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

Date: ___ / ___ / ___ Center: _____ Neurologist: _____

Code ID patient: _____ Birthdate: ____ / ____ / ____

Proband: ☐ Yes ☐ No Sex: ☐ female ☐ male

Initial 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?	☐	☐	
Other familial disease	☐	☐	Specify: _____
Consanguinity	☐	☐	Specify: _____
Parental inheritance of the disease	☐ Paternal ☐ 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	☐	☐	
Intellect. development	☐	☐	
Signs at onset	**Yes**	**No**	**At age**
• Unsteadiness	☐	☐	
• Dysarthria	☐	☐	
• Stiff legs	☐	☐	
• Cramps	☐	☐	
• Medical exam	☐	☐	
• Pain	☐	☐	
• Other :	☐	☐	

D. PREDOMINANT SIGNS at examination								
	SPASTICITY				**CEREBELLAR ATAXIA**			
	None	**Mild**	**Moder.**	**Severe**	**None**	**Mild**	**Moder.**	**Severe**
• Upper limbs	☐	☐	☐	☐	☐	☐	☐	☐
• Lower limbs*	☐	☐	☐	☐	☐	☐	☐	☐
• Gait	☐	☐	☐	☐	☐	☐	☐	☐
• Dysarthria	☐	☐	☐	☐	☐	☐	☐	☐
* at rest for spasticity and knee-heel for ataxia								

Please specify:

E- DISABILITY STAGE			
	At age		**At age**
☐ **0**: no functional handicap		☐ **4**: severe, walking with one stick	
☐ **1:** no functional handicap but signs at examination		☐ **5**: walking with two sticks	
☐ **2:** mild, able to run, walking unlimited		☐ **6**: unable to walk, requiring wheelchair	
☐ **3**: moderate, unable to run, limited walking without aid		☐ **7**: confined to bed	

F- OTHER CLINICAL SIGNS

1. Reflexes

	Normal	Increased	Diffused	Decreased	Absent	Clonus
• Jaw jerk	☐	☐	☐	☐	☐	☐
• Biceps	☐	☐	☐	☐	☐	☐
• Finger flexor	☐	☐	☐	☐	☐	☐
• Patellar	☐	☐	☐	☐	☐	☐
• Adductor	☐	☐	☐	☐	☐	☐
• Ankle	☐	☐	☐	☐	☐	☐

	Absent	Present
• Hoffmann's sign	☐	☐

	Flexor	Indifferent	Unilat. ↑	Bilat. ↑↑
• Plantar reflex	☐	☐	☐	☐

2. Motor deficit

	None	Mild 4/5	Moderat 2-3/5	Severe <2/5
• Facial palsy/ atrophy	☐	☐	☐	☐
• Proximal UL	☐	☐	☐	☐
• Distal UL	☐	☐	☐	☐
• Proximal LL	☐	☐	☐	☐
• Distal LL	☐	☐	☐	☐

3. Muscle wasting

	None	Mild	Moderate	Severe
• Proximal UL	☐	☐	☐	☐
• Distal UL	☐	☐	☐	☐
• Proximal LL	☐	☐	☐	☐
• Distal LL	☐	☐	☐	☐

4. Fasciculations or Myokymias (Facial contraction fasciculations) please circle

	No	Yes	Localisation:
	☐	☐	_____

5. Sensory deficit

	None	Mild	Moderate	Severe	Abolished
• Vibration sense ↓ (ankles)	☐	☐	☐	☐	☐
	(8/8)	(> 5/8)	(2-5/8)	(<2/8)	(0/8)

	No	Touch	Prick	Cold
• Superficial sensory loss	☐	☐	☐	☐

6. Skeletal abnormalities

	None	Mild	Moderate	Severe
• Scoliosis	☐	☐	☐	☐
• Pes cavus	☐	☐	☐	☐
• Other: _____	☐	☐	☐	☐

7. Facial dysmorphia

	No	Yes	Describe:
	☐	☐	_____

8. Sphincter and sexual disturbances

	None	Mild	Moderate	Severe
• Urinary urgency	☐	☐	☐	☐
• Urinary incontinence	☐	☐	☐	☐
• Urinary retention	☐	☐	☐	☐
• Anal incontinence	☐	☐	☐	☐
• Impaired sexual function	☐	☐	☐	☐
• Early menopause	☐ No	☐ Yes	At age:_____	

9. Extra-pyramidal symptoms

	None	Mild	Moderate	Severe	Specify:
• Resting tremor	☐	☐	☐	☐	
• Postural tremor	☐	☐	☐	☐	
• Chorea	☐	☐	☐	☐	
• Dystonia	☐	☐	☐	☐	
• Myoclonus	☐	☐	☐	☐	
• Hypokinesia	☐	☐	☐	☐	
• Rigidity	☐	☐	☐	☐	

10. Ophtalmological signs

	No	Yes	
• Diplopia	☐	☐	
• Ptosis	☐	☐	
• Eye lid retraction (bulging eyes)	☐	☐	
• Diminished visual acuity	☐	☐	**At age:** _____
**** Fundus***	**No**	**Yes**	
• Abnormal	☐	☐	☐ Optic atrophy ☐ *Retinis Pigmentosa* ☐ Other:

**** Oculomotor***	**No**	**Yes**	
• Nystagmus	☐	☐	**Describe:** _____
• Saccadic pursuit	☐	☐	**Describe:** _____
• Slow saccades	☐	☐	
• Ocular motor apraxia	☐	☐	
• Vertical ophthalmoplegia	☐	☐	**Describe:** _____
• Horizontal ophthalmoplegia	☐	☐	**Describe:** _____

11. Mental status

	No	Yes	
• Intellectual deterioration	☐	☐	**At age:** _____ **Type:**_____
• Mental retardation	☐	☐	**At age:** _____ **Type:**_____
• Psychiatric symptoms	☐	☐	**Describe:** _____

12. Other signs

	No	Yes	Describe
• Dysphagia	☐	☐	_____ **Severity:** _____
• Skin problems	☐	☐	_____
• Hearing impairment	☐	☐	_____
• Epilepsy	☐	☐	**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
- **SARA** (annex 2) : _____/ 40
- **CCFS** (annex 3) : _____
- **25 feet ambulatory test** (annex 4): _____sec
- **UHDRS** – functional part IV (annex 5): _____/ 25

H- CLINICAL DIAGNOSTIC CONCLUSION

Cerebellar ataxia

☐ Autosomal dominant	☐ Pure form	☐ Definitely affected
☐ Autosomal recessive	☐ Complicated form	☐ Probably affected (only dysarthria)
☐ Isolated case		☐ Possibly affected (only mild gait ataxia)
☐ X-linked		

Spastic paraparesis

☐ Autosomal dominant	☐ Pure form	☐ Definitely affected
☐ Autosomal recessive	☐ Complicated form	☐ Probably affected (enhanced or very brisk LL reflexes +/- Babinski)
☐ Isolated case		☐ Possibly affected (enhanced LL reflexes)
☐ X-linked		

I- MOLECULAR DIAGNOSIS

Genes/Loci to test:	Diagnosis:

J- COMPLEMENTARY INVESTIGATIONS

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
1. Cerebral MRI	☐	☐	☐	
				None Mild Moder. Severe
- Cerebrum				☐ ☐ ☐ ☐
- Cerebellum				☐ ☐ ☐ ☐
- Brainstem				☐ ☐ ☐ ☐
- Corpus callosum				☐ ☐ ☐ ☐

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
2. Medullar MRI	☐	☐	☐	
				ATROPHY
				None Mild Moder. Severe
- Upper spinal cord				☐ ☐ ☐ ☐

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
3. EMG + NCV UL	☐	☐	☐	
4. EMG + NCV LL	☐	☐	☐	
5. VEP	☐	☐	☐	
6. AEP	☐	☐	☐	
7. MEP	☐	☐	☐	
8. SEP	☐	☐	☐	

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
9. VLCFA	☐	☐	☐	
10. α-foetoprotein	☐	☐	☐	
11. Cholesterol	☐	☐	☐	
12. Serum protein electrophoresis	☐	☐	☐	
13. Vitamin E	☐	☐	☐	
14. Apolipo-protein A, B	☐	☐	☐	

EXAMINATION	NOT DONE	NOR-MAL	AB-NORMAL	SPECIFY
15. Muscle biopsy	☐	☐	☐	
16. Skin biopsy	☐	☐	☐	
17. ERG	☐	☐	☐	
18. Fundus examination	☐	☐	☐	
19. Neuropsycho-logical exam	☐	☐	☐	
- IQ				
20. Urodynamics	☐	☐	☐	
21. Urine density	☐	☐	☐	

K- STORED MATERIAL

	Yes	No
• DNA	☐	☐
• Immortalized cell lines	☐	☐
• Muscle tissue	☐	☐
• Skin biopsy	☐	☐
• Nerve biopsy	☐	☐
• Other: _____	☐	☐