PhenX Toolkit

Data Collection Worksheet

Please Note: The Data Collection Worksheet (DCW) is a tool to aid integration of a PhenX protocol into a study. The PhenX DCW is not designed to be a data collection instrument. Investigators will need to decide the best way to collect data for the PhenX protocol in their study. Variables captured in the DCW, along with variable names and unique PhenX variable identifiers, are included in the PhenX Data Dictionary (DD) files.

The Newcastle Mitochondrial Disease Adult Scale (NMDAS)

Name: _____

Date of birth: _____

Date of assessment: _____

1048576 **Height:** _____

Forced Vital Capacity (FVC):

1048576 **FVC - 1st attempt** _____

1048576 **FVC - 2nd attempt** _____

1048576 **FVC - 3rd attempt** _____

Section I- Current Function

Rate function over the preceding **4 week period, according to patient and/or caregiver** interview only. The clinician's subjective judgement of functional ability should **not** be taken into account.

1. **Vision** with usual glasses or contact lenses

    0. Normal.
    1. No functional impairment but aware of worsened acuities.
    2. Mild - difficulty with small print or text on television.
    3. Moderate - difficulty outside the home (eg bus numbers, road signs or shopping).
    4. Severe - difficulty recognising faces.
    5. Unable to navigate without help (eg carer, dog, cane).

2. **Hearing** with or without hearing aid

0. Normal.
1. No communication problems but aware of tinnitus **or** deterioration from prior ‘normal’ hearing.
2. Mild deafness (eg missing words in presence of background noise). **Fully** corrected with hearing aid.
3. Moderate deafness (eg regularly requiring repetition). **Not fully** corrected with hearing aid.
4. Severe deafness - poor hearing even with aid (see 3 above).
5. End stage - virtually no hearing despite aid. Relies heavily on non-verbal communication (eg lip reading) **or** has cochlear implant.

3. **Speech**

0. Normal.
1. Communication unaffected but patient or others aware of changes in speech patterns or quality.
2. Mild difficulties - usually understood and **rarely** asked to repeat things.
3. Moderate difficulties - poorly understood by strangers and **frequently** asked to repeat things.
4. Severe difficulties - poorly understood by family or friends.
5. Not understood by family or friends. Requires communication aid.

4. **Swallowing**

0. Normal.
1. Mild - sensation of solids ‘sticking’ (occasional).
2. Sensation of solids ‘sticking’ (most meals) **or** need to modify diet (eg avoidance of steak/salad).
3. Difficulty swallowing solids - affecting meal size or duration. Coughing, choking **or** nasal regurgitation infrequent (1 to 4 times per month) but more than peers.
4. Requires adapted diet - regular coughing, choking, **or** nasal regurgitation (more than once per week).
5. Requiring enteral feeding (eg PEG).

5. **Handwriting**

0. Normal.
1. Writing speed unaffected but aware of increasing untidiness.
2. Mild - Has to write slower to maintain tidiness/legibility.
3. Moderate - Handwriting takes at least twice as long **or** resorts to printing (must previously have used joined writing).
4. Severe - Handwriting mostly illegible. Printing very slow and untidy (eg ‘THE BLACK CAT’ takes in excess of 30 seconds).
5. Unable to write. No legible words.

6. **Cutting food and handling utensils** (irrespective of contributory factors - eg weakness, coordination, cognitive function etc. This is also true for questions 7-10)

0. Normal.
1. Slightly slow and/or clumsy but minimal effect on meal duration.
2. Slow and/or clumsy with extended meal duration, but no help required.
3. Difficulty cutting up food and inaccuracy of transfer pronounced. Can manage alone but avoids problem foods (eg peas) or carer typically offers minor assistance (eg cutting up steak).
4. Unable to cut up food. Can pass food to mouth with great effort or inaccuracy. Resultant intake minimal. Requires major assistance.
5. Needs to be fed.

7. **Dressing**

0. Normal.
1. Occasional difficulties (eg shoe laces, buttons etc) but no real impact on time or effort taken to dress.
2. Mild - Dressing takes longer and requires more effort than expected at the patient's age. No help required.
3. Moderate - Can dress unaided but takes at least twice as long and is a major effort. Carer typically helps with difficult tasks such as shoe laces or buttons.
4. Severe - Unable to dress without help but some tasks completed unaided.
5. Needs to be dressed.

8. **Hygiene**

0. Normal.
1. Occasional difficulties only but no real impact on time or effort required.
2. Mild - hygienic care takes longer but quality unaffected.
3. Moderate - bathes and showers alone with difficulty or needs bath chair / modifications. Dextrous tasks (eg brushing teeth, combing hair) performed poorly.
4. Severe - unable to bathe or shower without help. Major difficulty using toilet alone. Dextrous tasks require help.
5. Dependent upon carers to wash, bathe, and toilet.

9. **Exercise Tolerance**

0. Normal.
1. Unlimited on flat - symptomatic on inclines or stairs.
2. Able to walk < 1000m on the flat. Restricted on inclines or stairs - rest needed after 1 flight (12 steps).
3. Able to walk < 500m on the flat. Rest needed after 8 steps on stairs.
4. Able to walk < 100m on the flat. Rest needed after 4 steps on stairs.

5. Able to walk < 25m on the flat. Unable to do stairs alone.

10. **Gait stability**

0. Normal.
1. Normal gait - occasional difficulties on turns, uneven ground, or if required to balance on narrow base.
2. Gait reasonably steady. Aware of impaired balance. **Occasionally** off balance when walking.
3. Unsteady gait. **Always** off balance when walking. **Occasional** falls. Gait steady with support of stick or person.
4. Gait grossly unsteady without support. **High likelihood** of falls. Can only walk short distances (< 10m) without support.
5. Unable to walk without support. Falls on standing.

Section II - System Specific Involvement

Rate function according to patient and/or caregiver interview and consultation with the medical notes. Each inquiry should take into account the situation for the preceding **12 month period** only, unless otherwise stated in the question.

1. **Psychiatric**

0. None.
1. Mild & transient (eg reactive depression) - lasting **less** than 3 months.
2. Mild & persistent (lasting **more** than 3 months) **or** recurrent. Patient has consulted GP.
3. Moderate & warranting specialist treatment (e.g. from a psychiatrist) - eg. bipolar disorder or depression with vegetative symptoms (insomnia, anorexia, abulia etc).
4. Severe (eg self harm - psychosis etc).
5. Institutionalised or suicide attempt.

2. **Migraine Headaches** During **the last 3 months**, how many days have headaches prevented the patient from functioning normally at school, work, or in the home?

0. No past history.
1. Asymptomatic but past history of migraines.
2. One day per month.
3. Two days per month.
4. Three days per month.
5. Four days per month or more.

3. **Seizures**

0. No past history.

1. Asymptomatic but past history of epilepsy.
2. Myoclonic or simple partial seizures only.
3. Multiple absence, complex partial, or myoclonic seizures affecting function or single generalized seizure.
4. Multiple generalised seizures.
5. Status epilepticus.

4. **Stroke-like episodes** (exclude focal deficits felt to be of vascular aetiology)

0. None.
1. Transient focal sensory symptoms only (less than 24 hours).
2. Transient focal motor symptoms only (less than 24 hours).
3. Single stroke-like episode affecting one hemisphere (more than 24 hours).
4. Single stroke-like episode affecting both hemispheres (more than 24 hours).
5. Multiple stroke-like episodes (more than 24 hours each).

5. **Encephalopathic Episodes**

0. No past history.
1. Asymptomatic **but** past history of encephalopathy.
2. Mild - single episode of personality or behavioural change but retaining orientation in time/place/person.
3. Moderate - single episode of confusion or disorientation in time, place or person.
4. Severe - multiple moderate episodes (as above) **or** emergency hospital admission due to encephalopathy **without** associated seizures or stroke-like episodes.
5. Very severe - in association with seizures, strokes or gross lactic acidaemia.

6. **Gastro-intestinal symptoms**

0. None.
1. Mild constipation only **or** past history of bowel resection for dysmotility.
2. Occasional symptoms of ‘irritable bowel’ (pain, bloating or diarrhoea) with long spells of normality.
3. Frequent symptoms (as above) most weeks **or** severe constipation with bowels open less than once/week **or** need for daily medications.
4. Dysmotility requiring admission **or** persistent and/or recurrent anorexia/vomiting/weight loss.
5. Surgical procedures **or** resections for gastrointestinal dysmotility.

7. **Diabetes mellitus**

0. None
1. Past history of gestational diabetes or transient glucose intolerance related to intercurrent illness.

2. Impaired glucose tolerance (in absence of intercurrent illness).
3. NIDDM (diet).
4. NIDDM (tablets).
5. DM requiring insulin (irrespective of treatment at onset).

8. **Respiratory muscle weakness**

0. FVC normal (≥ 85% predicted).
1. FVC < 85% predicted.
2. FVC < 75% predicted.
3. FVC < 65% predicted.
4. FVC < 55% predicted.
5. FVC < 45% predicted **or** ventilator support for over 6 hours per 24 hr period (**not** for OSA alone).

9. **Cardiovascular system**

0. None.
1. Asymptomatic ECG change.
2. Asymptomatic LVH on echo **or** non-sustained brady/tachyarrhythmia on ECG.
3. Sustained or **symptomatic** arrhythmia, LVH **or** cardiomyopathy. Dilated chambers or reduced function on echo. Mobitz II AV block or greater.
4. Requires pacemaker, defibrillator, arrhythmia ablation, **or** LVEF < 35% on echocardiogram.
5. Symptoms of left ventricular failure **with** clinical and/or x-ray evidence of pulmonary oedema **or** LVEF < 30% on echocardiogram.

Section III - Current Clinical Assessment

Rate current status according to examination performed at **the time of** assessment

1. **Visual acuity** with usual glasses, contact lenses or pinhole.

0. CSD ≤ **12** (ie normal vision - 6/6, 6/6 or better).
1. CSD ≤ **18** (eg 6/9, 6/9).
2. CSD ≤ **36** (eg 6/12, 6/24).
3. CSD ≤ **60** (eg 6/24, 6/36).
4. CSD ≤ **96** (eg 6/60, 6/36).
5. CSD ≥ **120** (eg 6/60, 6/60 or worse).

2. **Ptosis**

0. None.
1. Mild ptosis - not obscuring **either** pupil.
2. Unilateral ptosis obscuring < 1/3 of pupil.
3. Bilateral ptosis obscuring < 1/3 **or** unilateral ptosis obscuring > 1/3 of pupil

or prior unilateral surgery.
4. Bilateral ptosis obscuring > 1/3 of pupils **or** prior bilateral surgery.
5. Bilateral ptosis obscuring >2/3 of pupils **or** >1/3 of pupils **despite** prior bilateral surgery.

3. **Chronic Progressive External Ophthalmoplegia**

0. None.
1. Some restriction of eye movement (any direction). Abduction complete.
2. Abduction of worst eye incomplete.
3. Abduction of worst eye below 60% of normal.
4. Abduction of worst eye below 30% of normal.
5. Abduction of worst eye minimal (flicker).

4. **Dysphonia/Dysarthria**

0. None.
1. Minimal - noted on examination only.
2. Mild - clear impairment but easily understood.
3. Moderate - some words poorly understood and infrequent repetition needed.
4. Severe - many words poorly understood and frequent repetition needed.
5. Not understood. Requires communication aid.

5. **Myopathy**

0. Normal.
1. Minimal reduction in hip flexion and/or shoulder abduction **only** (eg MRC 4+/5).
2. Mild but clear proximal weakness in hip flexion and shoulder abduction (MRC 4/5). Minimal weakness in elbow flexion and knee extension (MRC 4+/5 - both examined with joint at 90 degrees).
3. Moderate proximal weakness including elbow flexion & knee extension (MRC 4/5 or 4 -/5) **or difficulty** rising from a 90 degree squat.
4. Waddling gait. **Unable** to rise from a 90 degree squat (=a chair) unaided.
5. Wheelchair dependent **primarily** due to proximal weakness.

6. **Cerebellar ataxia**

0. None.
1. Normal gait but hesitant heel-toe.
2. Gait reasonably steady. Unable to maintain heel-toe walking **or** mild UL dysmetria.
3. Ataxic gait (but walks unaided) **or** UL intention tremor & past-pointing. Unable to walk heel-toe - falls immediately.
4. Severe - gait grossly unsteady without support **or** UL ataxia sufficient to affect feeding.

5. Wheelchair dependent **primarily** due to ataxia **or** UL ataxia **prevents** feeding.

7. **Neuropathy**

0. None.
1. Subtle sensory symptoms **or** areflexia.
2. Sensory impairment only (eg glove & stocking sensory loss).
3. Motor impairment (distal weakness) **or** sensory ataxia.
4. Sensory ataxia **or** motor effects severely limit ambulation.
5. Wheelchair bound **primarily** due to sensory ataxia or neurogenic weakness.

8. **Pyramidal Involvement**

0. None.
1. Focal or generalised increase in tone or reflexes only.
2. Mild **focal** weakness, sensory loss or fine motor impairment (eg cortical hand).
3. Moderate hemiplegia allowing unaided ambulation **or** dense UL monoplegia.
4. Severe hemiplegia allowing ambulation with aids **or** moderate tetraplegia (ambulant).
5. Wheelchair dependant **primarily** due to hemiplegia or tetraplegia.

9. **Extrapyramidal**

0. Normal.
1. Mild and unilateral. Not disabling (H&Y stage 1).
2. Mild and bilateral. Minimal disability. Gait affected (H&Y stage 2).
3. Moderate. Significant slowing of body movements (H&Y stage 3)
4. Severe. Rigidity and bradykinesia. Unable to live alone. Can walk to limited extent (H&Y stage 4).
5. Cannot walk or stand unaided. Requires constant nursing care (H&Y stage 5).

10. **Cognitition**

Patients undergo testing using WTAR, Symbol Search and Speed of Comprehension Test.

0. Combined centiles **100 or more.**
1. Combined centiles **60 - 99**
2. Combined centiles **30 - 59**
3. Combined centiles **15 - 29**
4. Combined centiles **5 - 14**
5. Combined centiles **4 or below.**

Protocol source: https://www.phenxtoolkit.org/protocols/view/220704