothy was sick with many health conditions. Her family thought they knew what she wanted, but they weren't 100% sure. Together the family chatted and Dorothy wrote an Advance Care Directive. This he ...</p> <p>Read more ></p>	<p>Benefits of ACP</p>  <p>Advance Care Planning reduces stress for your family, clarifies treatments and identifies the legal decision maker for medical staff, gives you peace of mind</p> <p>Read more ></p>	<p>When to do ACP</p>  <p>Anyone can do Advance Care Planning.</p> <p>Read more ></p>
<p>ACP in 3 Steps</p>  <p>Advance Care Planning can be broken down into 3 steps.</p> <p>Read more ></p>	<p>Resources</p>  <p>Resources</p> <p>Read more ></p>	<p>Contact Us</p>  <p>Contact Us</p> <p>Read more ></p>

ACCESS TO TREATMENTS AND TRIALS

Access to treatments

- Specialist only medications
- Pre-approval access
- Specialist facilities
- Pipeline drugs

SPINRAZA SOLUTION FOR INJECTION R_x (S4)

Nusinersen (heptadeca Na); clear, colourless, isotonic, preservative free; vial with bromobutyl rubber stopper

Dose: Allow vial contents to warm to room temp; removal of volume of CSF \equiv to dose prior to admin recommended. Admin intrathecal bolus dose by lumbar puncture over 1-3 min. Do not admin into infected, inflamed areas. Loading (initiate as early as poss after diagnosis): 4 doses of 12 mg (5 mL) on days 0, 14, 28, 63; if dose delayed or missed, admin as soon as poss with \geq 14 day interval. Maintenance: 12 mg (5 mL) every 4 mths; if delayed or missed, admin as soon as poss and continue every 4 mths. Complex, see full PI

Pack 12 mg/5 mL 5 mL [1] (AUSTR282522)

Section 100 (Highly Specialised Drugs) - Public - Authority (MP) (Rp 3)

PBS: \$110,000.00

Approved Indication(s) for Authority:

Symptomatic Type I, II or IIIa spinal muscular atrophy (SMA)

Treatment Phase: Initial treatment of symptomatic Type I, II or IIIa SMA - Loading doses

Treatment criteria:

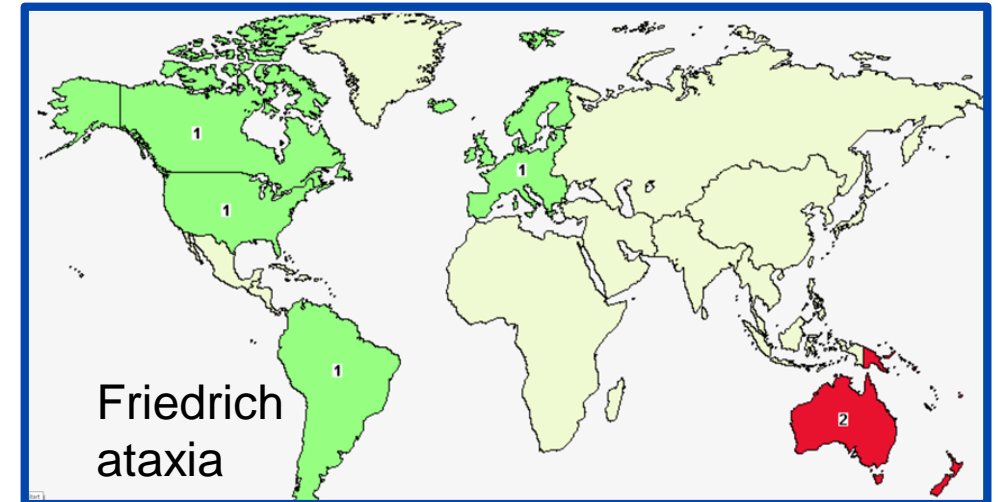
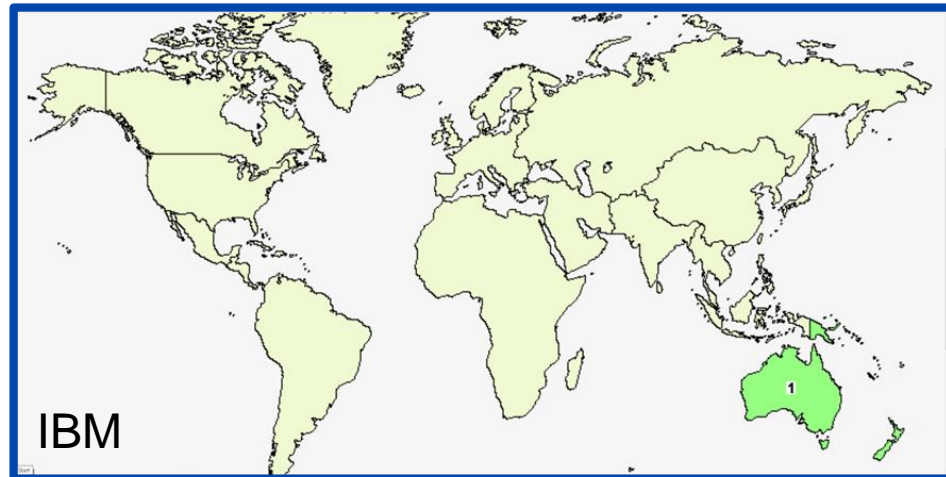
Must be treated by a specialist medical practitioner experienced in the diagnosis and management of SMA associated with a neuromuscular clinic of a recognised hospital in the management of SMA; or in consultation with a specialist medical practitioner experienced in the diagnosis and management of SMA associated with a neuromuscular clinic of a recognised hospital in the management of SMA.

Clinical criteria:

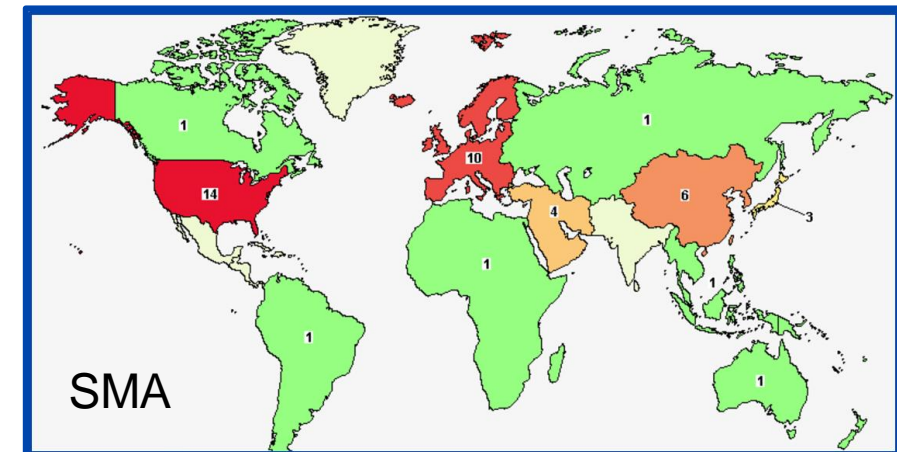
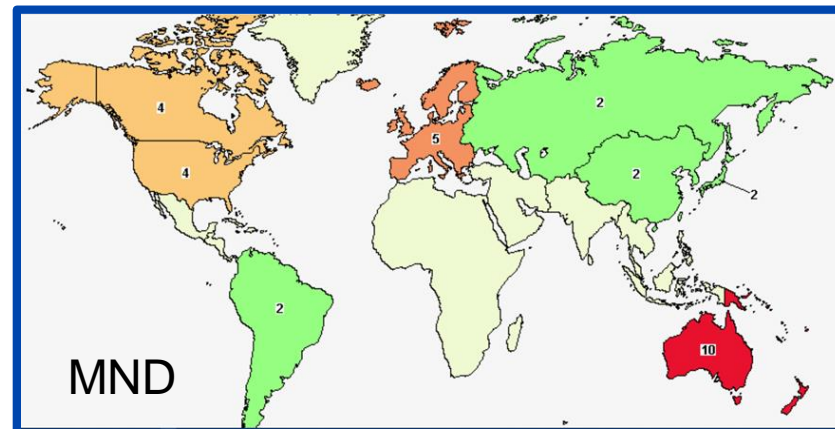
The condition must have genetic confirmation of 5q homozygous deletion of the survival motor neuron 1 (SMN1) gene; OR

The condition must have genetic confirmation of deletion of one copy of the SMN1 gene in addition to a pathogenic/likely pathogenic variant in the remaining single copy of the SMN1 gene,

Local interventional studies recruiting adults



- FSHD – 0
- Myotonic Dystrophy – 0
- DMD - 0
- CMT – 0
- LGMD – 0
- OPMD – 0
- Myotubular Myopathy – 0
- Bethlem Myopathy –
- Congenital Muscular Dystrophy -0



<https://www.australianmdregistry.org.au/>



register your interest



Home About us ▾ Type of diseases ▾ Clinical trial finder Contact us ▾

- Type of diseases
- DMD
- SMA
- FSHD
- Myotonic Dystrophy



Australian Neuromuscular Disease Registry
An Australia-wide registry of people
with a neuromuscular disease. It collects
important medical information from adult and child
patients across the country to improve the
understanding of neuromuscular disease and
accelerate the development of new therapies.

register your interest

