Pantothenate Kinase Associated Neurodegeneration Disease Rating Scale (PKAN-DRS)

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PKAN Disease Rating Scale (PKAN-DRS)

SC I. COGNITION	**SCORE** ☐
**1. ***Interview between doctor and patient or caregiver. Responses should refer to a period encompassing the prior month, including the day of assessment. Consider all types of altered cognitive functioning, including cognitive slowing, impaired reasoning, memory loss, deficits in attention, and orientation. Rate their impact on activities of daily living as perceived by the patient and/or caregiver. Ask for activities that do not depend on motor capability. Instructions to adult patients (and caregiver): Over the past month, have you had problems remembering things, following conversations, paying attention, or thinking clearly? Instructions to children (and caregiver): Over the past month, have you had problems paying attention in class, forgetting things, or playing games?	
0	No cognitive impairment.
1	Impairment appreciated by patient or caregiver with no concrete interference in the patient's ability to carry out normal activities and social interactions.
2	Clinically evident cognitive dysfunction, but only minimal interference with the patient's ability to carry out normal activities and social interactions.
3	Cognitive deficits interfere with but do not preclude the patient's ability to carry out normal activities and social interactions.
4	Cognitive dysfunction precludes the patient's ability to carry out normal activities and social interactions.

SC II. BEHAVIOR	**SCORE** ☐
2. ╪Interview between doctor and patient or caregiver. Responses should refer to a period encompassing the prior month, including the day of assessment. The examiner asks for the presence of any of the following 7 psychiatric symptoms: 1) **Sad/Mood:** feeling sad, sad voice/expression, tearfulness, inability to enjoy anything. 2) **Anxiety:** worries, anticipation of the worst, fearful anticipation. 3) **Suicidal Thoughts:** feels life is not worth living, has suicidal thoughts, active suicidal intent, preparation for the act. 4) **Disruptive or Aggressive behavior:** threatening behavior, physical violence, verbal outbursts, threatening, foul, or abusive language. 5) **Irritable Behavior:** impatient, demanding, inflexible, driven and impulsive, uncooperative.	

6) **Obsessions and Compulsions:** recurrent and persistent ideas, thoughts or images and/or repetitive, purposeful, and intentional behaviors. 7) **Hallucinations:** a perception without physical stimulus: auditory, visual, tactile, gustatory, or olfactory.	
0	No psychiatric symptoms.
1	One psychiatric symptom.
2	Two psychiatric symptoms.
3	Three psychiatric symptoms.
4	Four or more psychiatric symptoms.

SC III. DISABILITY Interview between doctor and patient or caregiver. Responses should refer to a period encompassing the prior month including the day of assessment.	
3. 1. SPEECH	**SCORE** ☐
0	Normal
1	Slightly involved, easily understood.
2	Some difficulty in understanding.
3	Marked difficulty in understanding.
4	Complete or almost complete anarthria.
3. 2. HANDWRITING	**SCORE** ☐
0	Normal.
1	Slight difficulty; legible.
2	Almost illegible.
3	Illegible.
4	Unable to grasp to maintain hold on pen.
3. 3. FEEDING	**SCORE** ☐
0	Normal.
1	Uses ‘tricks’; independent.
2	Can feed but not cut.
3	Finger food only.
4	Completely dependent.
3. 4. EATING/SWALLOWING	**SCORE** ☐
0	Normal.
1	Occasional choking (< 1 episode per month).

2	Chokes frequently; difficulty in swallowing (> 1episode per month).
3	Unable to swallow firm foods.
4	Marked difficulty swallowing soft foods and liquids or needs enteral feeding (nasogastric or gastric tube).
3. 5. HYGIENE	**SCORE** ☐
0	Normal.
1	Clumsy; independent.
2	Needs help with some activities.
3	Needs help with most activities.
4	Needs help with all activities.
3. 6. DRESSING	**SCORE** ☐
0	Normal.
1	Clumsy; independent.
2	Needs help with some activities.
3	Needs help with most activities.
4	Needs help with all activities.
3. 7. WALKING	**SCORE** ☐
0	Normal gait.
1	Slightly abnormal. Independent gait; hardly noticeable.
2	Moderately abnormal. Independent gait, occasional falls.
3	Considerably abnormal. Independent gait, frequent falls.
4	Needs assistance to walk (walk-device or living support).
5	Wheel-chair bound or bed bound.
3. 8. SCHOOL/WORK	**SCORE** ☐
0	Normal ability in mainstream classroom / able to engage in gainful employment in his/her accustomed work.
1	Marginal ability in mainstream classroom/ able to engage in any kind of gainful employment.
2	Requires special needs classroom/ able to engage in any kind of volunteer or non-gainful work.
3	Unable to attend special needs classroom/ unable to engage in any work
3. 9. VISION	**SCORE** ☐
0	Normal
1	Limited eye or head movement to large objects or parental face in visual field (children)/ finger counting only (adults).
2	No response to large objects or parental face in visual field.
3	No response to light.

SC IV. PARKINSONISM The investigator should "rate what you see", assuming that overlapping features of PKAN may interfere with individual items in the motor examination of parkinsonism.	
4. 1. *GLOBAL SPONTANEITY OF MOVEMENT (BODY BRADYKINESIA)	**SCORE** ☐
This global rating combines all observations on slowness, hesitancy, and small amplitude and poverty of movement in general, including a reduction of gesturing and of crossing the legs. This assessment is based on the examiner's global impression after observing for spontaneous gestures while sitting, and the nature of rising and walking.	
0	Normal. No problems.
1	Slight. Slight global slowness and poverty of spontaneous movements.
2	Mild. Mild global slowness and poverty of spontaneous movements.
3	Moderate. Moderate global slowness and poverty of spontaneous movements.
4	Severe. Severe global slowness and poverty of spontaneous movements.
4. 2. *RIGIDITY	**SCORE** ☐
Rigidity is judged on slow passive movement of major joints with the patient in a relaxed position and the examiner manipulating the limbs. First, test without an activation maneuver. Test limbs separately. If no rigidity is detected, use an activation maneuver, such as tapping fingers, fist opening/closing or heel tapping in a limb not being tested. Rate the most affected limb. Only live-rating is used for this test.	
0	Normal. No rigidity.
1	Slight. Rigidity only detected with activation maneuver.
2	Mild. Rigidity detected without the activation maneuver, but full range of motion is easily achieved.
3	Moderate. Rigidity detected without the activation maneuver; full range of motion is achieved with effort.
4	Severe. Rigidity detected without the activation maneuver and full range of motion not achieved.
4. 3. *FINGER-TAPPING	**SCORE** ☐
Each hand is tested separately. Demonstrate the task, but do not continue to perform the task while the patient is being tested. Instruct the patient to tap the index finger on the thumb 10 times as quickly AND as big as possible. Rate the most affected limb, evaluating speed, amplitude, hesitations, halts, and decrementing amplitude.	
0	Normal. No problems.
1	Slight. Any of the following: a) the regular rhythm is broken with one or two interruptions or hesitations of the tapping movement; b) slight slowing; c) the amplitude decrements near the end

	of the 10 taps.
2	Mild. Any of the following: a) 3 to 5 interruptions during tapping; b) mild slowing; c) the amplitude decrements midway into the 10-tap sequence.
3	Moderate. Any of the following: a) more than 5 interruptions during tapping or at least one longer arrest (freeze) in ongoing movement; b) moderate slowing; c) the amplitude decrements starting after the first tap.
4	Severe. Cannot or can only barely perform the task because of slowing, interruptions, or decrements.

4. 4. *LEG AGILITY	**SCORE** ☐

Have the patient sit in a straight-backed chair with arms. The patient should have both feet comfortably on the floor. Test each leg separately. Demonstrate the task, but do not continue to perform the task while the patient is being tested. Instruct the patient to place the foot on the ground in a comfortable position and then raise and stomp the foot on the ground 10 times as high and fast as possible. Rate the most affected limb, evaluating speed, amplitude, hesitations, halts and decrementing amplitude.

0	Normal. No problems.
1	Slight. Any of the following: a) the regular rhythm is broken with one or two interruptions or hesitations of the movement; b) slight slowing; c) the amplitude decrements near the end of the task.
2	Mild. Any of the following: a) 3 to 5 interruptions during the movements; b) mild slowness; c) the amplitude decrements midway into the task.
3	Moderate. Any of the following: a) more than 5 interruptions during the movement or at least one longer arrest (freeze) in ongoing movement; b) moderate slowing in speed; c) the amplitude decrements after the first tap.
4	Severe. Cannot or can only barely perform the task because of slowing, interruptions or decrements.

4. 5. *FREEZING OF GAIT	**SCORE** ☐

While assessing gait, also assess for the presence of any gait freezing episodes. Observe for start hesitation and stuttering movements especially when turning and reaching the end of the task. To the extent that safety permits, patients may not use sensory tricks during the assessment. In situations where it is absolutely impossible to test severity (e.g. in patients that cannot walk) the examiner should use the notation "UR" for Unable to Rate.

0	Normal. No freezing.

1	Slight. Freezes on starting, turning or walking through doorway with a single halt during any of these events, but then continues smoothly without freezing during straight walking.
2	Mild. Freezes on starting, turning or walking through doorway with more than one halt during any of these activities, but continues smoothly without freezing during straight walking.
3	Moderate. Freezes once during straight walking.
4	Severe. Freezes multiple times during straight walking.

4. 6. *POSTURAL AND/OR TRUNK STABILITY	**SCORE** ☐

Test retropulsion. Stand behind the patient and instruct the patient on what is about to happen. Explain that he/she is allowed to take a step backward to avoid falling. There should be a solid wall behind the examiner, at least 1-2 meters away to allow for the observation of the number of retropulsive steps. The first pull is an instructional demonstration, and is purposely milder and not rated. The second time the shoulders are pulled briskly and forcefully towards the examiner with enough force to displace the center of gravity so that the patient must take a step backwards. The examiner needs to be ready to catch the patient, but must stand sufficiently back so as to allow enough room for the patient to take several steps to recover independently. Observe for the number of steps backward or falling. In situations where it is absolutely impossible to test severity (e.g. in patients that cannot stand) the examiner should use the notation "UR" for Unable to Rate.

0	No problems. Recovers with one or two steps.
1	Slight. Three to 5 steps, but subject recovers unaided.
2	Mild. More than 5 steps, but subject recovers unaided.
3	Moderate. Stands safely, but with absence of postural response; falls if not caught by examiner.
4	Severe. Very unstable, tends to lose balance spontaneously or with just a gentle pull on the shoulders.

4. 7. *RESTING TREMOR AMPLITUDE	**SCORE** ☐

Test the resting tremor that may appear at any time during the exam, including when quietly sitting, during walking, and during activities when some body parts are moving but others are at rest. Score the maximum amplitude that is seen at any time as the final score. Rate only the amplitude and not the persistency or intermittency of the tremor. As part of this rating, the patient should sit quietly in a chair with the hands placed on the arms of the chair comfortably supported on the floor for ten seconds with no other directives. Rate the most affected limb.

0	Normal. No tremor.
1	Slight. Tremor <1 cm in maximal amplitude.

2	Mild. Tremor >1 cm but <3 cm in maximal amplitude.
3	Moderate. Tremor 3 - 10 cm in maximal amplitude.
4	Severe. Tremor >10 cm in maximal amplitude.

SC V. DYSTONIA	
5. 1. UPPER FACE	**SCORE** ☐
Rate ability to maintain eyes open. Rate excessive blinking or prolonged eye closure (blepharospasm).	
0	No dystonia present.
1	Slight. Occasional blinking.
2	Mild. Frequent blinking without prolonged eye closure.
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.
5. 2. EYES	**SCORE** ☐
Rate ability to gaze at the examiner. Rate dystonic eye deviation.	
0	No dystonia present.
1	Slight. Occasional eye deviation.
2	Mild. Frequent eye deviation but not interfering with gaze.
3	Moderate. Prolonged eye deviation, interfering with gaze.
4	Severe. Prolonged spasms of eye deviation, preventing the patient from looking at the examiner.
5. 3. LOWER FACE	**SCORE** ☐
Observe ability to make normal facial expressions during assessment. Rate grimacing, lip pursing.	
0	No dystonia present.
1	Slight. Occasional grimacing or lip pursing.
2	Mild. Grimacing or lip pursing present <50% of the time.
3	Moderate grimacing or lip pursing present most of the time.
4	Severe grimacing or lip pursing present most of the time.
5. 4. JAW AND TONGUE	**SCORE** ☐
Rate ability to chew during eating. Rate jaw-opening, jaw-clenching, tongue protrusion, tongue/lip biting	
0	No dystonia present.

1	Slight. Occasional jaw opening/clenching OR tongue movements.
2	Mild but obvious jaw-opening/clenching not interfering with eating OR mild tongue protrusion present <50% of the time.
3	Moderate jaw-opening/clenching interfering with eating, OR moderate tongue protrusion present most of the time.
4	Severe jaw-opening or jaw-clenching preventing chewing OR severe tongue protrusion present most of the time, including tongue/lip biting.

5. 5. NECK	**SCORE** ☐
Rate ability to maintain the head in midline position during lying, sitting, standing, walking or while performing upper limb motor tasks. Rate also ability to dissociate head to turn i.e. opposite to torticollis. Rate pulling of the neck in any direction	
0	No dystonia present.
1	Slight dystonia. Occasional pulling.
2	Mild but obvious pulling, not disabling.
3	Moderate pulling.
4	Extreme pulling.

5. 6. LEFT ARM	**SCORE** ☐
Rate dystonic posturing of the shoulders, arms and hands. Rate ability to write or grasp pencil/crayon while maintaining arms in normal range. In pre-school children or children with learning difficulties, rate functional bimanual use of hands during play activities. Rate both limbs separately.	
0	No dystonia present.
1	Slight dystonia. Clinically insignificant.
2	Mild but obvious dystonia, but capable of refined manual function.
3	Moderate. Able to grasp, capable of crude manual function.
4	Severe. No useful grasp.

5. 7. RIGHT ARM	**SCORE** ☐
0	No dystonia present.
1	Slight dystonia. Clinically insignificant.
2	Mild but obvious dystonia, but capable of refined manual function.

3	Moderate. Able to grasp, capable of crude manual function.
4	Severe. No useful grasp.

5. 8. TRUNK	**SCORE** ☐
Rate ability to sit, stand, and walk. Rate bending perceived due to active dystonic posturing	
0	No dystonia present.
1	Slight bending. Clinically insignificant (not even noted by patient).
2	Mild but obvious bending, not interfering with sitting, standing, or walking.
3	Moderate bending, interfering with sitting, standing, or walking
4	Severe bending of trunk preventing sitting, standing, or walking.

5. 9. LEFT LEG	**SCORE** ☐
Rate the ability to stand and walk. Rate dystonic posturing of the pelvis, leg, or foot. Torti-pelvis is usually a feature of trunk dystonia but if pelvic dystonia displaces the leg (i.e. into abduction, adduction, or rotation), it is considered to belong to the leg region. Rate both limbs separately.	
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	Unable to stand or walk on involved leg.

5. 10. RIGHT LEG	**SCORE** ☐
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	Unable to stand or walk on involved leg.

SC VI. OTHER NEUROLOGIC SIGNS	
6. 1. SPEECH	**SCORE** ☐
Ask questions regarding activities of daily living.	
0	Normal. No speech problems.

1	Slightly involved, no need to repeat to be understood.
2	Some difficulty in understanding speech, must repeat to be understood.
3	Marked difficulty in understanding speech, mostly incomprehensible.
4	Complete or almost complete anarthria

6. 2. GAIT	**SCORE** ☐
Make the patient walk away from and toward the examiner. The patient should walk at least 6 meters, then turn around and return to the examiner.	
0	Normal. No problems.
1	Slight. Independent walking with minor gait impairment.
2	Mild. Independent walking, but with substantial gait impairment.
3	Moderate. Requires an assistance device for safe walking (walking stick, walker) but not a person.
4	Severe. Cannot walk at all or only with another person's assistance.

6. 3. ╪CHOREA	**SCORE** ☐
Observe face, mouth, trunk and extremities during the physical exam. Rate the maximal chorea according to severity and duration during the time of assessment.	
0	Absent
1	Slight/intermittent
2	Mild/common or moderate/intermittent
3	Moderate/common
4	Marked/prolonged

6. 4. SPASTICITY	**SCORE** ☐
Judge on passive movement of major joints with patient relaxed in a sitting position; ignore cog wheeling. When testing a muscle that primarily flexes a joint (elbow and wrist in upper limb, knee in lower limb), place the joint in a maximally flexed position and move to a position of maximal extension over one second (count "one thousand one"). When testing a muscle that primarily extends a joint (ankle), place the joint in a maximally extended position and move to a position of maximal flexion over one second (count "one thousand one"). Rate the most affected joint. Only live rating is used for this test.	
0	No increase in muscle tone due to spasticity.
1	Slight increase in muscle tone, manifested by a catch and release or by minimal resistance at the

	end of the range of motion when the affected part(s) is moved in flexion or extension.
2	More marked increase in muscle tone through most of the range of movement, but affected part(s) easily moved.
3	Considerable increase in muscle tone. Passive movement difficult.
4	Affected part(s) rigid in flexion or extension.

6. 5. *ACTION OR POSTURAL TREMOR	**SCORE** ☐
Instruct the patient to stretch the arms out in front of the body with palms down. Observe this posture for 10 seconds. Rate the most affected area and the highest amplitude seen.	
0	Normal. No tremor.
1	Slight. Tremor is present, but less than 1 cm in amplitude.
2	Mild. Tremor is at least 1, but less than 3 cm in amplitude.
3	Moderate. Tremor is at least 3, but less than 10 cm in amplitude.
4	Severe. Tremor is at least 10 cm in amplitude.

6. 6. ┼OCULOMOTOR DYSFUNCTION	**SCORE** ☐
Eye movements are examined by asking the subject to follow slow horizontal finger movements of the examiner, to look laterally at the finger at different positions, and to perform saccades between two fingers in the vertical and horizontal level. In situations where it is absolutely impossible to test ocular movements (e.g. severe dystonic eye deviation, poor visual acuity, blepharospasm) the examiner should use the notation "UR" for Unable to Rate." The examiner assesses the following abnormal signs: 1) Broken-up smooth pursuit. 2) Gaze-evoked nystagmus. 3) Hypometric and slow horizontal saccades 4) Hypometric and slow vertical saccades 5) Strabismus	
0	None.
1	One abnormal ocular motor sign.
2	Two abnormal ocular motor signs.
3	Three abnormal ocular motor signs.
4	Four or more abnormal ocular motor signs.

* Items 1, 4.1, 4.2, 4.3, 4.4, 4.5, 4.6, 4.7 and 6.5 of the PKAN-DRS contain materials, both unchanged and modified, from the MDS-UPDRS with permission of the scale owner, the International Parkinson and Movement Disorder Society. ┼ Item 6.6 contains material from the UMSARS. ╪ Items 2 and 6.3 of the PKAN-DRS include materials from the Unified Huntington's Disease Rating Scale (UHDRS), which is owned exclusively by the Huntington Study Group, Ltd. (HSG). Materials from the UHDRS Behavioral Examination and chorea are included in the PKAN-DRS with permission from HSG.

APPENDIX 2.

PKAN-DRS video scoring protocol (8 segments)

Setting: - Camera to be set up on tripod in front of the patient. - Patient to be positioned at sufficient distance that patient's whole body can be captured in frame. - Scorers to be seated on either side of the camera to encourage midline posture and visual direction. - During seating position: to be seated 90:90 with feet supported. Remove straps and harnesses. Remove shoes and socks, trousers rolled up. - Video to be scored in segments as below. Endeavor to keep to the timings so that scoring is based on standardized clips of similar duration.	
1. Sitting at rest, with arms resting loosely on thighs: - Eyes open, eyes closed, forced eye blinks. - Tongue protrusion, open and close mouth 5 repetitions. - Turn head to right, then to left, then up and down, then tilt ear to shoulder on each side. - Rate rigidity and spasticity in the four limbs (proximal and distal limbs)	2 minutes
2. Ocular movements (zoom in to get a close view): - Rate smooth pursuit and vertical and horizontal saccades.	2 minutes
3. Speech: Ask questions regarding activities of daily living.	1 minute
4. Limb movements: - Stretch both arms out in front of the body. Both arms extended supinated, then pronated. - Place both arms flexed at elbow in front of chest. - Tap the index finger with the thumb 10 times as quickly and with as big a range of motion as	2 minutes

possible. Rate each arm separately. - Place the foot on the ground and then raise, and stomp the foot on the ground 10 times as high and as fast as possible. Rate each leg separately.	
5. Writing/drawing: - Patient seated with a height adjustable table positioned in front. The patient's arms must rest with elbows flexed at 90º. Place a single piece of paper at midline with pen to be placed to the same side of the paper as arm being tested. Allow 30 seconds for patient to attempt to pick up pen and, once grasped, up to 30 seconds to make their marks/drawings/writing on the paper. If patient is unable to pick pen up, score to offer pen held vertically to one side of the paper. If patient is unable to grasp pen held vertically, scorer to place pen in patient's hand. Always score each arm separately.	2 minutes
6. Standing and balance: - In standing, turn 90º facing forward, left, backward, right and forward again with the arms aside, and observe posture. Caregiver's assistance can be provided for balance support. - Test retropulsion.	2 minutes
7. Gait: Walk back and forth to and from the camera, at a distance where whole body is captured in the frame.	2 minutes
8. Feeding and eating-swallowing: - While seated, ask the patient to eat a biscuit	1 minute

