SCA living well March 2020

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Senior neurological clinicians
Duncan Foundation

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The Duncan Foundation

- Aim to identify and develop clinical services that will help improve the lives of New Zealanders living with neuromuscular conditions.
- Be recognised as a group of clinical leaders in the assessment and management of these conditions.
- Services accessable through accredited clinicians at main centres around New Zealand.
- We aim to provide a collaborative organisation that works hard to get maximum impact for people living with neuromuscular conditions.

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Duncan Objectives

- **National** network of accredited clinicians
- To increase nationwide therapists **understanding** of
  - the pathology of condition
  - the various presentation considerations
  - the effect of a condition on a whole person
  - treatment principles for management
- To provide clinical support for those living with a condition
- Latest research dissipated between the national network
- A hub of info – visibility and awareness

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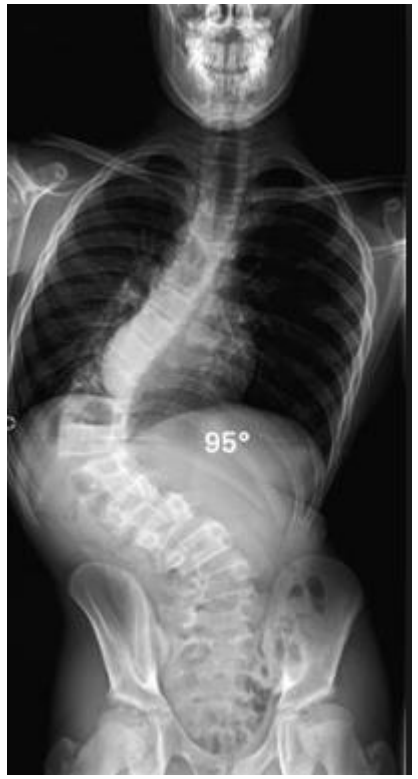
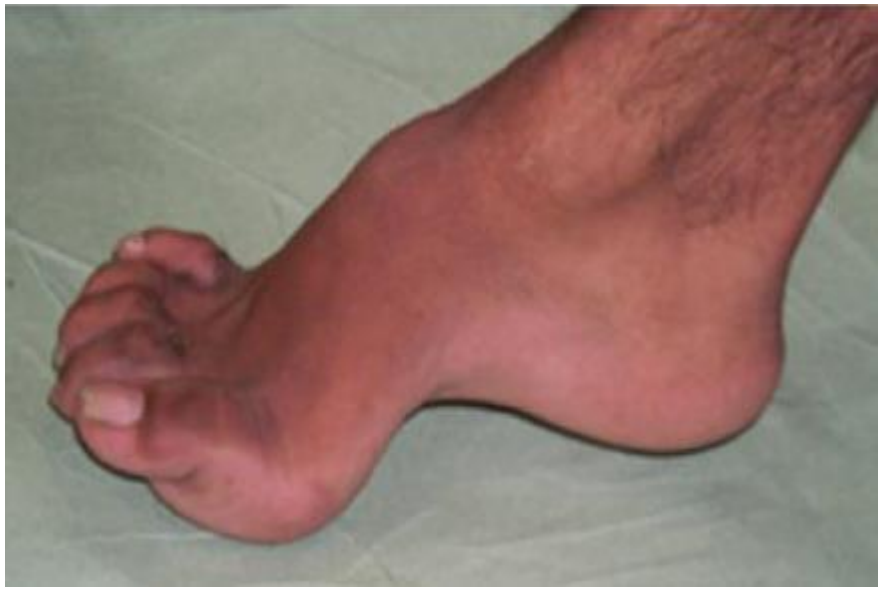
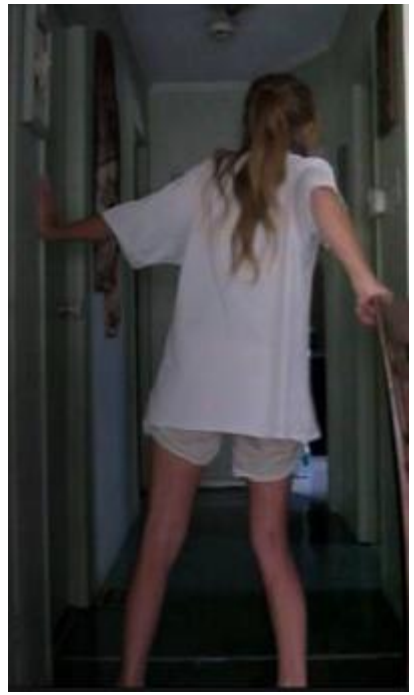


Affect function in everyday life





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Supports people living with a range of neuromuscular conditions - current focus on: Dystonia, Friedreich Ataxia, the Late Effects of Polio and Recently Diagnosed Parkinson's… and now SCA!!!

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Centre for Brain Research Neurogenetics Research Clinic!!!

What to expect

Contact from clinic coordinator – Kerry Walker

Schedule appt at Auckland Hospital

2 appointments Dr and Neurologist and then with PT and OT

Physical tests (hand function, walking) and questionnaire.

Issues presented further discussed and options for management

The team will hand over saves repeating info

Report and referrals made

Follow up

Duncan Registration/Lindsay Funding

Register Duncan Foundation

Contacted and Follow up outside clinic can be provided

Then …

Resources provided (advice, stretches, pacing, fatigue mgmt, sleep

Individual exc diagnosis specific considerations

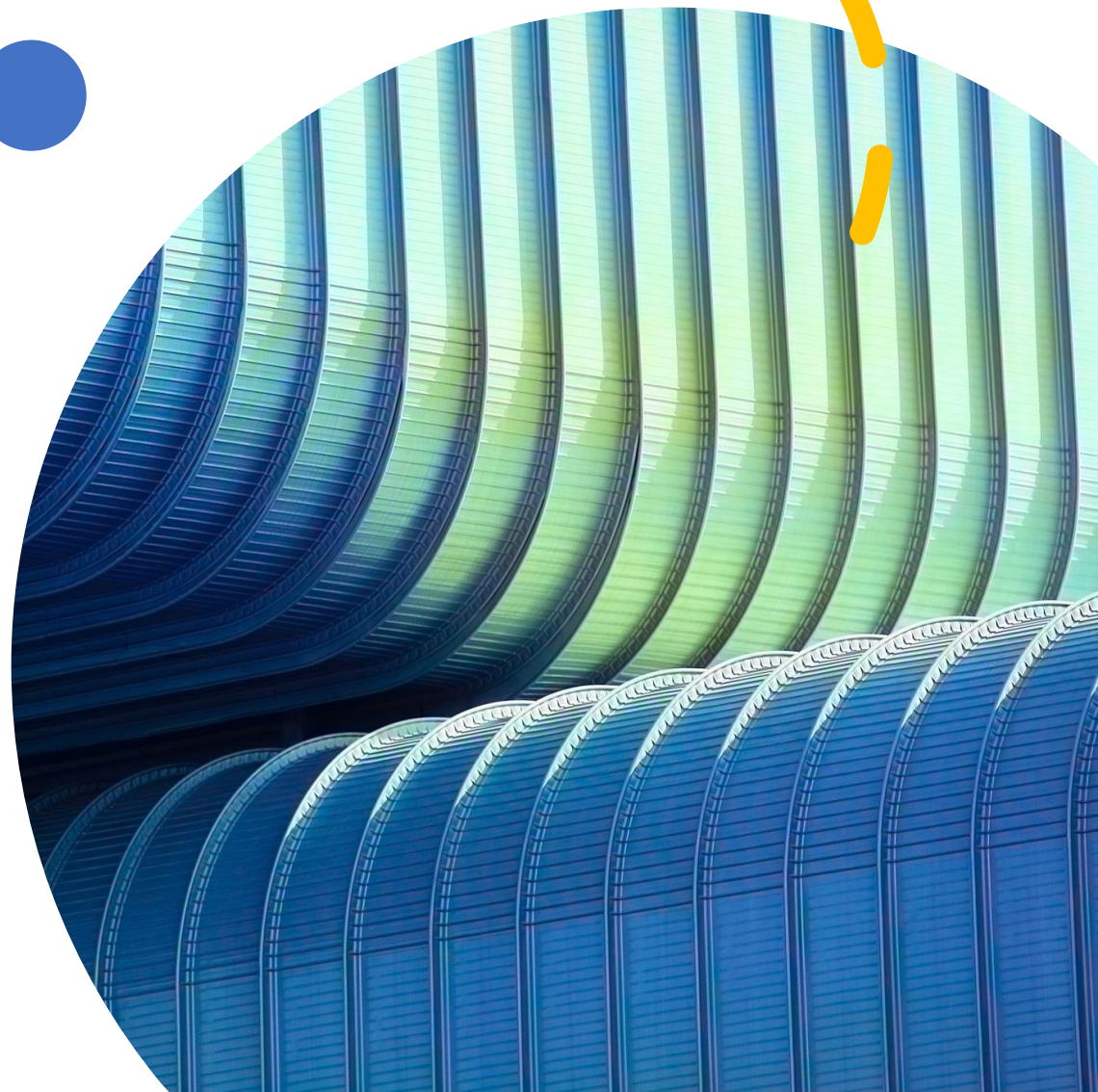
Referral to GP for ACC services

Referral to community OT?PT for rehab specific goals and measures provided

Referral for wheelchair review or application

Referral to orthotic for review or consideration of splint

Condition specific treatment following guidelines



Also may…

- Liaise with Local ACC provider if needed, (may recommend a neurological provider)
- Liaise with Local provider if needed, may recommend a neurological provider and provide support if identifies limited condition knowledge
- Following initial assessment may schedule a follow up to provide liaison or condition specific management and exc prescription maybe via clinic or ZOOM online meeting
- May be community visit for pool exc, gym visit,
- Specialised vertigo or visual treatment techniques
- FOLLOW UP ideally– AT 3 MONTHS or 6 MONTHS
- CAPTURE IMPACT OF INTERVENTION

Trying to understand the gaps from your perspective…

DELAYS TO DIAGNOSIS

DELAYS IN ACCESSING EQUIPMENT – HOME OR WALKING

ACCESS TO EXPERIENCED THERAPY STAFF

ACCESS TO GYM/POOL

NOT KNOWING WHAT TO DO – EXERCISE

IF WE KNOW WE CAN MAKE MORE OF AN IMPACT

What do you want?

- For us to know we are helping you in the way YOU want/need…
- How can we support your community?
- **Would you be happy to be interviewed by me to guide our support service for those living with a progressive neuro muscular condition?**

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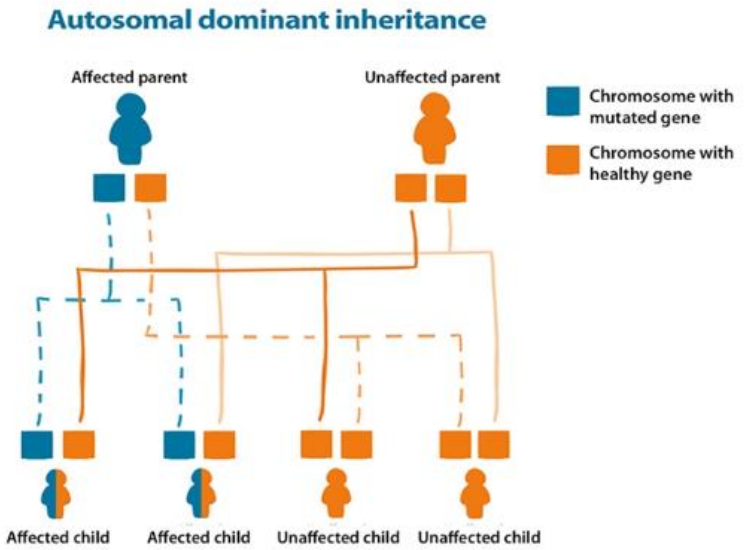
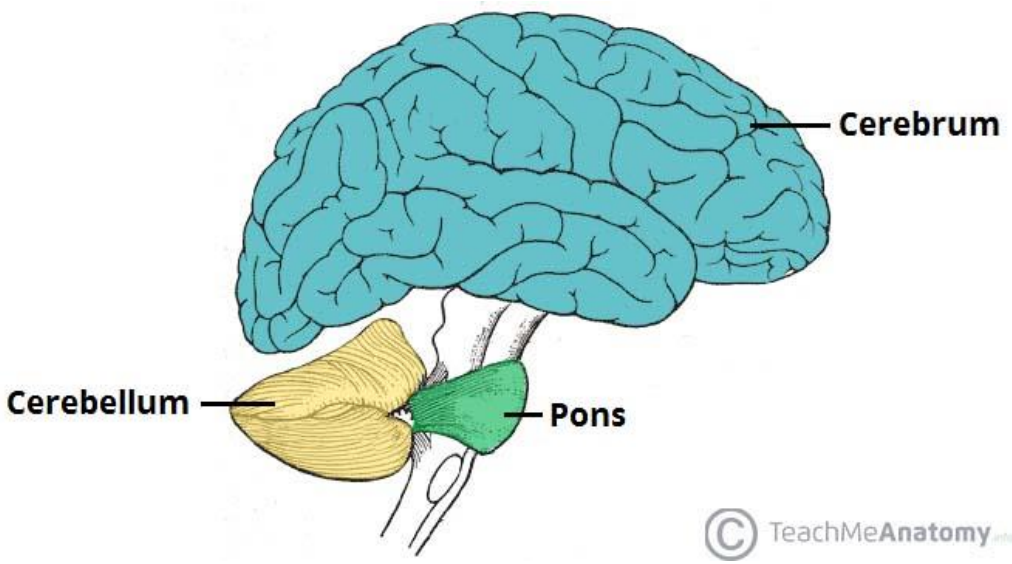
Spinocerebellar Ataxia

- SCA subset of hereditary cerebellar ataxias that are autosomal dominantly transmitted
- Progressive neurodegenerative disorder
- Degeneration of the cerebellum

- Ataxia- incoordination and balance dysfunction in movements without muscle weakness





Types of Spinocerebellar Ataxia

- Ataxia gene- SCA1-SCA36
- Age of onset
- Progression of the Ataxia
- Distinguishing features

SCA1	*ATXN1*	3rd–4th decade (<10 to >60)	15 years (10–28)	Pyramidal signs, peripheral neuropathy
SCA2	*ATXN2*	3rd–4th decade (<10 to >60)	10 years (1–30)	Slow saccadic eye movements, peripheral neuropathy, decreased DTRs, dementia
SCA3	*ATXN3*	4th decade (10–70)	10 years (1–20)	Pyramidal and extrapyramidal signs; lid retraction, nystagmus, decreased saccade velocity; amyotrophy fasciculations, sensory loss
SCA4	16q22.1	4th–7th decade (19–72)	Decades	Sensory axonal neuropathy, deafness; may be allelic with 16q22-linked SCA
SCA5	*SPTBN2*	3rd–4th decade (10–68)	>25 years	Early onset, slow course; first reported in descendants of Abraham Lincoln
SCA6	*CACNA1A*	5th–6th decade (19–71)	>25 years	Sometimes episodic ataxia, very slow progression
SCA7	*ATXN7*	3rd–4th decade (0.5–60)	20 years (1–45; early onset correlates with shorter duration)	Visual loss with retinopathy
SCA8	*ATXN8/ ATXN80S*	4th decade (1–65)	Normal life span	Slowly progressive, sometimes brisk DTRs, decreased vibration sense; rarely, cognitive impairment
SCA10	*ATXN10*	4th decade (12–48)	9 years	Occasional seizures; most families are of Native American background
SCA11	*TTBK2*	Age 30 years (15–70)	Normal life span	Mild, remain ambulatory
SCA12	*PPP2R2B*	4th decade (8–62)		Slowly progressive ataxia; action tremor in the 30s; hyperreflexia; subtle Parkinsonism possible; cognitive/psychiatric disorders including dementia
SCA13	*KCNC3*	Childhood or adulthood	Unknown	Mild intellectual disability, short stature
SCA14	*PRKCG*	3rd–4th decade (3–70)	Decades (1–30)	Early axial myoclonus
SCA15	*ITPR1*	4th decade (7–66)	Decades	Pure ataxia, very slow progression
SCA16	*SCA16*	Age 39 years (20–66)	1–40 years	Head tremor; one Japanese family
SCA17	*TBP*	4th decade (3–55)	>8 years	Mental deterioration; occasional chorea, dystonia, myoclonus,

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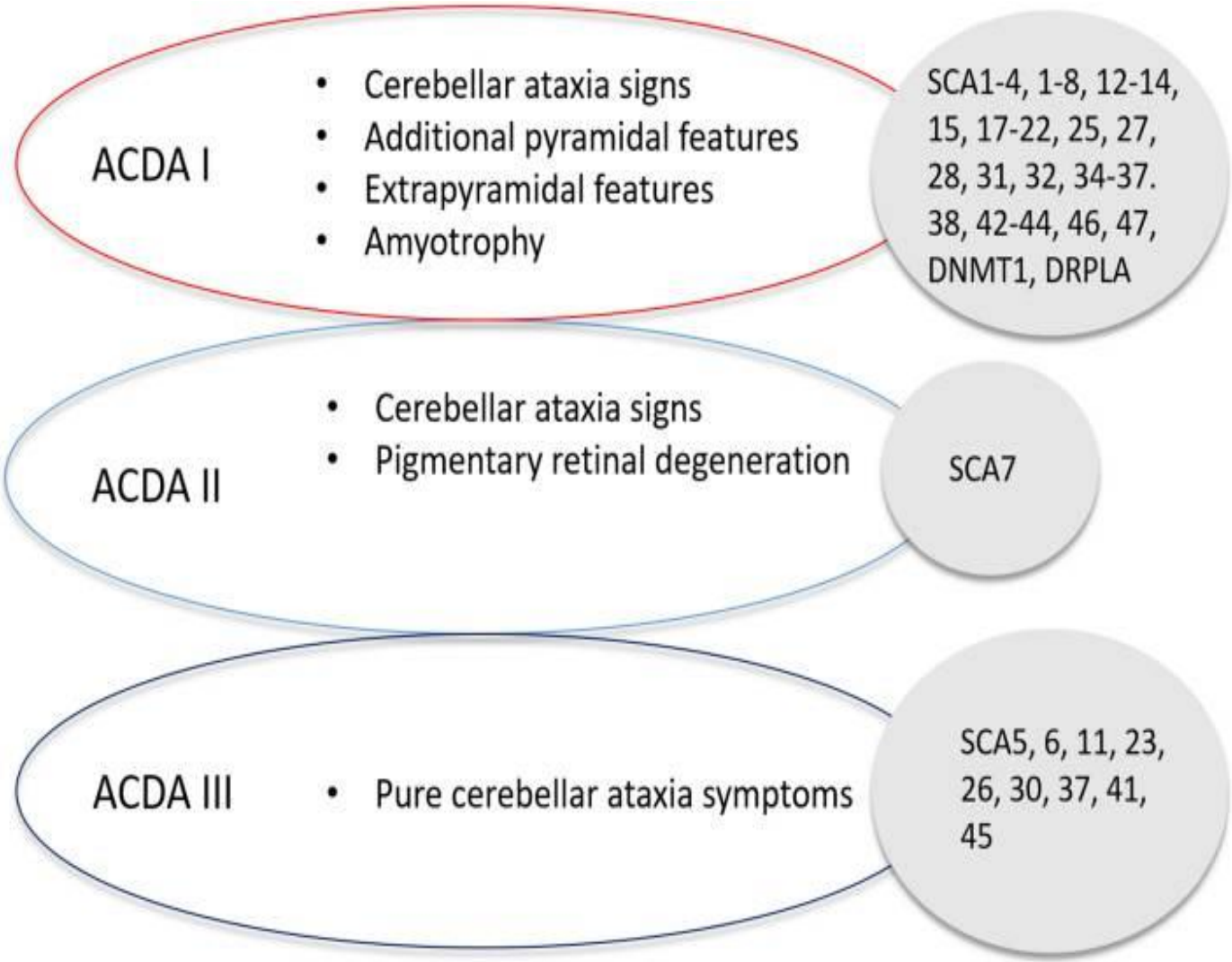

Associated clinical features	Genetic subtypes
Peripheral neuropathy	1, 2, 3, 4, 18, 25, 38, 43, 46
Pyramidal signs	1, 3, 7, 8, 10, 14, 15, 17, 35, 40, 43
Dystonia	3, 14, 17, 20, 35
Myoclonus	14
Parkinsonism	2, 3, 10, 14, 17, 19/22, 21
Tremor	12, 15, 27
Chorea	17, 27, DRPLA
Cognitive impairment	2, 8, 13, 17, 19/22, 21, 36, 44, DRPLA
Psychiatric symptoms	2, 17
Ophthalmoplegia	2, 3, 28, 40
Visual impairment	7
Face/tongue fasciculation	36
Ichthyosiform plaques	34
Seizures	10, 19/22, ATN1
Narcolepsy	DNMT1
Hearing loss	31, 36, DNMT1

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Common Symptoms…

Incoordination

clumsiness

Gait
Ataxia/*changes*
in walking

Fatigue

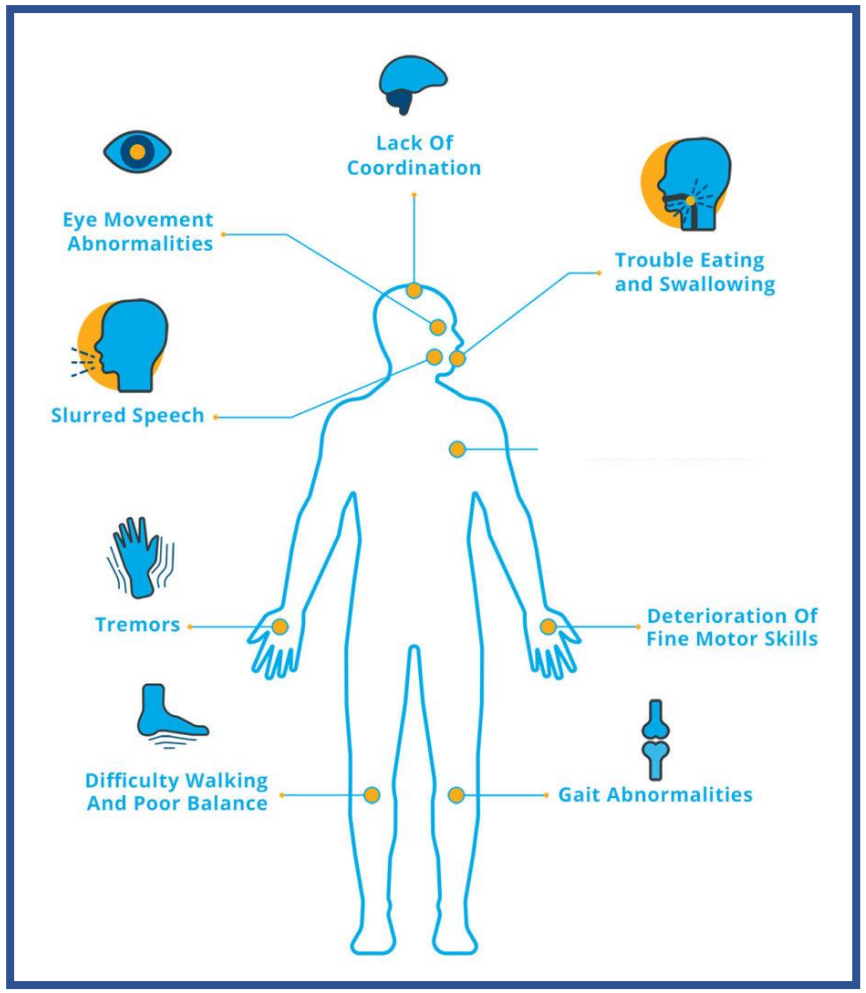
Reduced hand eye co-ordination

Difficulty with fine motor tasks such as buttons, writing

Falls

Changes in learning and processing information

Difficulty or changes in memory



Tremor

Changes in swallowing

Changes in speech

Changes in balance

Involuntary eye movement

Dizziness

Unsteadiness

Stiffness

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Decreased Coordination in Legs and Trunk

+ Impaired Balance

=

Variability of steps

Increased Falls

Fatigue

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How Can Therapists help?

- Interventions by therapists such as Physiotherapists, Occupational Therapists and Speech and Language Therapists play a vital role in the management of people with SCA
- All recommendations advise people with SCA should have access to the full range of therapies
- Ultimate aim to maintain function and activities of daily living
- Prevention of secondary complications
- Advice

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What does Physiotherapy help with?

- Restorative vs compensatory approach

- Balance / falls prevention
- Co-ordination
- Mobility
- Posture
- Muscle strength, length and tone
- Exercise (endurance)

NEURO PHYSIO

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Physiotherapy Approaches…

- Motor training (Synofzik et al, 2014)
- Dynamic task practice
- Gait and Balance training
- Strength and flexibility training

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Research into Physiotherapy and SCA

- Video based co-ordination training
- Treadmill training
- Balance and mobility aids
- Axial Weighting
- Lycra garments
- Upper limb rehabilitation
- General Exercise
- Intensity of therapy

Game Based Training

- Exergames
- Video Games

- Wang et al (2018)
- Schatton et al (2013)
- Synofzif et al (2013)

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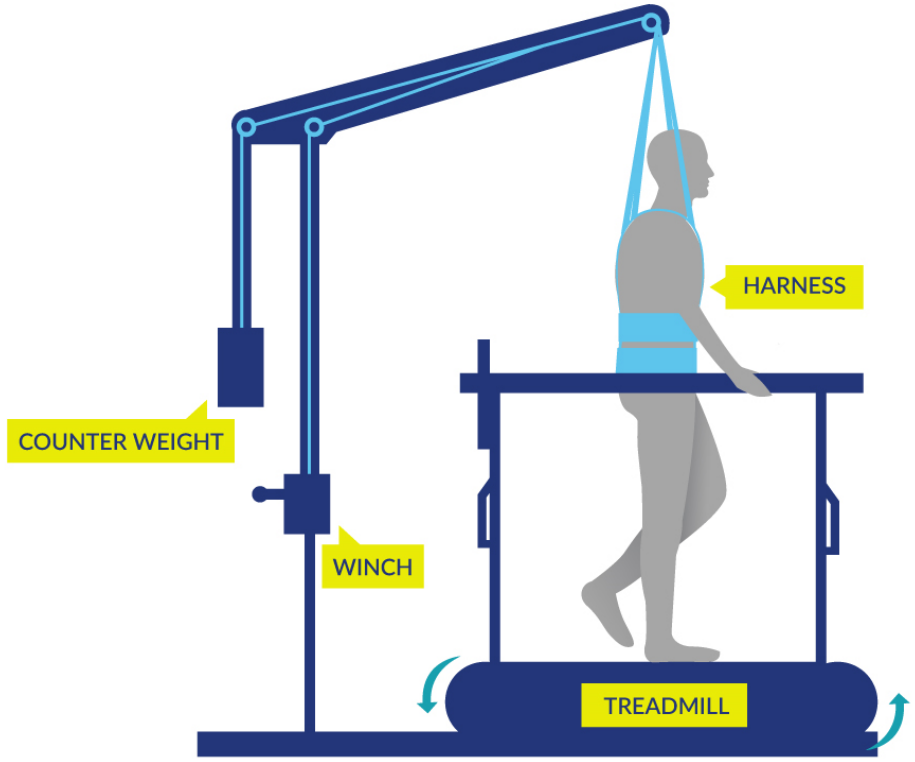
Treadmill Training

- Partial body weight support treadmill training

- Santos de Oilveira et al (2018)

- Ataxia after brain injury

- Cernak et al, 2008, Vaz et al, 2008



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Axial Weighting

- Ankle weights
- Trunk weights

Clopton et al (2003)
Perlmutter and Gregory (2003)

Upper Limb Rehabilitation

- Upper Body Symptoms – tremor (postural, kinetic), reduced co-ordination, abrupt movement, reduced ability to make accurate rapid or alternating movements.

- Wrist weighting
- Robotics
- Manipulation of visual information
- Cooling therapy
- Assistive devices

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Exercise

- Swimming/hydrotherapy,
- General fitness training
- Pilates
- Yoga
- Horse riding
- Climbing

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Balance and Walking Aids

- What type of aid?
- Should I be using a walking aid?
- Touch support vs upper body weight bearing
- Wheelchairs…
- What is available?

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Summary of the Research

- Those with SCA benefit from coordinative training through physiotherapy or on exergames
- Even people with advanced SCA will benefit from therapy
- Improvements with regular therapy are equal to regaining 1 or more years of disease progression
- Improvements in therapy have been shown to be ataxia specific
- Retention of training effects depends on the continuity of training often through home exercise programmes

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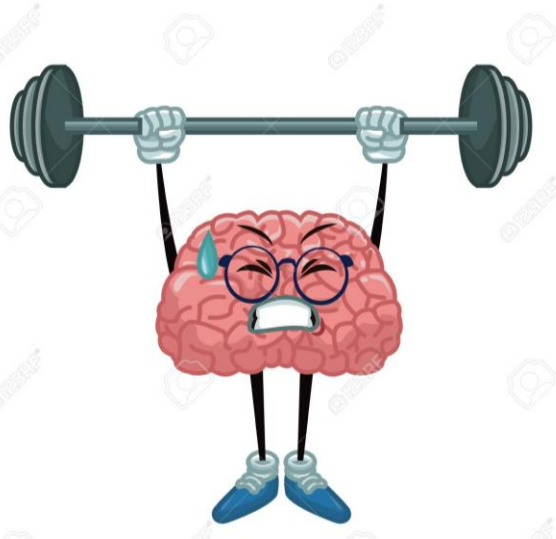
Intensity of Physiotherapy and SCA

- Intensive Physiotherapy what does this mean?
- Research into intensity *Miyai et al (2010), Ilg et all (2009, 2010)*
- Long term effect?



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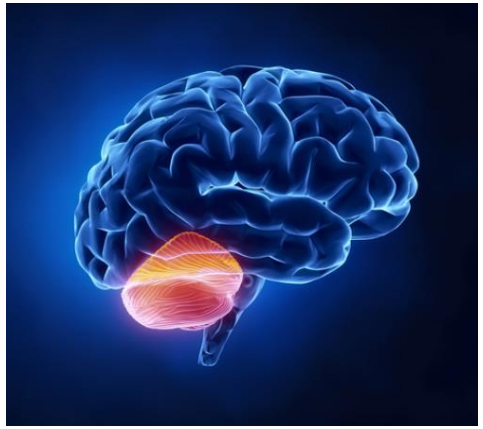
Exercise may help regain functional performance of one or more years of the disease progression!!!

- SARA scores may worsen 0.4 to 2.2 points per year depending on type of ataxia (Jacobi 2011)

- Intensive Coordination Therapy studies have shown SARA Scores improve up to 5.2 points = Gaining 2 or More YEARS of disease progression

  (Miyai, Ilg 2009,2010)



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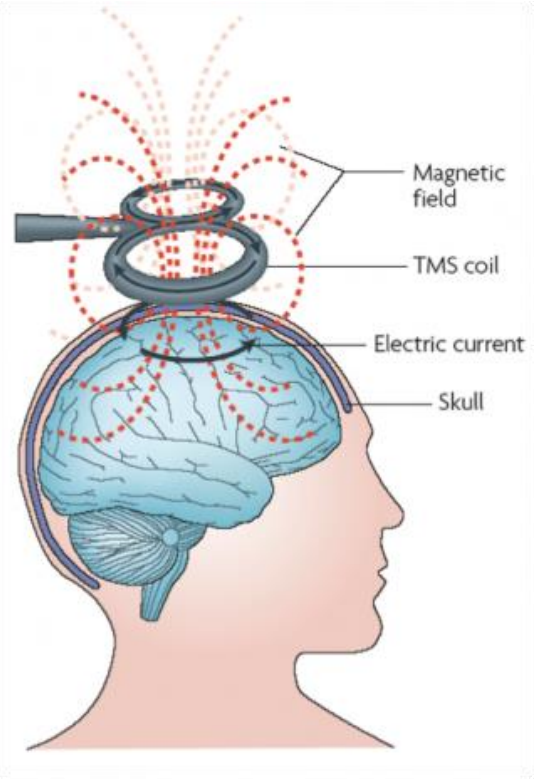
Clinical Trials

- Trans magnetic stimulation (Manor et al, 2017)
- Tai Chi
- Whole body vibration training
- Aerobic training vs balance training



www.clinicaltrials.gov

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Team management considerations

- Balance and gait retraining and
- Foot sensory stimulation ? Shakti Mats
- Prescribe walking aids – progressive
- HEP's
- Balance retraining
- Sitting/standing
- Hydro
- Spasticity management
- Gait re ed and strategies to prevent falling
- Awareness
- Proprioception
- Sensory/pressure
- Core/posture
- Manual therapy input

That's not even covering RLS, management, QOL, Mental health, considerations, respiratory

- General health
- Education
- Pain management
- Maintain Range of movement
- Postural alignment
- Stretches
- Strengthening antagonist
- Relaxation – direct effect or coping?
- Vestibular

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Julie Rope and Christine Tooke

wish to thank the Lindsay Foundation for their support in accessing the SCA community

www.duncanfoundation.org

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