



## 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)

0 - 24 months

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 \_\_\_\_ Y\_\_\_\_ N\_\_\_\_ unknown\_\_\_\_\_
- cardiomyopathy on antenatal scans \_\_\_\_ Y\_\_\_\_ N\_\_\_\_ unknown\_\_\_\_\_
- abnormalities on fetal anomaly scan \_\_\_\_ Y\_\_\_\_ N\_\_\_\_ unknown\_\_\_\_\_ Please specify \_\_\_\_\_
- other:

Neonatal information:

- gestational age \_\_\_\_\_ weeks

- delivery method (NVD vs instrumental vs C/S) \_\_\_\_\_
- birth weight \_\_\_\_\_ kg
- resuscitation and ventilation \_\_\_\_ Y\_\_\_\_ N\_\_\_\_ unknown\_\_\_\_\_

Please specify\_\_\_\_\_

Scores: Sections I-III:

Section IV:

#### ***Section I: Current Function***

Rate function during the **preceding 2 week period** only according to caregiver interview. Indicate the score that best fits patient's functional status independently of the nature of the signs.

##### **1. Vision**

0. **Normal.** No parental concerns
1. **Mild.** *Limited* eye or head movement to large objects or parental face in visual field
2. **Moderate.** No response to large objects or parental face in the visual field
3. **Severe.** No response to light

##### **2. Hearing**

0. **Normal**
1. **Mild.** Body, head or eye movement only to loud noise
2. **Moderate.** No reaction to loud noise
3. **Severe.** No hearing (even with aid)

##### **3. Communication** (assessed with appropriate regard for age)

0. **Normal.** Age appropriate communication
1. **Mild.** Delayed development of communication
2. **Moderate.** Communication unintelligible to parents or completely reliant on non-verbal communication
3. **Severe.** Not communicating effectively in any form

##### **4. Feeding**

0. **Normal**
1. **Mild.** Difficulties in sucking / coughing / anorexia / wheezy with feeds or occasional choking
2. **Moderate.** Supplementary enteral feeding or recurrent aspiration pneumonia

3. **Severe.** Exclusive enteral feeding (gastrostomy / NG tube). Nil by mouth

## 5. Mobility

0. **Normal.** No concerns. Age appropriate mobility
1. **Mild.** Clumsy age appropriate mode of mobility
2. **Moderate.** Mobile but through age inappropriate mode
3. **Severe.** Immobile

## *Section II: System Specific Involvement*

Rate system specific involvement during the **preceding 6 month period** only (or since birth if the child is less than 6 months old) unless otherwise stated in the question. Scores should be assigned according to caregiver interview, clinician's knowledge of the patient **and** clinical notes.

### 1. Seizures

0. **None**
1. **Mild.** Myoclonic or absence seizures only **or** < 1 generalised tonic-clonic seizure/month
2. **Moderate.** > 5 generalized tonic-clonic seizures/month **or** > 20 absence or myoclonic seizures/month
3. **Severe.** Status epilepticus or intractable seizures

### 2. Encephalopathy

0. **None**
1. **Mild.** Abnormal sleepiness / lethargy. Waking only for feeds
2. **Moderate.** Recurrent episodes of mild encephalopathy (> 2/year)
3. **Severe.** Life threatening encephalopathy - requires artificial ventilation

### 3. Gastrointestinal

0. **Normal.**
1. **Mild.** Constipation or unexplained vomiting / diarrhoea > 3/week
2. **Moderate.** Severe constipation (no relief with laxative treatment) **or** unexplained vomiting / diarrhoea every day **or** surgical intervention for dysmotility
3. **Severe** Malabsorption / Failure to thrive

### 4. 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)

## 5. Respiratory

0. **Normal**
1. **Mild.** Abnormal respiratory pattern not requiring therapy / hospitalization
2. **Moderate.** Abnormal respiration requiring oxygen flow or hospitalisation but **not** ventilation
3. **Severe.** Abnormal respiration requiring artificial ventilation

## 6. Cardiovascular- over preceding 12 months

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

## 7. 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

## 8. Liver

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

## 9. Blood

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

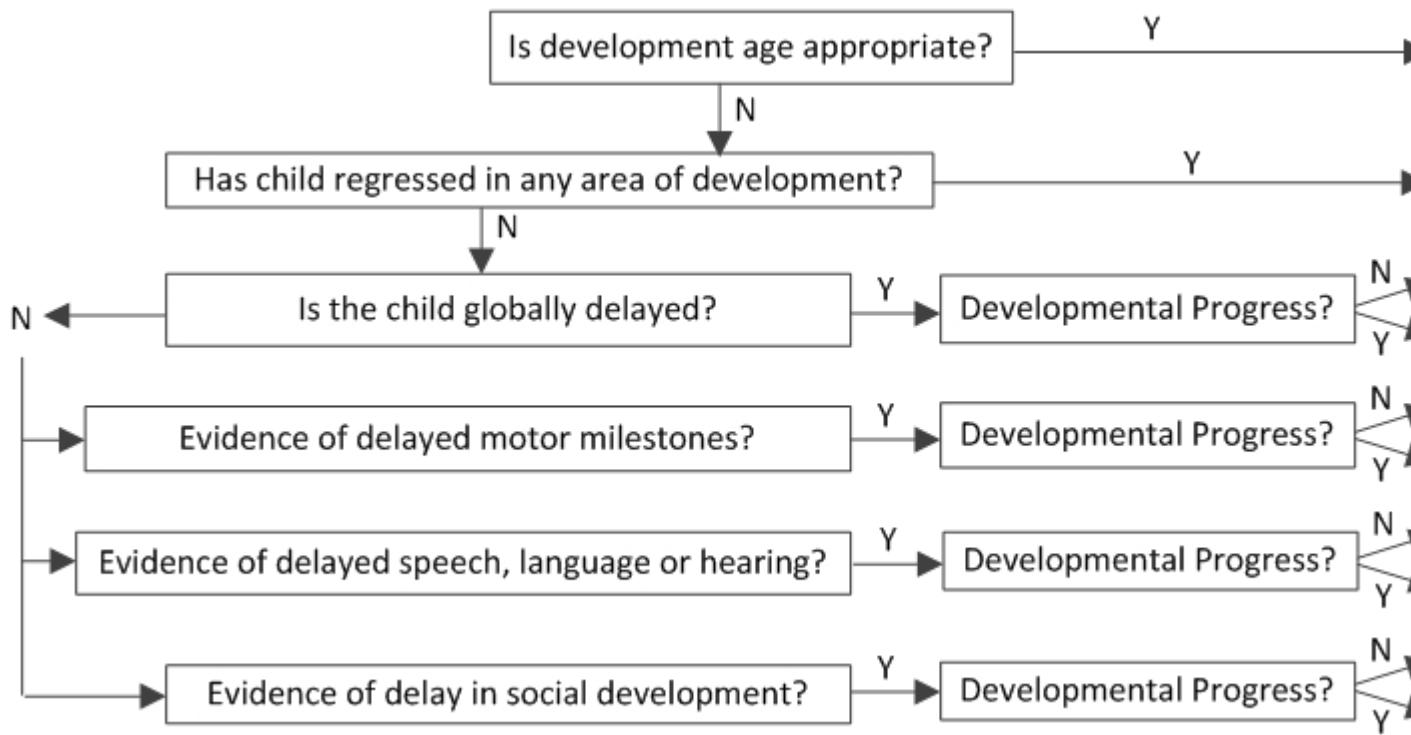
## ***Section III: Current Clinical Assessment***

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

## **1. Growth (weight) over preceding 6 months**

0. **Normal.** Following normal growth trajectory
1. **Mild.** Weight less than second centile but growing parallel to it
2. **Moderate.** Weight crossing one centile
3. **Severe.** Weight crossing  $\geq 2$  centiles **or** less than 2nd centile with divergent trajectory

## **2. Development over preceding 4 months Score: \_\_\_\_\_**



## **3. Vision**

0. **Normal.** Normal fixation and tracking
1. **Mild.** Impaired fixation and / or tracking of small objects
2. **Moderate.** Impaired fixation and / or tracking of familiar faces
3. **Severe.** No response to light or registered blind

## **4. Ptosis 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 functional movement
- 3. **Severe.** Wheelchair / carrier dependent **or** respiratory compromise due to *myopathy*.

## **6. Pyramidal**

- 0. **Normal**
- 1. **Mild.** Unilateral pyramidal signs but retaining functional movement
- 2. **Moderate.** Dense hemiplegia with little movement of affected side
- 3. **Severe.** Bilateral pyramidal weakness with little or no movement

## **7. Extrapyramidal**

- 0. **Normal.**
- 1. **Mild.** Unilateral extrapyramidal posturing and increased tone
- 2. **Moderate.** Bilateral extrapyramidal posturing and increased tone
- 3. **Severe.** Severe extrapyramidal posturing resulting in very little movement

## **8. 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**

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 moving or playing)?

Very much

Quite a lot

Somewhat

A little

Not at all

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

None

A little

Some

Quite a lot

Very much

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

Very much

Quite a lot

Some

A little

None

5) 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

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

with other people (e.g. with you, siblings or other children his / her age) compared with other children his / her age?

[ ] Very poor

[ ] Poor

[ ] Fair

[ ] Good

[ ] Very good

7) 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

8) 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

9) 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

10) 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

11) 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

12) 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

Protocol source: <https://www.phenxtoolkit.org/protocols/view/220701>