The clinical examination of the cerebellar ataxia patient

Thomas Klockgether

MDS-ES Online Course
Diagnosis and Treatment of Cerebellar Ataxias
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UNIVERSITÄT BONN

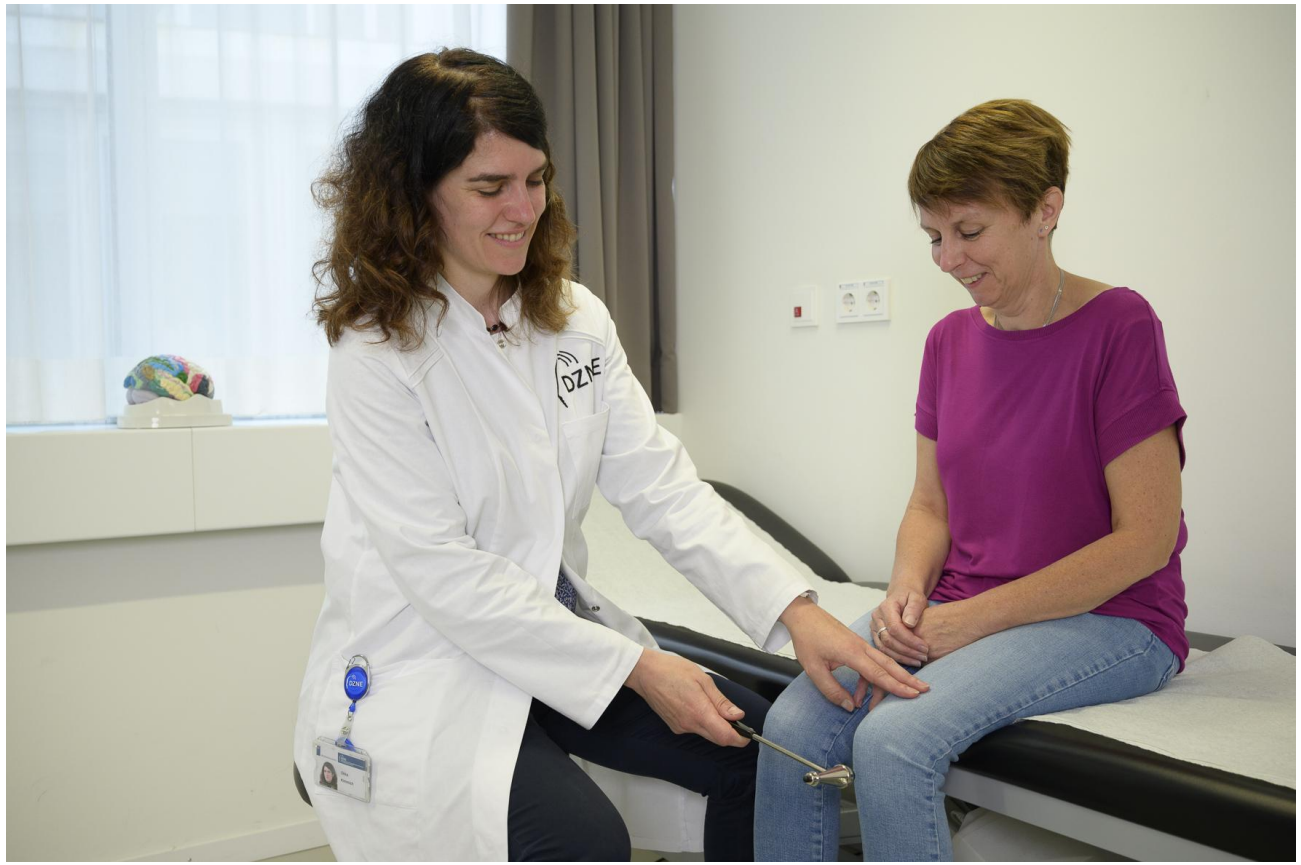
DZNE

Agenda

- Why perform a clinical examination?
- Perform a general physical examination!
- Perform a conventional neurological examination!
- Pay special attention to eye movements!
- Define the type of ataxia!
- Assess and rate ataxia!

Why perform a clinical examination?

Traditionally, the clinical examination has been the mainstay of diagnosing neurological diseases.



https://www.neurologiepsychiatrie.info/angebote/neurologischer-befund

Why perform a clinical examination?

SIGNS AND SYMPTOMS OF FRIEDREICH'S ATAXIA

PRIMARY SYMPTOMS AND SIGNS: (100% constant: obligatory for diagnosis)

1) Onset before end of puberty and never after age 20
2) Ataxia of gait
3) Progression of ataxia within last 2 years preceeding examination to all extremities, without remission
4) Dysarthria
5) Decrease in position and/or vibratory sense in lower limbs
6) Muscle weakness
7) Deep tendon areflexia in lower limbs

SECONDARY (PROGRESSIVE) SYMPTOMS AND SIGNS: (present in more than 90% cases, eventually 100%; not obligatory for diagnosis)

1) Babinski sign (extensor plantar response)
2) Pes cavus 3) Scoliosis 4) Cardiomyopathy

ACCESSORY SYMPTOMS AND SIGNS: (less than 50% of cases)

1) Decrease in visual acuity (usually optic atrophy)
2) Nystagmus
3) Paresthesias
4) Partial deafness
5) Essential type tremor
6) Vertigo
7) Spasticity
8) Pain
9) Decrease in I.Q.

LE JOURNAL CANADIEN DES SCIENCES NEUROLOGIQUES

Quebec Cooperative Study of
Friedreich's Ataxia

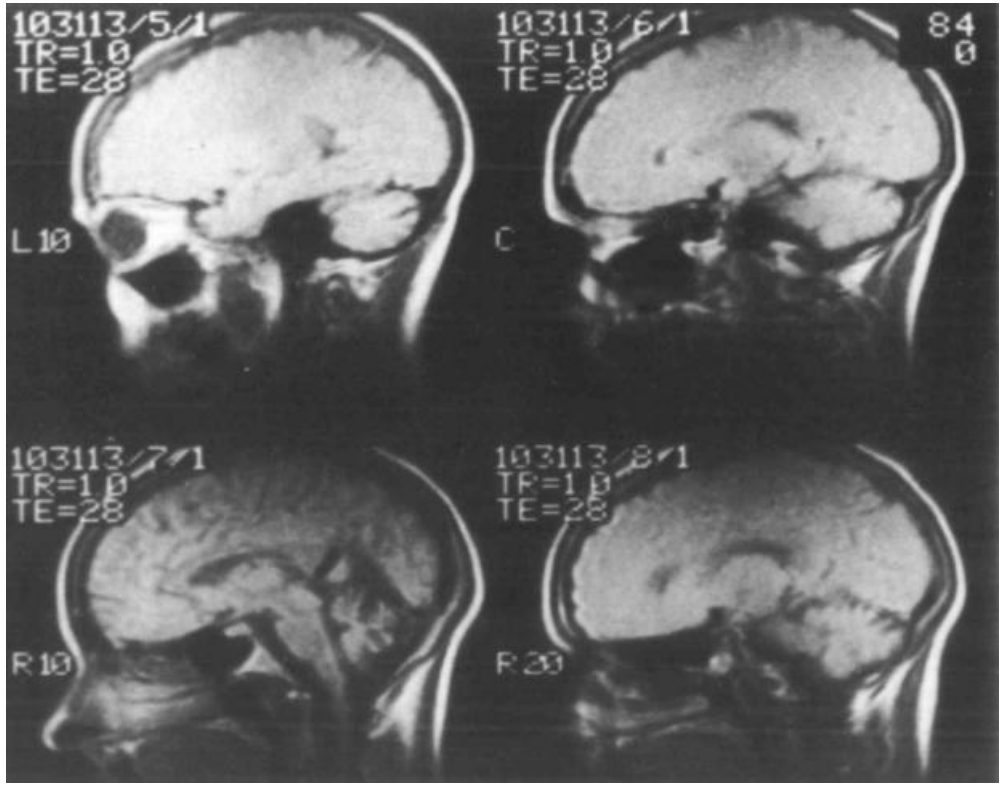
Clinical Description and Roentgenologic
Evaluation of Patients with Friedreich's Ataxia

G. GEOFFROY, A. BARBEAU, G. BRETON, B. LEMIEUX, M. AUBE,
C. LEGER AND J. P. BOUCHARD

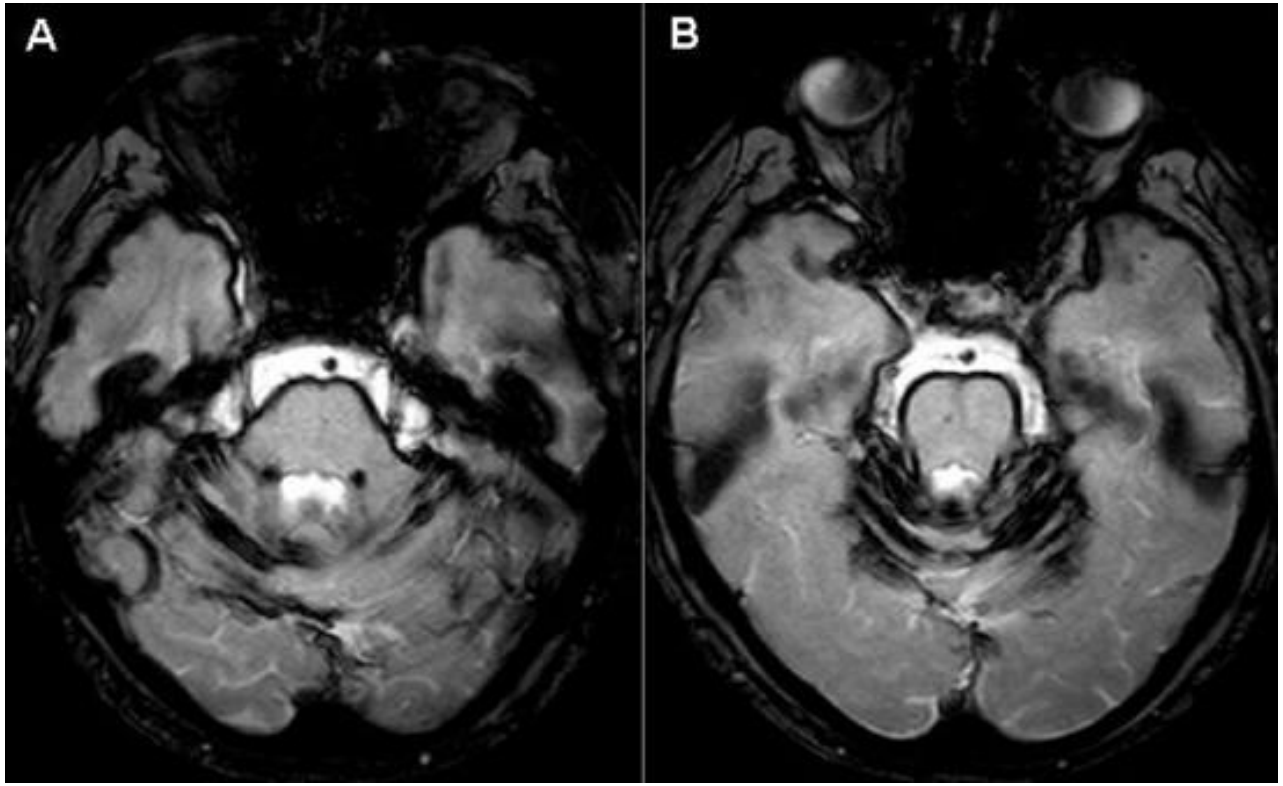
Can J Neurol Sci 1976;3:279-86

Why perform a clinical examination?

The MRI revolution



Sporadic late onset cerebellar ataxia
Baloh et al. Brain 1986;109:159-80



Superficial siderosis
Courtesy H. Urbach, Dept. Neuroradiology, University Hospital Bonn

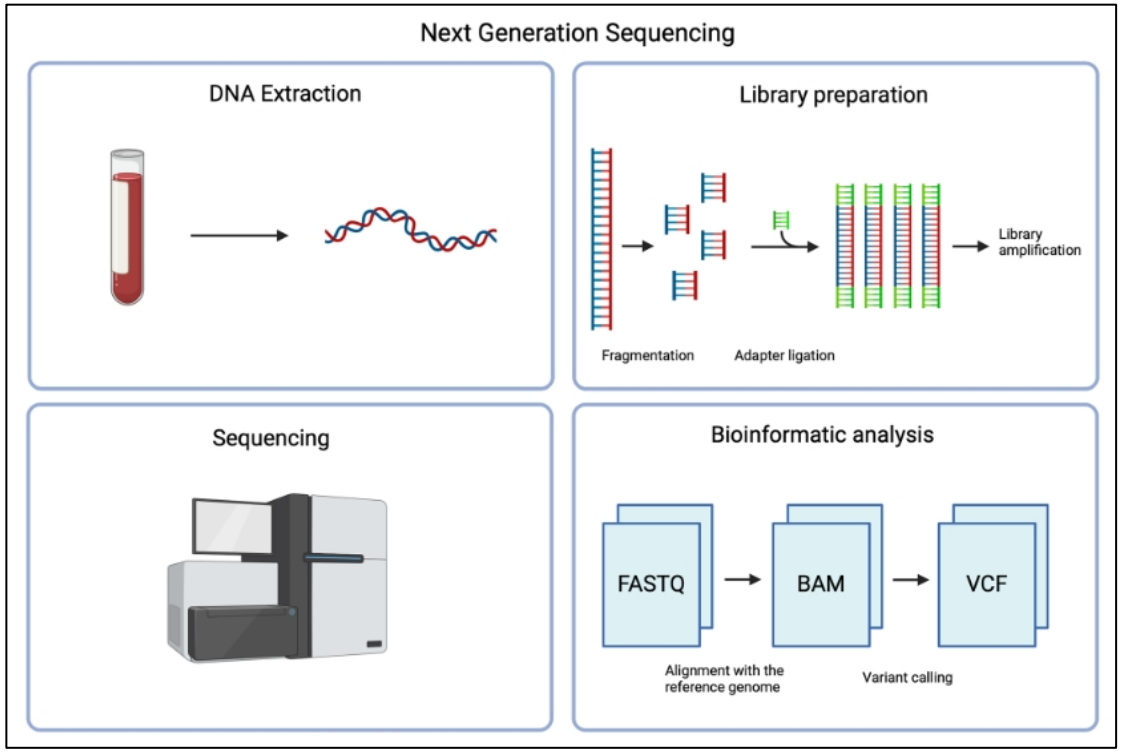
Why perform a clinical examination?

The molecular genetic revolution

Expansion of an unstable trinucleotide CAG repeat in spinocerebellar ataxia type 1

Harry T. Orr[1], Ming-yi Chung[1], Sandro Banfi[2], Thomas J. Kwiatkowski Jr.[3], Antonio Servadio[2], Arthur L. Beaudet[3,4], Alanna E. McCall[2], Lisa A. Duvick[1], Laura P. W. Ranum[1] & Huda Y. Zoghbi[2,3]

Nat Genet 1993;4:221-6



Lucain et al. J Neurol 2025;272:753

Why perform a clinical examination?

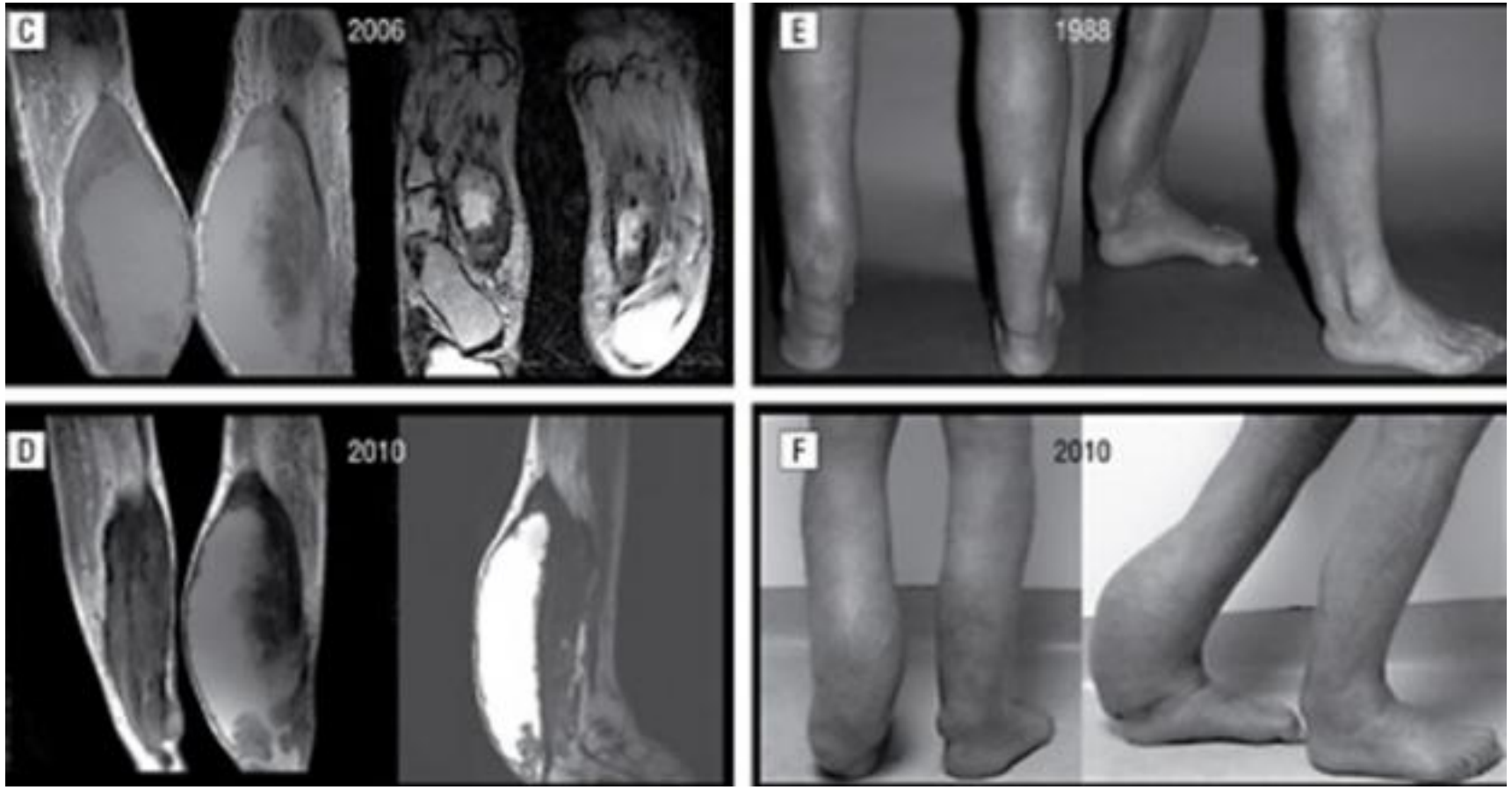
- The clinical examination strengthens the personal bond between the doctor and the patient and provides direct insight into the patient's specific problems.

- The clinical examination provides valuable clues for the diagnosis. For certain types of ataxia, a (nearly) definitive diagnosis can be made based solely on clinical findings. The results of the clinical examination are essential for interpreting ambiguous molecular genetic findings.

- A clinical examination is currently the only objective method for determining the severity of ataxia, monitoring its progression, and evaluating treatment effects.

Perform a general physical examination!

- Many ataxias have non-neurological signs, particularly affecting the heart, eye, skin, and skeleton.



Cerebrotendinous xanthomatosis (CTX)

Minnerop et al.
Arch Neurol 2012;69:782-3

Perform a general physical examination!

- In patients with sporadic late onset cerebellar ataxia, the physical examination must include blood pressure measurement in supine and upright position.

	Clinically established MSA	Clinically probable MSA
Core clinical features	1. Autonomic dysfunction defined as (at least one is required) ○ Unexplained voiding difficulties with post-void urinary residual volume ≥100 mL ○ Unexplained urinary urge incontinence ○ Neurogenic OH (≥20/10 mmHg blood pressure drop) within 3 minutes of standing or head-up tilt test and at least one of 1. Poorly L-dopa-responsive parkinsonism 2. Cerebellar syndrome (at least two of gait ataxia, limb ataxia, cerebellar dysarthria, or oculomotor features)	At least two of: 1. Autonomic dysfunction defined as (at least one is required): ○ Unexplained voiding difficulties with post-void urinary residual volume ○ Unexplained urinary urge incontinence ○ Neurogenic OH (≥20/10 mmHg blood pressure drop) within 10 minutes of standing or head-up tilt test 2. Parkinsonism 3. Cerebellar syndrome (at least one of gait ataxia, limb ataxia, cerebellar dysarthria, or oculomotor features)

Neurogenic orthostatic hypotension in MSA

Wenning et al.
Mov Disord. 2022;37:1131-48

Perform a conventional neurological examination!

- There are only few “purely cerebellar” ataxias. The majority of ataxia patients have additional non-ataxia signs.

	SCA1	SCA2	SCA3	SCA6	*p*
Hyperreflexia	67.5	13.2	40.1	21.9	<0.001
Areflexia	17.9	64.4	57.8	23.8	<0.001
Extensor plantar	50.5	31.0	41.9	2.0	<0.001
Spasticity	59.3	8.9	44.4	13.6	<0.001
Paresis	22.4	14.4	24.8	5.7	<0.001
Muscle atrophy	29.1	22.5	39.0	10.7	<0.001
Fasciculations	39.1	38.3	37.0	2.8	<0.001
Myoclonus	4.3	13.7	4.4	0.0	<0.001
Rigidity	1.7	7.4	10.3	5.7	NS
Chorea/dyskinesia	6.8	6.8	10.1	1.9	NS
Dystonia	12.8	14.2	23.9	4.7	<0.001
Resting tremor	6.8	14.9	3.6	1.9	<0.001
Sensory symptoms	62.4	68.4	65.6	48.0	NS
Urinary dysfunction	35.0	40.4	45.6	31.1	NS
Cognitive impairment	21.5	25.9	19.3	10.5	NS
Brainstem oculomotor signs	74.1	87.0	67.9	25.2	<0.001

EUROSCA cohort
Non-ataxia signs in SCA1, SCA2, SCA3, and SC6

Schmitz-Hübsch et al.
Neurology 2008;71:982-9

Multiple choice question (1)

Which of the following statements is correct?

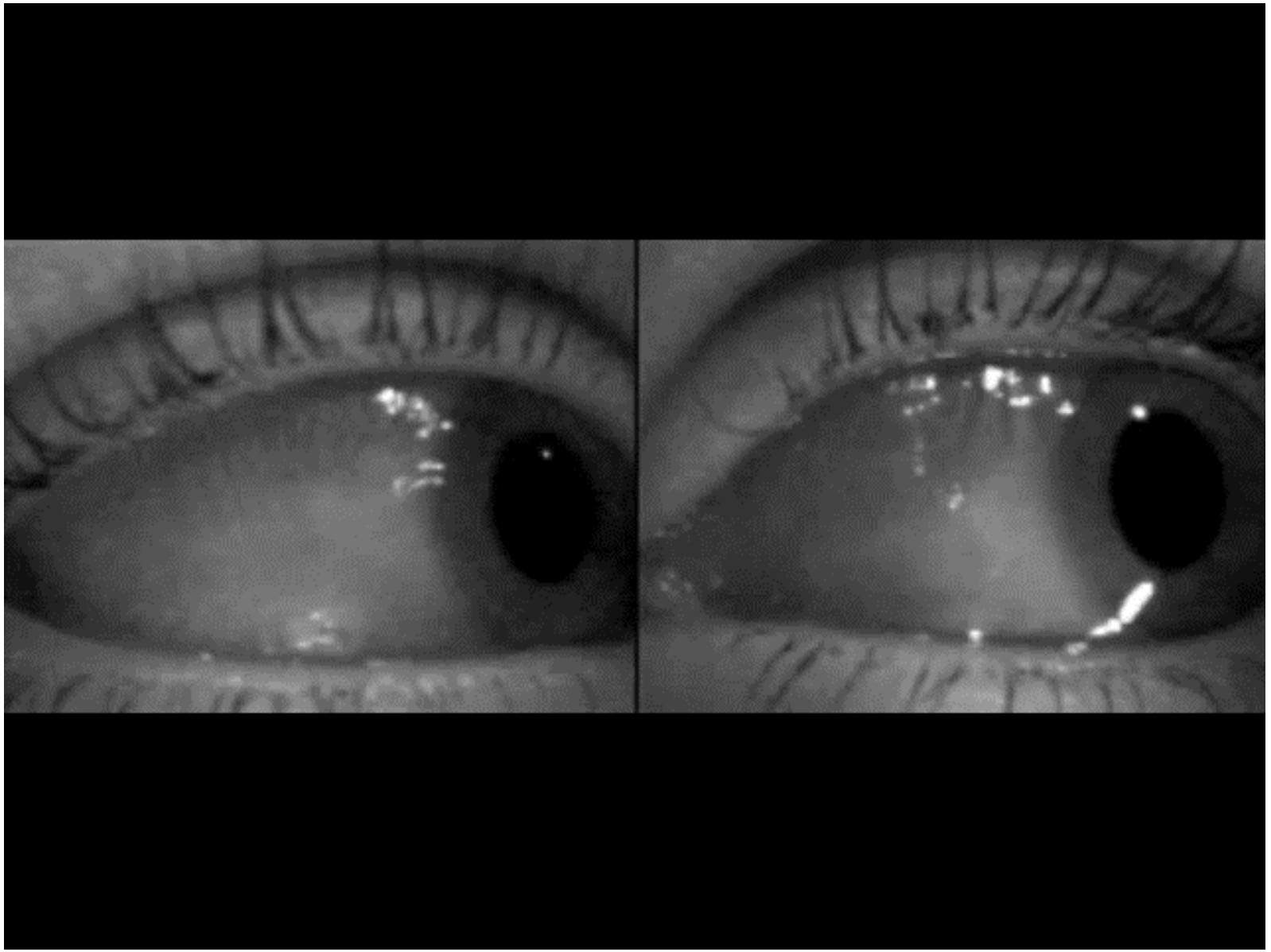
1. The clinical examination is not sensitive enough to detect ataxia progression.
2. The clinical examination plays only a minor role in the diagnosis of ataxias.
3. Molecular genetic tests always allow a definitive diagnosis, making it unnecessary to compare the results with clinical findings.
4. For certain types of ataxias, a (nearly) definitive diagnosis can be made based solely on clinical findings.
5. In most ataxias, digital motor assessment is more meaningful than clinical examination.

Pay special attention to eye movements!

Oculomotor sign	Oculomotor test	Affected structures
Gaze-evoked nystagmus	Eccentric gaze holding	Cerebellar flocculonodular lobule / Reticular formation
Saccadic hypermetria	Saccades	Cerebellar dorsal vermis
Downbeat nystagmus	Fixation and eccentric gaze holding	Cerebellar flocculonodular lobule
Vertical gaze palsy	Upward gaze	Mesencephalon
Saccadic slowing	Horizontal saccades	Pons
Square wave jerks	Fixation	Multiple structures outside the cerebellum
Vestibular areflexia	Head impulse test	Vestibular organs

Pay special attention to eye movements!

Gaze-evoked nystagmus (cerebellar disease)

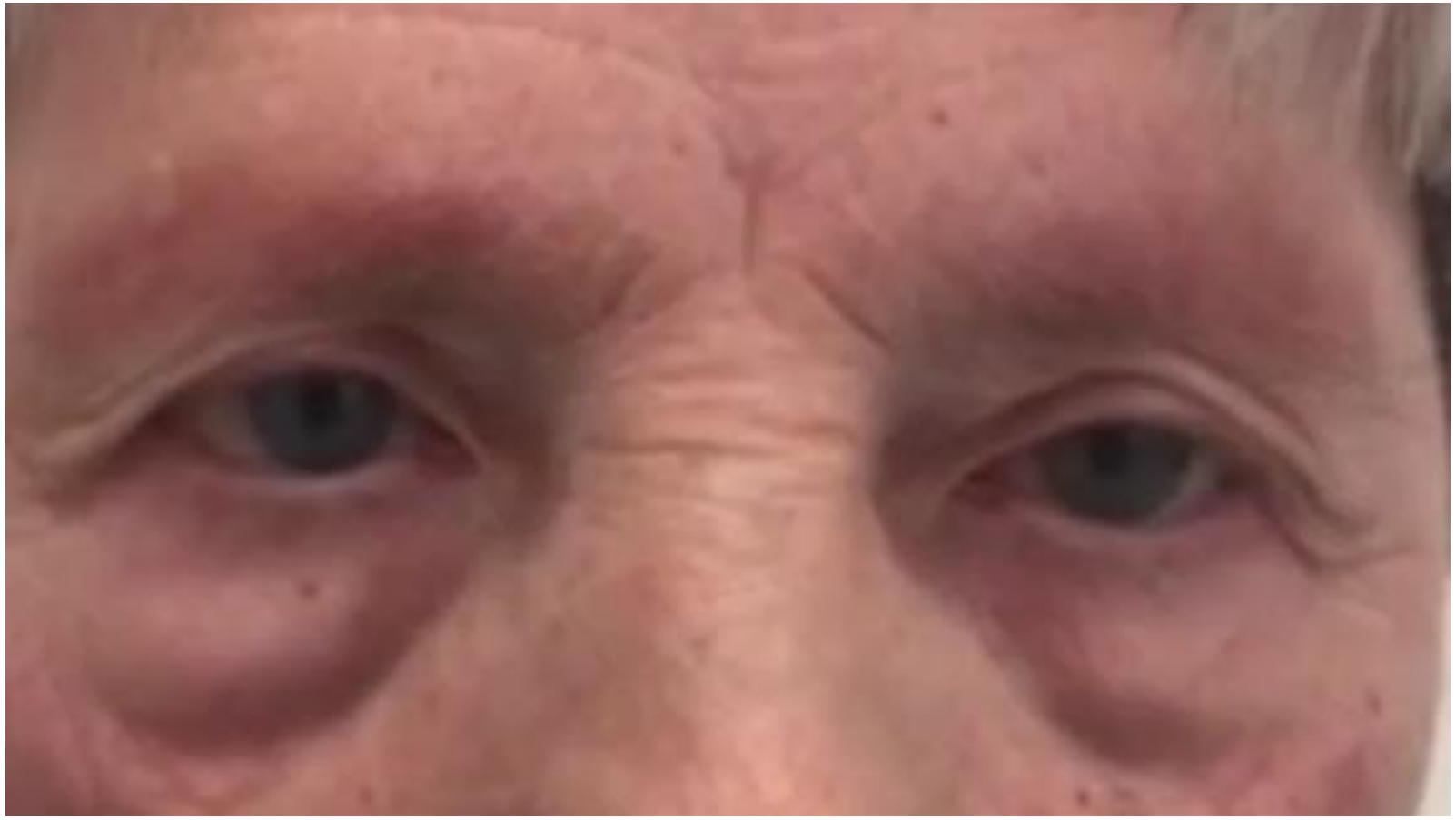


Pay special attention to eye movements!

Saccadic hypermetria
(cerebellar disease)

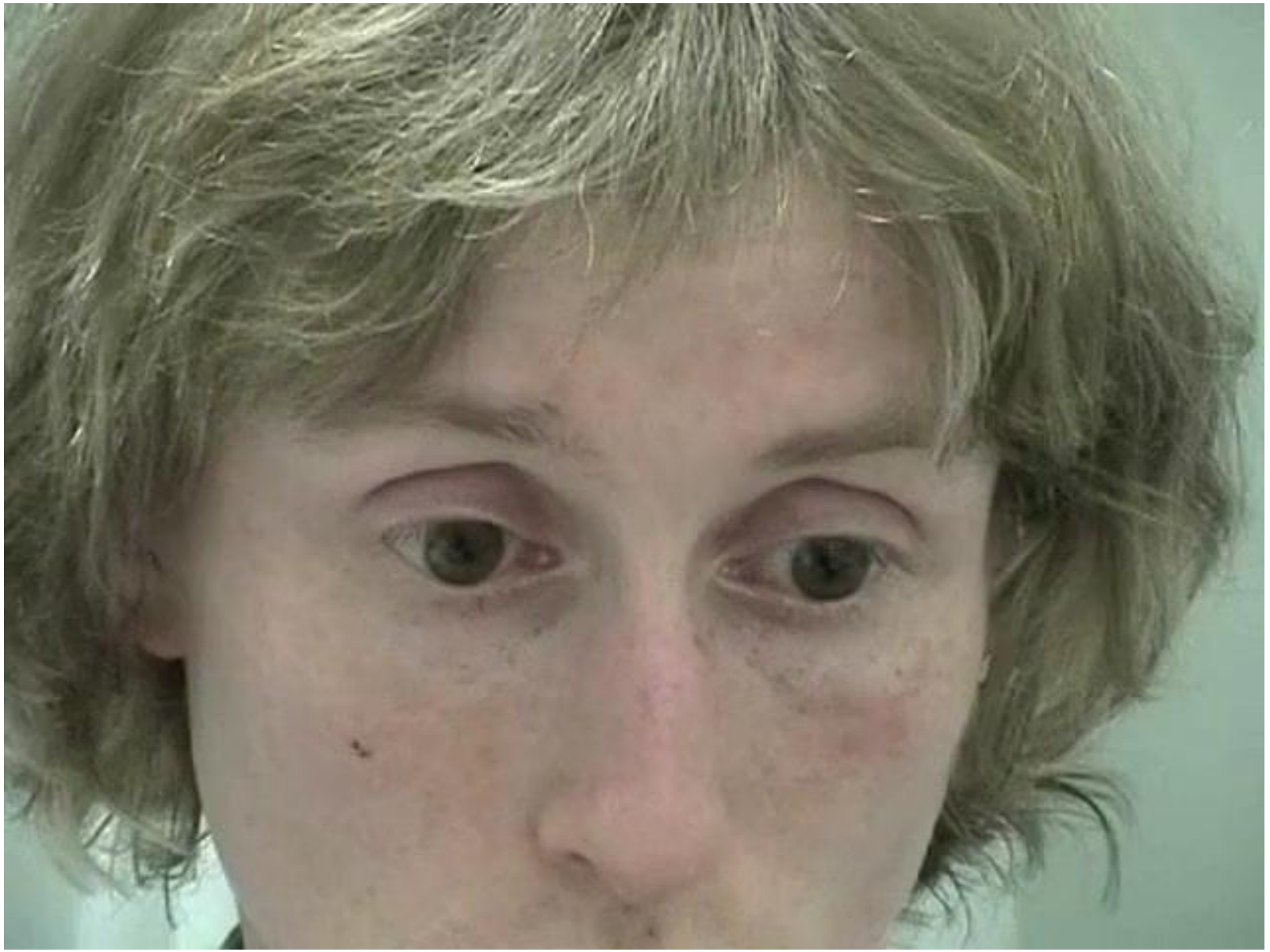
Pay special attention to eye movements!

Downbeat nystagmus (SCA27B)



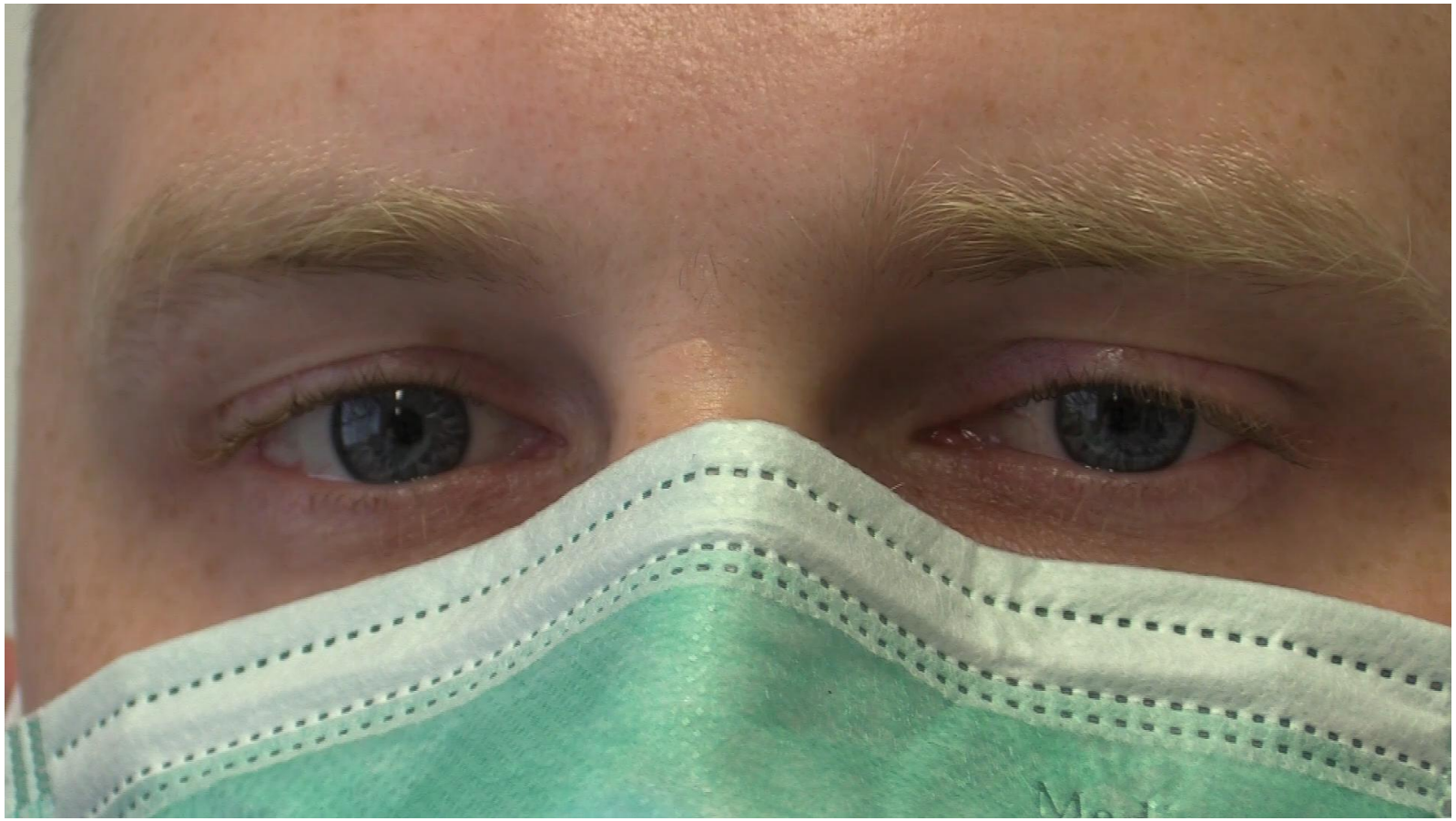
Pay special attention to eye movements!

Saccadic slowing (SCA2)



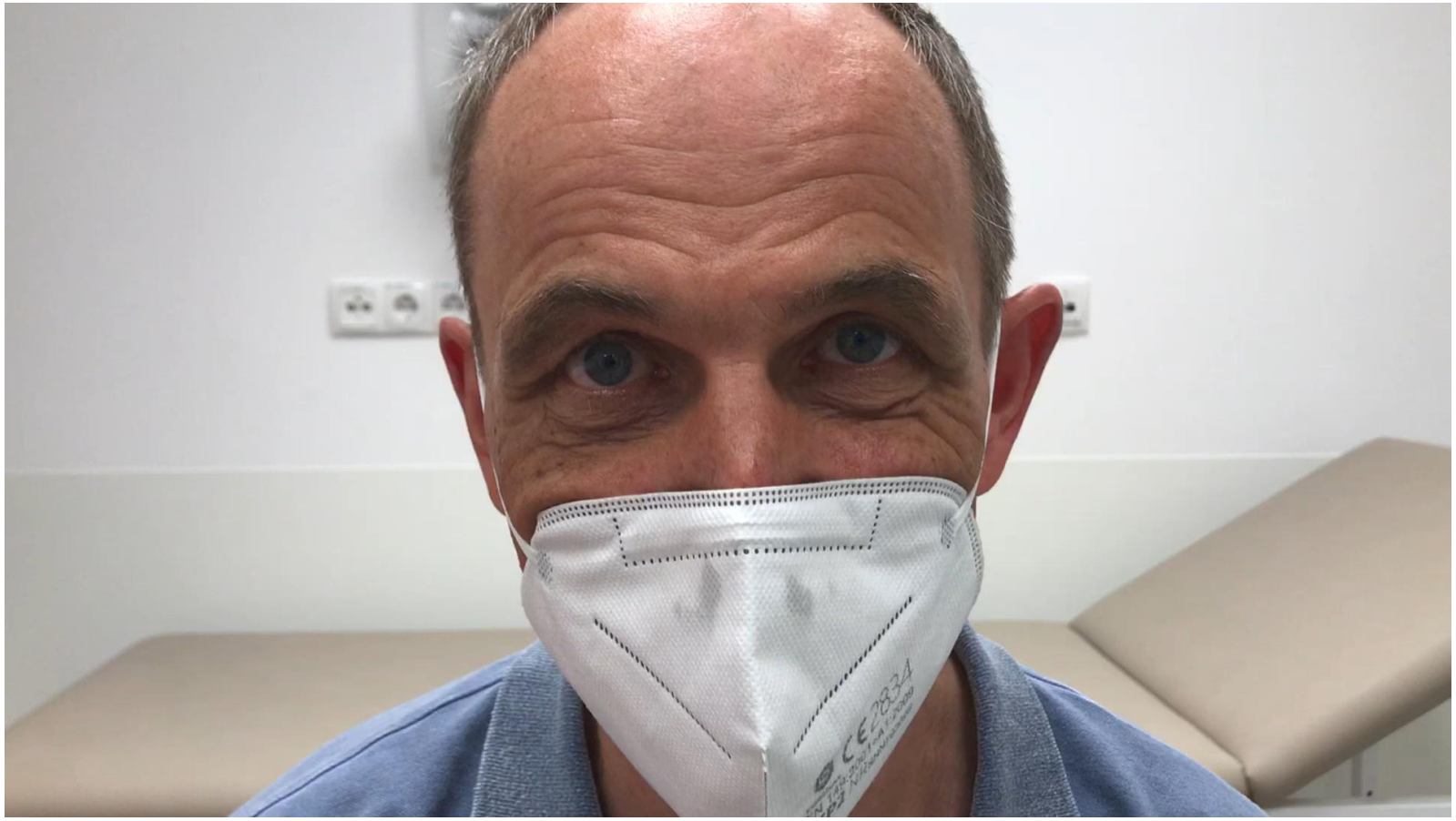
Pay special attention to eye movements!

Square wave jerks (FRDA)



Pay special attention to eye movements!

Vestibular areflexia
(RFC1 disease)

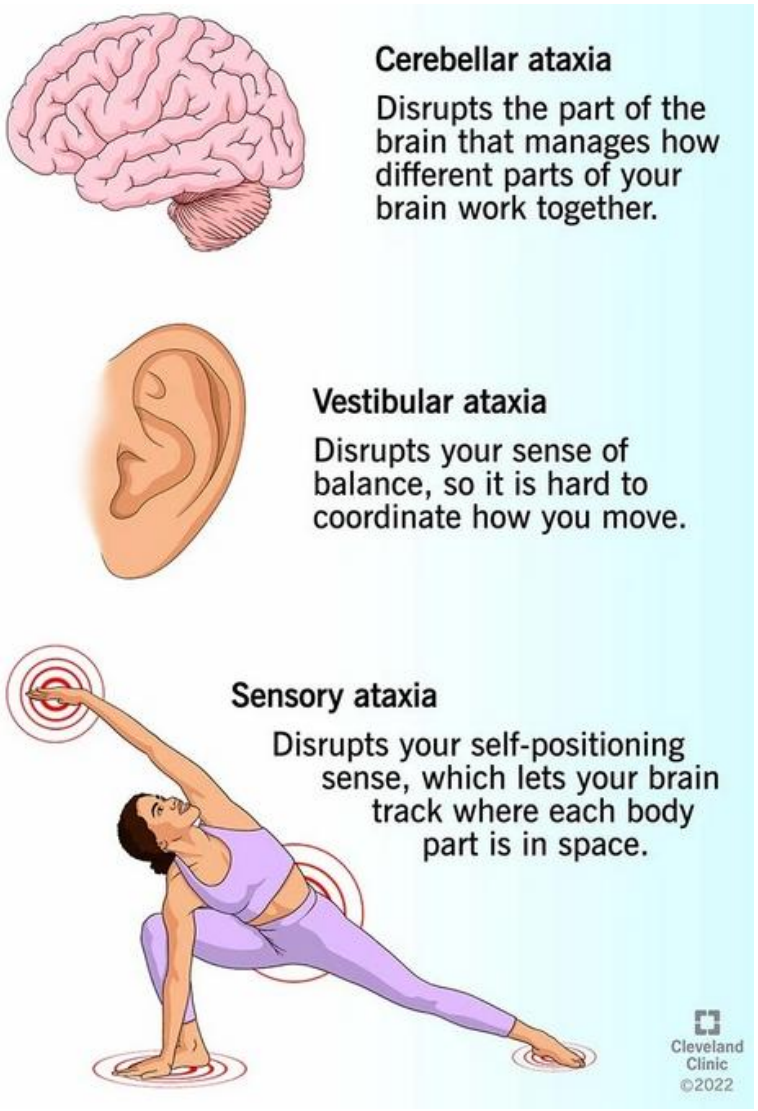


Multiple choice question (2)

Which of the following statements is wrong?

1. Square wave jerks are a frequent eye movement abnormality in Friedreich´s ataxia.
2. Slowing of saccades is often encountered in SCA2.
3. Vertical gaze palsy indicates dysfunction of the cerebellar flocculus.
4. Gaze-evoked nystagmus, saccadic hypermetria, and downbeat nystagmus are considered typical cerebellar eye movement signs.
5. The head-impulse test is used to test vestibular function.

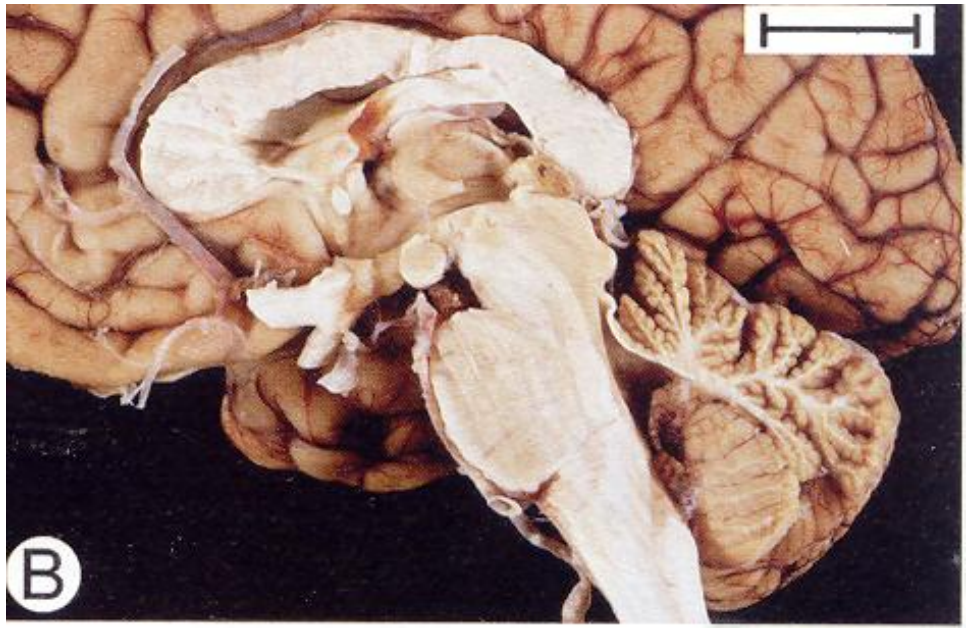
Define the type of ataxia!

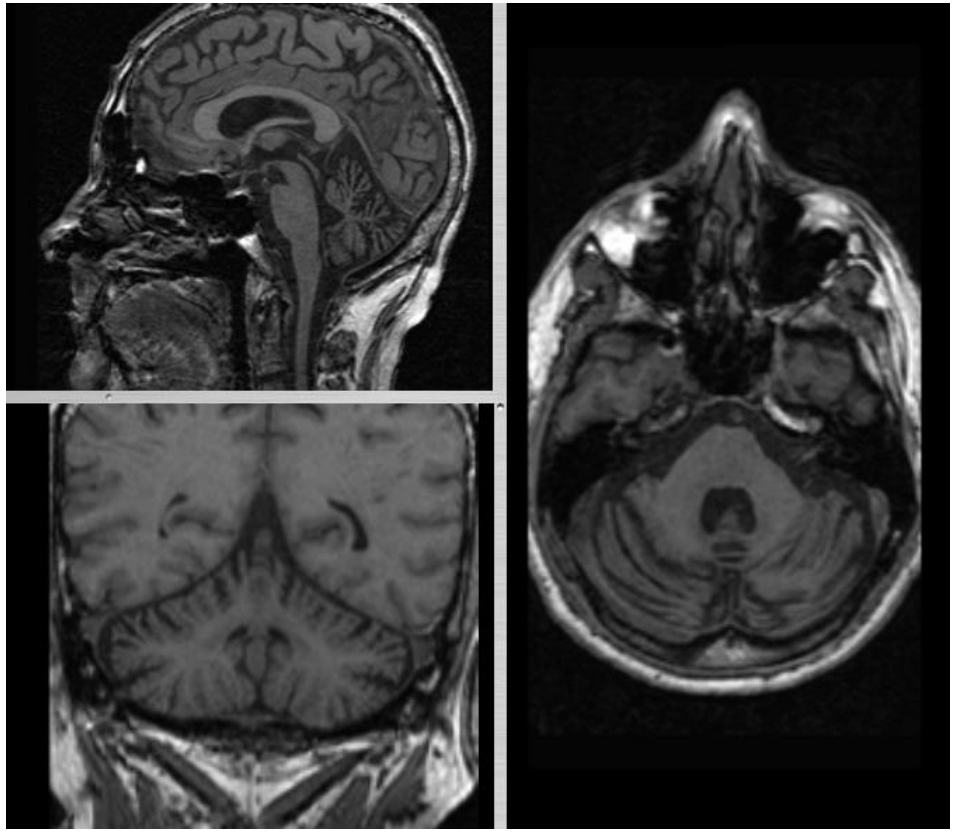


	Eye movements	Sensory signs	Stance
Cerebellar	Gaze-evoked nystagmus Saccadic hypermetria		
Vestibular	Abnormal head impulse test		
Sensory		Abnormal position sense Abnormal vibration sense	Positive Romberg´s sign

Define the type of ataxia!

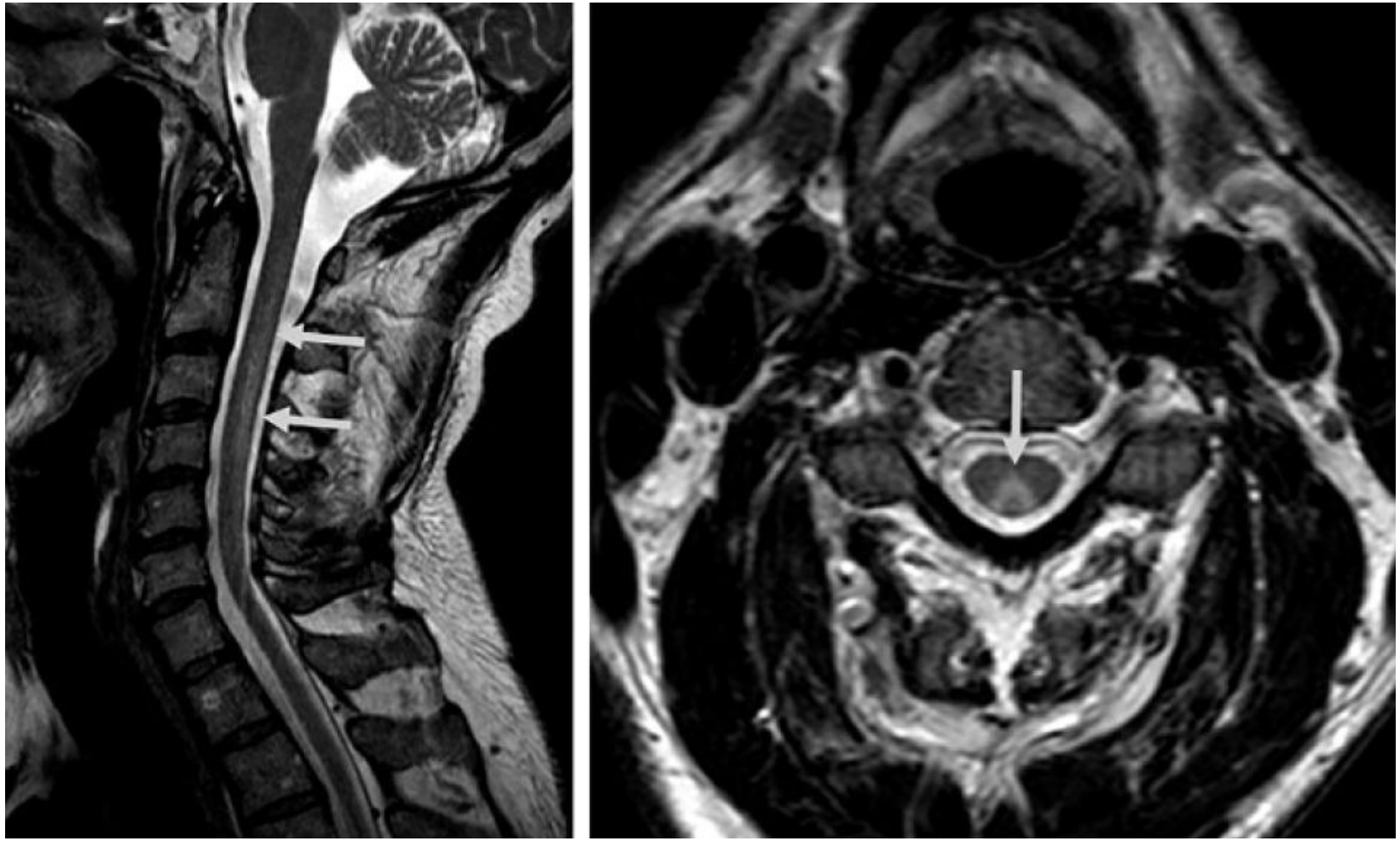
Cerebellar ataxia (SCA6)





Define the type of ataxia!

Sensory ataxia (Posterior column lesion due to nitrous oxide inhalation)



Meissner et al. Nervenarzt 2023:94:951-5

Define the type of ataxia!

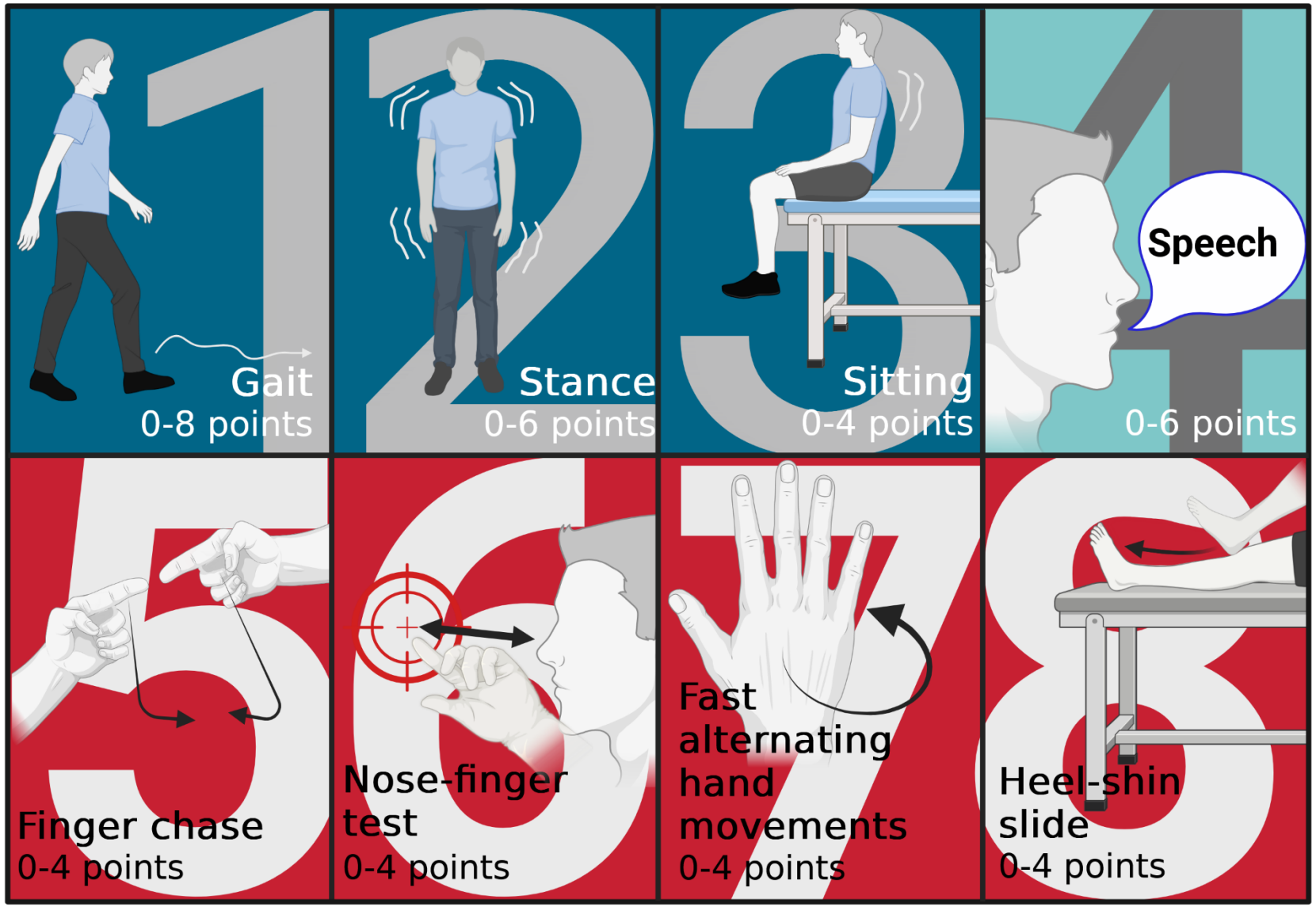
- Most of the more common forms of ataxia are characterized by a combination of cerebellar, vestibular and sensory ataxia.
- Examples are SCA3, FRDA, and RFC1 disease.

Multiple choice question (3)

Which of the following statements is correct?

1. SCA3 is characterized by a combination of cerebellar, vestibular and sensory ataxia.
2. RFC1 disease is a purely vestibular ataxia and does not have a cerebellar and sensory component.
3. Stance is particularly important to differentiate between cerebellar and vestibular ataxia.
4. Careful observation of gait allows differentiation of cerebellar, vestibular and sensory ataxia.
5. Vestibular areflexia always occurs in conjunction with deafness.

Assess and rate ataxia!



Scale for the Assessment and Rating of Ataxia (SARA)

Schmitz-Hübsch et al. Neurology 2006;66:1717-20

SARA Training Tool

https://ataxia-global-initiative.net/resources/sara-training-tool/

Assess and rate ataxia!

Item 1

Gait

Assess and rate ataxia!

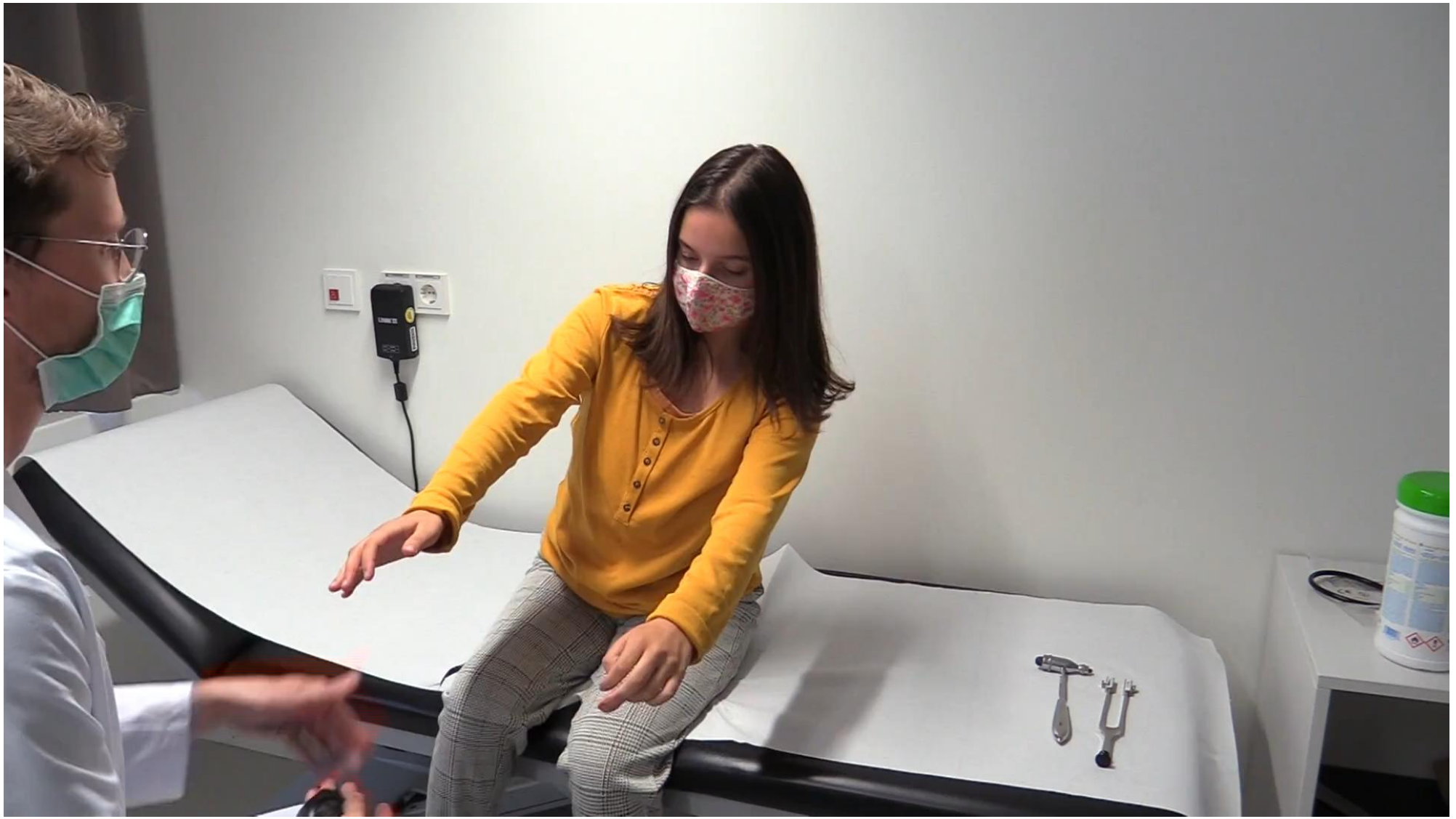
Item 2

Stance

Assess and rate ataxia!

Item 3

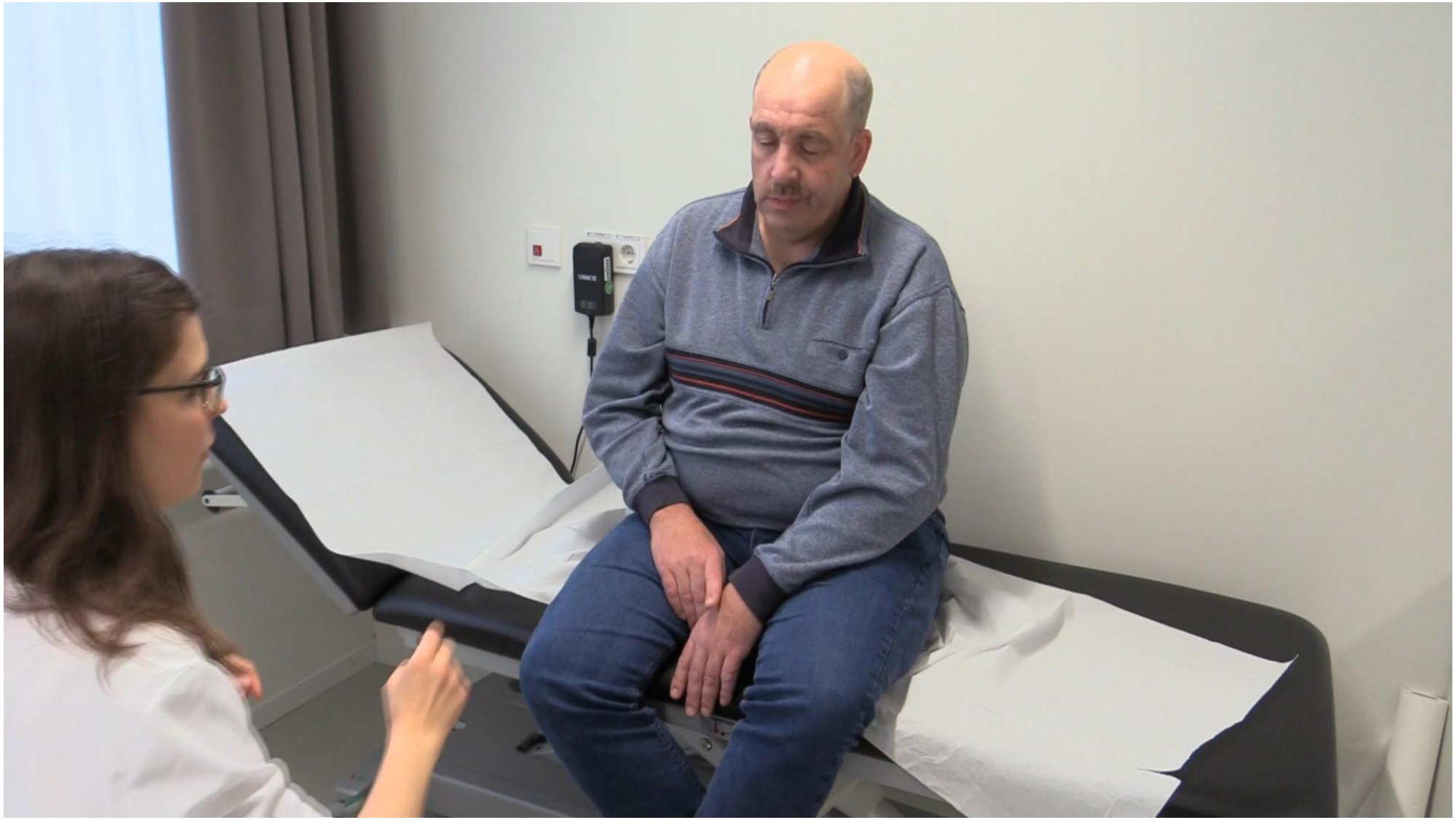
Sitting



Assess and rate ataxia!

Item 5

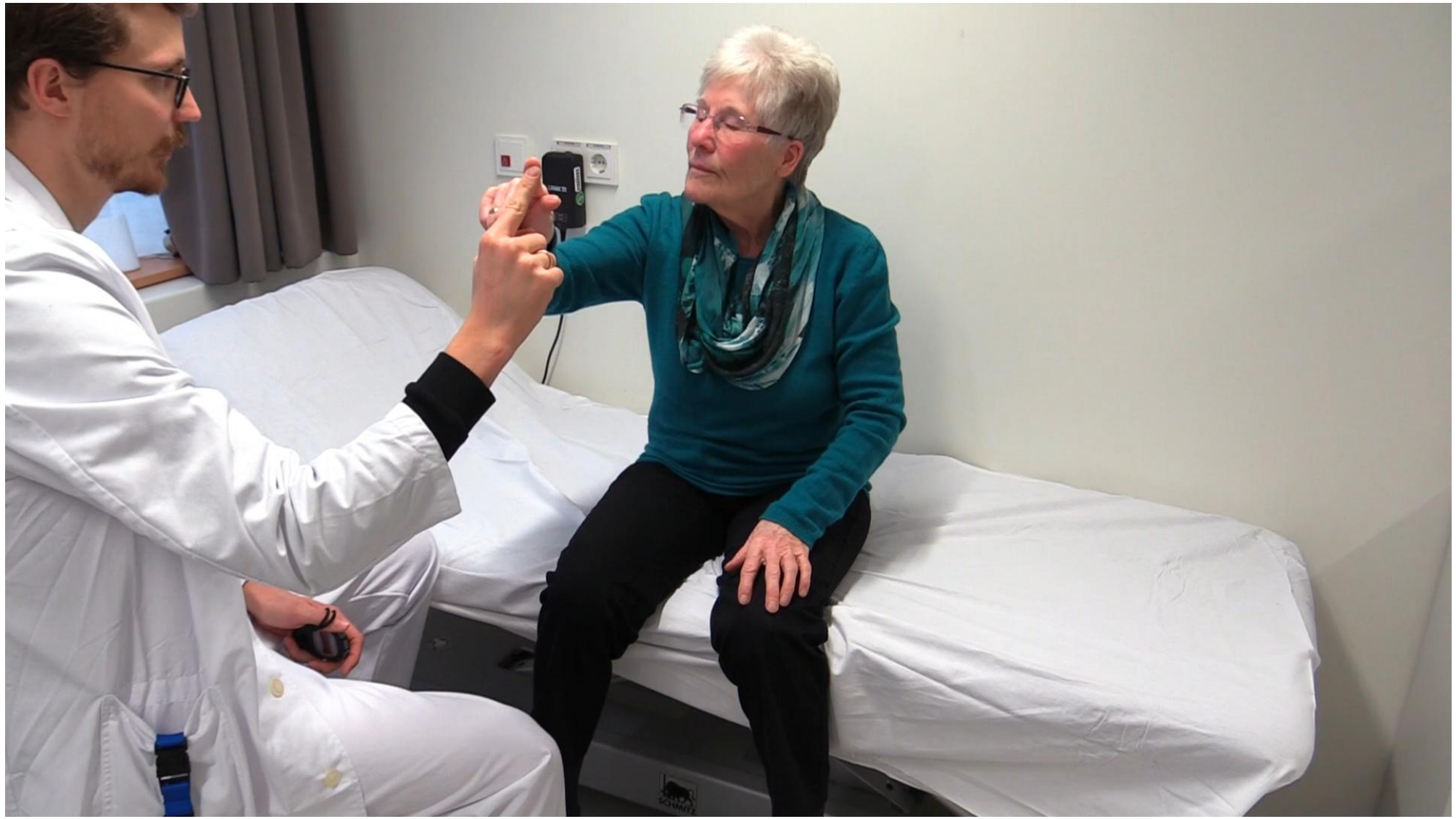
Finger chase



Assess and rate ataxia!

Item 6

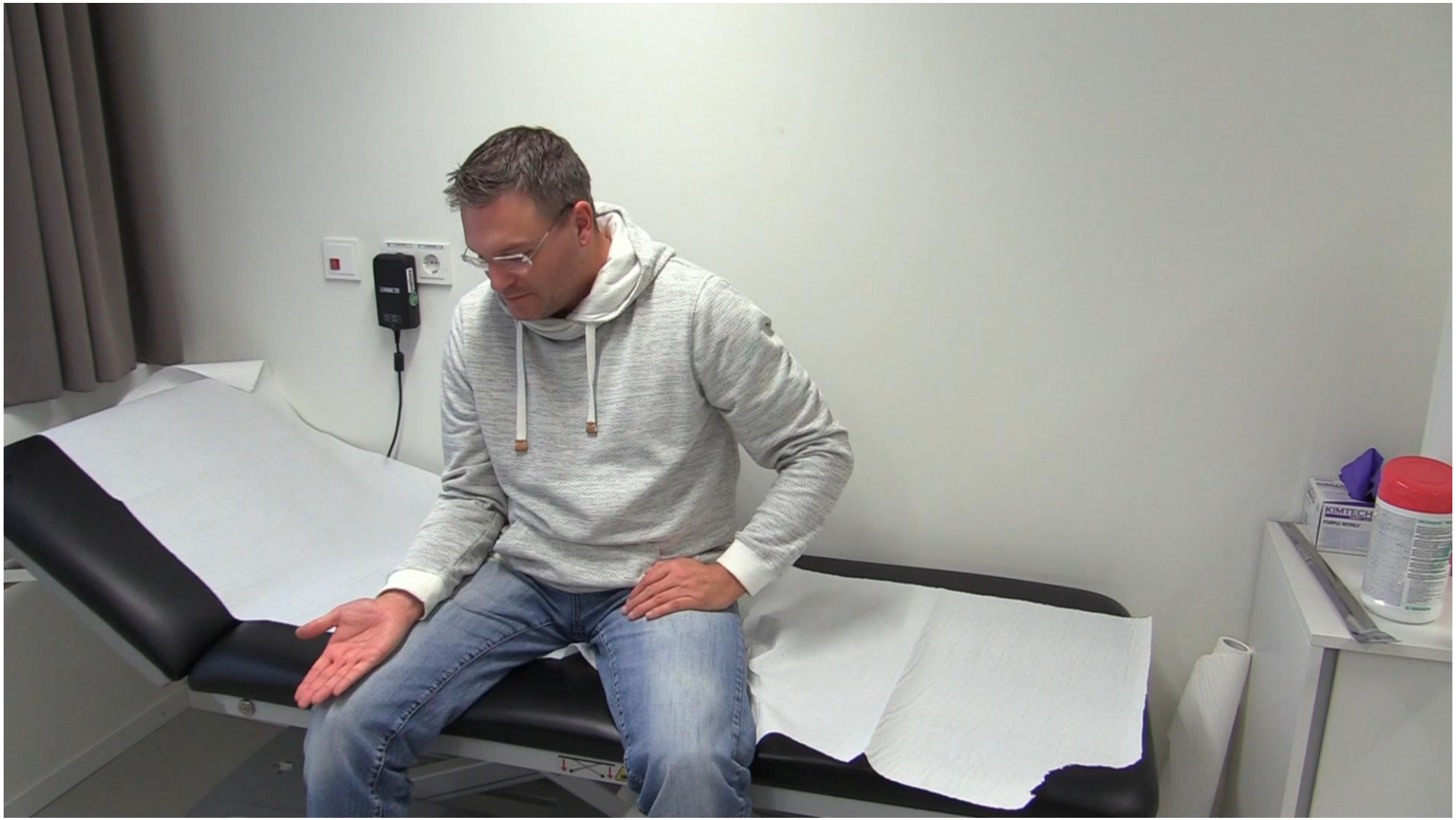
Nose-finger test



Assess and rate ataxia!

Item 7

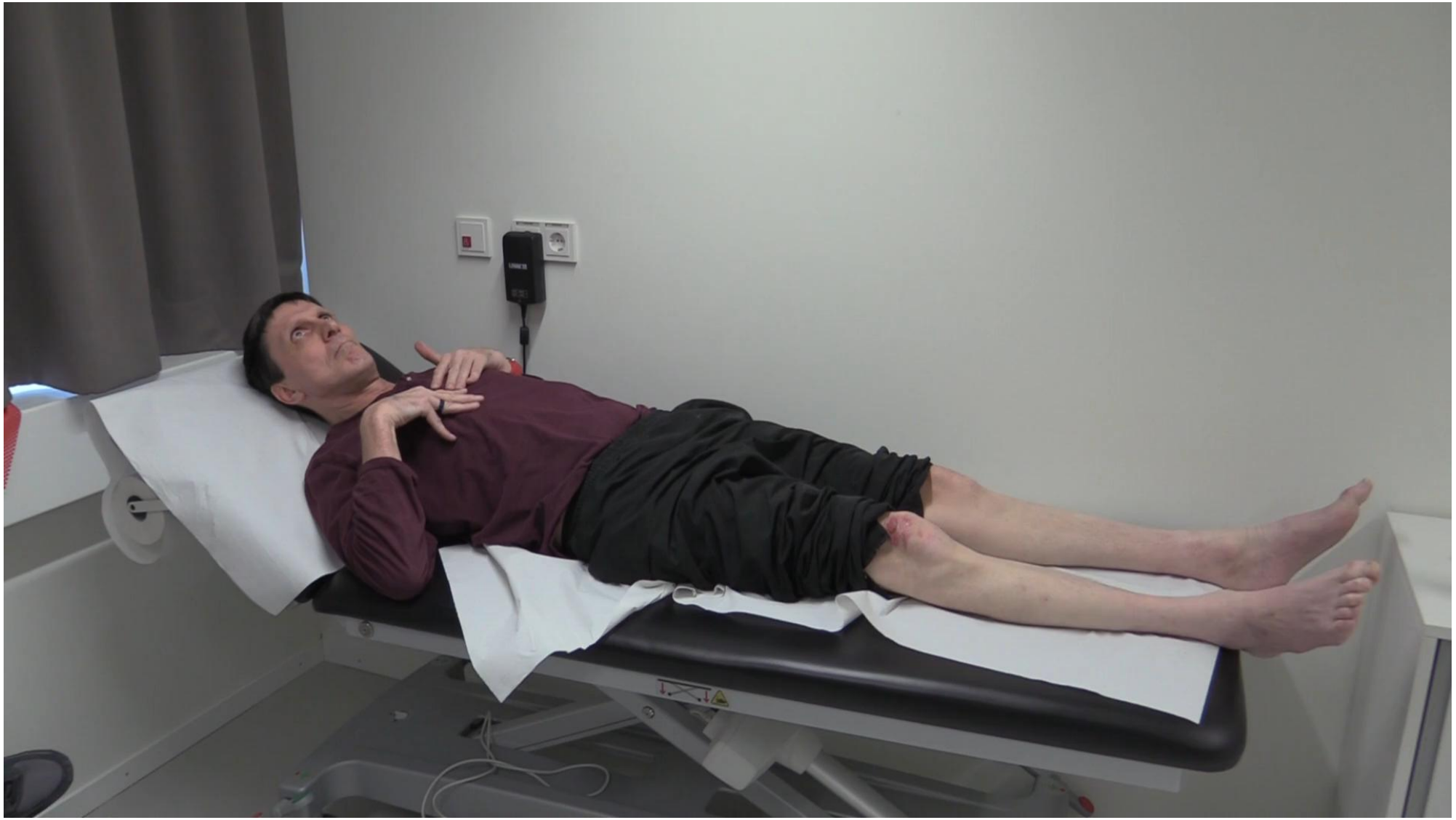
Fast alternating hand movements



Assess and rate ataxia!

Item 8

Heel-shin slide



Summary

- The clinical examination provides valuable clues for the diagnosis. For certain types of ataxia, a (nearly) definitive diagnosis can be made based solely on clinical findings.
- The clinical examination should include a general physical and conventional neurological examination.
- As many ataxias have characteristic eye movement abnormalities, you should pay special attention to eye movements.
- To define the (pathophysiological) type of ataxia, assessment of eye movements, sensory signs and stance are important.
- Application of ataxia scales allows for determining the severity of ataxia, monitoring its progression, and evaluating treatment effects.