

CAN HSP Questionnaire

1 of 5

CAN HSP ID *For the use of the research team*

ROU # *For the use of the research team*

Evaluation Date _____ / _____ / _____ (m/d/y) Age at evaluation _____
 Date sample sent _____ / _____ / _____ (m/d/y) Consent signed Yes No

A. PATIENT INFORMATION

Clinical Status Affected
 Not affected
 Possibly affected
 Gender Male Female
 Date of birth _____ / _____ / _____ (m/d/y)

B. FAMILY HISTORY (please include a pedigree)

Adopted? Yes No Consanguinity Yes No
 Impression of inheritance: Recessive X-linked Sporadic
 Dominant Mitochondrial Unknown
 Ancestral background (list all that apply) Maternal branch: _____
 Paternal branch: _____
 Ethno-religious background Ashkenazi Jewish Sephardic Jewish
 Amish / Mennonite Mormon
 Hutterite Other:
 No particular affiliation

C. CLINICAL INFORMATION

Developmental History
 Motor delay Yes No Unknown
 Achieved walking Age: _____ Never acquired Unknown
 Speech delay or abnormality Yes No Unknown
 Achieved speech Age: _____ Never acquired Unknown
 Learning disability Yes No Unknown
 Highest level of education (answer one or both) Number of years of education : _____
 Diploma / degree acquired: _____

Core symptoms / signs

Age of onset _____
 Lower Extremity weakness Yes No Unknown Extensor plantar responses Yes No Unknown
 Lower Extremity spasticity Yes No Unknown Abnormal bladder function Yes No Unknown
 Lower Extremity hyperreflexia Yes No Unknown Ankle clonus Yes No Unknown

Severity assessment

Walking aid Yes No Unknown
 Cane Age used cane : _____ Unknown Never
 Walker Age used walker : _____ Unknown Never
 Wheelchair Age used wheelchair : _____ Unknown Never

SPATAX-EUROSPA disability stage

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

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CAN HSP ID :

C. CLINICAL INFORMATION (cont'd)**Other associate symptoms/signs**

| | | | |
|--|--|---|--|
| Progressive cognitive deficits | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Retinopathy or optic atrophy | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Ocular movement abnormalities | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Deafness | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Jaw jerk | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Swallowing difficulty | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Dysarthria | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Upper extremity weakness | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Upper extremity hyperreflexia | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Myotrophy or lower motor neuron features | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Sensory abnormalities | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| - If sensory abnormalities : | <input type="checkbox"/> Mild vibratory loss | <input type="checkbox"/> Severe vibratory and proprioceptive loss | <input type="checkbox"/> loss of pain and/or temperature sensation |
| Peripheral neuropathy | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Pes cavus | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Ataxic gait | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Upper extremity ataxia | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Upper extremity intent tremor | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Lower extremity ataxia | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Lower extremity intent tremor | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Postural / action tremor | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Seizures | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Skeletal abnormalities | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Myoclonus | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |

Other comments / seemingly-related symptom(s):

D. INVESTIGATIONS

| | | | |
|--|---|--|--|
| Total spine MRI | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |
| Brain MRI | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |
| - If abnormal total spine or brain MRI: | | | |
| Thin corpus callosum | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Hydrocephalus | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Cerebellar atrophy | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Cerebral atrophy | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Leukodystrophy | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Leukoencephalopathy | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| EMG | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |
| - If abnormal EMG, changes are characterized as: | <input type="checkbox"/> Axonal / Demyelinating | <input type="checkbox"/> Sensory / Motor | |

Other pertinent findings:

E. BIOCHEMISTRY :

| | | | |
|----------------|---------------------------------|-----------------------------------|--|
| Vitamin B12 | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |
| Vitamin E | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |
| VLCFA | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |
| Phytanic acid | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |
| Pipecolic acid | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed |

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: ≥ 5 s) sec
- 2: Moderately reduced (10m: ≥ 10 s)
- 3: Severely reduced (10m: ≥ 20 s)
- 4: Unable to walk for a 10m distance or time ≥ 40 s

| |
|-----------------------------|
| 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 stopwatch*

- 0: Normal
- 1: Slightly reduced (≥ 5 s to perform task) sec
- 2: Moderately reduced (≥ 10 s to perform task)
- 3: Severely reduced (≥ 20 s 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
- 3: of motion
- 4: Considerable increase in muscle tone - passive movement is difficult
- 5: 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

CAN HSP ID :

F. GENETIC TESTING

Has genetic testing been performed?

Yes No Unknown

Check all that have been tested detail result

(N:normal; Mut:mutation; VUS: variant of unknown significance)

| | | | | | | | |
|--|----------------------------|------------------------------|------------------------------|--|----------------------------|------------------------------|------------------------------|
| <input type="checkbox"/> SPG 1 (L1CAM) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 27 (SPG27) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 2 (PLP1) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 28 (SPG28) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 3A (ATL1) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 29 (SPG29) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 4 (SPAST) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 30 (SPG30) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 5A (CYP7B1) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 31 (REEP1) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 6 (NIPA1) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 32 (SPG32) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 7 (CMAR) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 33 (ZFYVE27) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 8 (KIAA0196) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 34 (SPG34) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 9 (SPG9) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 35 (FA2H) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 10 (KIF5A) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 36 (SPG36) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 11 (K1AA1840) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 37 (SPG37) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 12 (RTN2) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 38 (SPG38) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 13 (HSPD1) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 39 (PNPLA6) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 14 (SPG14) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 41 (SPG41) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 15 (ZFYVE26) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 42 (SCL33A1) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 16 (SPG16) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 43 (SPG43) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 17 (BSCL2) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 44 (GJA12/GJC2) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 18 (SPG18) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 45 (SPG45) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 19 (SPG19) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 46 (SPG46) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 20 (SPG20) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 47 (SPG47) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 21 (ACP33) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SPG 48 (KIAA0415) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 22 (MCT8) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> FRDA (FXN) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 23 (SPG23) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> SCA1-8 | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 24 (SPG24) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> ARSACS (SACS) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 25 (SPG25) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> Other: _____ | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |
| <input type="checkbox"/> SPG 26 (SPG26) | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS | <input type="checkbox"/> Other: _____ | <input type="checkbox"/> N | <input type="checkbox"/> Mut | <input type="checkbox"/> VUS |

G. OTHER COMMENTS

Paternal ethnic background: _____ Maternal ethnic background: _____

| |
|--|
| |
| |
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| |
| |
| |
| |

Instructions:

1. Take a 3-generation pedigree
2. Clearly indicate the proband by an arrow
3. Indicate all individuals for whom a sample has been included into the CANHSP project, with their *individual #* on the pedigree, below their symbol
4. Mark the affected individuals according to the legend
5. Indicate maternal and paternal ancestry
6. Fill out all pertinent fields
7. Ensure that the pedigree is de-identified
8. Send the pedigree with the sample
9. If an additional person is subsequently recruited, please resend the pedigree with the additional individual #

CAN HSP#:

Site ## - Fam ### - Individual ##

Date pedigree taken:

Month / Day / Year

Date pedigree updated:

Month / Day / Year

Other notes:

Legend and Standardized Pedigree Symbols

| | | |
|--------------------|--|-------------------------------|
| Male | Female | DNA collected IND## |
| Deceased d. age | Adopted in | Proband |
| Mutation carrier | Asymptomatic/presymptomatic mutation carrier | Consanguineous union |

Clinical status

| | |
|--|---|
| Examined : E+ Dx of spastic paraplegia | Examined : E- No spastic paraplegia |
| Spastic paraplegia (by hearsay) | Examined : E? Possible spastic paraplegia |