

## DIAGNOSTIC FORM FOR SPINOCEREBELLAR DEGENERATION (ATAXIA AND/OR SPASTIC PARAPARESIS)

Date: \_\_\_ / \_\_\_ / \_\_\_    Center: \_\_\_\_\_    Neurologist: \_\_\_\_\_

Code ID patient: \_\_\_\_\_    Birthdate: \_\_\_ / \_\_\_ / \_\_\_

Proband:     Yes     No                      Sex:     female     maleInitial exam:     Yes     No            Follow up n°: \_\_\_\_\_

Stick the identification tag

### A. FAMILIAL HISTORY (add pedigree)

	No	Yes	
Spastic paraparesis and/or ataxia in the family?	<input type="checkbox"/>	<input type="checkbox"/>	
Other familial disease	<input type="checkbox"/>	<input type="checkbox"/>	Specify: _____
Consanguinity	<input type="checkbox"/>	<input type="checkbox"/>	Specify: _____
Parental inheritance of the disease	<input type="checkbox"/> Paternal <input type="checkbox"/> Maternal		
Geographical origin of the transmitting parent:	_____		

### B. AGE

Age at ONSET: \_\_\_\_\_

Age at examination: \_\_\_\_\_

### C. DEVELOPMENT AND SIGNS AT ONSET

	Normal	Delayed	Specify:
Motor development	<input type="checkbox"/>	<input type="checkbox"/>	
Intellect. development	<input type="checkbox"/>	<input type="checkbox"/>	
<b>Signs at onset</b>	<b>Yes</b>	<b>No</b>	<b>At age</b>
• Unsteadiness	<input type="checkbox"/>	<input type="checkbox"/>	
• Dysarthria	<input type="checkbox"/>	<input type="checkbox"/>	
• Stiff legs	<input type="checkbox"/>	<input type="checkbox"/>	
• Cramps	<input type="checkbox"/>	<input type="checkbox"/>	
• Medical exam	<input type="checkbox"/>	<input type="checkbox"/>	
• Pain	<input type="checkbox"/>	<input type="checkbox"/>	
• Other :	<input type="checkbox"/>	<input type="checkbox"/>	

### D. PREDOMINANT SIGNS at examination

	SPASTICITY				CEREBELLAR ATAXIA			
	None	Mild	Moder.	Severe	None	Mild	Moder.	Severe
• Upper limbs	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Lower limbs*	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Gait	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Dysarthria	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
* at rest for spasticity and knee-heel for ataxia								

Please specify:

### E- DISABILITY STAGE

	At age		At age
<input type="checkbox"/> 0: no functional handicap		<input type="checkbox"/> 4: severe, walking with one stick	
<input type="checkbox"/> 1: no functional handicap but signs at examination		<input type="checkbox"/> 5: walking with two sticks	
<input type="checkbox"/> 2: mild, able to run, walking unlimited		<input type="checkbox"/> 6: unable to walk, requiring wheelchair	
<input type="checkbox"/> 3: moderate, unable to run, limited walking without aid		<input type="checkbox"/> 7: confined to bed	

**F- OTHER CLINICAL SIGNS**

<b>1. Reflexes</b>						
	Normal	Increased	Diffused	Decreased	Absent	Clonus
• Jaw jerk	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Biceps	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Finger flexor	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Patellar	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Adductor	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Ankle	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<b>Absent</b>	<b>Present</b>				
• Hoffmann's sign	<input type="checkbox"/>	<input type="checkbox"/>				
	<b>Flexor</b>	<b>Indifferent</b>	<b>Unilat. ↑</b>	<b>Bilat. ↑↑</b>		
• Plantar reflex	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		

<b>2. Motor deficit</b>				
	None	Mild 4/5	Moderat 2-3/5	Severe <2/5
• Facial palsy/ atrophy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Proximal UL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Distal UL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Proximal LL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Distal LL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

<b>3. Muscle wasting</b>				
	None	Mild	Moderate	Severe
• Proximal UL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Distal UL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Proximal LL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Distal LL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

<b>4. Fasciculations or Myokymias (Facial contraction fasciculations) please circle</b>			
	No	Yes	Localisation:
	<input type="checkbox"/>	<input type="checkbox"/>	_____

<b>5. Sensory deficit</b>					
	None	Mild	Moderate	Severe	Abolished
• Vibration sense ↓ (ankles)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	(8/8)	(> 5/8)	(2-5/8)	(<2/8)	(0/8)
• Superficial sensory loss	<b>No</b>	<b>Touch</b>	<b>Prick</b>	<b>Cold</b>	
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

<b>6. Skeletal abnormalities</b>				
	None	Mild	Moderate	Severe
• Scoliosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Pes cavus	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Other: _____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

<b>7. Facial dysmorphia</b>			
	No	Yes	Describe:
	<input type="checkbox"/>	<input type="checkbox"/>	_____

<b>8. Sphincter and sexual disturbances</b>				
	None	Mild	Moderate	Severe
• Urinary urgency	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Urinary incontinence	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Urinary retention	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Anal incontinence	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Impaired sexual function	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Early menopause	<input type="checkbox"/> No	<input type="checkbox"/> Yes	At age: _____	

<b>9. Extra-pyramidal symptoms</b>					
	None	Mild	Moderate	Severe	Specify:
• Resting tremor	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
• Postural tremor	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
• Chorea	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
• Dystonia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
• Myoclonus	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
• Hypokinesia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
• Rigidity	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

10. Ophthalmological signs			
	No	Yes	
• Diplopia	<input type="checkbox"/>	<input type="checkbox"/>	
• Ptosis	<input type="checkbox"/>	<input type="checkbox"/>	
• Eye lid retraction (bulging eyes)	<input type="checkbox"/>	<input type="checkbox"/>	
• Diminished visual acuity	<input type="checkbox"/>	<input type="checkbox"/>	
* Fundus	No	Yes	
• Abnormal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/> Optic atrophy <input type="checkbox"/> Retinis Pigmentosa <input type="checkbox"/> Other:

* Oculomotor	No	Yes	
• Nystagmus	<input type="checkbox"/>	<input type="checkbox"/>	Describe: _____
• Saccadic pursuit	<input type="checkbox"/>	<input type="checkbox"/>	Describe: _____
• Slow saccades	<input type="checkbox"/>	<input type="checkbox"/>	
• Ocular motor apraxia	<input type="checkbox"/>	<input type="checkbox"/>	
• Vertical ophthalmoplegia	<input type="checkbox"/>	<input type="checkbox"/>	Describe: _____
• Horizontal ophthalmoplegia	<input type="checkbox"/>	<input type="checkbox"/>	Describe: _____

11. Mental status			
	No	Yes	
• Intellectual deterioration	<input type="checkbox"/>	<input type="checkbox"/>	At age: _____ Type: _____
• Mental retardation	<input type="checkbox"/>	<input type="checkbox"/>	At age: _____ Type: _____
• Psychiatric symptoms	<input type="checkbox"/>	<input type="checkbox"/>	Describe: _____

12. Other signs			
	No	Yes	Describe
• Dysphagia	<input type="checkbox"/>	<input type="checkbox"/>	_____ Severity: _____
• Skin problems	<input type="checkbox"/>	<input type="checkbox"/>	_____
• Hearing impairment	<input type="checkbox"/>	<input type="checkbox"/>	_____
• Epilepsy	<input type="checkbox"/>	<input type="checkbox"/>	At age: _____ Type: _____

13: Other medical complaints: \_\_\_\_\_

G- FUNCTIONAL CLINICAL EVALUATION - Please perform ALL tests listed in annexes and indicate scores below	
- SPRS (annex 1) : _____ / 52	- 25 feet ambulatory test (annex 4): _____ sec
- SARA (annex 2) : _____ / 40	- UHDRS – functional part IV (annex 5): _____ / 25
- CCFS (annex 3) : _____	

**H- CLINICAL DIAGNOSTIC CONCLUSION**

Cerebellar ataxia		
<input type="checkbox"/> Autosomal dominant	<input type="checkbox"/> Pure form	<input type="checkbox"/> Definitely affected
<input type="checkbox"/> Autosomal recessive	<input type="checkbox"/> Complicated form	<input type="checkbox"/> Probably affected (only dysarthria)
<input type="checkbox"/> Isolated case		<input type="checkbox"/> Possibly affected (only mild gait ataxia)
<input type="checkbox"/> X-linked		

Spastic paraparesis		
<input type="checkbox"/> Autosomal dominant	<input type="checkbox"/> Pure form	<input type="checkbox"/> Definitely affected
<input type="checkbox"/> Autosomal recessive	<input type="checkbox"/> Complicated form	<input type="checkbox"/> Probably affected (enhanced or very brisk LL reflexes +/- Babinski)
<input type="checkbox"/> Isolated case		<input type="checkbox"/> Possibly affected (enhanced LL reflexes)
<input type="checkbox"/> X-linked		

I- MOLECULAR DIAGNOSIS	
Genes/Loci to test:	Diagnosis:

**J- COMPLEMENTARY INVESTIGATIONS**

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
1. Cerebral MRI	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
				None Mild Moder. Severe
- Cerebrum				<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>
- Cerebellum				<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>
- Brainstem				<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>
- Corpus callosum				<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
2. Medullar MRI	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
				<b>ATROPHY</b>
				None Mild Moder. Severe
- Upper spinal cord				<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
3. EMG + NCV UL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
4. EMG + NCV LL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
5. VEP	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
6. AEP	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
7. MEP	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
8. SEP	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
9. VLCFA	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
10. $\alpha$ -foetoprotein	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
11. Cholesterol	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
12. Serum protein electrophoresis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
13. Vitamin E	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
14. Apolipoprotein A, B	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
15. Muscle biopsy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
16. Skin biopsy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
17. ERG	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
18. Fundus examination	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
19. Neuropsychological exam	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
- IQ				
20. Urodynamics	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
21. Urine density	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

**K- STORED MATERIAL**

	Yes	No
• DNA	<input type="checkbox"/>	<input type="checkbox"/>
• Immortalized cell lines	<input type="checkbox"/>	<input type="checkbox"/>
• Muscle tissue	<input type="checkbox"/>	<input type="checkbox"/>
• Skin biopsy	<input type="checkbox"/>	<input type="checkbox"/>
• Nerve biopsy	<input type="checkbox"/>	<input type="checkbox"/>
• Other: _____	<input type="checkbox"/>	<input type="checkbox"/>

## ANNEX 1: Spastic Paraplegia Rating Scale (SPRS)

**(1) Walking distance without pause**

*Due to history, walking aids allowed*

- 0: Normal, unlimited
- 1: Abnormal exhaustion due to spasticity after more than 500m
- 2: Walking distance less than 500m
- 3: Walking distance less than 10 m
- 4: Unable to walk

**(2) Gait quality**

*Patient is asked to walk as fast as possible a 10 meter distance including one turn*

- 0: Normal
- 1: Mild stiffness, running still possible
- 2: Clearly spastic gait, interfering with running
- 3: Spastic gait requiring use of canes/walker
- 4: Unable to walk for a 10 meter distance even with maximal support

**(3) Maximum gait speed**

*Time for a 10 meter distance including one turn, taken by stop watch*

- 0: Normal
- 1: Slightly reduced (10m:  $\geq$  5s)
- 2: Moderately reduced (10m:  $\geq$  10s)
- 3: Severely reduced (10m:  $\geq$  20s)
- 4: Unable to walk for a 10m distance or time  $\geq$  40s

**Timing:**

_____sec
----------

**(4) Climbing stairs**

*5 steps upstairs - turn - 5 steps downstairs*

- 0: Normal: needs no support of the banister
- 1: Mild impairment: needs intermittent support of the banister
- 2: Moderate impairment: needs permanent support of the banister
- 3: Severe impairment: needs support of another person or additional walking aid to perform task
- 4: Unable to climb stairs

**(5) Speed of stair climbing**

*Time for 5 steps upstairs - turn - 5 steps downstairs, taken by stop-watch*

- 0: Normal
- 1: Slightly reduced ( $\geq$  5s to perform task)
- 2: Moderately reduced ( $\geq$  10s to perform task)
- 3: Severely reduced ( $\geq$  20s to perform task)
- 4: Unable to climb stairs

**Timing:**

_____sec
----------

**(6) Arising from chair**

*Patient attempts to arise from a straight-back wood or metal chair with arms folded across chest*

- 0: Normal
- 1: Slow, or may need more than one attempt.
- 2: Pushes self up from arms of seat.
- 3: Tends to fall back and may have to try more than one time but can get up without help.
- 4: Unable to arise without help.

**(7) Spasticity -hip adductor muscles (Modified Ashworth scale)**

*Score more severely affected side*

- 0: No increase in muscle tone
- 1: Slight increase in muscle tone, manifested by a catch and release
- 2: More marked increase in muscle tone through most of the range of motion
- 3: Considerable increase in muscle tone - passive movement is difficult
- 4: Limb stiff in adduction

**(8) Spasticity -knee flexion (Modified Ashworth scale)**

*Score more severely affected side*

- 0: No increase in muscle tone
- 1: Slight increase in muscle tone, manifested by a catch and release
- 2: More marked increase in muscle tone through most of the range of motion
- 3: Considerable increase in muscle tone - passive movement is difficult
- 4: Limb stiff in flexion or extension

**(9) Weakness -hip abduction (Medical Research Council 1976)**

- 0: No weakness
- 1: Mild weakness (4/5)
- 2: Moderate weakness (3/5)
- 3: Severe weakness (1-2/5)
- 4: Plegia (0/5)

**(10) Weakness -foot dorsiflexion (Medical Research Council 1976)**

- 0: No weakness
- 1: Mild weakness (4/5)
- 2: Moderate weakness (3/5)
- 3: Severe weakness (1-2/5)
- 4: Plegia (0/5)

**(11) Contractures of lower limbs**

*Score in supine position*

- *Hip extension: lumbar spine and thighs touch the underlay. Hip abduction: abduction up to an angle of  $>60^\circ$  between the legs possible*
- *Knee extension: thigh and calf touch the underlay*
- *Ankle dorsal extension:  $> 10^\circ$  possible. Ankle pronation:  $> 10^\circ$  possible*
- 0: No contracture
- 1: Mild, not fixed abnormal position of one joint (unilaterally or bilaterally)
- 2: Fixed contracture of one joint (unilaterally or bilaterally)
- 3: Fixed contracture of two joints (unilaterally or bilaterally)
- 4: Fixed contracture of more than two joints (unilaterally or bilaterally)

**(12) Pain due to SP related symptoms**

- 0: None
- 1:  $\leq 50\%$  of waking day present AND intensity 0 - 3 points on visual analogue scale
- 2:  $\leq 50\%$  of waking day present AND intensity 4 - 10 points on visual analogue scale
- 3:  $> 50\%$  of waking day present AND intensity 0 - 3 on visual analogue scale
- 4:  $> 50\%$  of waking day present AND intensity 4 - 10 points on visual analogue scale

**(13) Bladder and bowel function**

- 0: Normal bladder and bowel function
- 1: Urinary or fecal urgency (difficulties to reach toilet in time)
- 2: Rare and mild urge incontinence (no nappy required)
- 3: Moderate urge incontinence (requires nappy or catheter when out of the house)
- 4: Permanent catheterization or permanent nappy

**Total SPRS Score:      /52**

**ANNEX 2: 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)
- 1  Slight difficulties, only visible when walking 10 consecutive steps in tandem
- 2  Clearly abnormal, tandem walking >10 steps not possible
- 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 with supported

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 : \_\_\_\_\_

**3) Sitting**

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

- 0  Normal, no difficulties sitting > 10 sec
- 1  Slight difficulties, intermittent sway
- 2  Constant sway, but able to sit for > 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 : \_\_\_\_\_

**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    \_\_\_\_\_

**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    \_\_\_\_\_

**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    \_\_\_\_\_

**8) Heel-shin slide (Rated separately for each side)**

**Proband lies on examination bed, without vision 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 to lay the leg back on the examination bed. The task is performed 3 times. Slide-down movements should be performed within 1 s.**

- 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    \_\_\_\_\_

ANNEX 3: Composite Cerebellar Functional Severity Score (CCFS)

Dominant hand  Right  Left

**Nine-hole Pegboard test – dominant hand**

The patient is seated and holds nine dowels (9mm in diameter and 32-mm long) in one hand and places them randomly, one by one, with the other hand in a board with nine holes. Timing begins when the first peg is placed in a hole and ends when the last peg is placed. The examiner holds the board steady on the table during the test. The trial is performed once only with the dominant hand. If the patient drops a peg the examiner stops the timer and the patient starts the test again once from the beginning.

Timing dominant hand: \_\_\_\_\_sec

**Click test – dominant hand**

The patient is seated facing the examiner across a table on which is placed a device composed of two mechanical counters fixed on a wooden board 39 cm apart. The patient uses his index finger to press the buttons on the counters alternately 10 times. Timing begins when the first button is pressed and stops when the second counter reaches 10. The trial is performed once only with the dominant hand.

Timing dominant hand: \_\_\_\_\_sec

$$Z \text{ pegboard dominant hand} = \text{Pegboard DH} - (13.4 - 0.16 * \text{age} + 0.002 * \text{age}^2) = \underline{\hspace{2cm}}$$

$$Z \text{ click dominant hand} = \text{click DH} - (8 + 0.05 * \text{age}) = \underline{\hspace{2cm}}$$

$$\text{CCFS} = \log_{10} (7 + Z \text{ pegboard dominant hand} / 10 + 4 * Z \text{ click dominant hand} / 10) = \underline{\hspace{2cm}}$$

(Mean normal values  $0.85 \pm 0.05$  (0.64 – 0.94))

ANNEX 4: 25 feet ambulatory test  
(from the Friedreich Ataxia Rating Scale)

To test gait, place markers 25 feet apart in hallway with no furniture within reach of 1 m/3 ft. and no loose carpet. Patient walks 7.62 m/25 ft **at normal pace**, turns around using single step pivot and return to start. The activity is timed. Note if the gait was achieved with or without helping device and serial examinations should be done with the same device as in the first examination.

Helping device  No  Yes Describe: \_\_\_\_\_

Time: \_\_\_\_\_ sec

**ANNEX 5: Unified Huntington's Disease Rating Scale (UHDRS)**  
**part IV: Functional Assessment**

<b>IV. UHDRS - FUNCTIONAL ASSESSMENT</b>	<b>NO</b>	<b>YES</b>
43. Could subject engage in gainful employment in his/her accustomed work?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
44. Could subject engage in any kind of gainful employment?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
45. Could subject engage in any kind of volunteer or non gainful work?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
46. Could subject manage his/her finances (monthly) without any help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
47. Could subject shop for groceries without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
48. Could subject handle money as a purchaser in a simple cash (store) transaction?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
49. Could subject supervise children without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
50. Could subject operate an automobile safely and independently?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
51. Could subject do his/her own housework without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
52. Could subject do his/her own laundry (wash/dry) without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
53. Could subject prepare his/her own meals without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
54. Could subject use the telephone without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
55. Could subject take his/her own medications without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
56. Could subject feed himself/herself without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
57. Could subject dress himself/herself without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
58. Could subject bathe himself/herself without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
59. Could subject use public transportation to get places without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
60. Could subject walk to places in his/her neighbourhood without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
61. Could subject walk without falling?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
62. Could subject walk without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
63. Could subject comb hair without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
64. Could subject transfer between chairs without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
65. Could subject get in and out of bed without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
66. Could subject use toilet/commode without help?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
67. Could subject's care still be provided at home?	<input type="checkbox"/> 0	<input type="checkbox"/> 1
<b>Functional Assessment Score: _____/25</b>		
<b>68. Information Sources</b> <div style="display: flex; justify-content: space-between; align-items: flex-start;"> <div style="width: 60%;">                     Was the Functional Assessment information obtained from:                 </div> <div style="width: 35%;"> <input type="checkbox"/> Subject only  <input type="checkbox"/> Subject and family/companion                 </div> </div>		