



Ataxia

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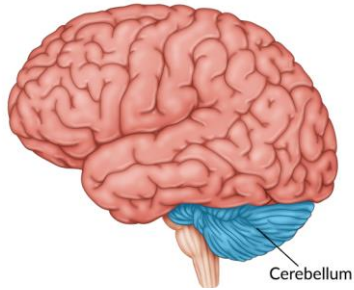
Cerebellum and gait

Causes of ataxia

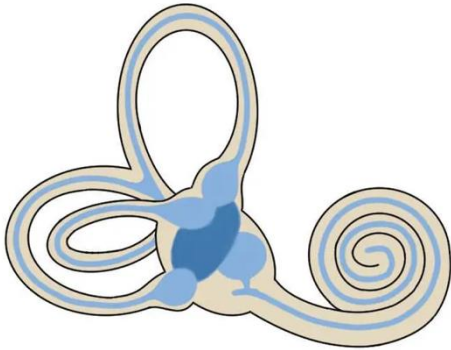
Evaluation of ataxia and ataxia syndromes

Management of ataxia/cerebellar disorders

Ataxia – disorder of balance and coordination



Cerebellar ataxia - Gait ataxia (wide-based), limb ataxia, oculomotor deficits, dysarthria



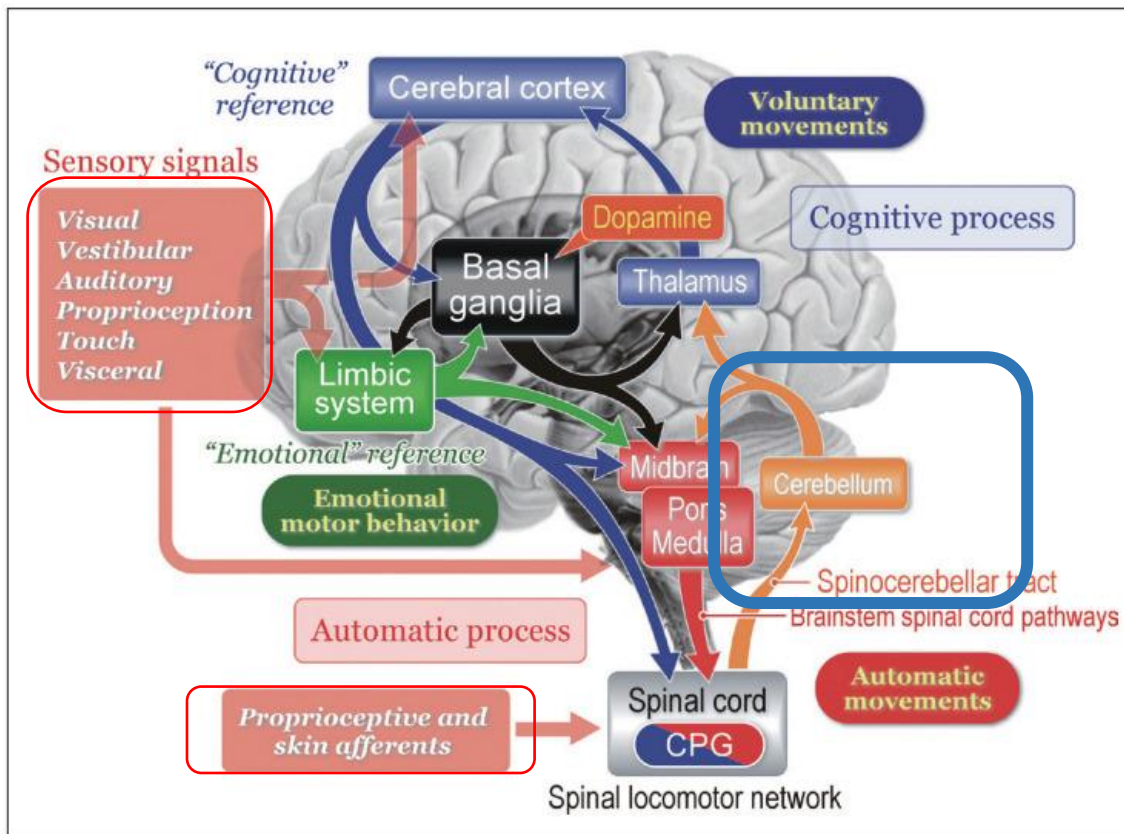
Vestibular ataxia – vestibular neuronopathy/ vestibular dysfunction (motion oscillopsia, gait ataxia, abnormal VOR on Head Impulse Test)



Sensory ataxia – proprioceptive loss, peripheral neuropathy (gait ataxia with positive Romberg's sign)

Many ataxia disorders – have overlap of cerebellar + vestibular + peripheral nerve dysfunction

Gait control is very complex!!!



Cerebellum is a center for integration of vestibular information (balance)

Gait problems is one the first manifestations of cerebellar syndrome!!!!

Takakusaki K. Functional Neuroanatomy for Posture and Gait Control. J Mov Disord. 2017;10(1):1-17.

Topo-diagnostic representation of locomotor control within the cerebellum

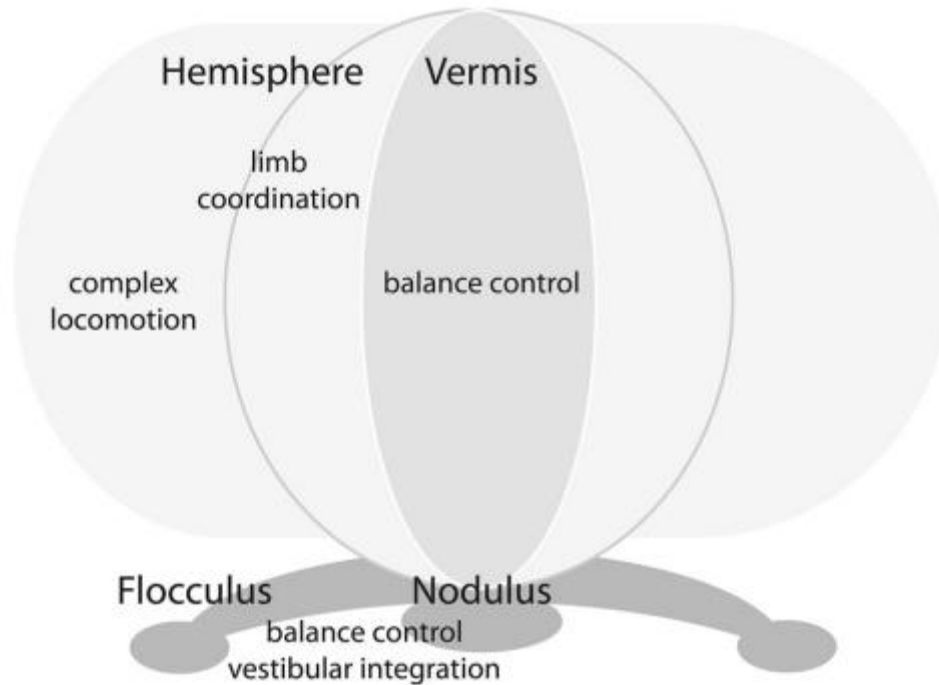
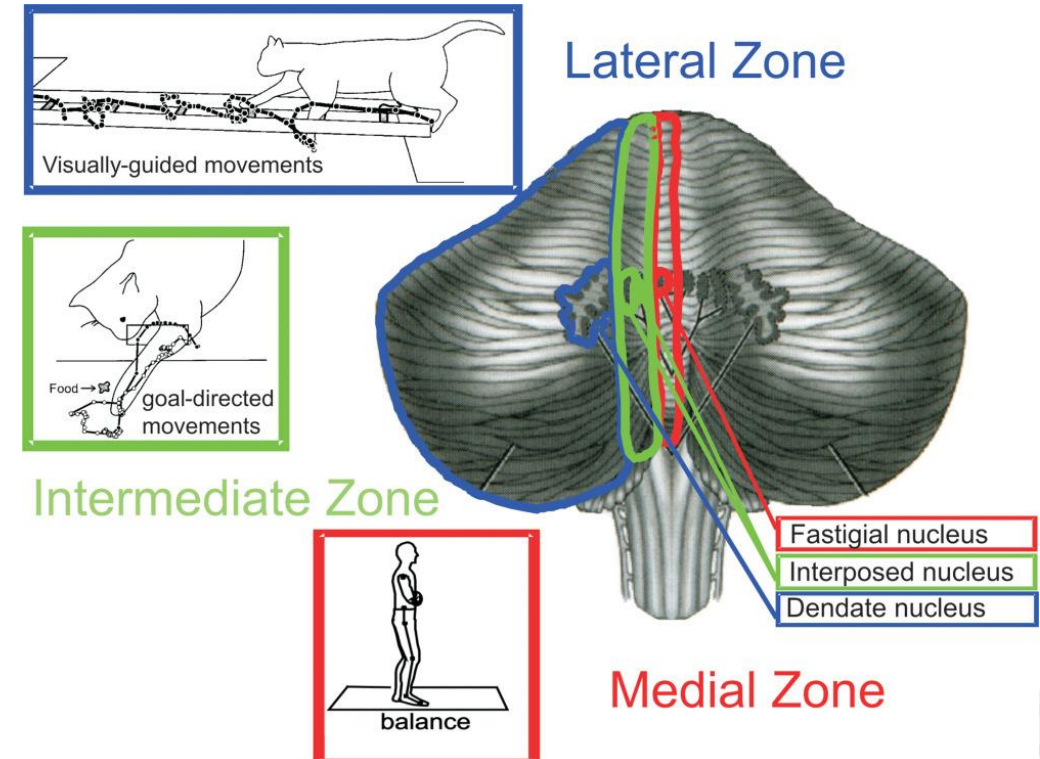


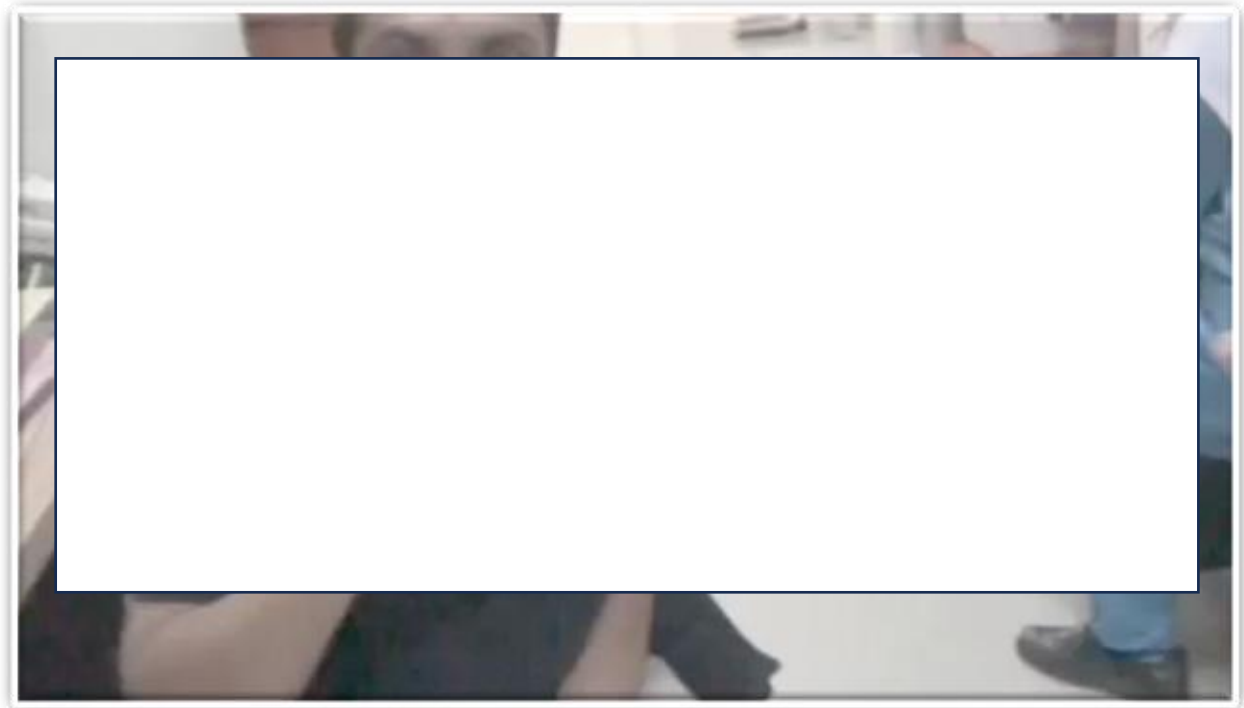
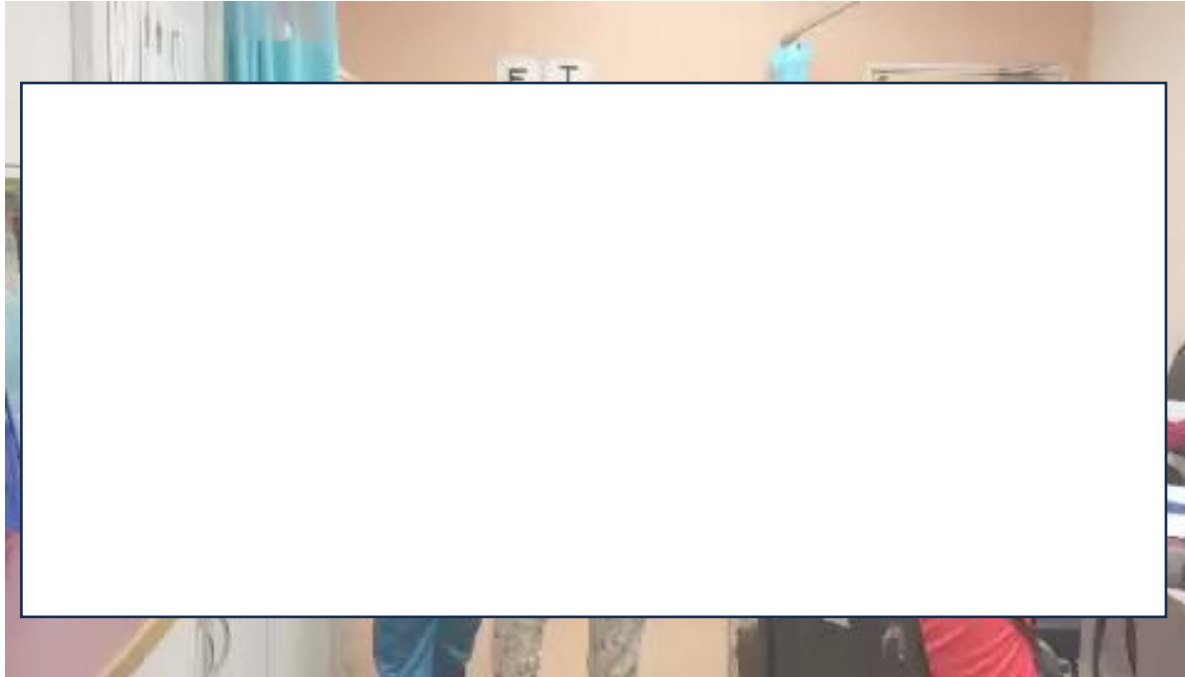
Fig. 2 Topodiagnostic scheme for the different locomotor control functions within the cerebellum. Midline structures (vermis, nodulus,

- *Midline (vermis, nodulus, flocculus) – upright posture*
- *Flocculus – integration of vestibular information into locomotor network*
- *Intermediate zone – Feedforward control of multi-joint movement and important for intra-limb and inter-limb coordination during walking*
- *Lateral zones – higher locomotor function such as target-guided locomotion and visually guided stepping*

Ataxia

- Depends on the cerebellar structure or pathways affected
- Truncal ataxia and Gait Ataxia (**Midline-vermis/paravermis involvement**)
- Limb Ataxia – Appendicular Ataxia (**Cerebellar hemisphere involvement**)
- Nystagmus and eye movement abnormalities (**Flocculonodular lobe**)
- Speech disturbances (**Dysarthria**)





Gait ataxia (Truncal ataxia) – unsteadiness – Wide based

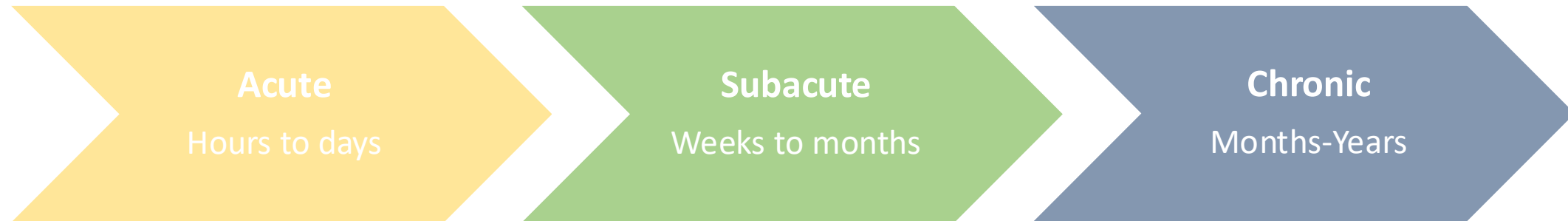
- Present in all three ataxia types... (not very discriminative –unless very severe ataxia)
- More severe in cerebellar disorders (vermis)

Cerebellar dysfunction

Appendicular ataxia- Dysmetria and irregularity of rapid alternating movement (complex movement)

Causes of Ataxia – many!!!

Onset and Clinical Course



- Vascular
- Miller-Fisher
- Infective (cerebellitis)
- Demyelinating
- SOL

- Nutritional (Vitamin deficiency)
- Toxic – ethanol, alcohol
- Drugs - anticonvulsants
- Paraneoplastic /Immune mediated encephalitis
- GAD Ataxia*
- Prionopathies

- Degenerative
 - Sporadic
 - Inherited
- Immune mediated
 - Gluten Ataxia*

Blood investigations , Neuroimaging, Antibody testing, Vitamin B1,B12
Autoimmune/paraneoplastic antibodies (anti-Yo, Ma, Ri)

Genetic Ataxia



Recessive Ataxia*

< 20
years

Later
onset

FRDA commonest
AOA types 1 and 2
Ataxia Telangiectasia

**Ataxia with Vitamin E Def
(AVED)*
**Wilson's disease*
**CTX (cerebro tendinous
xanthomasosis)*
Refsum's disease

CANVAS
Late onset Freidreich's
Ataxia (LOFA)

MIRAS (mitochondrial)
SPG-7
Rarer Ataxia syndromes
(Sialidosis, NCL)



Dominant Ataxias
Mostly >20 years

Common SCA mutations
SCA 1,2,3,6,7,17, DRPLA
SCA 31 (Japan), SCA 12 (India)

Other SCAs



Mitochondrial

NARP

X-linked

FXTAs (adult males)
Adrenoleuko-
dystrophy

Non Ataxia features – may influence gait in genetic ataxias (SCA)

Table 1

SCA subtypes with associated clinical signs that feature prominently with cerebellar ataxia

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

ATN1 atrophin 1, mutation responsible for dentatorubral–pallidoluysian atrophy, *DNA methyltransferase 1*, mutation responsible for ADCA-deafness and narcolepsy

Chorea = SCA 17

Pyramidal signs
SCA 1,2,3,7,8,10,15,17,35,40,43

Ophthalmoplegia
SCA 2,3,28,40

Myoclonus
SCA 14

There are 48 genes

Onset in adulthood, 30s

Autosomal dominant,
some may appear
sporadic (SCA 6)

Non ataxia features may
predominate

Characteristics of Gait Ataxia (Pure Ataxia)

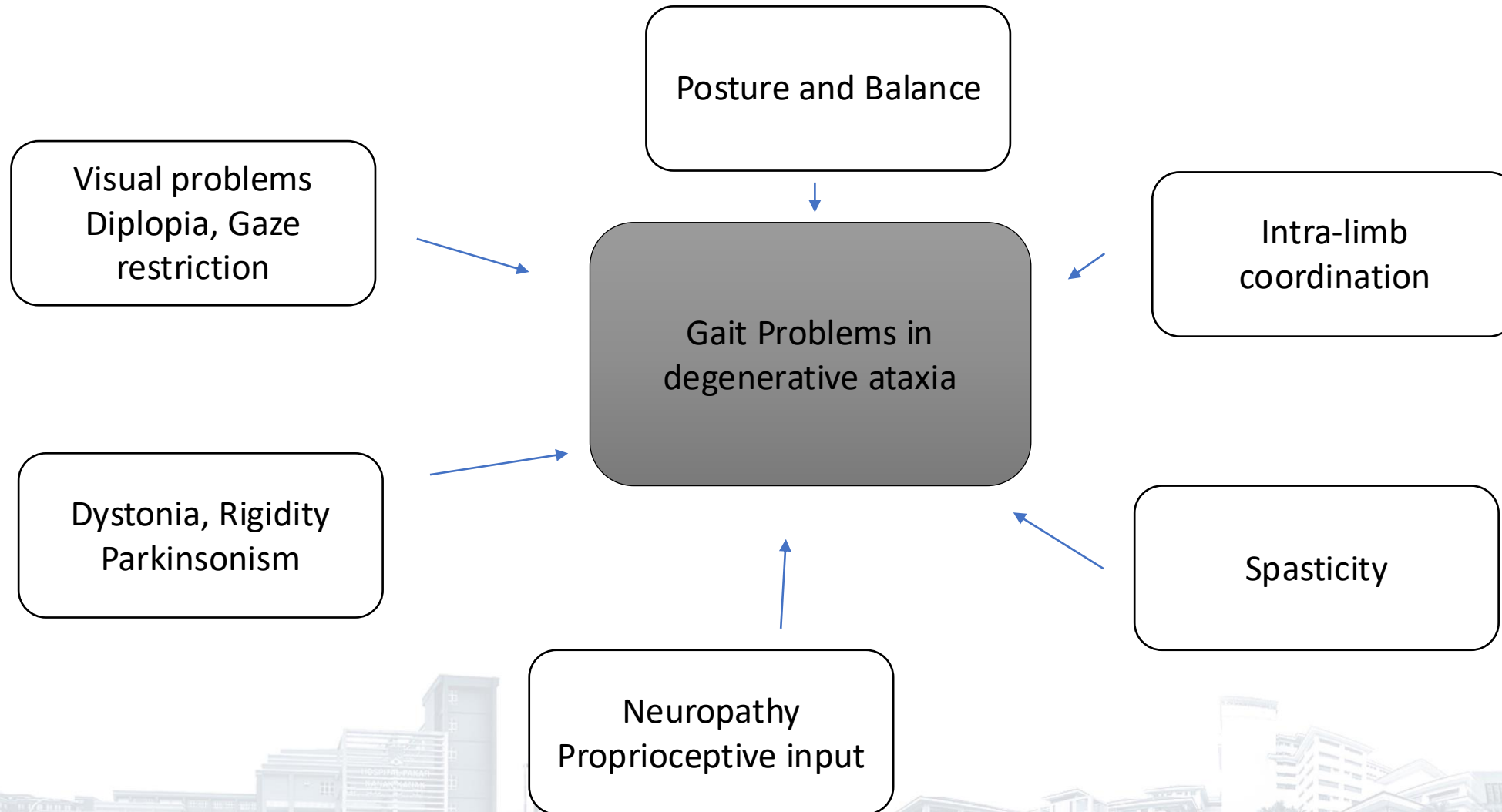
Clinically

Gait ataxia = (Unsteady gait)

- Wide-based stance with truncal instability and irregular lurching steps
- Increased risk of falls
- Maybe accompanied or predominated by other symptoms (depending on the ataxia subtype)
- **Unstable stumbling walking path, increased step width and high spatio-temporal variability of gait**

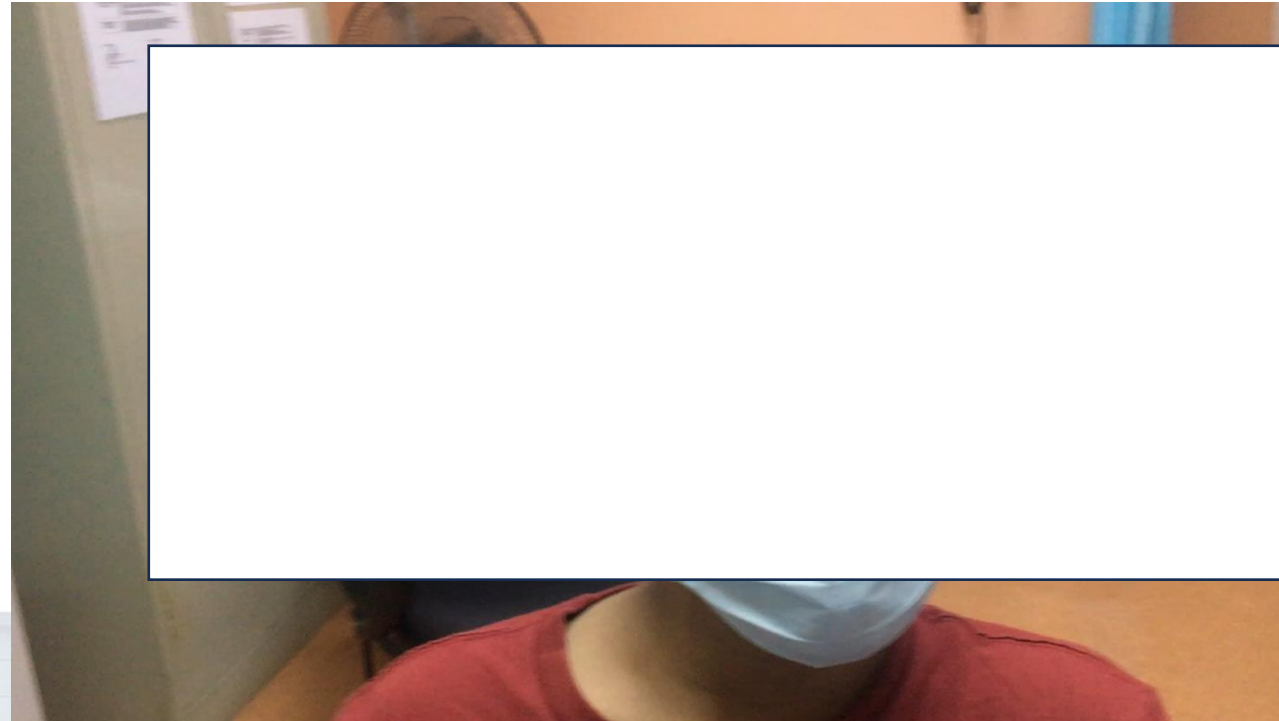
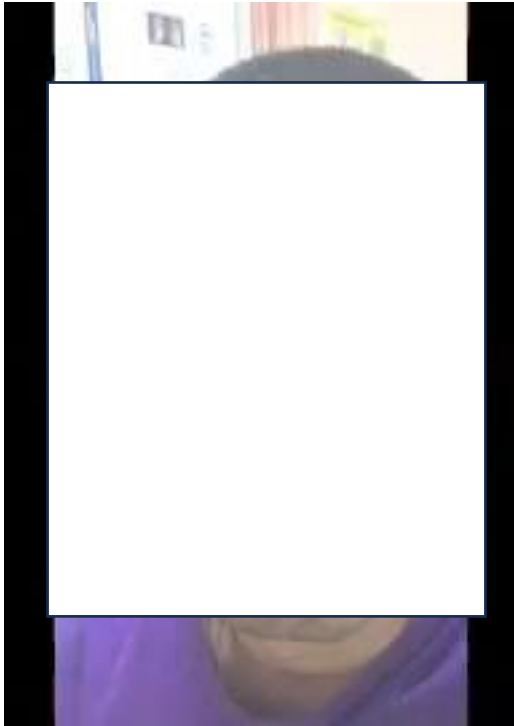
Cerebellar Disorders – Heterogeneous gait

- Pure Cerebellar gait
- Mixed pattern (cerebellar gait plus)
 - Pyramidal (Spasticity) Multiple System Atrophy; SCA 1,2,3
 - Dystonia
 - Dominant and Recessive Ataxias, Wilson's disease
 - Parkinsonism
 - MSA; SCA 1,2,3
 - Neuropathy
 - Freidreich's Ataxia, SCAN etc (mixture of high stepping gait)



Other features of cerebellar syndrome

Oculomotor deficits – quite early in the disease



Square wave jerks
Ophthalmoparesis
Hypermetric saccades
Hypometric saccades
Saccadic slowing
Saccadic pursuit

Other features of cerebellar syndrome

Oculomotor deficits – quite early in the disease

Square wave jerks
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Hypometric saccades
Saccadic slowing
Saccadic pursuit

Other movement disorders may predominate (SCA3)

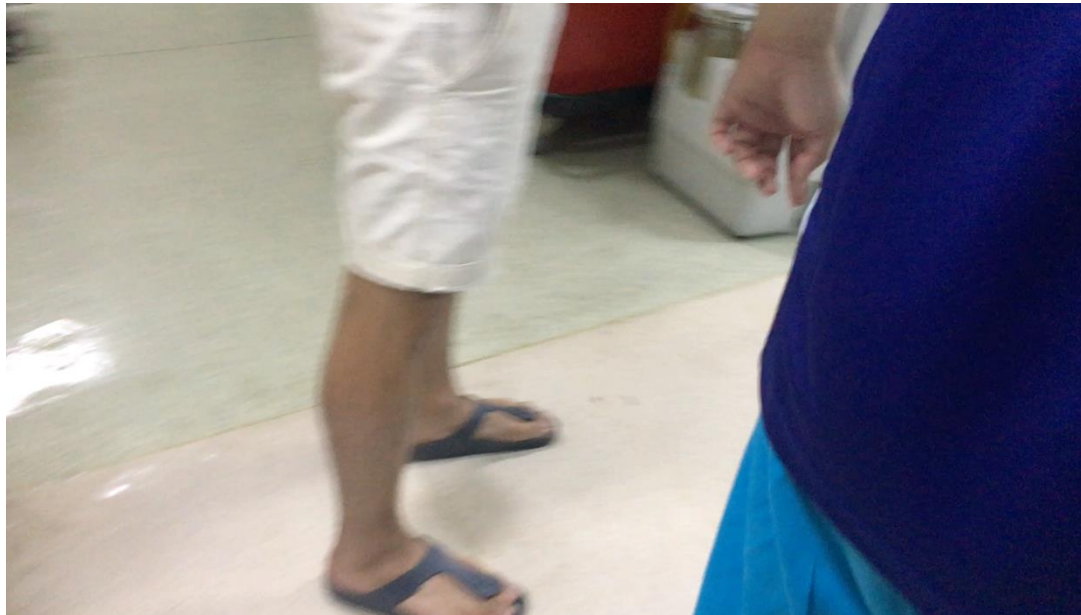
- Heterogeneous with intra-familial and interfamilial variability



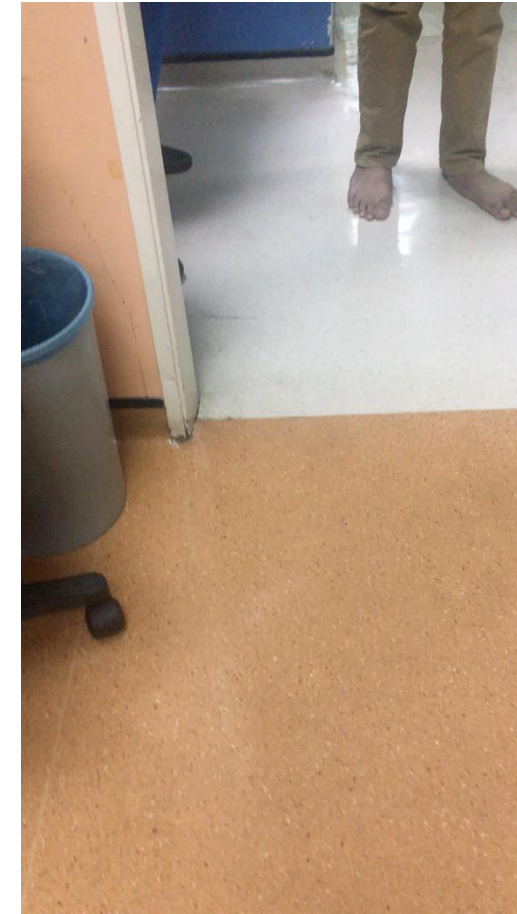
Mixed
movement
disorders

Dystonia +
Parkinsonism+
Pyramidal
syndrome

GAIT DIFFICULTIES IN DA

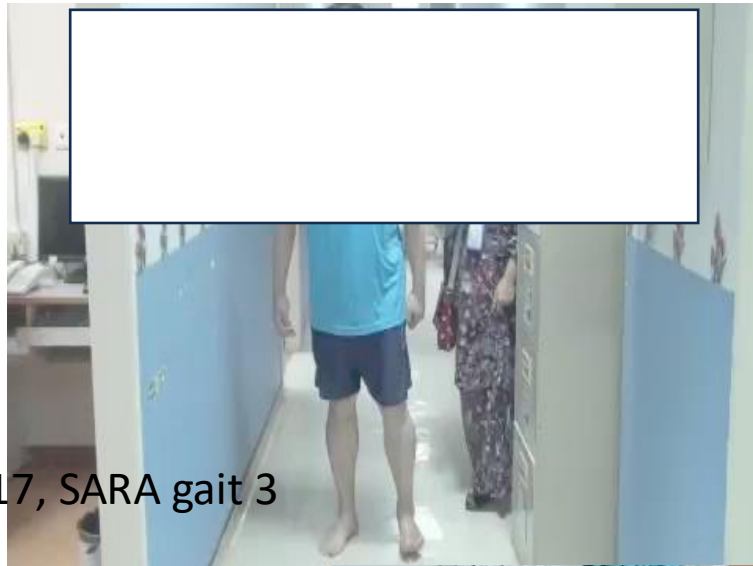


Pure cerebellar ataxia (self-paced)



Mixed ataxia + spasticity + dystonia
(SCA3)

Gait Progression in degenerative ataxia – Individualized



2017, SARA gait 3

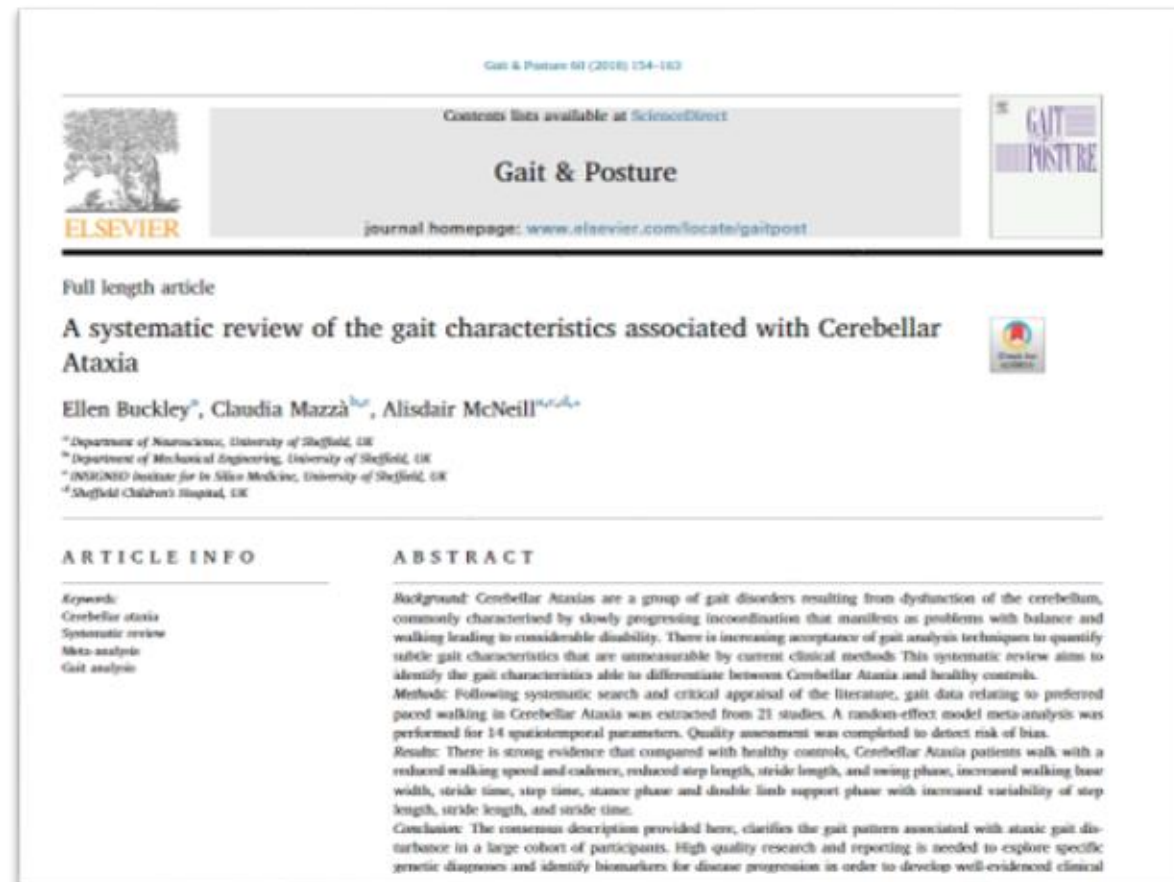


2019, SARA gait 5



Dec 2020: SARA gait 7

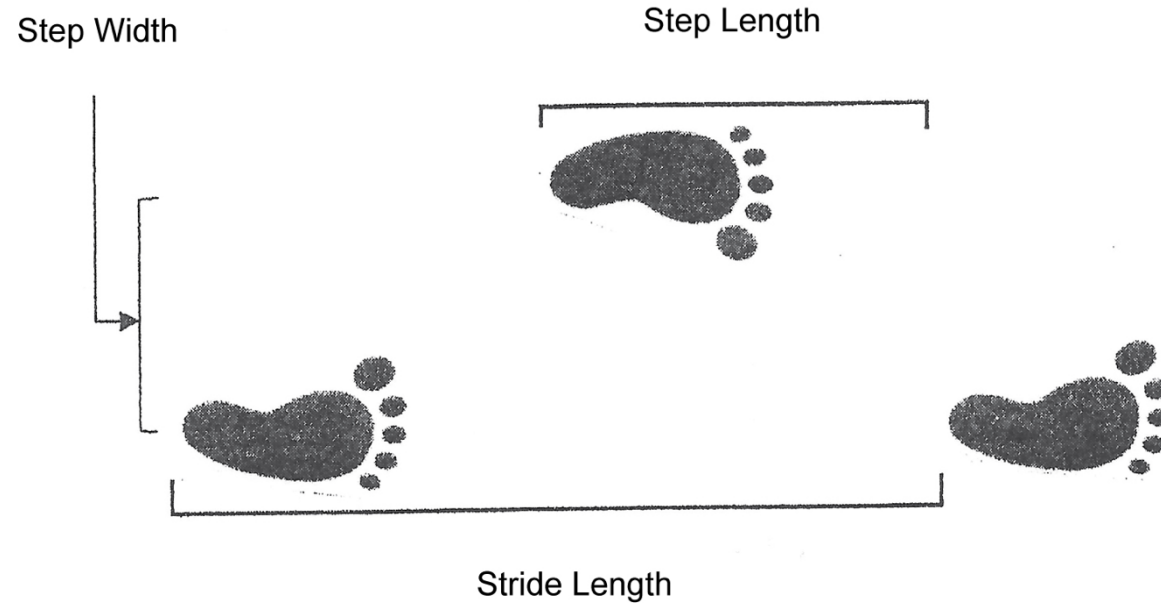
Video obtained with
permission



Cerebellar Ataxia patients walk with a reduced walking speed and cadence, reduced step length, stride length, and swing phase,

increased walking base width, stride time, step time, stance phase and double limb support phase

with increased variability of step length, stride length, and stride time.



Gait Cycle

“gait to be a highly complex skill requiring the integration of external sensory information within elaborate neural networks in the central nervous system”

Stance Phase (60%)

- Initial Contact
- Loading Response
- Midstance
- Preswing

Swing Phase (40%)

- Initial Swing
- Midswing
- Terminal swing

Nnodim JO, Nwagwu CV, Nnodim Opara I (2020) Gait Disorders in Older Adults - A Structured Review and Approach to Clinical

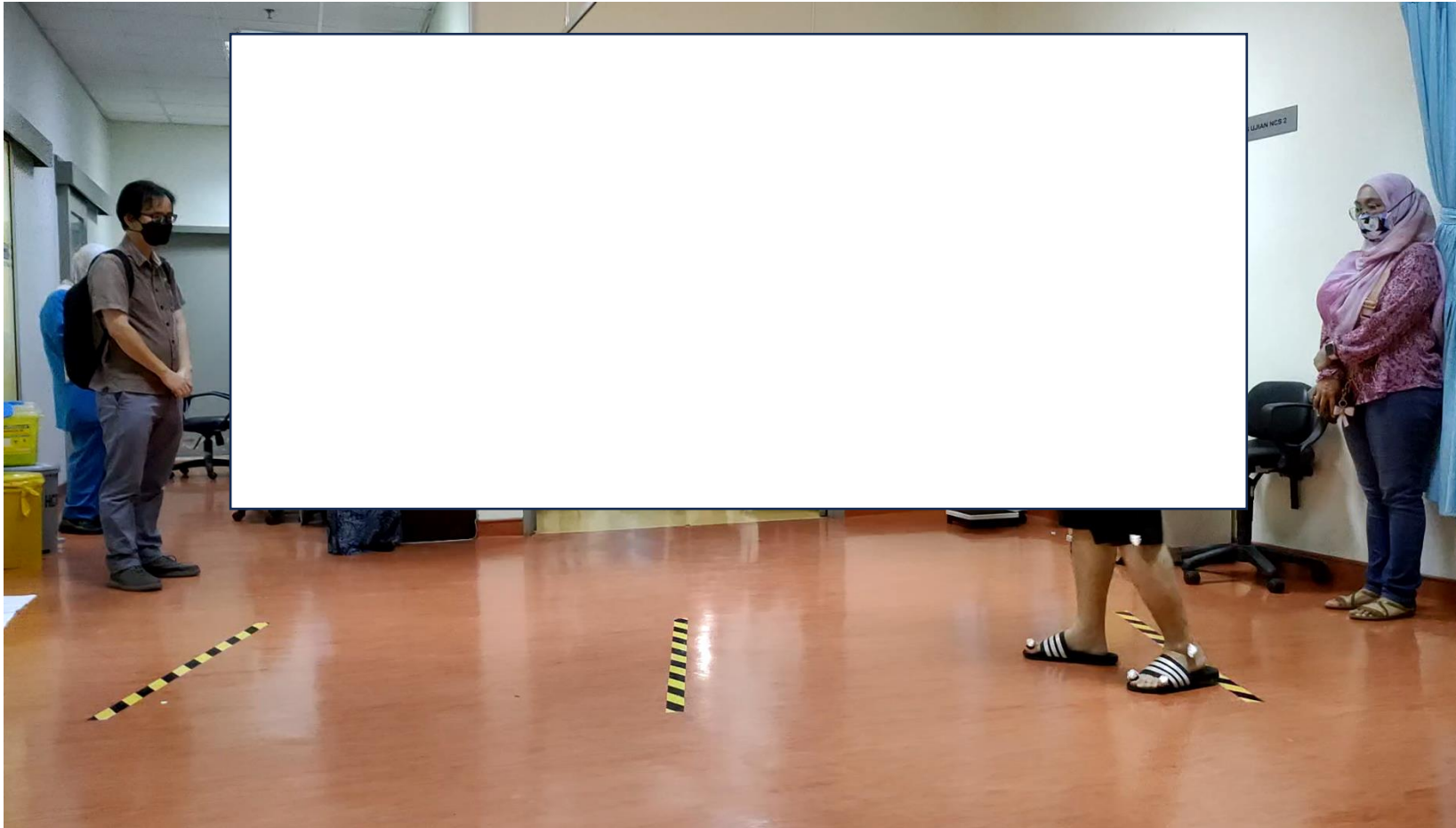
Assessment. J Geriatr Med Gerontol. 6:101. doi: 10.23937/2469-5858/1510101

Pioneering Medicine, Nurturing Scholars.

Mempelopori Perubatan, Melahirkan Cendekiawan.

Case Examples

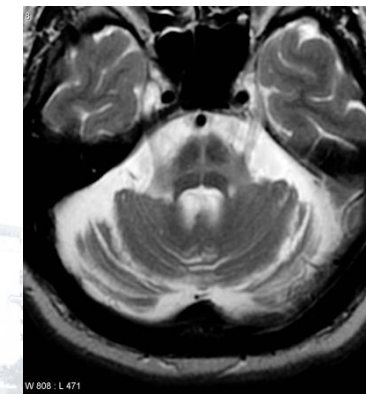




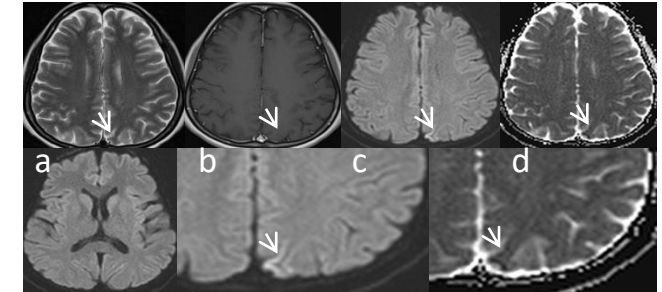
43 yr old with problems with
balance for 3 years, progressively
worsening,
REM sleep behavior disorder,
'noisy breathing',
Inspiratory sighs,
Postural BP drop, urinary urgency
+ urge incontinence
Hyperreflexia



Multiple System
Atrophy



Hot-cross
bun sign



42 year old
Progressive **Ataxia + Chorea** +
Weight Loss + Cognitive difficulties
+Seizures (> 2 years) – No FHx



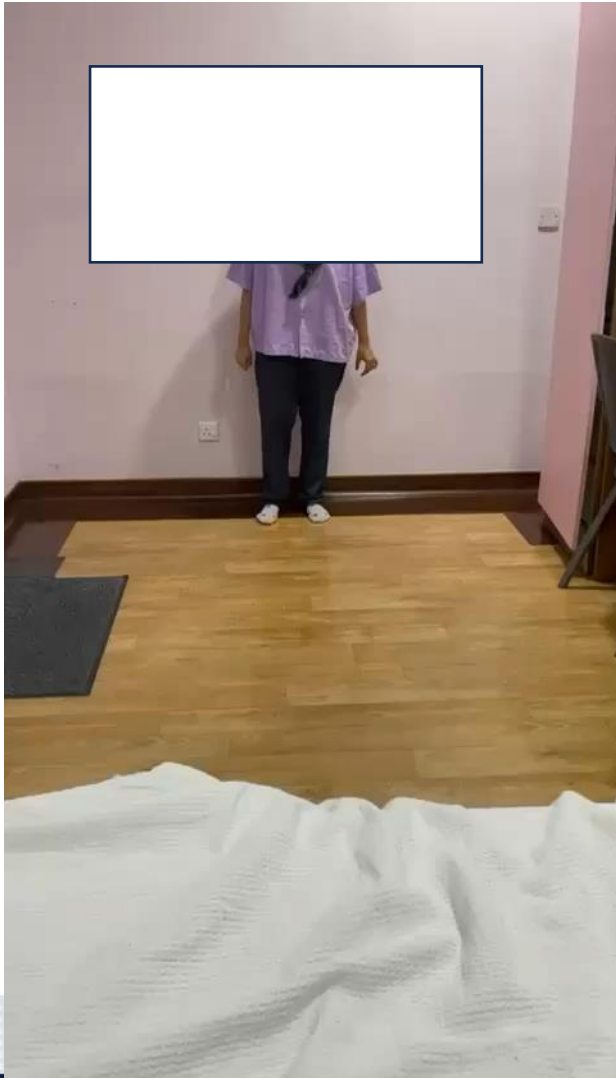
Negative autoimmune /paraneoplastic
antibodies
Negative for SCA 1,2,3,6,7
EEG – unremarkable
WES – pathogenic mutation (PRNP gene)
NM_000311.5(PRNP):c.532G>A (p.Asp178Asn)



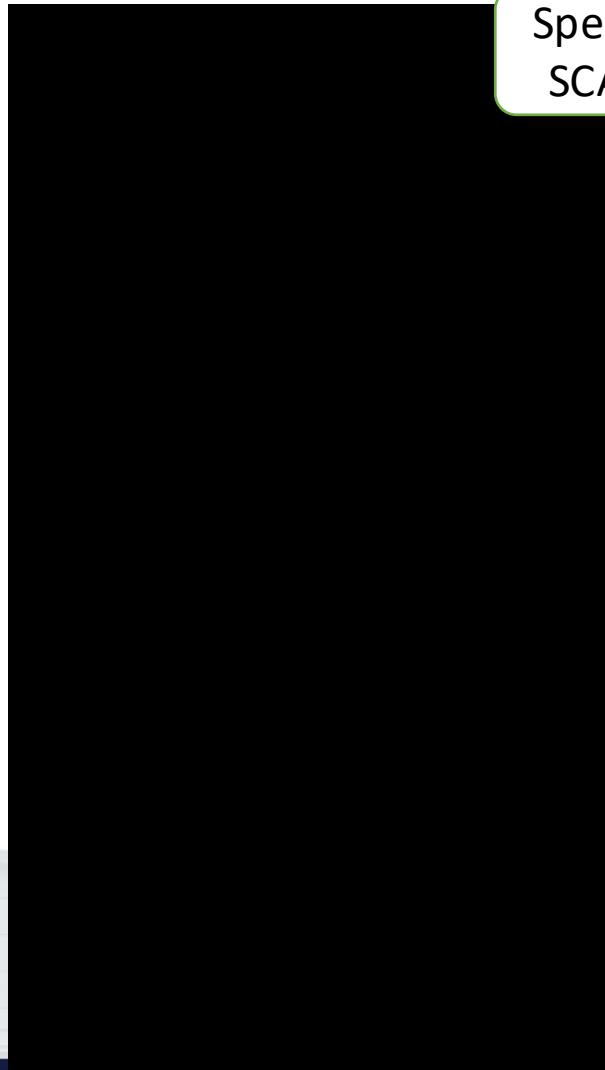
**Genetic Prion
Disease**

27 year of female with subacute onset of progressive bilateral cerebellar syndrome

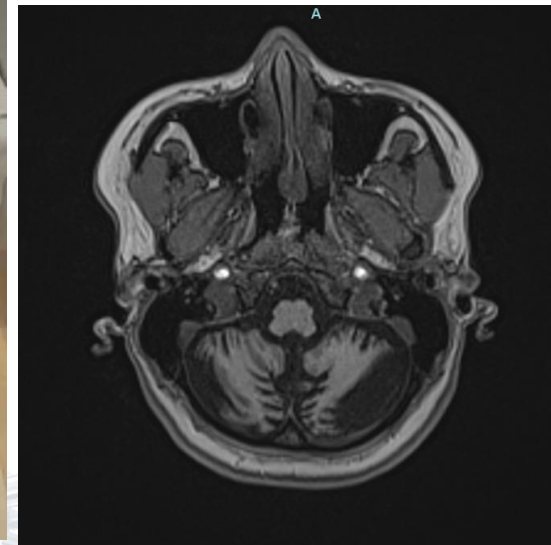
Investigations revealed positive anti-Yo Ab in serum



Speech
SCAFI



Finger
chase



Methods to evaluate gait in CA

- **Clinical Evaluation**
 - Ask patient to stand in **normal station** (base) and **in tandem**
 - Walk at own pace (self-paced), in tandem (mild ataxia) and include a turn
 - Fast paced and slow paced to distinguish between types of ataxia
 - Presence of other movement disorders (dystonia, spasticity, bradykinesia)
- **Ataxia rating scales (Severity/Dynamic Gait)**
 - SARA – 8 subtests (easily administered in clinic)
 - ICARS – 4 components (**time consuming -12-15 minutes**)
 - Speed tests – Timed Up and Go Test; 8MWT
 - Disease specific functional/rating scales - SCA Functional Index (SCA), FARS (FA),
 - Balance Test – Bergs Balance Test
- **Instrumented Gait Analysis (sensors, pressurized soles, balance mat)**

Scale for the assessment and rating of ataxia (SARA)

1) Gait

Proband is asked (1) to walk at a safe distance parallel to a wall including a half-turn (turn around to face the opposite direction of gait) and (2) to walk in tandem (heels to toes) without support.

- 0 Normal, no difficulties in walking, turning and walking tandem (up to one misstep allowed)
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- 3 Considerable staggering, difficulties in half-turn, but without support
- 4 Marked staggering, intermittent support of the wall required
- 5 Severe staggering, permanent support of one stick or light support by one arm required
- 6 Walking > 10 m only with strong support (two special sticks or stroller or accompanying person)
- 7 Walking < 10 m only with strong support (two special sticks or stroller or accompanying person)
- 8 Unable to walk, even supported

Score

3) Sitting

Proband is asked to sit on an examination bed without support of feet, eyes open and arms outstretched to the front.

- 0 Normal, no difficulties sitting >10 sec
- 1 Slight difficulties, intermittent sway
- 2 Constant sway, but able to sit > 10 s without support
- 3 Able to sit for > 10 s only with intermittent support
- 4 Unable to sit for >10 s without continuous support

Score

2) Stance

Proband is asked to stand (1) in natural position, (2) with feet together in parallel (big toes touching each other) and (3) in tandem (both feet on one line, no space between heel and toe). Proband does not wear shoes, eyes are open. For each condition, three trials are allowed. Best trial is rated.

- 0 Normal, able to stand in tandem for > 10 s
- 1 Able to stand with feet together without sway, but not in tandem for > 10s
- 2 Able to stand with feet together for > 10 s, but only with sway
- 3 Able to stand for > 10 s without support in natural position, but not with feet together
- 4 Able to stand for >10 s in natural position only with intermittent support
- 5 Able to stand >10 s in natural position only with constant support of one arm
- 6 Unable to stand for >10 s even with constant support of one arm

Score

4) Speech disturbance

Speech is assessed during normal conversation.

- 0 Normal
- 1 Suggestion of speech disturbance
- 2 Impaired speech, but easy to understand
- 3 Occasional words difficult to understand
- 4 Many words difficult to understand
- 5 Only single words understandable
- 6 Speech unintelligible / anarthria

Score

5) Finger chase

Rated separately for each side

Proband sits comfortably. If necessary, support of feet and trunk is allowed. Examiner sits in front of proband and performs 5 consecutive sudden and fast pointing movements in unpredictable directions in a frontal plane, at about 50 % of proband's reach. Movements have an amplitude of 30 cm and a frequency of 1 movement every 2 s. Proband is asked to follow the movements with his index finger, as fast and precisely as possible. Average performance of last 3 movements is rated.

- 0 No dysmetria
- 1 Dysmetria, under/ overshooting target <5 cm
- 2 Dysmetria, under/ overshooting target < 15 cm
- 3 Dysmetria, under/ overshooting target > 15 cm
- 4 Unable to perform 5 pointing movements

Score

Right

Left

mean of both sides (R+L)/2

7) Fast alternating hand movements

Rated separately for each side

Proband sits comfortably. If necessary, support of feet and trunk is allowed. Proband is asked to perform 10 cycles of repetitive alternation of pro- and supinations of the hand on his/her thigh as fast and as precise as possible. Movement is demonstrated by examiner at a speed of approx. 10 cycles within 7 s. Exact times for movement execution have to be taken.

- 0 Normal, no irregularities (performs <10s)
- 1 Slightly irregular (performs <10s)
- 2 Clearly irregular, single movements difficult to distinguish or relevant interruptions, but performs <10s
- 3 Very irregular, single movements difficult to distinguish or relevant interruptions, performs >10s
- 4 Unable to complete 10 cycles

Score

Right

Left

mean of both sides (R+L)/2

6) Nose-finger test

Rated separately for each side

Proband sits comfortably. If necessary, support of feet and trunk is allowed. Proband is asked to point repeatedly with his index finger from his nose to examiner's finger, which is in front of the proband at about 90 % of proband's reach. Movements are performed at moderate speed. Average performance of movements is rated according to the amplitude of the kinetic tremor.

- 0 No tremor
- 1 Tremor with an amplitude < 2 cm
- 2 Tremor with an amplitude < 5 cm
- 3 Tremor with an amplitude > 5 cm
- 4 Unable to perform 5 pointing movements

Score

Right

Left

mean of both sides (R+L)/2

8) Heel-shin slide

Rated separately for each side

Proband lies on examination bed, without sight of his legs. Proband is asked to lift one leg, point with the heel to the opposite knee, slide down along the shin to the ankle, and lay the leg back on the examination bed. The task is performed 3 times. Slide-down movements should be performed within 1 s. If proband slides down without contact to shin in all three trials, rate 4.

- 0 Normal
- 1 Slightly abnormal, contact to shin maintained
- 2 Clearly abnormal, goes off shin up to 3 times during 3 cycles
- 3 Severely abnormal, goes off shin 4 or more times during 3 cycles
- 4 Unable to perform the task

Score

Right

Left

mean of both sides (R+L) / 2

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Score

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- 0 Normal, able to stand in tandem for > 10 s
- 1 Able to stand with feet together without sway, but not in tandem for > 10s
- 2 Able to stand with feet together for > 10 s, but only with sway
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- 4 Able to stand for >10 s in natural position only with intermittent support
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Score

SARA Gait (0-8)

1. Self Paced Walking with Half Turn 2. Tandem Walk – distinguishes normal and abnormal (10 steps)

- Score 0 : Normal - Able to tandem walk
- Score 1-3 : Gait ataxia but still walking without support
- Score 4-5 : Moderate ataxia (require light support)
- Score 6-7 : Require strong support
- Score 8 : Unable to walk

Assessment of Stance (Tandem, Feet Together and Natural Position – 10 seconds)



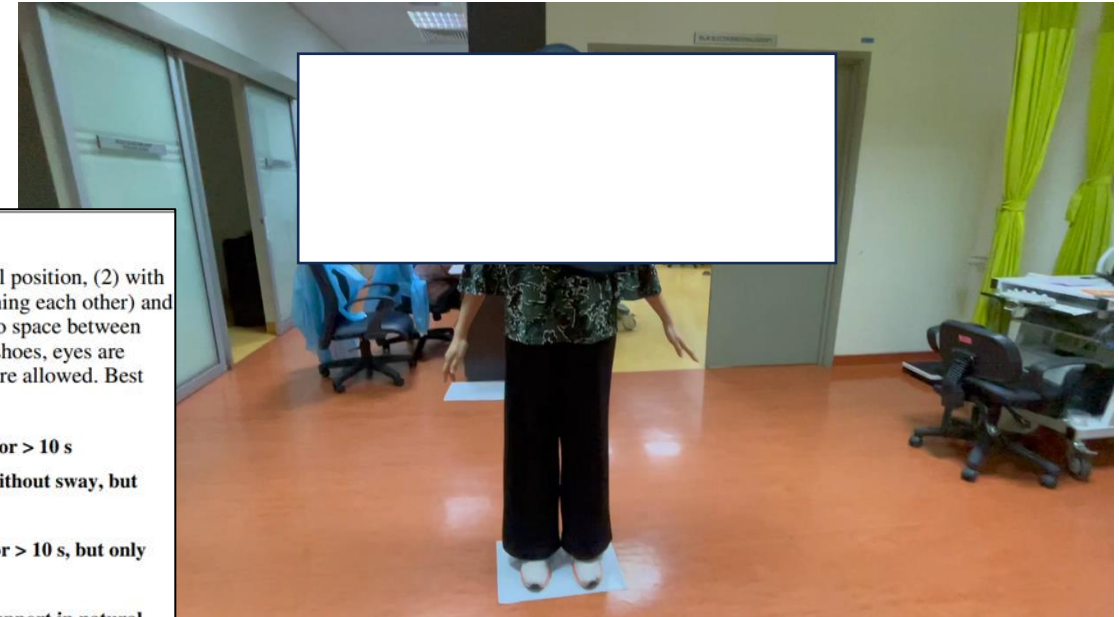
Not able to stand in tandem, but in natural position without sway

SARA Stance score : 1

2) Stance

Proband is asked to stand (1) in natural position, (2) with feet together in parallel (big toes touching each other) and (3) in tandem (both feet on one line, no space between heel and toe). Proband does not wear shoes, eyes are open. For each condition, three trials are allowed. Best trial is rated.

- 0 Normal, able to stand in tandem for > 10 s
- 1 Able to stand with feet together without sway, but not in tandem for > 10s
- 2 Able to stand with feet together for > 10 s, but only with sway
- 3 Able to stand for > 10 s without support in natural position, but not with feet together
- 4 Able to stand for >10 s in natural position only with intermittent support
- 5 Able to stand >10 s in natural position only with constant support of one arm
- 6 Unable to stand for >10 s even with constant support of one arm



Stand in natural position with sway

SARA Stance score : 2

Mid-adulthood – mild ataxia

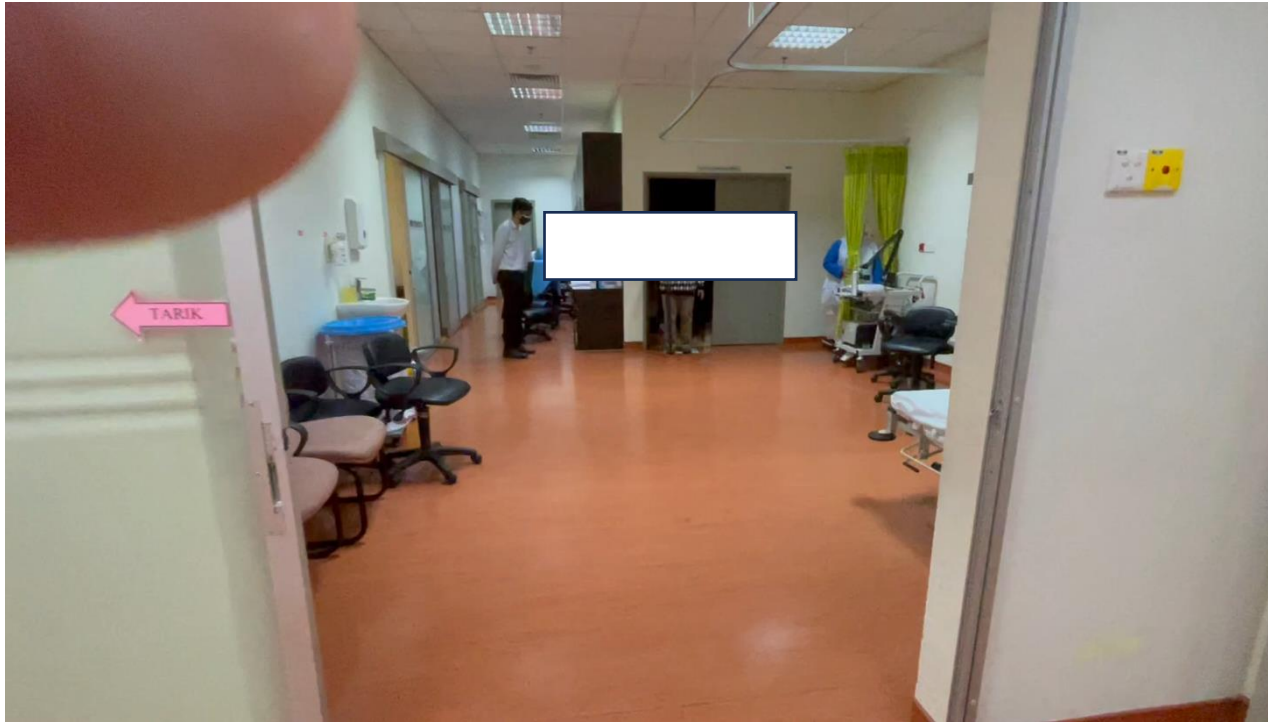


Unable to tandem walk but able to walk unsupported, wide based, with no difficulties with half-turn (SARA gait score 2)

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Wide based
Staggering side-to side for stability
Reduced step length and stride length
Increased stride time and step time

SARA gait 2/3

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- 1 Slight difficulties, only visible when walking 10 consecutive steps in tandem
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- 8 Unable to walk, even supported

Gait and Balance in Degenerative Ataxia - dynamic

Functional Assessment

8MW (SCA Functional Index),
Berg Balance, TUGT

Wheelchair
dependence

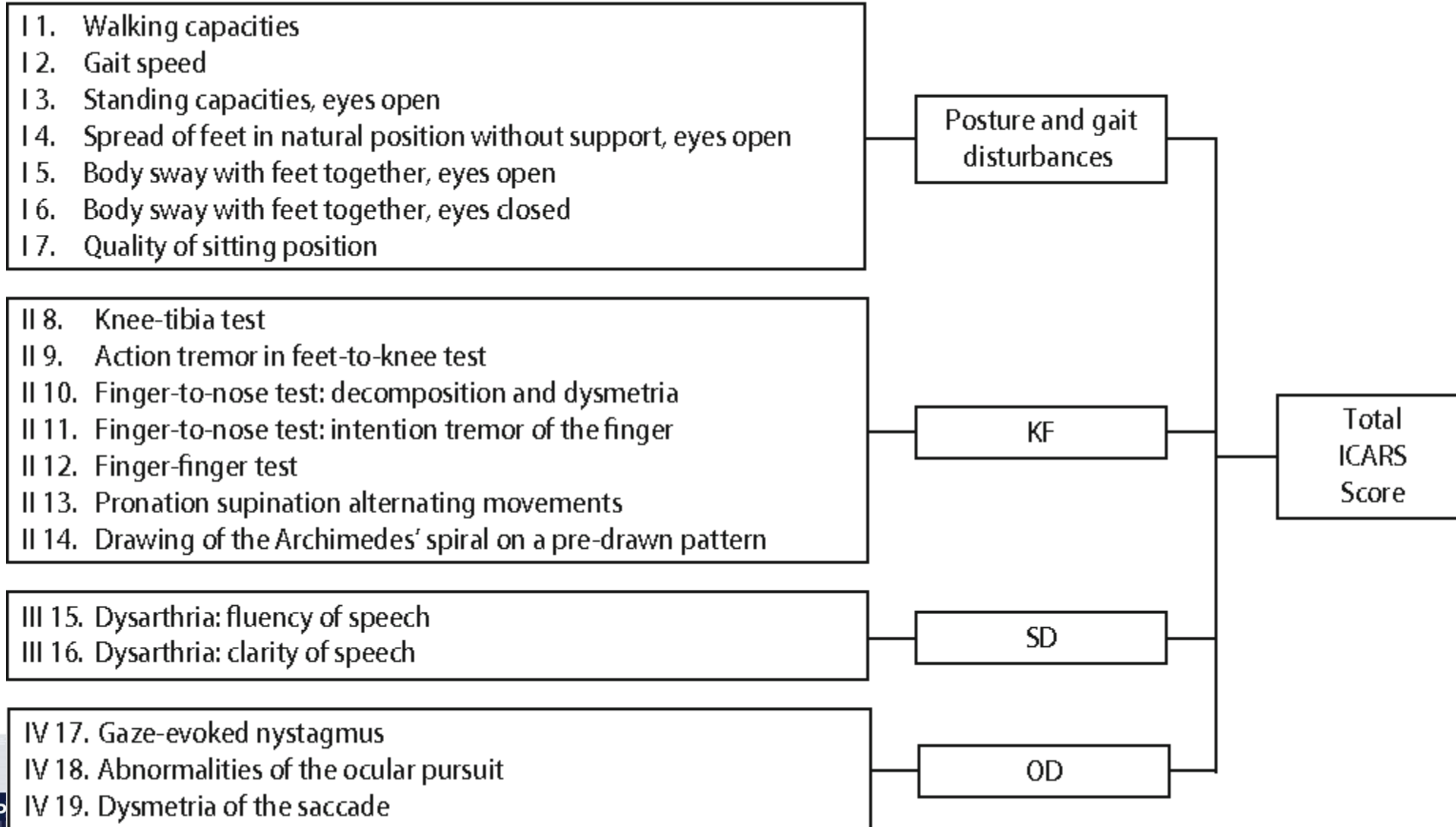
Walking Frame

Early falls

Ataxic Gait
Parkinsonism
Spastic Gait
Dystonic Gait
Neuropathy
Any combination

Physiotherapy
Intrathecal baclofen
Botulinum Toxin
Injection for severe
spasticity

ICARS



Other measures

- Dynamic Gait Index (DGI)
 - 8 items (scored 0-3; 0 =more severe disease)

- G
- G
- G
- G
- G
- G
- G
- S

Dynamic Gait Index (DGI)

Patient's full name: _____ Patient's age: _____

Patient's gender: _____ Date of assessment: _____

What you need:

- A 20-foot walkway, should also be 15 inches wide; shoebox; 2 cones; an area with stairs

1. Gait level surface

Instructions: Walk at your normal speed from her to the next mark (20 feet).

Grading: Mark the lowest category that applies.

Score	Level of function	Criteria
☐ 0	Severe impairment	Cannot walk 20 ft. without assistance, severe gait deviations, or imbalance.
☐ 1	Moderate impairment	Walks 20 ft., slow speed, abnormal gait pattern, evidence for imbalance.
☐ 2	Mild impairment	Walks 20 ft., uses assistive devices, slower speed, mild gait deviations.
☐ 3	Normal	Walks 20 ft., no assistive devices, good speed, no evidence of imbalance, normal gait pattern.

- SCA Functional Index
 - 8 Meter walking time
 - 9 Hole Peg Test
 - PATA repeats

What do we in clinic for DA? Practical Aspects

Ataxia Severity

Use validated scoring system – SARA, BARS, ICARS, UMSARs, FRDA rating scales

SARA 8- domains – most sensitive to change

Other non-ataxia symptoms

Inventory of Non-Ataxia Signs (INAS)

Spasticity
Cramps
Myoclonus
Dystonia/Rigidity

Functional Ability and QoL milestones

Gait – walking function (timed up and go or 8MWT)

Complications associated with ataxia

Depression
CCAS

Diabetes Mellitus
Neuropathy
Scoliosis
Cardiac Abnormalities
Visual Impairment

PROBLEMS WITH CURRENT METHOD USING CLINICAL SCALES



Inter-rater
variability



Floor or Ceiling
Effects (reach the
end of scale)



Time consuming



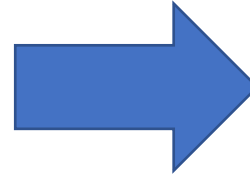
Require clinic visits



Does not capture
true function at
home

Instrumented Gait Analysis - more objective

- 3D Motion Capture
- Pressure Sensitive Walkways,
- Triaxial inertial sensors,
- Pressure sensitive insoles,
- Force plates Pressure sensitive treadmill



Spatiotemporal characteristics

- reduced cadence, step and length, gait velocity,
- increased step and stride time and stance and swing phases

Project : Assessment of gait in cerebellar ataxia video kinematics and deep learning

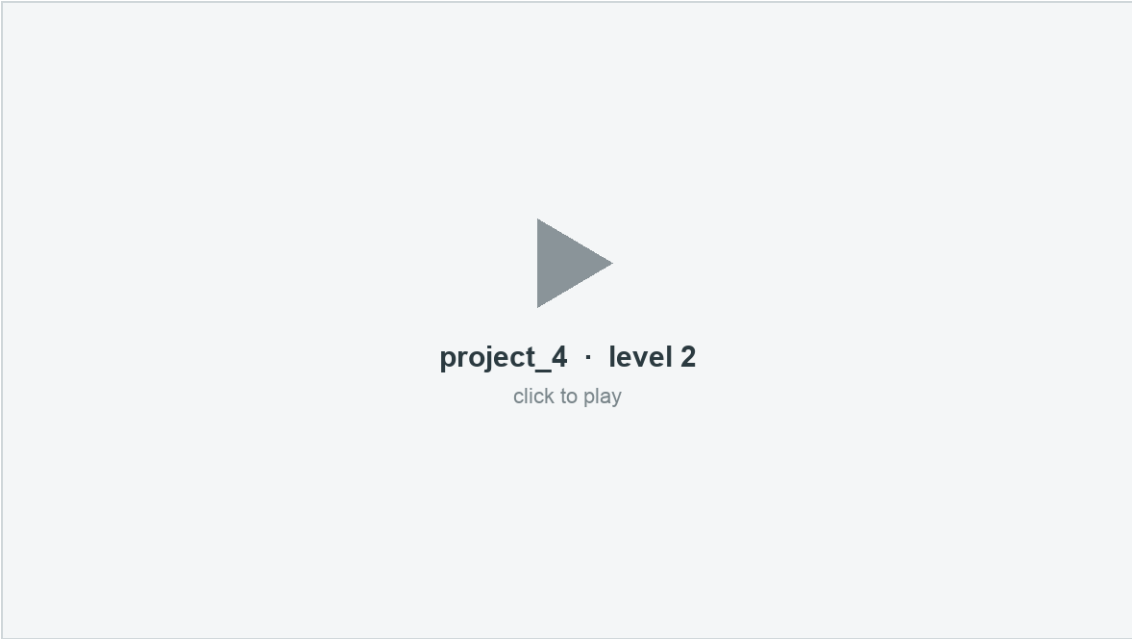
Used video capture and extracting keypoints (Smartphone app) and cloud based algorithm to extract gait specific parameters



Healthy control and mild

The recorded walk for each subject. Same walkway, same camera setup across the four.

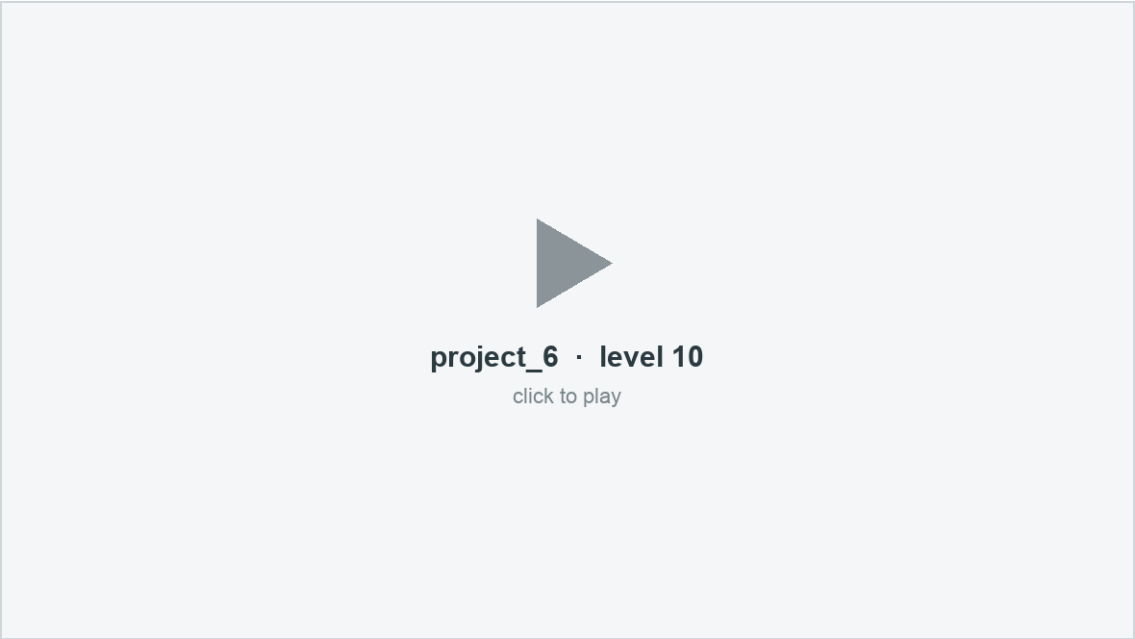
Healthy control — project_4, level 2



Reference walk: full stride excursion, contacts evenly spaced.

Gait speed 0.95 m/s (stride) · stride 1.19 m · step-time CV 15.7% · 4.5 s

Mild — project_6, level 10



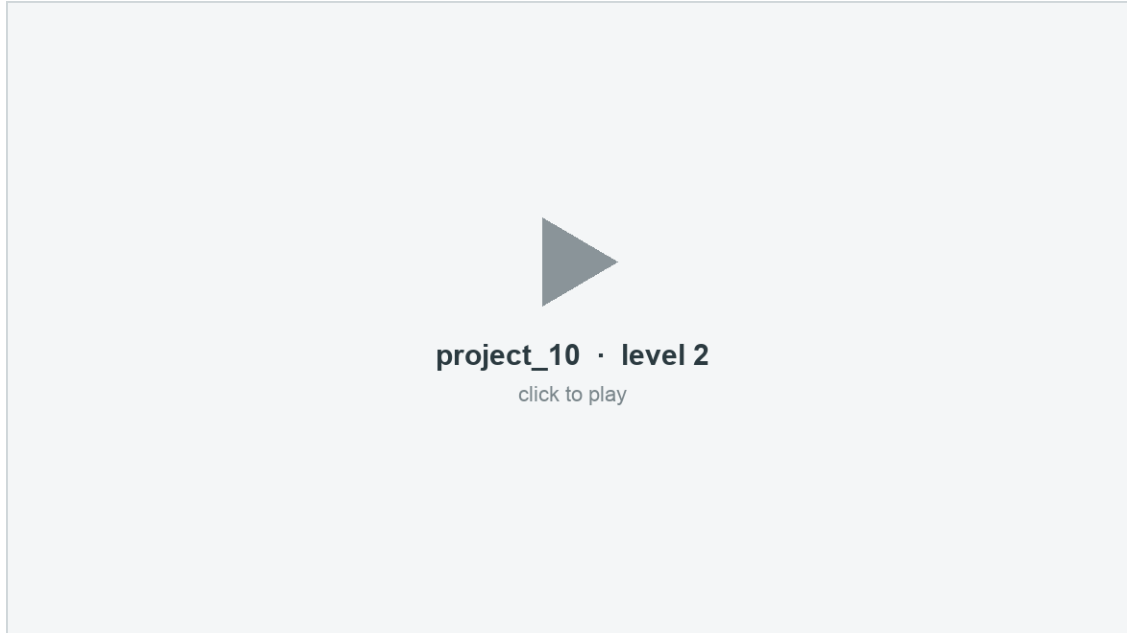
Stride and rhythm at or above the healthy control's — the clearest example of why single features need not be ordered.

Gait speed 0.95 m/s (stride) · stride 1.25 m · step-time CV 2.6% · 4.7 s

Moderate and severe

The recorded walk for each subject. Same walkway, same camera setup across the four.

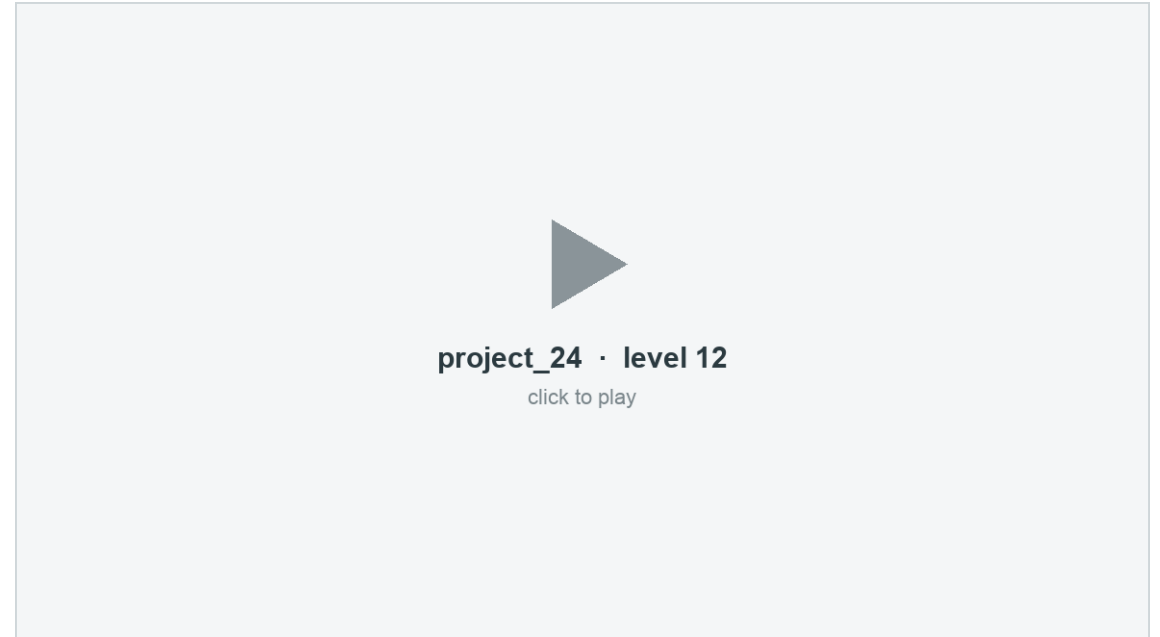
Moderate — project_10, level 2



Shorter steps and slower cadence; the cycle-to-cycle rhythm starts to drift.

Gait speed 0.66 m/s (stride) · stride 0.85 m · step-time CV 12.0% · 6.2 s

Severe — project_24, level 12



Marked slowing with a shortened, irregular stride; the trial lasts nearly twice as long as the healthy control's.

Gait speed 0.41 m/s (stride) · stride 0.59 m · step-time CV 18.1% · 8.7 s

All measured features

Every per-trial parameter, for the four trials.

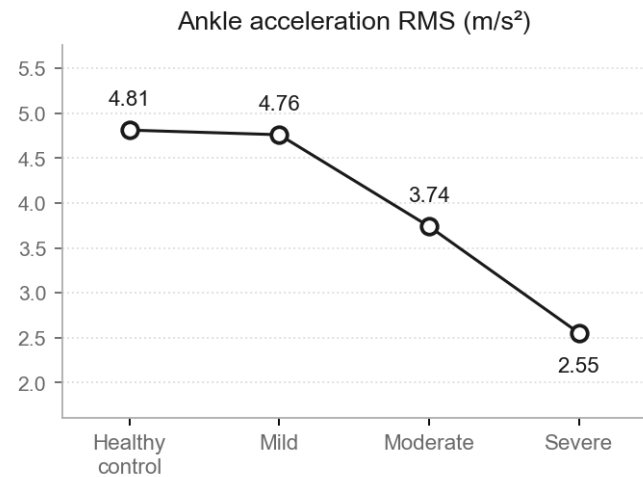
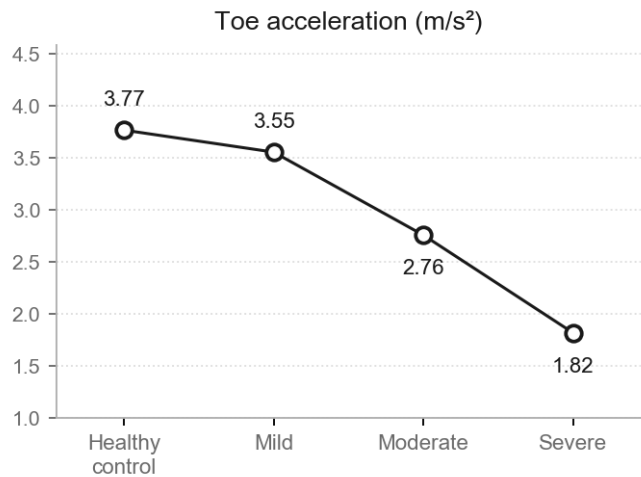
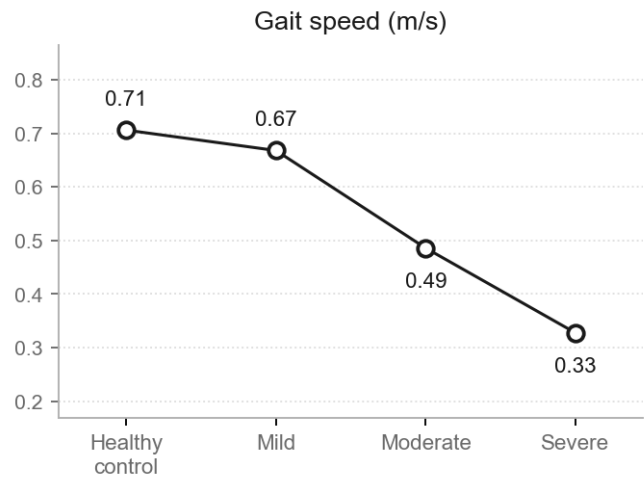
Feature	Healthy control	Mild	Moderate	Severe
Step time L (s)	0.550	0.650	0.578	0.767
Step time R (s)	0.733	0.650	0.717	0.692
Stride time L (s)	1.283	1.300	1.317	1.383
Stride time R (s)	1.200	1.333	1.267	1.475
Stance time L (s)	0.867	0.883	0.917	0.958
Stance time R (s)	0.767	0.917	0.922	1.050
Swing time L (s)	0.417	0.417	0.400	0.425
Swing time R (s)	0.433	0.433	0.383	0.425
Double support L→R (s)	0.133	0.233	0.200	0.267
Double support R→L (s)	0.267	0.267	0.344	0.267
Step time variability (s)	0.101	0.017	0.076	0.132
Step time variability L (s)	0.050	0.017	0.042	0.167
Step time variability R (s)	0.033	0.017	0.017	0.036
Step time CV (%)	15.75	2.56	12.00	18.06
Step time CV L (%)	9.09	2.56	7.20	21.83
Step time CV R (%)	4.55	2.56	2.33	5.25
Double support variability (s)	0.069	0.037	0.072	0.041
Double support CV (%)	38.53	14.91	25.05	15.31

Feature	Healthy control	Mild	Moderate	Severe
Step time asymmetry (s)	0.183	0.000	0.139	0.075
Step time asymmetry index (%)	28.57	0.00	21.46	10.29
Step length L (m)	0.534	0.568	0.358	0.164
Step length R (m)	0.524	0.452	0.374	0.330
Stride length L (m)	1.210	1.228	0.850	0.574
Stride length R (m)	1.175	1.278	0.850	0.610
Gait speed (step) (m/s)	0.826	0.803	0.578	0.337
Gait speed (stride) (m/s)	0.954	0.949	0.658	0.414
Stance phase L (%)	67.54	67.95	69.67	69.29
Stance phase R (%)	63.89	67.50	69.75	70.73
Swing phase L (%)	32.46	32.05	30.33	30.71
Swing phase R (%)	36.11	32.50	30.25	29.27
Double support phase L→R (%)	10.36	17.95	15.21	19.14
Double support phase R→L (%)	22.22	17.50	26.33	18.27
Heel strikes, left (count)	3	3	3	5
Heel strikes, right (count)	2	2	3	5
Toe offs, left (count)	2	2	3	5
Toe offs, right (count)	2	3	4	4

Four walking trials, one per severity grade

	Healthy control	Mild	Moderate	Severe
Subject	project_4	project_6	project_10	project_24
Trial	level 2	level 10	level 2	level 12
Recording length	4.5 s	4.7 s	6.2 s	8.7 s
Foot contacts detected	9	10	13	19

Three features that fall steadily from healthy control to severe:

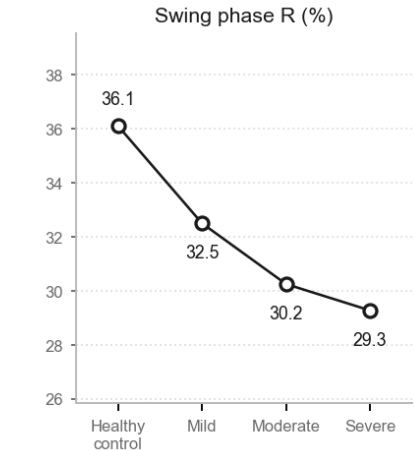
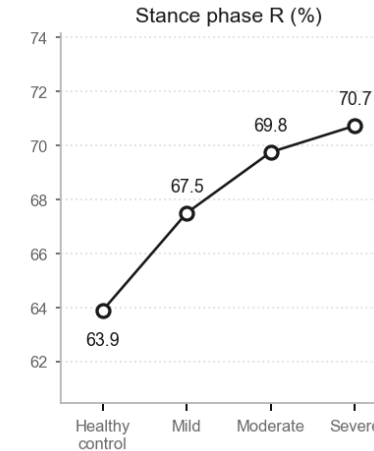
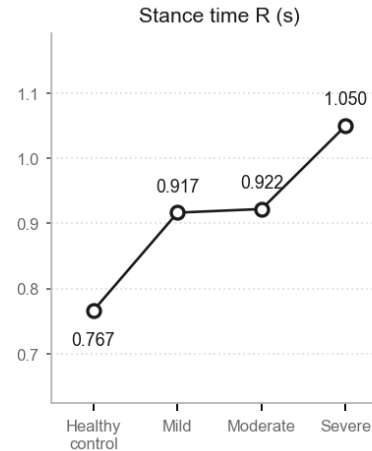
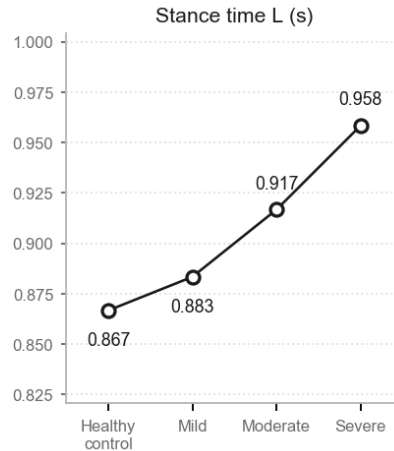
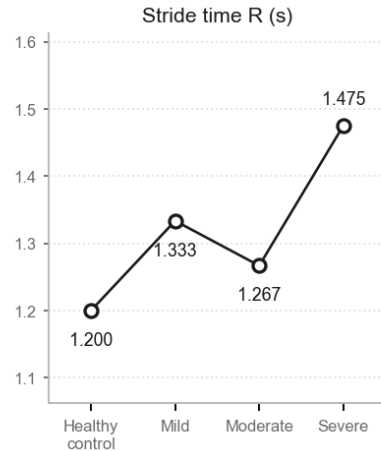
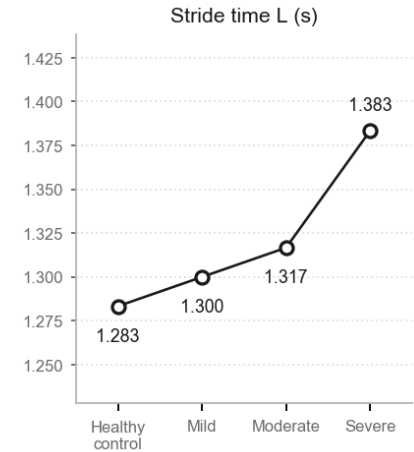
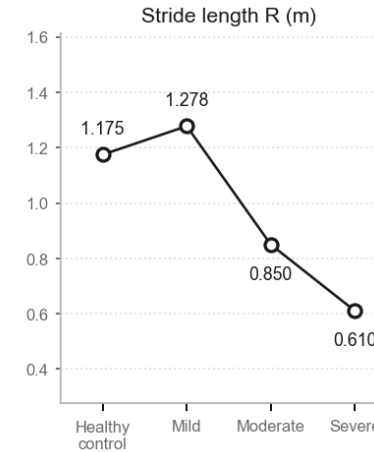
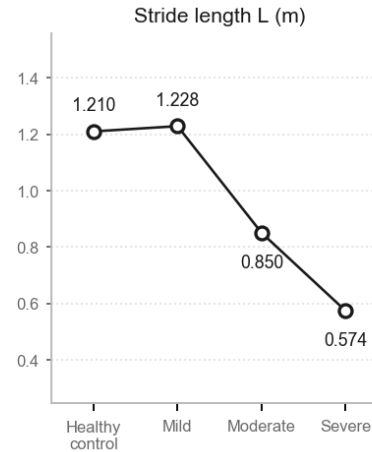
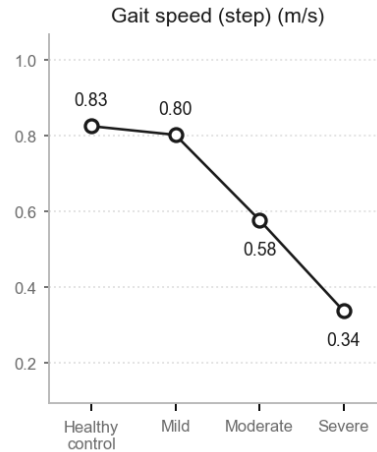
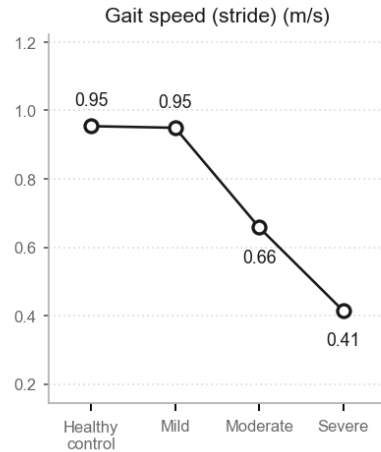


Not every feature is monotone across the four subjects, and that is expected.

These are individual cases, not group averages, and nothing is normalised for height, weight, age or sex. A few percent of individual difference can outweigh the severity effect at any single feature, so read the features as a family rather than as a strict ordering of any one measure.

The features that track severity most closely

Ten features, ranked by how closely their four values follow the severity order.
Seven are strictly ordered; stride length and stride time differ only at one step.



Management of Degenerative Ataxia : multidisciplinary team involvement

Ataxia & Non Ataxia Symptoms

Riluzole
4 aminopyridine
Baclofen
Gabapentin
Benzhexol
Clonazepam

Rehabilitation

Falls prevention

Maintain
independence

Swallowing and
speech
management

Cerebellar Affective and Cognitive Symptoms

Anti depressants
CBT / Psychological
coaching

Pharmacological management of Ataxia

- Treat reversible causes (immune mediated, structural, vitamin deficiency, metabolic causes)
- Inherited Ataxias
 - AVED – supplementation with **Vitamin E (800-1100mg/day)** – reversed ataxia symptoms and stabilized disease
 - Ataxia with CoEQ10 deficiency – **replace with CoEQ10**
 - Wilsons disease – (decreased ceruloplasmin)- **Chelation RX**
 - Cerebrotendinous xanthomasosis – (↑cholestanol)- **Chenodeoxycholic acid 750mg**
 - Refsum's disease (↑ phytanic acid) – **PEX or dietary restriction**
 - Adrenoleukodystrophy (very long chain fatty acids) – **Stem cell transplantation**
 - Ataxia-Telangiectasia – RCT- **oral beclomethasone** (ICARS score improvement)

Pharmacological Management of Degenerative Ataxias (EBM)

Episodic Ataxia and Downbeat Nystagmus

4- aminopyridine
3,4 diaminopyridine
Acetazolamide

Friedreichs Ataxia

Idebenone – cardiac hypertrophy (negative RCT trial findings), but overall
?improvement in neurological function

Dominant Ataxias

Riluzole 50mg bd (RCT) – improved SARA over 12 months
Varenicline 1mg (RCT) ? outcome

Na valproate 1200 mg (RCT) – improvement in SARA
Zn Sulphate 50mg (RCT)- SCA 2, Cuban
IV Trehalose 15-30mg weekly (open label controlled)
Acetazolamide 250mg
Branch chain aminoacid (RCT)
Tetrabiopterin
Sulphamethaxole/Trimethoprim (RCT)
Lamotrigine 25mg bd (improved 6 weeks, but not sustained)

Nerve growth factor injection
MSC transplantation

Oral trehalose 100g daily (single arm)
Gene therapy (ASO)-SCA 2, SCA3

Rehabilitation Therapy in genetic DA

- A number of systematic reviews (2014, 2017, 2020) – **consistently** showed that various therapies are useful for maintaining gait and balance
 - Conventional Physiotherapy and OT (RCT)
 - Virtual Reality/ Exercise games
 - Adapted physical therapy
 - Wearables with robotics technology
 - TMS over cerebellum (RCT) – improved gait
 - Transcranial direct current stimulation (RCT)- improved balance and coordination

Summary

- Ataxia may be due to acquired and degenerative causes
- Onset of ataxia – gives a clue to underlying diagnosis
- Gait ataxia is the earliest manifestation of cerebellar syndrome
- Identify treatable causes
- Rehabilitation and multidisciplinary input –important for degenerative ataxia