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 frombelow list) | Number of AffectedFamily 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)