1. \*Source from which medical and family history obtained?

Participant  Family, specify relation:  Unknown

2. \*Is the participant adopted?  Yes  No  Unknown

Indicate whether the participant’s first and second degree relatives have a history of the following conditions.

| Condition | Family History? | Relationship of Family Member to Participant  [[1]](#footnote-1)(Choose all that apply from  below list) | Number of Affected  Family Members |
| --- | --- | --- | --- |
| Alzheimer’s disease/ dementia | Yes  No  Unknown | Relationship: | #: |
| Ataxia\* | Yes  No  Unknown | Relationship: | #: |
| Autism\* | Yes  No  Unknown | Relationship: | #: |
| Attention-deficit/hyperactivity disorder (ADHD)\* | Yes  No  Unknown | Relationship: | #: |
| Depression | Yes  No  Unknown | Relationship: | #: |
| Schizophrenia | Yes  No  Unknown | Relationship: | #: |
| Developmental delays\* | Yes  No  Unknown | Relationship: | #: |
| Speech and language delays\* | Yes  No  Unknown | Relationship: | #: |
| Gross motor delays\* | Yes  No  Unknown | Relationship: | #: |
| Fine motor delays\* | Yes  No  Unknown | Relationship: | #: |
| Intellectual disability\* | Yes  No  Unknown | Relationship: | #: |
| Behavioral problems\* | Yes  No  Unknown | Relationship: | #: |
| Cognitive, language, motor and/or social regression\* | Yes  No  Unknown | Relationship: | #: |
| Epilepsy\* | Yes  No  Unknown | Relationship: | #: |
| Febrile seizures | Yes  No  Unknown | Relationship: | #: |
| Recurrent encephalopathy (Leigh syndrome)\* | Yes  No  Unknown | Relationship: | #: |
| Stroke or stroke-like episodes\* | Yes  No  Unknown | Relationship: | #: |
| Microcephaly:  Congenital | Yes  No  Unknown | Relationship: | #: |
| Microcephaly:  Acquired/postnatal onset | Yes  No  Unknown | Relationship: | #: |
| Learning disability\* | Yes  No  Unknown | Relationship: | #: |
| Memory loss\* | Yes  No  Unknown | Relationship: | #: |
| Migraine headaches\* | Yes  No  Unknown | Relationship: | #: |
| Cerebral palsy | Yes  No  Unknown | Relationship: | #: |
| Spasticity | Yes  No  Unknown | Relationship | #: |
| Toe walking | Yes  No  Unknown | Relationship: | #: |
| Muscle disease: Congenital\* | Yes  No  Unknown | Relationship: | #: |
| Muscle disease: Acquired/postnatal onset\* | Yes  No  Unknown | Relationship: | #: |
| Exercise intolerance\* | Yes  No  Unknown | Relationship: | #: |
| Recurrent myoglobinuria\* | Yes  No  Unknown | Relationship: | #: |
| Neuromuscular junction: Congenital | Yes  No  Unknown | Relationship: | #: |
| Neuromuscular junction: Postnatal onset | Yes  No  Unknown | Relationship: | #: |
| Peripheral neuropathy \* | Yes  No  Unknown | Relationship: | #: |
| Mitochondrial disease\* | Yes  No  Unknown | Relationship: | #: |
| Miscarriages and pregnancy-related complications | Yes  No  Unknown | Relationship: | #: |
| Sudden Infant Death Syndrome (SIDS)/Apparent Life-threatening Event (ALTE) | Yes  No  Unknown | Relationship: | #: |
| Child-onset diseases | Yes  No  Unknown | Relationship: | #: |
| Blindness\* | Yes  No  Unknown | Relationship: | #: |
| Ptosis/ophthalmoplegia\* | Yes  No  Unknown | Relationship: | #: |
| Sensorineural hearing loss\* | Yes  No  Unknown | Relationship: | #: |
| Failure to thrive\* | Yes  No  Unknown | Relationship: | #: |
| Short stature\* | Yes  No  Unknown | Relationship: | #: |
| Cardiomyopathy (hypertrophic)\* | Yes  No  Unknown | Relationship: | #: |
| Cardiac conduction block\* | Yes  No  Unknown | Relationship: | #: |
| Liver disease/failure\* | Yes  No  Unknown | Relationship: | #: |
| Renal disease (Fanconi syndrome)\* | Yes  No  Unknown | Relationship: | #: |
| Intestinal pseudo-obstruction\* | Yes  No  Unknown | Relationship: | #: |
| Sideroblastic anemia\* | Yes  No  Unknown | Relationship: | #: |
| Diabetes mellitus\* | Yes  No  Unknown | Relationship: | #: |
| Hypoparathyroidism\* | Yes  No  Unknown | Relationship: | #: |
| Known genetic syndrome | Yes  No  Unknown | Relationship: | #: |
| Other, specify: | Yes  No  Unknown | Relationship: | #: |
| **Pattern of inheritance** | | | |
| Autosomal recessive\* | Yes  No  Unknown | Relationship: | #: |
| Autosomal dominant\* | Yes  No  Unknown | Relationship: | #: |
| X-linked recessive\* | Yes  No  Unknown | Relationship: | #: |
| Maternal inheritance\* | Yes  No  Unknown | Relationship: | #: |
| Sporadic\* | Yes  No  Unknown | Relationship: | #: |

Recorder Signature: Date:

## General Instructions

Information on each disease is gathered for blood relatives based on self-report from the participant or family member.

Important note: Most of the data elements are classified as Core (i.e., strongly recommended for all mitochondrial diease clinical studies to collect) as indicated by asterisks below.

\*Element is classified as Core

The remaining data elements are classified as Supplemental (i.e., non-Core) and should only be collected if the research team considers them appropriate for their study.

Please see the Data Dictionary for element classifications.

## Specific Instructions

Please see the Data Dictionary for definitions for each of the data elements included in this CRF Module.

* Family history – If there is a history of this condition in the first or second degree family, indicate Yes, otherwise choose No, or Unknown.
* Relationship of family member to participant – Select the relationship from the options of the family members listed in the “relationship” of family member to “participant” column. Record/choose more than one family member, if applicable.
* Number of affected family members – Record the total number of family members affected by condition.
* Other condition, specify – If a family member has a condition not listed, specify the condition under "Other".
* Memory loss – Should be considered relative to age-expected norms. This condition should be marked if there is an unexpected or sudden loss of memory, which may or may not be accompanied by neurological deterioration.
* Date/time should be recorded to the level of granularity known (e.g., year, year and month, complete date plus hours and minutes, etc.) and in an unambiguous format acceptable to the study database like DD-MMM-YYYY. When date/time data are prepared for aggregation or sharing, they should be converted to the format specified by [ISO 8601](https://www.iso.org/iso-8601-date-and-time-format.html);  YYYY-MM-DD T:hh:mm:ss.

1. Mother;Father;Full sibling;Half sibling;Child;Maternal grandmother;Paternal grandmother;Maternal grandfather;Paternal grandfather;Maternal aunt;Paternal aunt;Maternal uncle;Paternal uncle;Maternal niece/nephew;Paternal niece/nephew;Grandchild;Maternal cousin;Paternal cousin;Great-grandchild;Other, specify:; [↑](#footnote-ref-1)