

CMT-FOM

Charcot-Marie-Tooth disease Functional Outcome Measure (CMT-FOM)

Equipment and training resource manual

ACT-CMT Study Group | Melissa R. Mandarakas, Kate Eichinger, Paula Bray, Kayla M.D. Cornett, Michael E. Shy, Mary M. Reilly, Gita Ramdharry, Steven S. Scherer, Timothy Estilow, Davide Pareyson, Marnee J. McKay, David N. Herrmann, Joshua Burns

CMT-FOM

Contents	Page
CMT-FOM Data Collection Form	3
List of Equipment	4
General Instructions	5
Item Instructions	6
1. Hand Grip Strength	6
2. Foot Plantarflexion Strength	7
3. Foot Dorsiflexion Strength	8
4. Functional Dexterity Test	9
5. Nine-Hole Peg Test	10
6. Stair climb	11
7. 30-second Chair Stand Test	12
8. Standing with feet apart on a line with eyes open	14
9. Standing with feet apart on a line with eyes closed	15
10. Single leg stance with eyes closed	16
11. Timed Up and Go	17
12. Six-Minute Walk Test	18
Patient profile	19
Calibration tasks	27
References	28

CMT-FOM

Initial Evaluation ☐ Re-Evaluation ☐ Date: _____ Time: _____ Evaluator: _____

Patient Profile											
ID:			D.O.B:		Age (yrs):		Sex: <input type="checkbox"/> Male <input type="checkbox"/> Female				
Height (m):		Weight (kg):		Dominant Side: L <input type="checkbox"/> R <input type="checkbox"/>		Diagnosis:					
Ankle-Foot Orthoses (AFOs): <input type="checkbox"/> Yes <input type="checkbox"/> No Type: <input type="checkbox"/> Solid <input type="checkbox"/> Hinged <input type="checkbox"/> Leaf Spring <input type="checkbox"/> Carbon <input type="checkbox"/> Other:						Footwear: <input type="checkbox"/> Barefoot <input type="checkbox"/> Walking shoes <input type="checkbox"/> Study shoes <input type="checkbox"/> Other:					
Other Assistive devices: <input type="checkbox"/> Yes <input type="checkbox"/> No Required for testing: <input type="checkbox"/> Cane <input type="checkbox"/> Walker <input type="checkbox"/> Walking stick <input type="checkbox"/> Wheelchair <input type="checkbox"/> Other:											
Symptoms:	Foot pain <input type="checkbox"/>		Leg cramps <input type="checkbox"/>		Unsteady ankles <input type="checkbox"/>		Daily trips and/or falls <input type="checkbox"/>		Hand pain <input type="checkbox"/>		
	Hand weakness <input type="checkbox"/>		Hand tremor <input type="checkbox"/>		Sensory symptoms (e.g., pins and needles, tingling, numbness, prickling) <input type="checkbox"/>						
Lunge test (degrees)						Left:		Right:			
Foot Posture Index	Talar head palpation										
	Curves above and below lateral malleolus										
	Inversion/eversion of the calcaneus										
	Bulge in the region of the talonavicular joint										
	Congruence of the medial longitudinal arch										
	Abd/adduction of forefoot on rearfoot (too-many-toes)										
	Total (-12 to 12)										
Gait	Foot drop: No <input type="checkbox"/> Some <input type="checkbox"/> Yes <input type="checkbox"/>				Difficulty heel walking: No <input type="checkbox"/> Some <input type="checkbox"/> Yes <input type="checkbox"/>		Difficulty toe walking: No <input type="checkbox"/> Some <input type="checkbox"/> Yes <input type="checkbox"/>				
Sensation	0	1	2	3	4	Score					
Pinprick	Normal	Decreased below or at ankle bones	Decreased at or below midline of calf	Decreased above calf midline up to and including knee	Decreased above knee (above top of patella)						
Vibration	Normal	Reduced at first metatarsal bone	Reduced at ankle	Reduced at knee (tibial tuberosity)	Absent at knee and ankle						
Strength		Trial 1		Trial 2		Trial 3		Average			
1. Hand grip (N)								x2:			
2. Foot plantarflexion (N)											
3. Foot dorsiflexion (N)											
Hand Dexterity											
4. Functional Dexterity Test (sec)						5. Nine-Hole Peg Test (sec)					
Lower Extremity Function		Assistive device required:				<input type="checkbox"/> No <input type="checkbox"/> Yes, Describe:					
6. Stair climb (sec)		Handrail used? <input type="checkbox"/> Yes <input type="checkbox"/> No				7. 30-second Chair Stand Test (#)					
Balance		Assistive device required:		<input type="checkbox"/> No <input type="checkbox"/> Yes, Describe:							
8. Stance with feet apart on line with eyes open (10 sec)		T1:		9. Stance with feet apart on line with eyes closed (20 sec)		T1:		10. Single leg stance with eyes closed (20 sec)		T1:	
		T2:				T2:				T2:	
Mobility		Assistive device required:				<input type="checkbox"/> No <input type="checkbox"/> Yes, Describe:					
11. Timed Up and Go (sec)						12. Six-Minute Walk Test (m)					
Total Score (0-100)											

CMT-FOM Equipment

List of Equipment

1. **Hand Grip Strength:** Citec hand-hand dynamometer with grip strength applicator, spare batteries (6V) C.I.T. Technics, Haren, The Netherlands: www.citec.nu
2. **Foot Plantarflexion strength:** Citec hand-hand dynamometer incl. Grip Strength Applicator, spare batteries (6V)
3. **Foot Dorsiflexion Strength:** Citec hand-hand dynamometer incl. Grip Strength Applicator, spare batteries (6V)
4. **Functional Dexterity Test:** Functional Dexterity Test (FDT) and digital stopwatch
5. **Nine-Hole Peg Test:** Rolyan® 9-Hole Peg Test Kit and digital stopwatch
6. **Stair Climb:** Free standing: 6" (15 cm) high, 10" (25 cm) deep, with a 24" (61 cm) x 30" (76 cm) platform and digital stopwatch
7. **30-Second Chair Stand Test:** Height adjustable bench, <http://kayeproducts.com/kaye-adjustable-benches/> and Digital stopwatch
8. **Standing with feet apart on a line with eyes open:** Digital stopwatch and tape to mark a line (or designate a line on the flooring)
9. **Standing with feet apart on a line with eyes closed:** Digital stopwatch and tape to mark a line (or designate a line on the flooring)
10. **Single leg stance with eyes closed:** Digital stopwatch and tape to mark a line (or designate a line on the flooring)
11. **Timed Up and Go:** Chair with armrests approximately 46 cm in height and a stopwatch
12. **Six-minute Walk Test:** Two traffic cones, lap counter, stopwatch, tape measure, 30-meter course

Other equipment for Patient Profile

- **Neurotips™** (100): Owen Mumford Ltd, Oxford, UK: www.owenmumford.com
- **Inclinometer** Baseline Digital Inclinometer: <http://www.bpp2.com/> or iHandy Level App (be sure to calibrate before use)
- **Tuning Fork** Rydel Seiffer tuning fork, C 64 Hz / c 128 Hz detachable clamps Arno Barthelmes & Co. GmbH - Tuttlingen - Germany: www.barthelmes.info

General Instructions

- Participants should be asked to wear comfortable clothing and walking shoes. If they wear AFOs, they should be instructed to bring shoes that they can wear without their AFOs.
- Timed items should be recorded to the nearest tenth of a second (1 decimal point).
- Lower extremity function, balance and mobility items of the CMT-FOM (Stair Climb, 30-second Chair Stand Test, Timed Up and Go, Standing with feet apart on a line with eyes open, Standing with feet apart on a line with eyes closed, Single leg stance with eyes close, Six-Minute Walk Test) should be performed in the participant's own walking shoes.
 - No ankle-foot orthoses (AFOs) or other assistive devices such as crutches/walking stick/walker should be used during assessment of these items.
 - If AFOs or assistive devices are absolutely required for any of the items, assign the raw values below so a CMT-FOM total score can still be calculated, and the item does not need to be performed.
 - If an inappropriate style of shoe is worn (e.g., flip-flops or high heels) or the participant's walking shoes do not fit without AFOs, barefoot is permitted if safe to do so and institutionally permissible.
- If the participant is unable to perform an item due to disease severity, assign the following raw values so a CMT-FOM total score can still be calculated:
 - 150 for the following items: Functional Dexterity Test, Nine-Hole Peg Test, Stair Climb, Timed Up and Go.
 - 0 for the following items: Hand Grip, Foot Plantarflexion and Foot Dorsiflexion, 30-second Chair Stand Test, Standing with feet apart on a line with eyes open, Standing with feet apart on a line with eyes closed, Single leg stance with eyes closed, Six-Minute Walk Test.
- The CMT-FOM can be collected in approximately 30 min, with training resources and scoring against normative reference values^{1,2} available online via ClinicalOutcomeMeasures.org.

Strength Assessment Instructions

- Hand Grip, Foot Plantarflexion and Foot Dorsiflexion strength of the dominant side is assessed using a Citec hand-held dynamometer (HHD). The Citec HHD should be calibrated before use following the procedures found at the end of this manual.
- The maximum force that can be applied to the Citec HHD is 500 Newtons.
- Perform up to 5 trials and record 3 valid trials which should reflect maximum strength.
- A 10 second rest is given between trials.

1. Grip strength

Background/Purpose: Quantitative muscle testing of hand grip strength using standardized procedures has demonstrated high reliability and validity in adults and children.^{3,4} Grip strength is quantified using the Citec hand-held dynamometer with the grip applicator attached (Citec dynamometer CT 3001, CIT Technics, Groningen, the Netherlands). Please note, the grip applicator must be screwed on all the way, this may result in the visual display facing in different directions.

Assessment Position: The participant is seated comfortably in a chair with their feet supported. The arm being assessed is positioned with the shoulder adducted and in neutral rotation. The elbow is in 90° of flexion, forearm in neutral, and the wrist in 0 to 30° of extension and 0 to 15° of ulnar deviation.⁵

Assessment Procedure: The participant is asked to grasp the Citec hand-held dynamometer with the fingers wrapped around the handle. The clinical evaluator instructs the participant to perform a maximal contraction lasting 3-5 seconds. The dominant hand is assessed and determined as the hand used to write.



The clinical evaluator says:

"I am now going to measure the strength in your hand. When I say go I want you to squeeze the handle as hard as you can. Ready, set, go."

Scoring: Three (3) valid measures of the dominant hand are recorded to the nearest Newton. In accordance with the Citec hand-held dynamometer manufacturer's instructions, the displayed grip values must be multiplied by 2, as the grip applicator of the citec measures strength in a 1:2 ratio.

For those unable to perform grip strength due to disease severity, score 0 Newtons.

2. Foot Plantarflexion Strength

Background/Purpose: Strength measurements are frequently employed in the clinical setting to assess muscle performance, enabling clinicians to diagnose weakness or impairment, evaluate treatment efficacy, and implement further interventions. Using standardized assessment procedures, measurement of maximal voluntary isometric contraction using hand-held dynamometry has demonstrated good reliability and validity in children and^{5,6} adults.^{7,8} In this protocol, strength is quantified using the Citec hand-held dynamometer (Citec dynamometer CT 3001, CIT Technics, Groningen, the Netherlands).

Hand-held dynamometer set-up: The Citec hand-held dynamometer can be used with the small square or concave attachment. Choose according to size/shape of the foot and comfort for the participant. The Citec hand-held dynamometer should be zeroed before placing it against the participant's dominant foot, determined as the foot used to kick a ball.

Assessment position: The participant is seated on the plinth in long sitting with feet off the edge. Participant may support trunk by leaning back on upper limbs. The clinical evaluator stabilizes the participant's lower leg against the plinth with one hand. With the other hand the clinical evaluator positions the foot in mid-range plantarflexion and then places the Citec hand-held dynamometer against the plantar surface of the foot just proximal to the metatarsal heads

Assessment Procedure: Each participant is assessed using the 'make' technique to measure strength, whereby the participant exerts a maximal force against the applicator of the Citec hand-held dynamometer. After a practice trial at submaximal effort, the participant will be instructed to perform 3-5 maximal voluntary contractions lasting 3-5 seconds each. A resting period of 10 seconds in between each contraction is given. Standardized verbal encouragement is used.



The clinical evaluator says:

"I am now going to measure your foot strength. I want you to push against the device as if trying to point your foot towards the ground as hard as you can for several seconds. Ready, set, go."

Scoring: Three valid measures for each muscle group of the dominant foot are recorded to the nearest Newton and averaged. For those unable to perform plantarflexion strength due to disease severity, score 0 Newtons.

3. Foot Dorsiflexion Strength

Hand-held dynamometer set-up: The Citec hand-held dynamometer can be used with the small square or concave attachment. Choose according to size/shape of the foot and comfort for the participant. The Citec hand-held dynamometer should be zeroed before placing it against the participant's dominant foot, determined as the foot used to kick a ball.

Assessment Position: The participant is positioned in long sitting with their heel off the edge of the plinth. Participant may support trunk by leaning back on upper limbs. The clinical evaluator stabilizes the participant's lower leg against the plinth with one hand. Be certain not to squeeze the muscle that you are testing when stabilizing the lower leg. With the other hand the clinical evaluator positions the foot in mid-range and then places the Citec hand-held dynamometer against the dorsal surface of the foot just proximal to the 2nd-4th metatarsal heads.

Assessment Procedure: Each participant is assessed using the 'make' technique to measure strength, whereby the participant exerts a maximal force against the applicator of the Citec hand-held dynamometer. After a practice trial at submaximal effort, the participant will be instructed to perform 3-5 maximal voluntary contractions lasting 3-5 each. A resting period of 10 seconds in between each contraction is given. Standardized verbal encouragement is used.



The clinical evaluator says:

"I am now going to take a measure of your foot strength. I want you to only use the muscles in your lower leg, try not to move the rest of your leg or body. I want you to pull against the device as if trying to bend your foot up towards you as hard as you can for several seconds. Ready, set, go."

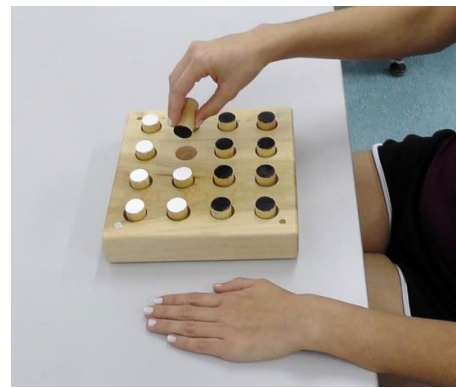
Scoring: Three valid measures for each muscle group of the dominant foot are recorded to the nearest Newton and averaged. For those unable to perform dorsiflexion strength due to disease severity, score 0 Newtons.

4. Functional Dexterity Test

Background/Purpose: The Functional Dexterity Test (FDT) is an assessment of hand dexterity that provides information regarding the use of the fingers and hands for daily functional tasks. The FDT requires 3-jaw chuck prehensions, also referred to as palmar pinch, pencil pinch or tripod grip. The FDT has demonstrated good validity and reliability in healthy individuals.^{9,10} The test may be performed with either the black or the white side of the pegs facing upwards.

Assessment Position: The participant is seated comfortably on a chair with their elbows in 90° of flexion, in front of the pegboard. The frontal edge of the pegboard is 10cm from the edge of the table placed at navel height. The non-dominant hand is placed next to the pegboard with the palm resting on the table.

Assessment Procedure: Starting with their hand flat next to the board; on the word "go" the participant turns over all the pegs as quickly as possible with their dominant hand, starting at the top at the opposite side of the board and continuing in a zigzag manner. The clinical evaluator demonstrates by turning over four (4) pegs, and the participant is then given a practice trial turning those 4 pegs. The dominant hand is assessed. It is important to demonstrate that the wrist should not supinate past the neutral position when manipulating the peg.



The clinical evaluator says:

"This is an assessment of your hand and finger function. I want you to turn over all the pegs as quickly as possible one at a time using one hand. I don't want you to turn your hand towards the ceiling, touch the board, touch the peg with your other hand, allow the peg to touch your chest, or drop a peg. Watch me and then you can have a practice trial."

After the practice trial the clinical evaluator says:

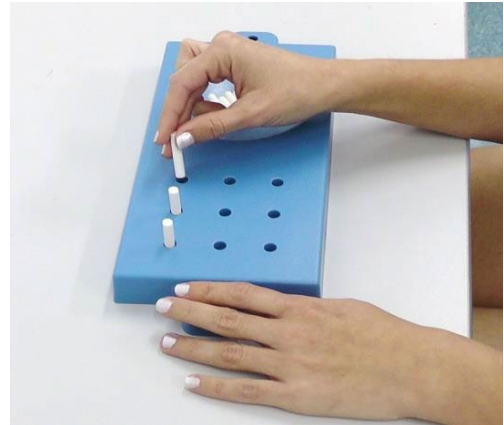
"I want you to now turn over all the pegs as fast as you can. Ready, set, go." The clinical evaluator starts the stopwatch at "Go" and stops timing when the participant releases the last peg.

Scoring: This protocol has been modified to exclude time penalties. If the participant supinates or touches the board the timer is paused, peg returned to the unturned position and the assessment resumed. If a peg is dropped the timer is paused, the participant retrieves the peg and places it in the unturned position, and then continues to turn the pegs, starting with the returned peg. The timer is re-started from the point it was stopped (not reset and restarted). The time to complete the task is recorded in seconds to the nearest tenth of a second. If the participant is unable to perform the item due to disease severity, assign the maximum time of 150 seconds.

5. Nine-Hole Peg Test

Background/Purpose: The Nine-Hole Peg Test is a timed assessment in which nine pegs are inserted and removed from nine holes in a pegboard with one hand. The Nine-Hole Peg Test provides a measure of hand function and examines fine motor ability, dexterity and hand-eye coordination. The Nine-Hole Peg Test has demonstrated very high inter-rater reliability in pediatric and adult populations.^{11,12}

Assessment Position: The participant is seated comfortably at a desk with their feet supported on the floor. The Nine-Hole Peg Test is placed 10 cm from the edge of the table at navel height, centered in front of the participant with the container for the pegs on the same side as the hand being assessed. The dominant hand is assessed, while the non-dominant hand stabilizes the peg board using fingers placed on the small extension of the board.



Assessment Procedure: Starting with their hand flat next to the board; on the word "go" the participant places all 9 pegs, one at a time, with their dominant hand in the holes and then removes them, one at a time, as quickly as possible. The clinical evaluator demonstrates the assessment using 3 pegs. The participant has a practice trial with 3 pegs using their dominant hand.

The clinical evaluator says:

"This is a timed assessment to see how fast you can put the pegs in and take them back out. Pick up each peg one at a time. Ready, set go."

When the participant puts the last peg into the hole, remind them to continue the test by saying "now, remove the pegs".

Scoring: The clinical evaluator starts the stopwatch at "Go" and stops timing when the last peg is returned to the container. If a peg is dropped off the board, the timer is stopped and the peg is replaced to its initial position. The participant may 'fumble' the peg, but can keep going if they are able to without too much delay (<1 sec). The time taken to insert all 9 pegs and remove them again is recorded in seconds to the nearest tenth of a second.

If the participant is unable to perform the item due to disease severity, assign the maximum time of 150 seconds.

6. Stair climb

Background/Purpose: This test measures how fast the participant ascends 4-steps.¹³ Equipment required is a digital stopwatch and 4 stairs with railings. The steps are free standing with each step being 6" (15 cm) high and 10" (25 cm) deep with a 24" by 30" (61cm by 76 cm) platform.

Assessment Position: The participant begins the test standing at the bottom of the stairs with their hands at their sides.

Assessment Procedure: Before testing the participant on the stairs, assess comfort level and ability on stairs via a practice trial. If the participant is unable or unsafe doing stairs, do not attempt the test. Record the max score on the data entry form (see below).



The clinical evaluator says:

"This is an assessment to see how fast you can safely go up these 4 stairs. You can use the railing if you want to. Ready, set, go."

Scoring: The clinical evaluator starts recording the time on the stopwatch at "Go" and stops timing when they have completed 4 steps and their feet are together within their base of support. Notate if the handrail was used to ensure consistency during reassessment. If the participant is unable to perform the item due to disease severity, assign the maximum time of 150 seconds.

7. 30-Second Chair Stand Test

Background/Purpose: The 30-Second Chair Stand Test is used by researchers and clinicians as an assessment of functional lower limb strength.¹⁴ Using a standardized protocol, which includes one practice stand by the participant, the 30-Second Chair Stand Test has demonstrated good inter-tester and intra-tester reliability in adult populations.¹⁵

Assessment Position: A height adjustable flat chair/bench is placed against a wall. The participant sits comfortably, with their sacrum in the middle of the chair. The height of the seat is adjusted so that the participant sits with their hips and knees flexed at 90° and their feet hip width apart. The participant is asked to cross their arms across their chest (hands at shoulders), and not to rest against the back of the chair or wall during the assessment. They are not allowed to use their arms to generate momentum or to push up on the armrests, chair seat or any part of their body and must extend fully when standing. The clinical evaluator stands to the side of the participant holding a stopwatch.



Assessment Procedure: The evaluator demonstrates the task and then the participant is asked to perform one practice stand. The participant is then asked to perform as many repetitions as possible in 30 seconds.

The clinical evaluator says:

“This assessment records how many times you can sit and stand as quickly as possible from the chair in 30 seconds. You must fully sit down with your bottom touching the seat and stand up for the repetition to be counted. When I say go you will start the assessment, you can stop at any time as you need to. Ready, set, go.”

If they do not come to a fully extended (erect) position, remind them that they need to come to a fully extended position for the repetition to count.

Scoring: Repetitions where the arms are used for support are not counted. The number of full sit to stands performed in the 30 seconds is recorded. If the timer reaches 30 seconds when the participant is in full stand, it is counted as this test is measuring the number of sit to stand transitions. For those unable to perform this item due to disease severity, score 0 repetitions.

Note: participant’s own walking shoes should be worn unless an inappropriate style of shoe is worn (e.g., flip-flops or high heels) or the participant’s walking shoes do not fit without AFOs, then barefoot is permitted if safe to do so and institutionally permissible.



Balance General Instructions

Background/Purpose: These items are performed to assess static standing balance under different conditions and include standing with feet apart on a line with eyes open, standing with feet apart on a line with eyes closed and single leg stance with eyes closed

Set-up:

- Place a target on the wall at the appropriate level for the participant's height, with the bottom of the target at the participant's eye level.
- Using a piece of tape, make a straight line on the floor
- For each item, conduct a second trial only if the participant does not earn the maximum score on their first trial.
- Before administering each item, teach the task to the participant using verbal and non-verbal directions as necessary to ensure understanding of the task.
- The entire foot should be placed along the line, unless foot deformity precludes this testing position e.g. if foot is fixed in abduction they may use this position for testing.
- The participant must attain the starting position on their own. They may not use a wall or lean on the clinical evaluator for stability.
- For those unable to perform balance items (including getting into the test position), a score of 0 is given.



8. Standing with feet apart on a line with eyes open

Assessment procedure: The participant stands with feet together, dominant foot on the line with their hands on their hips. The participant will be asked to take one natural step forward, placing their non-dominant foot along the line, and to look at the target. Dominance is determined by the foot they kick a ball with. Teach the task to the participant, giving verbal directions along with a visual demonstration. Then allow the participant a practice trial and provide any necessary feedback. If the participant does not earn the maximum score of 10 seconds, conduct a second trial. If necessary, re-teach the task after you say “Let’s try it again.”



The clinical evaluator says:

“This assessment is testing your balance. I want you to stand on the line with your (state dominant) foot and take a step forward with your (state non dominant) foot so that you are standing with both feet on the line. I want you to hold this position. as long as you can or until I tell you to stop.”

Scoring: Start timing when the participant has attained proper form. Record the number of seconds, to nearest tenth of a second, that the participant maintains proper form, up to 10 seconds. Stop after 10 seconds or if the participant steps off the line or fails to keep hands on hips. For those unable to perform this item (including getting into the test position), a score of 0 is given.

9. Standing with feet apart on a line with eyes closed

Assessment procedure: The participant stands with feet together, dominant foot along the line with their hands on their hips. The participant will be asked to take one natural step forward, placing their non-dominant foot along the line and close their eyes. Teach the task to the participant, giving verbal directions along with a visual demonstration. Then allow the participant a practice trial and provide any necessary feedback. If the participant does not earn the maximum score of 20 seconds, conduct a second trial. If necessary, re-teach the task after you say, "Let's try it again."

The clinical evaluator says:

"This assessment is testing your balance when you close your eyes. I want you to stand on the line with your (state dominant) foot and take a step forward with your (state non dominant) foot so that you are standing with both feet on the line. I want you to hold this position. as long as you can or until I tell you to stop."

Scoring: Start timing when the participant has attained proper form and closed their eyes. Record the number of seconds, to nearest tenth of a second, that the participant maintains proper form, up to 20 seconds. Stop after 20 seconds or if the participant steps off the line, fails to keep hands on hips or opens their eyes.



For those unable to perform this item (including getting into the test position), a score of 0 is given.

10. Single leg stance with eyes closed

Assessment procedure: The participant stands with feet together, dominant foot along the line with their hands on their hips. The participant will be asked to raise their non-dominant leg behind them, with their knee bent to 90° and shin parallel to the floor and to close their eyes. Teach the task to the participant, giving verbal directions along with a visual demonstration. Then allow the participant a practice trial and provide any necessary feedback. If the participant does not earn the maximum score of 20 seconds, conduct a second trial. If necessary, re-teach the task after you say, "Let's try it again."

The clinical evaluator says:

"This assessment is testing your balance when you close your eyes and stand on one leg. Can you please bend your left or right knee (state the non-dominant foot) and close your eyes. Try and hold this position as long as you can or until I tell you to stop."



Scoring: Start timing when the participant has attained proper form and closed their eyes. Record the number of seconds, to nearest tenth of a second, that the participant maintains proper form, up to 20 seconds. Stop after 20 seconds or if the participant steps off the line, fails to keep hands on hips or opens their eyes. For those unable to perform this item (including getting into the test position), a score of 0 is given.

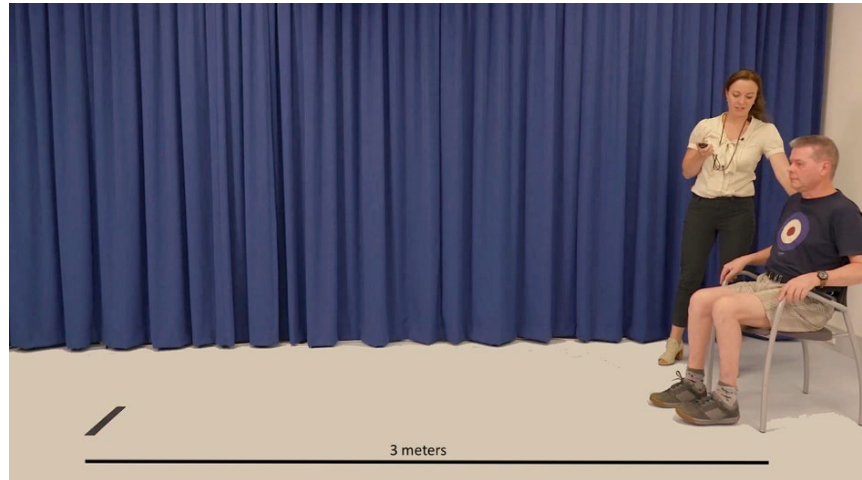
Note: reminder that participant's own walking shoes should be worn unless an inappropriate style of shoe is worn (e.g., flip-flops or high heels) or the participant's walking shoes do not fit without AFOs, then barefoot is permitted if safe to do so and institutionally permissible.

11. Timed Up and Go

Background/Purpose: This is a test of mobility, balance, walking, as well as falls risk.¹⁶

Assessment Position: Measure a 3-meter walkway by placing tape on the floor. Place a standard chair with arm rests at the beginning of the walkway, with the front legs on the start line and ensure that the line is easily seen by the participant. To begin the test, the participant sits comfortably with their back and hips against the back of the chair.

Assessment Procedure: The participant is allowed to use the arm rests during the sit–stand and stand–sit movements. Take care to ensure that the chair is stable and will not move when the participant moves from sit to stand. The participant should wear their regular footwear. There is no time limit, and they may stop and rest (but not sit down) if they need to.

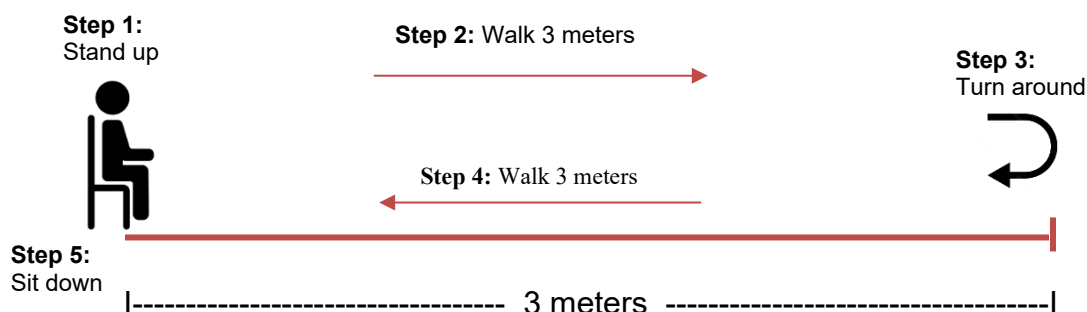


The participant is given a practice trial that is not timed and provided feedback as necessary.

The clinical evaluator says:

“This assessment is testing your mobility, balance and walking, and is not a test of speed. When I say GO, you will stand up, walk to the line on the floor (not past), turn around and walk back to the chair and sit down. You can use the arm rests if you need to and it is important that you perform this test at a comfortable, safe pace.”

Scoring: The clinical evaluator starts recording the time on the stopwatch on the word “GO” and stops timing when the participant’s bottom touches the chair. If the participant is unable to perform the item due to disease severity, assign the maximum time of 150 seconds.



12. Six-Minute Walk Test

Background/Purpose: The Six-Minute Walk Test (6MWT) is a measure of sub-maximal endurance capacity.^{17,18} High reliability and validity has been established in pediatric and adult populations.^{19,20} Standardized instructions are used, as encouragement can enhance 6MWT performance by up to 30%.¹⁷ Performance of a single 6MWT has been shown to be as effective as two repeated 6MWTs for outcome measurement and exercise prescription.²¹ The American Thoracic Society provides guidelines for administering the 6MWT.²² The 6MWT is performed indoors, along a long, flat, straight, firm surface. The walking course is 25 meters long and the turnaround points are marked with cones (with the edge on the inside of the course). The direction the participant turns has no effect on the outcome.²³ The participant performs the assessment in comfortable walking shoes and does not have anyone accompanying them during this test.

Assessment Position: The clinical evaluator is positioned at the start of the course with a stopwatch and counter in each hand. The participant stands at the starting line of the course.

Assessment Procedure: The clinical evaluator says:

“This item measures how far you can walk in six minutes. When I say go I want you to walk as fast as you comfortably can, without running, for six minutes. I will count your laps and tell you how much time is remaining at each minute. You can stop to take rests if you need to and then keep going, or you can cease the assessment if you can’t go any further. Ready, set, go.”



At each minute-split, the timer announces the number of minutes completed and the number remaining with standardized encouragement (Ex: you are doing well, keep up the good work).

Scoring: Each time the participant rounds a cone, record the lap on the tally (lap) counter. The extra distance walked in the final lap is measured using a tape measure along the track and added to the total. The final 6MWT distance is recorded to the nearest meter. For those unable to perform the item due to disease severity, score 0 meters.

Patient Profile

Complete: ID, DOB, Age, Sex, Height, Weight, Dominant Hand/Foot, Diagnosis.

Symptoms

Common complaints in participants with CMT include foot pain, hand pain, leg cramps, hand weakness, unsteady ankles during walking, daily trips and/or falls hand tremor, sensory symptoms (e.g. pins and needles, tingling, numbness, prickling).^{24,25} Each participant is asked about the presence or absence of each symptom and marked as present or absent.

Note: Look for tremor throughout the assessment and ask about symptom history if observed.

Lunge test (degrees)

Flexibility of ankle joint dorsiflexion is measured weight bearing using the lunge test. This technique has been shown to be reliable and valid in children and adults.²⁶

Assessment Position: The participant is asked to place their foot perpendicular to a wall and lunge their knee toward the wall.²⁷ The foot is progressively moved away from the wall until the maximum angle of ankle joint dorsiflexion is obtained without the heel lifting. Pronation and supination of the subtalar and midtarsal joints is restricted by ensuring that the foot is positioned perpendicular to the wall, and the participant lunges directly over the midline of the foot (second toe). Participants are instructed to hold onto the wall for balance and the contralateral leg is placed in a comfortable position. While the participant is positioned in maximum dorsiflexion range, a digital inclinometer is aligned with the midline of the Achilles tendon. The number of degrees is recorded.



Equipment: Baseline Digital Inclinometer:
<http://www.bpp2.com/> or iHandy Level App (be sure to calibrate before use)



Foot Posture Index

Foot structure of each foot of all participants is assessed using the Foot Posture Index (FPI), a diagnostic tool that evaluates the multisegmental and multiplanar aspects of the foot using six criteria that together enable the foot to be scored along a continuum of cavus (supinated) to planus (pronated) features.²⁸

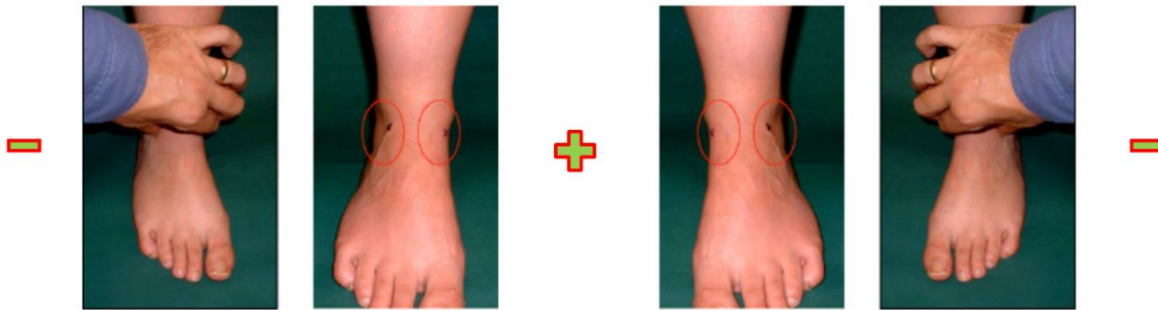
The FPI allocates a score between -2 and +2 to each of the six criteria:

1. Talar head palpation
2. Curves above and below the lateral malleolus
3. Inversion/eversion of the calcaneus
4. Prominence in the region of the talonavicular joint
5. Congruence of the medial longitudinal arch
6. Abduction/adduction of the forefoot on the rearfoot

Scores are allocated for each criterion with a score of 0 denoting a neutral position, -2 for clear signs of supination, and +2 for clear signs of pronation. The aggregated score ranges from -12 (extremely supinated/pes cavus) to +12 (extremely pronated/pes planus).

Test Position: All observations of the FPI are made with the participant in a relaxed stance with double limb support. They are asked to take several steps on the spot and then to relax and stand still, with their arms by their side and looking straight ahead. Ensure the participant does not swivel to see what is happening as this will change the foot posture. The FPI should take 2 minutes. Scoring information and instructions are detailed below.

1.Talar head palpation



Right Foot

Left Foot

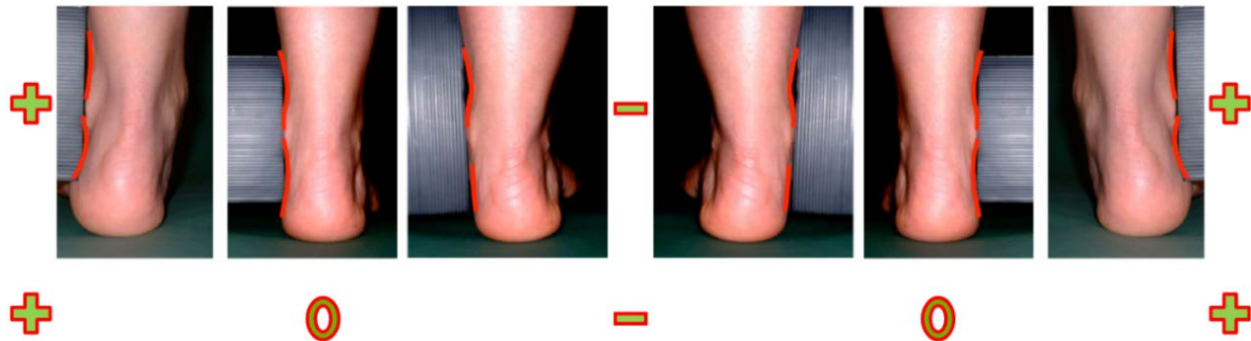
-2	-1	0	+1	+2
Talar head palpable on lateral side/but not on medial side	Talar head palpable on lateral side/ slightly palpable on medial side	Talar head equally palpable on lateral and medial side	Talar head palpable on medial side/slightly palpable on lateral side	Talar head palpable on medial side/but not on lateral side

+2	+1	0	-1	-2
Talar head palpable on medial side/but not on lateral side	Talar head palpable on medial side/slightly palpable on lateral side	Talar head equally palpable on lateral and medial side	Talar head palpable on lateral side/ slightly palpable on medial side	Talar head palpable on lateral side/but not on medial side

2.Curves above and below the lateral malleolus

Left Foot

Right Foot



+2	+1	0	-1	-2
Curve below malleolus markedly more concave than curve above malleolus	Curve below malleolus more concave than curve above malleolus	Both infra and supra malleolar curves roughly equal	Curve below the malleolus concave but flatter/ more shallow than the curve above malleolus	Curve below the malleolus either straight or convex

-2	-1	0	+1	+2
Curve below the malleolus either straight or convex	Curve below the malleolus concave but flatter/ more shallow than the curve above malleolus	Both infra and supra malleolar curves roughly equal	Curve below malleolus more concave than curve above malleolus	Curve below malleolus markedly more concave than curve above malleolus

3. Inversion/eversion of the calcaneus

Left Foot

+

0

-

+2	+1	0	-1	-2
More than an estimated 5° everted (valgus)	Between vertical and an estimated 5° everted (valgus)	Vertical	Between vertical and an estimated 5° inverted (varus)	More than an estimated 5° inverted (varus)

Right Foot

+

0

-

-2	-1	0	+1	+2
More than an estimated 5° inverted (varus)	Between vertical and an estimated 5° inverted (varus)	Vertical	Between vertical and an estimated 5° everted (valgus)	More than an estimated 5° everted (valgus)

4. Bulge in the region of the talonavicular joint

Left Foot

+

0

-

+2	+1	0	-1	-2
Area of TNJ bulging markedly	Area of TNJ bulging slightly	Area of TNJ flat	Area of TNJ slightly, but definitely concave	Area of TNJ markedly concave

Right Foot

+

0

-

-2	-1	0	+1	+2
Area of TNJ markedly concave	Area of TNJ slightly, but definitely concave	Area of TNJ flat	Area of TNJ bulging slightly	Area of TNJ bulging markedly

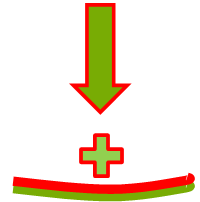
5. Congruence of the medial longitudinal arch



Supinated foot (-2)



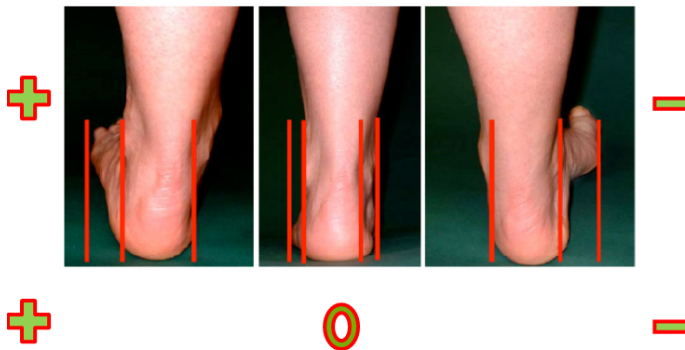
Pronated foot (+2)



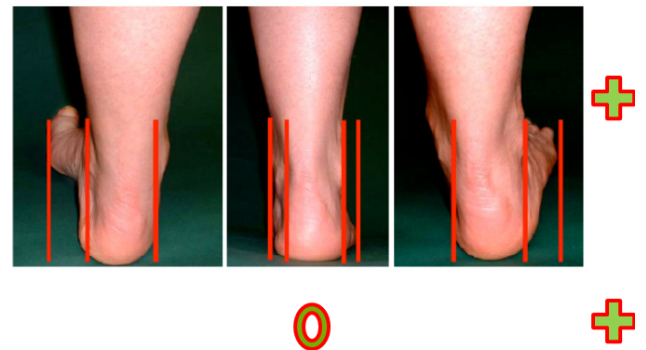
-2	-1	0	+1	+2
Arch highly acutely angled towards the posterior end of the medial arch	Arch moderately high and slightly acute posteriorly	Arch height normal and concentric ally curved	Arch lowered with some flattening in the central portion	Arch very low with severe flattening in the central portion – arch making ground contact

6. Abduction/adduction of forefoot and rearfoot (too-many-toes)

Left Foot



Right Foot



+2	+1	0	-1	-2
No medial toes visible . Lateral toes clearly visible	Lateral toes clearly more visible than medial	Medial and lateral toes equally visible	Medial toes clearly more visible than lateral	No lateral toes visible . Medial toes clearly visible

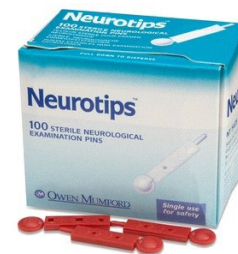
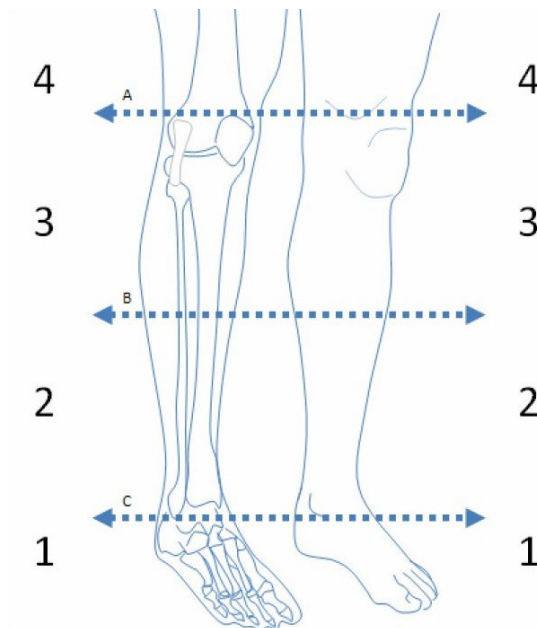
-2	-1	0	+1	+2
No lateral toes visible . Medial toes clearly visible	Medial toes clearly more visible than lateral	Medial and lateral toes equally visible	Lateral toes clearly more visible than medial	No medial toes visible . Lateral toes clearly visible

Pinprick

Test Position: Dominant lower limb only is tested. Participants should have their eyes closed during the test.

Testing Procedures: The first important part of this exam is to determine if the participant is able to feel the sharp side of the Neurotip™. This test should be first performed in a region with “expected” normal sensation (distal thigh, above top of the patella). If the participant does not feel pain, score 4 and go to the next test. If he/she does feel pain, the test will be performed in the lower limbs (distal-to-proximal direction) and graded according to the 4 levels shown below from the *CMT Neuropathy Score* (CMTNS-2nd, 2010)²⁹:

- 0 Normal
- 1 Decreased below or at ankle bones (Note: Below the line passing at ankle malleoli)
- 2 Decreased at or below the midline of the calf
- 3 Decreased above the calf midline, up to and including knee (Knee = Top of the patella.)
- 4 Decreased above the knee (above top of the patella)



Neurotips™ (100): Owen Mumford Ltd, Oxford, UK: www.owenmumford.com

Vibration

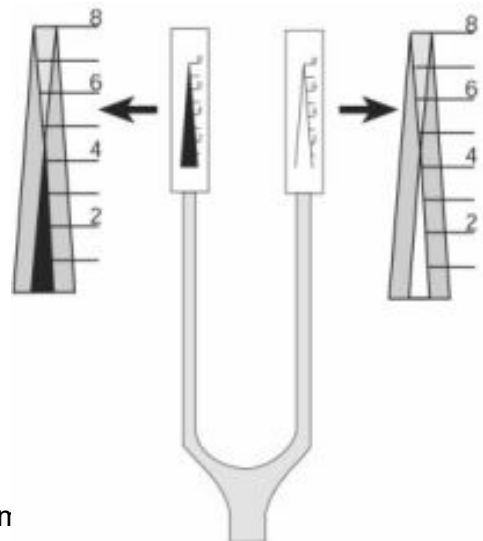
Test Position: Dominant lower limb only is tested. All participants should have their eyes closed during the test.

Testing positions (start distally):

1. Foot (dorsum of first metatarsophalangeal joint)
2. Ankle (medial malleolus)
3. Knee (tibial tuberosity)

Testing Procedures: Determine if the participant is able to feel the tuning fork vibrating on a bony region with “expected” normal sensation (e.g., sternum, forehead). Set the tuning fork into motion by compressing the prongs (tines) in a finger snapping motion. As the prongs start to oscillate, the illusion of two triangles is visible on each damper. The participant is asked to indicate the moment when they can no longer perceive the decreasing vibratory stimulus. Read off the black triangle. If the participant does not feel any vibration at the “expected” normal region, score 4 and go to the next test. If the participant does feel vibration, the test will be performed in the lower limbs (distal-to-proximal direction) and graded according to the 4 levels described below from the *CMT Neuropathy Score* (CMTNS-2nd, 2010)²⁹:

- | | |
|----------|---|
| 0 | Normal (≥ 5) ³⁰ |
| 1 | Reduced at great toe (dorsum 1 st MPJ) |
| 2 | Reduced at ankle |
| 3 | Reduced at knee (tibial tuberosity) |
| 4 | Absent at knee and ankle |



Rydel Seiffer tuning fork, C 64 Hz / c 128 Hz detachable clan
Arno Barthelmes & Co. GmbH - Tuttlingen - Germany: www.barthelmes.info

Gait

Background/Purpose: Difficulty toe walking is a gross indicator of plantarflexion weakness, difficulty heel walking is a gross indicator of dorsiflexion weakness (and Achilles tendon shortening), and the presence of foot drop is a sign of dorsiflexion weakness during gait.²⁴

Foot Drop Testing Procedure

The participant is asked to walk 10 steps and the clinical evaluator assesses the presence of foot drop. Foot drop is defined as plantarflexion during the mid-swing phase of gait, often with a forefoot strike/slap on loading.

Scoring

No: If the participant demonstrates NO forefoot strikes/slaps on all steps

Some: If the participant demonstrates SOME forefoot strikes/slaps during gait or lands flatfooted

Yes: If the participant demonstrates forefoot strikes/slaps on ALL steps

Detecting foot drop can be subtle, so ask the participant to walk additional steps or confirm during the Six-Minute Walk Test if needed

Difficulty Heel Walking Testing Procedure

The participant is asked to heel walk (forefoot lifted with medial and lateral border off the floor) for 10 steps.

Scoring:

No: If the participant demonstrates NO difficulty heel walking (10 steps)

Some: If the participant demonstrates SOME difficulty heel walking (1-9 steps)

Yes: If the participant demonstrates difficulty heel walking on ALL steps (0 steps)

Difficulty Toe Walking Testing Procedure Testing

The participant is asked to tip-toe walk (heel lifted) for 10 steps.

Scoring:

No: If the participant demonstrates NO difficulty tip-toe walking (10 steps)

Some: If the participant demonstrates SOME difficulty tip-toe walking (1-9 steps)

Yes: If the participant demonstrates difficulty tip-toe walking on ALL steps (0 steps)

Calibration Tasks

Citec hand-held dynamometer

The Citec hand-held dynamometer should be calibrated regularly with a known weight (e.g. 5 kilograms) which has been verified by a biomedical department or calibrated scales. Calibrating it before use is ideal but if this is not possible it should be calibrated at least monthly with a calibration record kept.

Using the technique shown below, whereby a 5-kilogram known weight (49 Newtons) is applied to the Citec hand-held dynamometer.

For alternate weight, use Force Converter website:

www.unitconversion.org/unit_converter/force.html



References

1. McKay MJ, Baldwin JN, Ferreira P, et al. Normative reference values for strength and flexibility of 1,000 children and adults. *Neurology* 2017; 88(1): 36-43.
2. McKay MJ, Baldwin JN, Ferreira P, et al. Reference values for developing responsive functional outcome measures across the lifespan. *Neurology* 2017; 88(16): 1512-9.
3. Merlini L, Mazzone ES, Solari A, Morandi L. Reliability of hand-held dynamometry in spinal muscular atrophy. *Muscle & Nerve* 2002;26:64-70.
4. Solari A, Laura M, Salsano E, Radice D, Pareyson D. Reliability of clinical outcome measures in Charcot-Marie-Tooth disease. *Neuromuscular Disorders* 2008;18:19-26.
5. Burns J, Ouvrier R, Estilow T, et al. Validation of the Charcot-Marie-Tooth disease pediatric scale as an outcome measure of disability. *Annals of Neurology* 2012;71:642-652.
6. Rose KJ, Burns J, Ryan MM, Ouvrier RA, North KN. Reliability of quantifying foot and ankle muscle strength in very young children. *Muscle & Nerve* 2008;37:626-631.
7. Beenakker EA, van der Hoeven JH, Fock JM, Maurits NM. Reference values of maximum isometric muscle force obtained in 270 children aged 4-16 years by hand-held dynamometry. *Neuromuscular disorders* 2001;11:441-446.
8. Burns J, Redmond A, Ouvrier R, Crosbie J. Quantification of muscle strength and imbalance in neurogenic pes cavus, compared to health controls, using hand-held dynamometry. *Foot & Ankle International* 2005;26:540-544.
9. Aaron DH, Jansen CW. Development of the Functional Dexterity Test (FDT): construction, validity, reliability, and normative data. *Journal of Hand Therapy* 2003;16:12-21.
10. Sartorio F, Bravini E, Vercelli S, et al. The Functional Dexterity Test: test-retest reliability analysis and up-to date reference norms. *Journal of Hand Therapy* 2013;26:62-67; quiz 68.
11. Oxford Grice K, Vogel KA, Le V, Mitchell A, Muniz S, Vollmer MA. Adult norms for a commercially available Nine Hole Peg Test for finger dexterity. *The American Journal of Occupational Therapy* 2003;57:570-573.
12. Poole JL, Burtner PA, Torres TA, et al. Measuring dexterity in children using the Nine-hole Peg Test. *Journal of Hand Therapy* 2005;18:348-351.
13. Piscosquito G, Reilly MM, Schenone A, et al. Responsiveness of clinical outcome measures in Charcot-Marie-Tooth disease. *European Journal of Neurology* 2015;22:1556-1563.
14. Jones CJ, Rikli RE, Beam WC. A 30-s chair-stand test as a measure of lower body strength in community-residing older adults. *Research quarterly for exercise and sport* 1999;70:113-119.
15. Gill S, McBurney H. Reliability of performance-based measures in people awaiting joint replacement surgery of the hip or knee. *Physiotherapy Research International* 2008;13:141-152.
16. Podsiadlo D, Richardson S. The timed "Up & Go": a test of basic functional mobility for frail elderly persons. *Journal of the American Geriatrics Society* 1991;39:142-148.
17. Camarri B, Eastwood PR, Cecins NM, Thompson PJ, Jenkins S. Six minute walk distance in healthy subjects aged 55-75 years. *Respiratory Medicine* 2006;100:658-665.
18. Enright PL. The six-minute walk test. *Respiratory Care* 2003;48:783-785.
19. de Groot JF, Takken T. The six-minute walk test in paediatric populations. *Journal of Physiotherapy* 2011;57:128.
20. Kervio G, Carre F, Ville NS. Reliability and intensity of the six-minute walk test in healthy elderly subjects. *Medicine and Science in Sports and Exercise* 2003;35:169-174.
21. Bellet RN, Francis RL, Jacob JS, et al. Repeated six-minute walk tests for outcome measurement and exercise prescription in outpatient cardiac rehabilitation: a longitudinal study. *Archives of Physical Medicine and Rehabilitation* 2011;92:1388-1394.
22. ATS statement: guidelines for the six-minute walk test. *American Journal of Respiratory and Critical Care Medicine* 2002;166:111-117.

23. Ng SS, Yu PC, To FP, Chung JS, Cheung TH. Effect of walkway length and turning direction on the distance covered in the 6-minute walk test among adults over 50 years of age: a cross-sectional study. *Physiotherapy* 2013;99:63-70.
24. Burns J, Ryan MM, Ouvrier RA. Evolution of foot and ankle manifestations in children with CMT1A. *Muscle & Nerve* 2009;39:158-166.
25. Burns J, Bray P, Cross LA, North KN, Ryan MM, Ouvrier RA. Hand involvement in children with Charcot-Marie-Tooth disease type 1A. *Neuromuscular disorders* 2008;18:970-973.
26. Khan K, Roberts P, Nattrass C, et al. Hip and ankle range of motion in elite classical ballet dancers and controls. *Clinical Journal of Sport Medicine* 1997;7:174-179.
27. Rose KJ, Burns J, North KN. Factors Associated With Foot and Ankle Strength in Healthy Preschool-Age Children and Age-Matched Cases of Charcot-Marie-Tooth Disease Type 1A. *Journal of Child Neurology* 2010;25:463-468.
28. Redmond AC, Crosbie J, Ouvrier RA. Development and validation of a novel rating system for scoring standing foot posture: the Foot Posture Index. *Clinical Biomechanics* 2006;21:89-98.
29. Murphy SM, Herrmann DN, McDermott MP, et al. Reliability of the CMT neuropathy score (second version) in Charcot-Marie-Tooth disease. *Journal of the Peripheral Nervous System* 2011;16:191-198.
30. Hilz MJ, Axelrod FB, Hermann K, Haertl U, Duetsch M, Neundorfer B. Normative values of vibratory perception in 530 children, juveniles and adults aged 3-79 years. *Journal of the Neurological Sciences* 1998;159:219-225.