

## Supplementary Material Study III

### Reliability and validity of the Dyskinesia Impairment Scale in children and young adults with inherited or idiopathic dystonia

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## Supplementary material A

### Dyskinesia Impairment Scale, video protocol

A: General view	1. entering the room (walking or with wheelchair)	F - general view
B: Sitting - comfort position	2. sitting in rest (chair or wheelchair) 3. sitting in rest (chair or wheelchair) 4. eyes tracking movement 5. eyes blinking (10x) 6. opening and closing mouth (10x) 7. speech -interview <sup>a</sup> 8. turn head to right then to left (5x) 9. lateroflexion of the head to the left (5x) and right (5x) (=bring your ear to your shoulder on each side) 10. elevate arms sideways (5x) 11. alternating heel to toe taps right (5x) 12. alternating heel to toe taps left (5x) 13. take a cup with your right hand on the left corner and bring it to the right corner of the table and bring it back to the left corner of the table (2x) 14. take a cup with your left hand on the right corner and bring it to the left corner of the table and bring it back to the right corner of the table (2x) 15. take a pen with your right hand on the left corner and bring it to the right corner of the table and bring it back to the left corner of the table (2x) 16. take a pen with your left hand on the right corner and bring it to the left corner of the table and bring it back to the right corner of the table (2x)	F - general view F - close-up F - close-up F - close-up F - close-up F - bust F - bust F - bust F - general view F - close-up lower limbs F - close-up lower limbs F - general view F - general view F - general view F - general view
C: Sitting - active position	17. active sitting on a bench 18. bend trunk forwards and back (5x)	F - general view P - general view P - general view
D: Lying position	19. lying on a mat in rest 20. grasping/reaching a pen with right arm from lying position (4x: aside / over the head / over the midline / to the limbs) 21. grasping/reaching a pen with left arm from lying position (4x: aside / over the head / over the midline / to the limbs) 22. rolling over right 23. rolling over left	F - general view F - general view F - general view F - general view
E: Standing position	24. stand upright 25. stand upright 26. stand upright	F - general view P - general view right P - general view left

Abbreviations: F=frontal view; P=profile view  
Duration of recording for each task is 30 seconds

Practice trial(1x)

<sup>a</sup>Speech interview (standard questions) - e.g. - What is your name? How do you come to school? By car or by bus? Is your teacher a man male or female? Do you sleep at school? On which days?

## Dyskinesia Impairment Scale, scoring protocol

The Dyskinesia Impairment Scale is developed to evaluate dystonia and choreoathetosis in dyskinetic cerebral palsy.<sup>1</sup> The DIS consists of dystonia (DIS-D) and choreoathetosis (DIS-CA) subscales discriminating the presence and rating the severity (amplitude and duration) of either movement disorder in twelve body regions during activity and rest.

### Definitions:

**Definition Dyskinetic Cerebral Palsy (CP):** Dyskinetic CP is characterized by involuntary, uncontrolled, recurring occasionally stereotyped movements, where the primitive reflex patterns predominate, and the muscle tone is varying. Two movement disorder patterns are dominantly present: dystonia and choreoathetosis

**Dystonia in CP** refers to abnormal postures and/or involuntary or distorted voluntary twisting and repetitive movements due to sustained or intermittent muscle contractions.

**Choreoathetosis in CP** is dominated by hyperkinesia and tone fluctuating (but mainly decreased). It can be differentiated in chorea, i.e. rapid involuntary, jerky and often fragmented movements, and athetosis, i.e. slower, constantly changing, writhing or contorting movements.

### Region descriptions in the Dyskinesia Impairment Scale:

Region	DYSTONIA	CHOREOATHETOSIS
Eye	Dystonia around the eyes, eyelids, eyebrow, forehead: e.g. sustained muscle contractions (blepharospasms) around the eyes and/or the eyelid (open/closed) and/or forced eye movement deviations for example during eye tracking movement of fixation.	Choreoathetosis around the eyes, eyelids, eyebrows, forehead: e.g. constantly, fragmented movements around the eyes and/or blinking eyelid (open/closed) and/or variable (saccadic) eye movements for example during eye tracking movement of fixation.
Mouth	Dystonia around the lips, jaw, cheeks, tongue: e.g. sustained muscle contraction resulting in grimacing movement, clenched or deviated jaw, forced open mouth and/or forceful tongue thrusting.	Choreoathetosis lips, jaw, cheeks; tongue: e.g. constantly changing, fragmented movements in the lower face like grimacing, mouth movements and tongue protrusion movements.
Neck	Dystonia in the neck: sustained muscle contraction resulting in pulling neck movements and/or posture into any plane of motion: extension, flexion, lateral flexion, rotation.	Choreoathetosis in the neck: e.g., constantly changing fragmented or contorting neck movements (wagging) into any plane of motion: extension, flexion, lateral flexion, rotation.
Trunk	Dystonia in the trunk: e.g. sustained muscle contraction resulting in pulling trunk movements and/or posture into any plane of motion: extension, flexion, lateral flexion, rotation.	Choreoathetosis in the trunk: e.g. constantly changing fragmented or contorting trunk movements (wagging) into any plane of motion: extension, flexion, lateral flexion, rotation.
Arm Proximal	Dystonia in the shoulder girdle, upper arm, elbow: e.g. sustained muscle contractions causing abnormal posturing, involuntary and/or distorted voluntary movements of the proximal arm.	Choreoathetosis in the shoulder girdle, upper arm, elbow: e.g. constantly changing fragmented or contorting movements of the proximal arm: jerky, stormy (chorea) and/or wriggling, contorting (athetosis).
Arm Distal	Dystonia in the forearm, wrist, hand: e.g. sustained muscle contractions causing abnormal posturing, involuntary and/or distorted voluntary movements of the distal arm.	Choreoathetosis in the forearm, wrist, hand: e.g. constantly changing fragmented or contorting neck movements of the distal arm: jerky, stormy (chorea) and/or wriggling, contorting (athetosis).
Leg Proximal	Dystonia in the hip girdle, upper leg, knee: e.g. sustained muscle contractions causing abnormal posturing and/or distorted voluntary movements of the proximal leg.	Choreoathetosis in the hip girdle, upper leg, knee: e.g. constantly changing fragmented or contorting movements of the proximal leg: jerky, stormy (chorea) and/or wriggling, contorting (athetosis).
Leg Distal	Dystonia in the lower leg, ankle, foot: e.g. sustained muscle contractions causing abnormal posturing and/or distorted voluntary movements of the distal leg.	Choreoathetosis in the lower leg, ankle, foot: e.g. constantly changing fragmented or contorting movements of the distal leg: jerky, stormy (chorea) and/or wriggling, contorting (athetosis).

Duration factor	Amplitude factor
0 =D/CA is absent	0 =D/CA is absent
1 =D/CA is occasionally present ≤10%	1 =D/CA in small range of motion ≤10%
2 =D/CA is frequently present ≥10%<50%	2 =D/CA in moderate range of motion ≥10%<50%
3 =D/CA is mostly present ≥50%<90%	3 =D/CA in submaximal range of motion ≥50%<90%
4 =D/CA is always present ≥90%	4 =D/CA in maximal range of motion ≥90%

Region	Action			Rest		
	Single activity	Duration factor	Amplitude factor	Single position	Duration factor	Amplitude factor
Eye Dystonia	eye tracking eye blinking	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
Eye Choreo-athetosis	eye tracking eye blinking	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
Mouth Dystonia	mouth open/close speech	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
Mouth Choreo-athetosis	mouth open/close speech	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
Neck Dystonia	lateroflexion right/left rotation left/right	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
Neck Choreo-athetosis	lateroflexion right/left rotation left/right	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
Trunk Dystonia	active sitting position forward flexion	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
Trunk Choreo-athetosis	active sitting position forward flexion	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
R Arm prox Dystonia	arm abduction grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
R Arm prox Choreo-athetosis	arm abduction grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
L Arm prox Dystonia	arm abduction grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
L Arm prox Choreo-athetosis	arm abduction grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4

Region	Action			Rest		
	Single Activity	Duration factor	Amplitude factor	Single Position	Duration factor	Amplitude factor
R Arm dist Dystonia	grasp and move a cup grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
R Arm dist Choreo-athetosis	grasp and move a cup grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
L Arm dist Dystonia	grasp and move a cup grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
L Arm dist Choreo-athetosis	grasp and move a cup grasp and move a pen	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	sitting position	0-1-2-3-4	0-1-2-3-4
R leg prox Dystonia	rolling standing	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4
R leg prox Choreo-athetosis	rolling standing	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4
L leg prox Dystonia	rolling standing	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4
L leg prox Choreo-athetosis	rolling standing	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4
R leg dist Dystonia	rolling heel/toe raising	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4
R leg dist Choreo-athetosis	rolling heel/toe raising	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4
L leg dist Dystonia	rolling heel/toe raising	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4
L leg dist Choreo-athetosis	rolling heel/toe raising	0-1-2-3-4 0-1-2-3-4	0-1-2-3-4 0-1-2-3-4	lying position	0-1-2-3-4	0-1-2-3-4

Abbreviations score form: D, dystonia; CA, choreoathetosis; R, right; L, left; prox, proximal; dist, distal

## Supplementary material B

### Burke-Fahn-Marsden Dystonia Rating Scale, Movement scale, scoring protocol

Region	Provoking factor	X	Severity factor	Weight	Product
Eyes	0-4	x	0-4	0.5	0-8
Mouth	0-4	x	0-4	0.5	0-8
Speech/swallowing	0-4	x	0-4	1.0	0-16
Neck	0-4	x	0-4	0.5	0-8
R arm	0-4	x	0-4	1.0	0-16
L arm	0-4	x	0-4	1.0	0-16
Trunk	0-4	x	0-4	1.0	0-16
R leg	0-4	x	0-4	1.0	0-16
L leg	0-4	x	0-4	1.0	0-16
Sum					0-120

#### I. Provoking factor

##### A. general

- 0 – no dystonia
- 1 – dystonia on particular action
- 2 – dystonia on many actions
- 3 – dystonia on distant part of body or intermittently at rest
- 4 – dystonia present at rest

##### B. speech and swallowing

- 1 – occasional, either or both
- 2 - frequent either
- 3 – frequent one and occasional other
- 4 – frequent both

#### II. Severity factor

##### Eyes

- 0 – no dystonia
- 1 – slight, occasionally blinking
- 2 – mild, frequently blinking
- 3 – moderate, prolonged spasms of eyelid closure, but eyes open most of the time
- 4 – severe, prolonged spasms of eyelid closure, with eyes closed at least 30% of the time

## Mouth

0 - no dystonia

1 – slight, occasionally grimacing or other mouth movements (eg jaw open or clenched)

2 – mild, movement present less than 50% of the time

3 – moderate, dystonic movements or contractions present most of the time

4 – severe dystonic movements or contractions present most of the time

## Speech and swallowing

0 – normal

1 – slightly involved; speech easily understood or occasionally choking

2 – some difficulty in understanding speech or frequent choking

3 – marked difficulty in understanding speech or inability to swallow firm foods

4 – complete or almost complete anarthria or marked difficulty swallowing soft foods and liquid

## Neck

0 – no dystonia present

1 – slight, occasionally pulling

2 – obvious torticollis, but mild

3 – moderate pulling

4 – extreme pulling

## Arm

0 – no dystonia present

1 – slight dystonia, clinically insignificant

2 – mild, obvious dystonia but not disabling

3 – moderate, able to grasp, with some manual function

4 – severe, no useful grasp

## Trunk

0 – no dystonia present

1 – slight bending, clinically insignificant

2 – definite bending, but not interfering with standing or walking

3 – moderate bending interfering with standing or walking

4 – extreme bending of trunk preventing standing or walking

## Leg

0 – no dystonia present

1 – slight dystonia, but not causing impairment clinically insignificant

2 – mild dystonia, walks briskly and unaided

3 – moderate dystonia, severely impairs walking or requires assistance

4 – severe, unable to stand or walk on involved leg

## Supplementary material C

**Table S1;** Patient characteristics

Patient number	Sex	Age at visit (y, m)	Age at onset (y, m)	GMFCS-ER	MACS
1	M	16 y 8 m	6 m	V	V
2	F	19 y 8 m	6 m	IV	III
3	F	18 y 8 m	5 y	II	II
4	M	21 y 7 m	4 m	V	V
5	F	8 y 9 m	6 m	I	I
6	F	19 y 10 m	5 m	IV	V
7	M	16 y 6 m	2 y	II	II
8	F	14 y 9 m	9 y	I	I
9	M	6 y 5 m	5 y 5 m	II	II
10	M	12 y 8 m	6 y 5 m	II	II
11	M	15 y 10 m	1 y	III	III
12	F	11 y 12 m	9 m	V	V
13	M	23 y 7 m	6 y	III	III
14	F	22 y 4 m	11 y	V	V
15	M	17 y 3 m	3 m	V	V
16	M	8 y 1 m	6 m	IV	V
17	F	24 y 1 m	15 y	I	I
18	M	10 y 8 m	1 y 6 m	IV	V
19	M	18 y 4 m	3 m	IV	IV
20	F	15 y 10 m	3 y	III	IV

M=male; F=female; y=years; m=months; GMFCS-ER=gross motor function classification system-expanded and revised; MACS>manual ability classification system

**Table S2;** Genetic information

Patient number	Sex	Diagnosis, clinical or genetically confirmed	Genetic tests performed in participants with idiopathic dystonia
1	M	Idiopathic dystonia	Karyotyping, <i>PANK2</i> gene Last testing: 2019
2	F	Idiopathic dystonia	Dystonia panel Last testing: 2019
3	F	Idiopathic dystonia	Dystonia panel Last testing: 2019
4	M	Idiopathic dystonia	Micro array, dystonia panel Last testing: 2019
5	F	Benign hereditary chorea <i>TTF-1/Nkx2.1</i> gene c.673delG p.Ala225ProfsX3	/
6	F	Idiopathic dystonia	Micro array
7	M	Idiopathic dystonia	Analysis of mitochondrial DNA 2005, <i>SURF1</i> gene 2005, <i>SLC19A3</i> gene 2009, WES with analysis of metabolic and mitochondrial panels 2014
8	F	DYT-THAP1 dystonia <i>THAP1</i> gene c.377_378delCT p.Pro126Argfs*71	/
9	M	Rapid onset parkinsonism dystonia <i>ATPIA3</i> gene c.2351C>T p.Thr784Ile	/
10	M	Idiopathic dystonia	Micro array and <i>FMRI</i> gene 2018, WES with analysis of dystonia, intellectual disability and neuromuscular panels 2019
11	M	Glutaric aciduria type 1 <i>GCDH</i> gene c.892G>A p.Ala298Thr	/
12	F	Glutaric aciduria type 1 <i>GCDH</i> gene c.892G>A p.Ala298Thr	/
13	M	DYT-THAP1-dystonia <i>THAP1</i> gene c.173T>C p.Phe58Ser	/
14	F	Rapid onset parkinsonism dystonia <i>ATPIA3</i> gene c.1838C>T p.Thr613Met	/
15	M	Lesch-Nyhans syndrome <i>HPRT</i> gene del exon 6-9	/
16	M	Hypomyelination with atrophy of the basal ganglia and cerebellum <i>TUBB4A</i> gene c.745G>A p.Asp249Asn	/
17	F	Idiopathic dystonia	No genetic testing performed
18	M	Spastic paraplegia type 49/56, with primarily dystonic features <i>CYP2U1</i> gene c.947A>T p.Asp316Val	/
19	M	Lesch-Nyhans syndrome <i>HPRT</i> gene del exon 1	/
20	F	Leigh syndrome <i>SURF1</i> gene c.703A>G p.Met235Val c.312-321delTCTGCCAGCCinsAT p.Pro104Argfs*2	/

M=male; F=female; WES=whole exome sequencing; \*=stop codon