



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 Paediatric Mitochondrial Disease Scale (NPMDS)

12 - 18 years

Date of assessment:

Age at assessment:

Parental consanguinity:

Age at presentation:

Age at clinical diagnosis:

Clinical diagnosis:

Genotype if known:

Biochemical phenotype if known:

Basis of clinical diagnosis e.g. MRI, blood / CSF lactate

Information regarding pregnancy

- reduced fetal movements _____
- cardiomyopathy on antenatal scans _____
- abnormalities on fetal anomaly scan _____
- other:

Neonatal information:

- gestational age _____

- delivery method (NVD vs instrumental vs C/S) _____
- birth weight _____
- resuscitation and ventilation _____

Scores: Sections I-III:

Section IV:

Section I: Function

Rate function during the **preceding 4 week period** only according to patient and / or caregiver interview. Indicate the score that best fits patient's functional status independently of the nature of the signs.

1. Vision (with usual glasses)

0. **Normal.** No parental / patient concerns
1. **Mild.** Difficulty with small print or text on the television
2. **Moderate.** Difficulty outside the home (e.g. bus numbers, road signs or shopping)
3. **Severe.** Difficulty recognising faces or unable to navigate without help or registered blind

2. Hearing

0. **Normal**
1. **Mild.** Mild deafness (e.g. missing words in presence of background noise)
2. **Moderate.** Hearing impaired but fully corrected with hearing aid
3. **Severe.** Poor hearing even with aid or reliant on non-verbal communication

3. Communication

0. **Normal**
1. **Mild.** Mild speech difficulties but easily understood and rarely asked to repeat things
2. **Moderate.** Speech poorly understood by strangers and frequently asked to repeat things
3. **Severe.** Speech not understood by family or friends and / or requires communication aid

4. Feeding

0. **Normal**
1. **Mild.** Choking / vomiting / anorexia resulting in reduced intake or swallowing difficulties requiring dietary adaptations

2. **Moderate.** Supplementary enteral feeding or recurrent aspiration pneumonia
 3. **Severe.** Exclusive enteral feeding (gastrostomy / NG tube). Nil by mouth
5. **Self-care** (personal hygiene, dressing, utensil use e.g. for feeding)
0. **Normal**
 1. **Mild.** Slow and / or clumsy self-care requiring extra time or effort. Requires help with *some* tasks
 2. **Moderate.** Requires help with *most* tasks but retains some independent selfcare function
 3. **Severe.** Requires one to one assistance
6. **Mobility**
0. **Normal**
 1. **Mild.** Difficulty walking up stairs or inclines
 2. **Moderate.** Requires support (stick / frame / callipers) to walk on the flat
 3. **Severe.** Wheelchair dependent
7. **Educational Achievement**
0. **Normal.** Academic achievement comparable to peers in mainstream school
 1. **Mild.** Struggling to remain in mainstream school
 2. **Moderate.** Attending special school
 3. **Severe.** Not attending school *primarily due to illness*

Section II: System Specific Involvement

Rate system specific involvement during the **preceding 12 month period** only according to patient and / or caregiver, clinician's knowledge **and** clinical notes.

1. **Seizures**
 0. **None**
 1. **Mild.** Myoclonic or absence seizures only or < 1 generalised tonic-clonic seizure each month
 2. **Moderate.** > 5 generalized tonic-clonic seizures/month or > 20 absence or myoclonic seizures/month
 3. **Severe.** Status epilepticus
2. **Encephalopathy**
 0. **None**
 1. **Mild.** Single episode of personality change, excessive sleepiness, confusion or disorientation
 2. **Moderate.** Obtunded or >2 encephalopathic episodes/year

3. **Severe.** Life-threatening encephalopathy- requires artificial ventilation

3. Stroke-like Episodes

0. **None**

1. **Mild.** Transient motor symptoms lasting < 24 hours

2. **Moderate.** Single stroke-like episode (> 24 hours)

3. **Severe.** Multiple stroke-like episodes (> 24 hours each)

4. Gastrointestinal

0. **Normal**

1. **Mild.** Mild constipation or unexplained vomiting / diarrhoea < 1/week

2. **Moderate.** Moderate constipation (some relief with laxative treatment) or unexplained vomiting / diarrhoea > 3/week

3. **Severe.** Severe constipation (no relief with laxative treatment) or unexplained vomiting / diarrhoea every day or surgical intervention for dysmotility

5. Endocrine

0. **Normal**

1. **Mild.** Biochemical evidence of impaired function

2. **Moderate.** Endocrine failure requiring replacement therapy

3. **Severe.** Endocrine decompensation (e.g. diabetic ketoacidosis, Addisonian crisis)

6. Respiratory

0. **Normal**

1. **Mild.** Abnormal respiration not requiring hospitalization

2. **Moderate.** Abnormal respiration requiring hospitalisation but **not** ventilation

3. **Severe.** Abnormal respiration requiring artificial ventilation

7. Cardiovascular

0. **Normal**

1. **Mild.** Asymptomatic ECG change

2. **Moderate.** Abnormal echocardiogram (e.g. cardiomegaly) or sustained / symptomatic arrhythmia on ECG

3. **Severe.** Decompensated cardiomyopathy or requiring pacing device / defibrillator / ablation

8. Renal

0. **Normal**

1. **Mild.** Impaired function but no change in diet or therapy required

2. **Moderate.** Impaired function requiring restricted protein diet
3. **Severe.** Failure requiring transplant / dialysis

9. Liver

0. **Normal**
1. **Mild.** Mildly impaired Liver Function Tests (LFTs). No symptoms of hepatic failure
2. **Moderate.** Impaired LFTs with symptoms (e.g. jaundice, oedema)
3. **Severe.** Failure requiring hospitalisation and / or transplantation

10. Blood

0. **Normal**
1. **Mild.** Anaemia only
2. **Moderate.** Asymptomatic pancytopenia
3. **Severe.** Pancytopenia requiring regular transfusion / transplantation

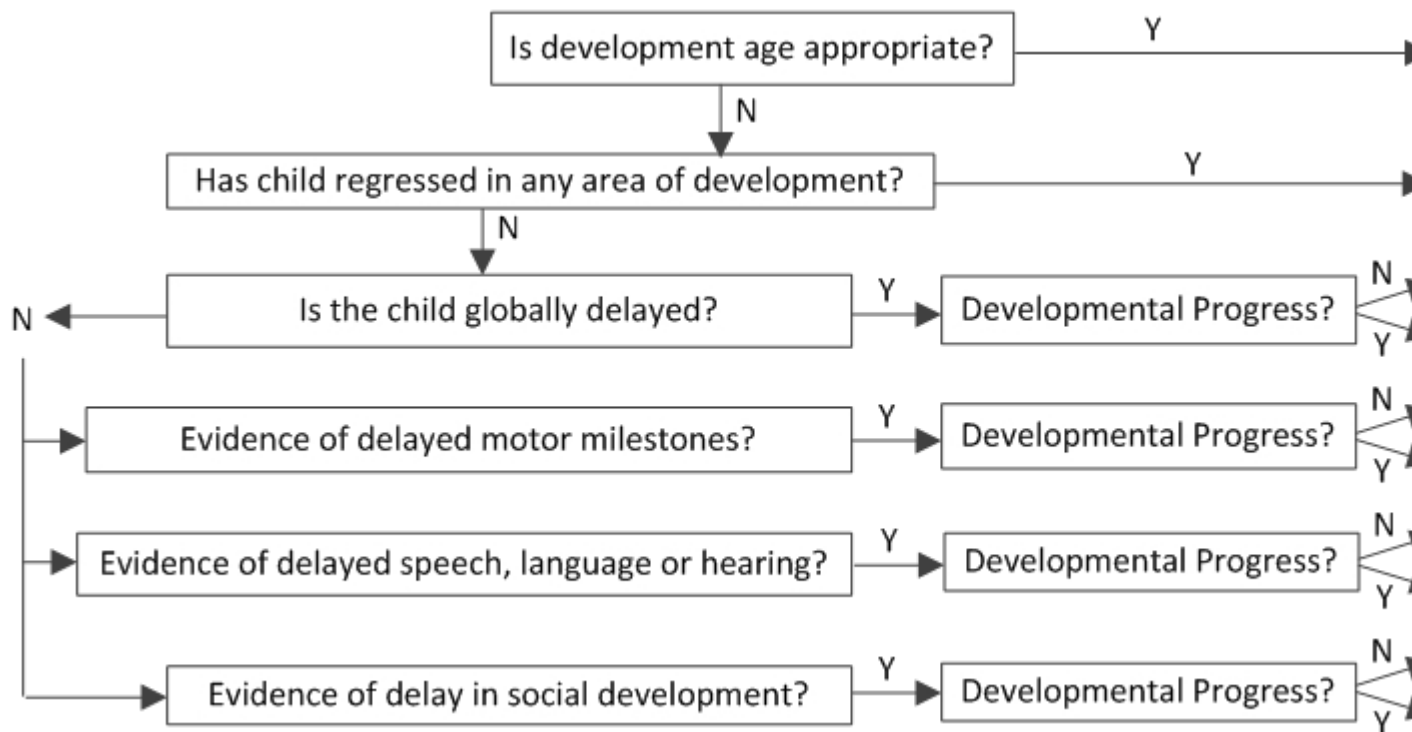
Section III: Current Clinical Assessment

Rate current status according to the clinician's examination performed **at the time of assessment** unless otherwise stated in the question.

1. Growth (ht and weight) over preceding 6 months

0. **Normal.** Following normal growth trajectory
1. **Mild.** Height or weight or both less than 2nd centile but growing parallel to it
2. **Moderate.** Height or weight or both crossing one centile
3. **Severe.** Height or weight or both crossing ≥ 2 centiles or less than 2nd centile with divergent trajectory

2. Development over preceding 12 months Score:_____



3. Vision with usual glasses. Acuity is based on vision in the **better** eye

0. **Normal.** Visual acuity better than or equal to 6/12 or normal fixation and tracking
1. **Mild.** Acuity worse than 6/12 but better than or equal to 6/18 or no fixation on small objects
2. **Moderate.** Acuity worse than 6/18 but better than or equal to 6/60 or impaired fixation on large brightly coloured objects
3. **Severe.** Acuity worse than 6/60 or no response to light or visual threat or unable to finger count

4. Ptoxis and Eye Movement

0. **Normal**
1. **Mild.** Gaze evoked nystagmus or unilateral ptosis or impaired eye movement at extremities
2. **Moderate.** Intermittent nystagmus at rest or bilateral ptosis not obscuring pupils or restriction of >50% eye movement
3. **Severe.** Continuous nystagmus at rest or bilateral ptosis obscuring pupils or only a flicker of eye movement

5. Myopathy

0. **Normal**
1. **Mild.** Mild symmetrical weakness of hip and / or shoulder girdle only
2. **Moderate.** Moderate symmetrical weakness (proximal > distal) limiting

mobility

3. **Severe.** Wheelchair dependent or respiratory compromise *due to myopathy*

6. Ataxia

0. **Normal**

1. **Mild.** Ataxic gait but walks unaided or mild upper limb dysmetria
2. **Moderate.** Gait abnormality requiring assistance or severe upper limb dysmetria
3. **Severe.** Wheelchair dependent or unable to feed *due to ataxia*

7. Pyramidal

0. **Normal**

1. **Mild.** Mild hemiplegia allowing unaided ambulation
2. **Moderate.** Moderate hemiplegia allowing ambulation with aids
3. **Severe.** Wheelchair dependent *due to hemi / tetraplegia*

8. Extrapyramidal

0. **Normal**

1. **Mild.** Focal dystonia or unilateral extrapyramidal tremor / bradykinesia
2. **Moderate.** Generalised dystonia or bilateral extrapyramidal tremor / bradykinesia
3. **Severe.** Wheelchair dependent *due to extrapyramidal disorder*

9. Neuropathy

0. **Normal.**

1. **Mild.** Areflexia only
2. **Moderate.** Sensory ataxia or motor impairment (distal weakness) but mobile
3. **Severe.** Reliant on mobility aids primarily due to neuropathy

Section IV: Quality of Life- Parent Completed

This survey asks for your views about your child's recent health. Please answer every question by marking an 'x' in the box next to the phrase which best describes your answer.

- 1) During the **past 4 weeks**, how would you rate your child's overall health?

Very poor

Poor

Fair

Good

Very good

2) During the **past 4 weeks**, how much did your child's physical health problems limit

their physical activities (such as walking or playing with friends)?

Very much

Quite a lot

Somewhat

A little

Not at all

3) During the **past 4 weeks**, how much did your child's health problems limit their everyday life (such as attending school, caring for themselves) compared with a person of the same age in perfect health?

Very much

Quite a lot

Somewhat

A little

Not at all

4) During the **past 4 weeks**, how much energy did your child have?

None

A little

Some

Quite a lot

Very much

5) During the **past 4 weeks**, how much bodily pain/discomfort did your child have?

Very much

Quite a lot

Some

A little

None

6) During the **past 4 weeks**, how much was your child bothered by emotional problems?

Very much

Quite a lot

Somewhat

A little

Not at all

7) During the **past 4 weeks**, how satisfied did your child feel with their abilities, looks,

relationships with other people and life in general?

Very dissatisfied

Quite dissatisfied

Neither dissatisfied or satisfied

Quite satisfied

Very satisfied

8) During the **past 4 weeks**, how would you rate your child's behaviour compared with other children his / her age?

Very poor

Poor

Fair

Good

Very good

9) During the **past 4 weeks**, how would you rate your child's ability to interact with

other people (e.g. making friends, talking to other children / strangers) compared

with

other children his / her age?

Very poor

Poor

Fair

Good

Very good

10) During the **past 4 weeks**, how much were you (the parent / carer) bothered by emotional problems (e.g. feelings of anxiety, sadness) as a **result of your child's illness?**

Very

Quite a lot

Somewhat

A little

Not at all

11) During the **past 4 weeks**, how much was your time limited as a **result of your child's illness?**

Very

Quite a lot

Somewhat

A little

Not at all

12) During the **past 4 weeks**, how much were your family's activities limited or interrupted as a **result of your child's illness?**

Very

Quite a lot

Somewhat

A little

Not at all

13) During the **past 6 months**, what has been the financial cost of your child's illness?

Very expensive

Quite expensive

Moderately expensive

Little additional cost

No additional cost

14) During the **past 4 weeks**, how would you rate your family's ability to get along with one another?

Very poor

Poor

Fair

Good

Very good

15) During the **past 4 weeks**, how often did your child's illness have a positive effect

on your child, you or your family (e.g. being treated well due to illness, meeting new

people)?

Never

Occasionally

Sometimes

Quite a lot

Most of the time

Section D: Quality of Life- Self Completed

12-16 years

This survey asks about how you have been feeling recently. Please answer every question by marking an 'x' in the box next to the phrase which best describes your answer.

1) During the **past 4 weeks**, how would you rate your overall health?

Very poor

Poor

Fair

Good

Very good

2) During the **past 4 weeks**, how much did your physical health problems limit your

physical activities (such as walking or going out with friends)?

Very much

Quite a lot

Somewhat

A little

Not at all

3) During the **past 4 weeks**, how much did your health problems limit your everyday

life (such as going to school, caring for yourself) compared with a person of your age

in perfect health?

Very much

Quite a lot

Somewhat

A little

Not at all

4) During the **past 4 weeks**, how much energy did you have?

None

A little

Some

Quite a lot

Very much

5) During the **past 4 weeks**, how much pain or discomfort did you have?

Very much

Quite a lot

Some

A little

None

6) During the **past 4 weeks**, how much were you bothered by emotional problems (e.g. feeling sad or frightened)?

Very much

Quite a lot

Somewhat

A little

Not at all

7) During the **past 4 weeks**, how satisfied did you feel with your abilities, looks, relationships with other people and life in general?

Very dissatisfied

Quite dissatisfied

Neither dissatisfied or satisfied

Quite satisfied

Very satisfied

8) During the **past 4 weeks**, how would you rate your behaviour compared with other

people your age?

Very poor

Poor

Fair

Good

Very good

9) During the **past 4 weeks**, how easy did you find it to get on with other people (e.g.

making friends, talking to friends / strangers) compared with other people your age?

Very hard

Hard

OK

Easy

Very easy

10) During the **past 4 weeks**, how would you rate your family's ability to get along with one another?

Very poor

Poor

Fair

Good

Very good

11) During the **past 4 weeks**, how often did your illness have a positive effect on you

or your family (e.g. being treated well because of your illness, meeting new people)?

Never

Occasionally

Sometimes

Quite a lot

Most of the time

Protocol source: <https://www.phenxtoolkit.org/protocols/view/220703>