## Genetic Information

1. What mitochondrial disease diagnosis is present? *(Specify):*
2. Is there a known genetic diagnosis?

[ ]  Yes

[ ]  No

1. Is this a nuclear or mitochondrial DNA variant/deletion?

[ ]  Nuclear

[ ]  Mitochondrial

1. Nuclear variant *(Specify)*:
2. Mitochondrial DNA variant *(Specify)*:
3. Mitochondrial DNA deletion *(Specify)*:

## Informant Information

1. Who is the informant for this questionnaire?

1. How are they related to the person with mitochondrial disease?

[ ]  The person with mitochondrial disease

[ ]  Mother

[ ]  Father

[ ]  Guardian (not a relative)

[ ]  Grandmother

[ ]  Grandfather

[ ]  Sister

[ ]  Brother

[ ]  Spouse

[ ]  Other, specify:

1. Weight in kg *(Specify)*:
2. Height in cm *(Specify)*:
3. Mother’s height in cm *(Specify)*:
4. Father’s height in cm *(Specify)*:
5. Has the participant had any of the following problems?

[ ]  Vomiting

[ ]  GERD, heartburn, mid-line chest pain, esophageal burning

[ ]  Oral regurgitation

[ ]  Burping

[ ]  Difficulty swallowing

[ ]  Pain with swallowing

[ ]  Feeding difficulties

[ ]  Abdominal pain

[ ]  Early satiety (Early fullness)

[ ]  Abdominal bloating or distension

[ ]  Constipation

[ ]  Diarrhea

[ ]  Straining

[ ]  Pain with bowel movements

[ ]  Blood in the stool

[ ]  Poor appetite affecting growth

[ ]  Cyclic vomiting

[ ]  Nausea

[ ]  Post-prandial distress

[ ]  Fecal incontinence

## Specific Questions About How the Participant Eats

1. How does the participant eat? (Choose all that apply)

[ ]  Eats by oneself (regular diet)

[ ]  Infant formula (not specialized)

[ ]  Infant formula (specialized)

[ ]  Liquid diet (non-infant)

[ ]  Gastrostomy (G-tube)

[ ]  Jejunostomy (J-tube)

[ ]  Gastrojejunostomy (G-J tube) gastrojejunostomy is a surgical anastomosis; jejunal feeding tube via gastrostomy

[ ]  Nasogastric tube (NG tube)

[ ]  They do not take any food by mouth or feeding tube

[ ]  TPN (total parenteral nutrition)

1. Name of formula? *(Specify)*: *Please add as much detail as possible.*
2. Why was this formula chosen? (*Specify*):
3. Who recommended this formula?

[ ]  Registered dietician

[ ]  Doctor

[ ]  Internet/social media

[ ]  Parent

[ ]  Other, specify:

1. What liquid diet does the participant use? *(Specify)*:
2. Is the participant currently on a restricted/specialized diet?

[ ]  Yes

[ ]  No

1. What type of specialized diet does the participant eat?

[ ]  Ketogenic diet

[ ]  Celiac diet / gluten free

[ ]  Vegetarian

[ ]  Vegan

[ ]  Organic

[ ]  Food allergen avoidance (shellfish, eggs, nuts, etc.)

[ ]  Lactose free

[ ]  Sucrose free

[ ]  Fructose free

[ ]  Low fat

[ ]  Low carbohydrate

[ ]  Low protein

[ ]  High fat

[ ]  High carbohydrate

[ ]  High protein

[ ]  Low Fermentable Oligo-Di-Monosaccharides and Polyols (Low FODMAP)

[ ]  Enteral feed

[ ]  Other, specify:

1. Why is the participant on a specialized diet? *(Specify)*:
2. Have the participant’s symptoms changed since the diet started? *(Specify)*:
3. Has the participant ever been on a restricted/specialized diet in the past?

[ ]  Yes

[ ]  No

1. What type of specialized diet was the participant on?

[ ]  Ketogenic diet

[ ]  Celiac diet / gluten free

[ ]  Vegetarian

[ ]  Vegan

[ ]  Organic

[ ]  Food allergen avoidance (shellfish, eggs, nuts, etc.)

[ ]  Lactose free

[ ]  Sucrose free

[ ]  Fructose free

[ ]  Low fat

[ ]  Low carbohydrate

[ ]  Low protein

[ ]  High fat

[ ]  High carbohydrate

[ ]  High protein

[ ]  Low Fermentable Oligo-Di-Monosaccharides and Polyols

[ ]  Enteral feed

[ ]  Other, specify:

1. Why did the participant stop?
2. Did the diet affect symptoms?

## Vomiting

1. Does the participant have vomiting currently or in the past?

[ ]  Present

[ ]  Past

[ ]  Never

1. What was the participant’s age when the problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. For the current vomiting episodes, were any of the following triggers present at the time symptoms began?

[ ]  Right after birth

[ ]  Fever

[ ]  Upper respiratory infection

[ ]  Surgery

[ ]  Diarrhea

[ ]  Prolonged fasting (i.e., not eating well)

[ ]  After starting a medicine

[ ]  During exercise

[ ]  After a seizure

[ ]  After trauma

[ ]  History of recent COVID-19 infection

[ ]  I don’t know

[ ]  Other, specify:

1. Are the symptoms pretty consistent from day to day?

[ ]  Yes

[ ]  No

1. How often does the participant vomit?

[ ]  Multiple times a day

[ ]  Once a day

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

1. When the participant has vomiting, is it only a single episode or do they usually vomit repeatedly?

[ ]  Single episode

[ ]  Vomit more than once

[ ]  Vomit repeatedly until there is nothing left

1. Characteristics of vomiting:

[ ]  Effortless regurgitation

[ ]  Projectile or expel regurgitant

1. Does the vomiting happen during a specific time of the day?

[ ]  Morning (wake up until 12pm)

[ ]  Afternoon (12pm - 5pm)

[ ]  Evening (5pm- Bed)

[ ]  Overnight (wakes from sleep)

[ ]  All times of day

[ ]  I don't know

1. Are there particular triggers for vomiting episodes?

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. What triggers vomiting episodes?
2. Do any of the following occur frequently before vomiting starts?

[ ]  Fasting more than 12 hours

[ ]  Viral illness/fever

[ ]  Taking medicine

[ ]  Right after or within 30 minutes of feeding

[ ]  Seizure

[ ]  Headache/migraine/light sensitivity

[ ]  Exercise

[ ]  Stress

1. Does the participant ever have bright green or yellow vomiting?

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. How often has green or yellow vomiting occurred?

[ ]  Only once

[ ]  Less than half of vomiting episodes

[ ]  Most vomiting episodes

[ ]  All vomiting episodes

1. Does the participant ever vomit blood?

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. What color is the blood?

[ ]  Bright red

[ ]  Dark red

[ ]  Brown

[ ]  Black

[ ]  I don’t know

1. Does the participant ever see blood clots in what they vomit?

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. Does the participant have black stool? Note: If the participant answers yes, confirm that stool was really black and not dark brown or dark green.

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. Does the participant have abdominal pain before vomiting?

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. Which symptom is most severe with vomiting episodes?

[ ]  Abdominal pain

[ ]  Nausea

[ ]  Both

1. Please provide additional details about current vomiting symptoms.

## Past Vomiting

1. How old was the participant when they first started having issues with vomiting?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. How old was the participant when the vomiting stopped?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What stopped the vomiting?
2. What did the participant use that failed to stop vomiting?
3. In the past, were the symptoms consistent from day to day?

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. During that period, how often did the participant vomit?

[ ]  Multiple times a day

[ ]  Once a day

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

1. In the past, when the participant had vomiting, was it only a single episode or did the participant usually vomit repeatedly?

[ ]  Single episode

[ ]  Vomited more than once

[ ]  Vomited repeatedly until there was nothing left

1. Did the vomiting happen during a specific time of the day?

[ ]  Morning (wake up until 12pm)

[ ]  Afternoon (12pm - 5pm)

[ ]  Evening (5pm- Bed)

[ ]  Overnight (wakes from sleep)

[ ]  All times of day

[ ]  I don't know

1. In the past, were there any particular triggers for vomiting episodes?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. In the past, what had triggered vomiting episodes?
2. Did any of the following occur frequently before the vomiting started?

[ ]  Fasting more than 12 hours

[ ]  Viral illness/Fever

[ ]  Taking medicine

[ ]  Right after feeding

[ ]  Seizure

[ ]  Headache/migraine/light sensitivity

[ ]  Exercise

[ ]  Stress

1. Did the participant ever have bright green or yellow vomiting?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. How often did the green or yellow vomiting occur?

[ ]  Only once

[ ]  Less than half of vomiting episodes

[ ]  Most vomiting episodes

[ ]  All vomiting episodes

1. Did the participant ever vomit blood?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. What color was the blood?

[ ]  Bright red

[ ]  Dark red

[ ]  Brown

[ ]  Black

[ ]  I don't know

1. Did the participant ever see blood clots in what they vomited?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Did the participant ever have black stools?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Did the participant have abdominal pain before vomiting?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Which symptom was most severe with past vomiting episodes?

[ ]  Abdominal pain

[ ]  Nausea

[ ]  Both

1. In past episodes of vomiting, were any of the following triggers present at the time symptoms began?

[ ]  Right after birth

[ ]  Fever

[ ]  Upper respiratory infection

[ ]  Surgery

[ ]  Diarrhea

[ ]  Prolonged fasting (i.e., not eating well)

[ ]  After starting a medicine

[ ]  During exercise

[ ]  After a seizure

[ ]  After trauma

[ ]  I don’t know

[ ]  Other, specify:

1. Please provide additional details about past vomiting symptoms.

## Cyclic Vomiting or Abdominal Migraine

1. Has the participant ever been diagnosed with cyclic vomiting or abdominal migraine?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Does the participant have a history of migraines?

[ ]  Yes

[ ]  No

1. Does the participant have a family history of migraines?

[ ]  Yes

[ ]  No

1. Has the participant ever experienced vision changes or auras?

[ ]  Vision changes

[ ]  Auras

[ ]  Both

[ ]  Neither

## GERD, Heartburn, Mid-line Chest Pain, Esophageal Burning

1. Has the participant had GERD, heartburn, mid-line chest pain, esophageal burning in the past or currently?

[ ]  Present

[ ]  Past

1. For the current GERD, heartburn, mid-line chest pain, esophageal burning episodes, what was the participant’s age when the problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. Are the symptoms pretty consistent from day to day?

[ ]  Yes

[ ]  No

1. How often does the participant have GERD, heartburn, mid-line chest pain, esophageal burning?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don’t know

1. Timing in relation to meals:

[ ]  Within 5-10 minutes

[ ]  After 20-30 minutes

[ ]  After an hour

1. Does GERD, heartburn, mid-line chest pain, esophageal burning disrupt activity?

[ ]  Yes

[ ]  No

1. Does GERD, heartburn, mid-line chest pain, esophageal burning wake the participant from sleep?

[ ]  Yes

[ ]  No

1. Please provide additional details about the participant’s GERD, heartburn, mid-line chest pain, esophageal burning symptoms.
	1. Triggers:
	2. Relief:
	3. Seasonal variation:

## Past GERD, Heartburn, Mid-line Chest Pain, Esophageal Burning

1. For past GERD, heartburn, mid-line chest pain, esophageal burning episodes, what was the participant’s age when the problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. How old was the participant when the symptoms stopped?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What stopped the GERD, heartburn, mid-line chest pain, and esophageal burning?
2. In the past, were the symptoms pretty consistent from day to day?

[ ]  Yes

[ ]  No

1. How often did the participant have GERD, heartburn, mid-line chest pain, esophageal burning?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don’t know

1. Did past GERD, heartburn, mid-line chest pain, esophageal burning disrupt activity?

[ ]  Yes

[ ]  No

1. Did past GERD, heartburn, mid-line chest pain, esophageal burning wake the participant from sleep?

[ ]  Yes

[ ]  No

1. Please provide additional details about the participant’s past GERD, heartburn, mid-line chest pain, esophageal burning symptoms.

## Oral Regurgitation

1. Does the participant have oral regurgitation or have they had oral regurgitation in the past?

[ ]  Present

[ ]  Past

1. For the current oral regurgitation episodes, what was the participant’s age when the problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. Are the symptoms pretty consistent from day to day?

[ ]  Yes

[ ]  No

1. How often does the oral regurgitation occur?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don’t know

1. Is the oral regurgitation before, during, or after meals?

[ ]  Before

[ ]  During

[ ]  After

[ ]  Variable

1. Please provide additional details about the participant’s oral regurgitation symptoms.

## Past Oral Regurgitation

1. In past oral regurgitation episodes, what was the participant’s age when the problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. How old was the participant when the symptoms stopped?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What stopped the oral regurgitation?
2. Were the symptoms pretty consistent from day to day?

[ ]  Yes

[ ]  No

1. In the past, how often did the oral regurgitation occur?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don’t know

1. In the past, was the oral regurgitation before, during, or after meals?

[ ]  Before

[ ]  During

[ ]  After

[ ]  Variable

1. Please provide additional details about the participant’s past oral regurgitation symptoms.

## Burping

1. Does the participant have burping, or have they had burping in the past?

[ ]  Present

[ ]  Past

1. For the current burping problems, what was the participant’s age when the problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. Is the burping pretty consistent from day to day?

[ ]  Yes

[ ]  No

1. Does the burping occur with reflux or abdominal distension?

[ ]  Reflux

[ ]  Abdominal distension

[ ]  Both

1. Does the burping occur with any specific drinks or food?

[ ]  Yes

[ ]  No

1. What food or drink?
2. Please provide additional details about the participant’s burping symptoms.

## Past Burping

1. For the past burping problems, what was the participant’s age when the problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What was the participant’s age when the burping problem stopped?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What stopped the burping?
2. Was the burping pretty consistent from day to day?

[ ]  Yes

[ ]  No

1. Did the burping occur with reflux or abdominal distension?

[ ]  Reflux

[ ]  Abdominal distension

[ ]  Both

1. Did the burping occur with any specific drinks or food?

[ ]  Yes

[ ]  No

1. What food or drink? *(Specify)*:
2. Please provide additional details about the participant’s past burping symptoms.

## Difficulty and Pain with Swallowing

1. Does the participant have difficulty with swallowing?

[ ]  Yes

[ ]  No

1. What was the participant’s age when difficulty with swallowing began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. Does the difficulty with swallowing occur with liquids or solids?

[ ]  Solids only

[ ]  Liquids and solids

[ ]  Liquids only

[ ]  I don’t know

1. How often do problems with swallowing occur?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don’t know

1. Has the difficulty in swallowing changed over time?

[ ]  Yes

[ ]  No

1. Please provide additional details about the participant’s swallowing problems.
2. Does the participant have pain with swallowing?

[ ]  Yes

[ ]  No

1. What was the participant’s age when pain with swallowing began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. Does the pain with swallowing occur with liquids or solids?

[ ]  Solids only

[ ]  Liquids and solids

[ ]  Liquids only

[ ]  I don’t know

1. How often does pain with swallowing occur?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don't know

1. Does pain with swallowing disrupt activity or wake the participant from sleep?

[ ]  No

[ ]  Pain wakes participant from sleep

[ ]  Pain disrupts activity

[ ]  Pain wakes participant and disrupts activity

[ ]  I don't know

1. Has the pain with swallowing been consistent day to day?

[ ]  Yes

[ ]  No

If NO, has the pain with swallowing:

[ ]  Worsened

[ ]  Improved

1. Please provide additional comments about pain with swallowing.
2. Does the participant feel food get stuck in their esophagus?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Where does the participant feel things get stuck?

[ ]  Back of the throat

[ ]  Neck

[ ]  Upper chest

[ ]  Lower chest

1. Has the participant ever had food or a pill removed from the esophagus?

[ ]  Yes

[ ]  No

1. How many times?
2. Has the participant ever been diagnosed with any of the following:

[ ]  Achalasia

[ ]  Eosinophilic esophagitis

[ ]  Esophageal stricture

## Feeding Difficulties

1. Does the participant have a good appetite?

[ ]  Yes

[ ]  No

1. Has there been a recent change in appetite?

[ ]  Yes

[ ]  No

1. Does the participant have early satiety or get full fast?

[ ]  Yes

[ ]  No

1. Does the participant choke with liquids?

[ ]  Yes

[ ]  No

1. Does the participant choke with solids?

[ ]  Yes

[ ]  No

1. Does the participant’s swallowing change with position changes?

[ ]  Yes

[ ]  No

1. Does the participant cough while eating?

[ ]  Yes

[ ]  No

1. Does the participant get tired while eating?

[ ]  Yes

[ ]  No

1. Is the participant distracted while eating?

[ ]  Yes

[ ]  No

1. Does the participant have difficulty breathing while eating?

[ ]  Yes

[ ]  No

1. Does the participant drool?

[ ]  Yes

[ ]  No

1. Does the participant have a change in voice with eating?

[ ]  Yes

[ ]  No

1. Has the participant received a diagnosis of

[ ]  Oral phase dysphagia

[ ]  Pharyngeal phase dysphagia or esophageal phase dysphagia

1. Has the participant had a swallow study done? [ ]  Yes [ ]  No

If YES, did it show:

[ ]  Penetration without aspiration

[ ]  Penetration with trace aspiration

[ ]  Aspiration with protective cough

[ ]  Silent aspiration

1. Has the participant been evaluated for a feeding and swallowing center/feeding therapist?

[ ]  Yes

[ ]  No

If YES, what was the recommendation:

[ ]  Participant placed on recommendation to stop oral feed

[ ]  Participant placed on recommendation to continue oral feed without any diet modification

1. Has the participant had a fiberoptic endoscopic evaluation of swallowing (FEES) procedure done by an ENT?

[ ]  Yes

[ ]  No

## Abdominal Pain

1. Has the participant experienced abdominal pain in the past or currently?

[ ]  Present

[ ]  Past

1. What was the participant’s age when abdominal pain began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What indicates that the participant has abdominal pain?

[ ]  The participant tells me

[ ]  Holds belly

[ ]  Holds belly and cries or looks uncomfortable

[ ]  Cries without another obvious cause

[ ]  Cries all the time

[ ]  Cries with meals

[ ]  Wakes up from sleep

[ ]  Irritability

[ ]  Other, specify:

[ ]  I don't know

1. Where is the abdominal pain located? Choose all that apply.

[ ]  Upper abdomen midline (between umbilicus and bottom of ribs)

[ ]  Upper abdomen on the right

[ ]  Upper abdomen on the left

[ ]  Entire upper abdomen

[ ]  Near the belly button

[ ]  Right lower quadrant (below umbilicus)

[ ]  Left lower quadrant

[ ]  Lower abdomen midline

[ ]  Entire lower abdomen

[ ]  Entire abdomen

[ ]  Variable locations

[ ]  I don't know

1. How often does abdominal pain occur?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don't know

1. How long does the abdominal pain last when it occurs?

[ ]  Less than a minute

[ ]  One minute to 30 minutes

[ ]  31 minutes to many hours

[ ]  All day

[ ]  Day and night

[ ]  Until the participant falls asleep

[ ]  Until the participant vomits

[ ]  Until the participant eats

[ ]  Until the participant has a bowel movement

[ ]  Until the participant urinates

[ ]  Until the participant passes gas

[ ]  Variable

[ ]  None of the above

[ ]  I don't know

1. Please provide additional comments about the duration of abdominal pain.
2. Do any of the following occur frequently before abdominal pain starts?

[ ]  Fasting more than 12 hours

[ ]  Viral illness/fever

[ ]  Taking medicine

[ ]  Right after feeding

[ ]  Seizure

[ ]  Headache/migraine/light sensitivity

[ ]  Exercise

[ ]  Stress

[ ]  Depression

[ ]  Anxiety

[ ]  Other psychological issues

[ ]  Particular foods

[ ]  Other, specify:

1. Please provide additional details about abdominal pain triggers.
2. Does abdominal pain disrupt activity?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Does abdominal pain wake the participant from sleep?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Is there anything specific that makes the pain better?
2. Is there anything specific that makes the pain worse?
3. Please provide additional details about present abdominal pain.

## Past Abdominal Pain

1. What was the participant’s age when past abdominal pain began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What was the participant’s age when the abdominal pain stopped?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What stopped the abdominal pain?
2. What indicated that the participant had abdominal pain?

[ ]  The participant told me

[ ]  Held belly

[ ]  Held belly and cried or looked uncomfortable

[ ]  Cried without another obvious cause

[ ]  Cried all the time

[ ]  Cried with meals

[ ]  Woke up from sleep

[ ]  Irritability

[ ]  Other, specify:

[ ]  I don't know

1. Please provide additional details if desired about past indications that the participant had abdominal pain.
2. Where was the past abdominal pain located? Choose all that apply.

[ ]  Upper abdomen midline (between umbilicus and bottom of ribs)

[ ]  Upper abdomen on the right

[ ]  Upper abdomen on the left

[ ]  Entire upper abdomen

[ ]  Near the belly button

[ ]  Right lower quadrant (below umbilicus)

[ ]  Left lower quadrant

[ ]  Lower abdomen midline

[ ]  Entire lower abdomen

[ ]  Entire abdomen

[ ]  Variable locations

[ ]  I don't know

1. How often did past abdominal pain occur?

[ ]  Daily

[ ]  At least once a week, but not daily

[ ]  One to 4 times per month

[ ]  Less than once a month, more than once a year

[ ]  About once a year

[ ]  Only once

[ ]  Other, specify:

[ ]  I don't know

1. How long did the past abdominal pain last when it occurred?

[ ]  Less than a minute

[ ]  One minute to 30 minutes

[ ]  31 minutes to many hours

[ ]  All day

[ ]  Day and night

[ ]  Until the participant fell asleep

[ ]  Until the participant vomited

[ ]  Until the participant ate

[ ]  Until the participant had a bowel movement

[ ]  Until the participant urinated

[ ]  Until the participant passed gas

[ ]  Variable

[ ]  None of the above

[ ]  I don't know

1. Please provide additional comments about the duration of past abdominal pain.
2. Did any of the following triggers occur before abdominal pain started?

[ ]  Fasting more than 12 hours

[ ]  Viral illness/fever

[ ]  Taking medicine

[ ]  Right after feeding

[ ]  Seizure

[ ]  Headache/migraine/light sensitivity

[ ]  Exercise

[ ]  Stress

[ ]  Depression

[ ]  Anxiety

[ ]  Other psychological issues

[ ]  Particular foods

[ ]  Other, specify:

1. Please provide additional details about past abdominal pain triggers.
2. Did past abdominal pain disrupt activity?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Did past abdominal pain wake the participant from sleep?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Is there anything specific that made the pain better?
2. Is there anything specific that made the pain worse?
3. Please provide additional details about past abdominal pain.

## Early Satiety

1. Does the participant experience early satiety after eating a small amount of food?

[ ]  Yes

[ ]  No

## Abdominal Bloating and Distension

1. Does the participant have bloating or abdominal distension or have they had it in the past?

[ ]  Present

[ ]  Past

1. What was the participant’s age when the abdominal bloating/ distension problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. Is the bloating or abdominal distension worse at any particular time of day?

[ ]  Morning (wake up until 12pm)

[ ]  Afternoon (12pm - 5pm)

[ ]  Evening (5pm - Bed)

[ ]  Overnight (wakes from sleep)

[ ]  All times of the day

[ ]  I don't know

1. Is current abdominal distension caused by air or stool?

[ ]  Air

[ ]  Stool

[ ]  Both

[ ]  I don't know

1. Is the participant an air swallower?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Please provide additional details about abdominal bloating and distension.

## Past Abdominal Bloating and Distension

1. What was the participant’s age when the past abdominal bloating/ distension problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What was the participant’s age when the past abdominal bloating/ distension stopped?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. What stopped the abdominal bloating/ distension?
2. Was the past bloating or abdominal distension worse at any particular time of day?

[ ]  Morning (wake up until 12pm)

[ ]  Afternoon (12pm - 5pm)

[ ]  Evening (5pm - Bed)

[ ]  Overnight (wakes from sleep)

[ ]  All times of the day

[ ]  I don't know

1. Was past abdominal distension caused by air or stool?

[ ]  Air

[ ]  Stool

[ ]  Both

[ ]  I don't know

1. Please provide additional details about past abdominal bloating and distension.

## General Stool Questions

1. How often does the participant have bowel movements?

[ ]  More than once a day

[ ]  Once a day

[ ]  Every other day

[ ]  A few times a week

[ ]  Once a week

[ ]  Less than one a week

[ ]  Only after a suppository

[ ]  Only after an enema/irrigation/suppository

[ ]  Requires disimpaction with a finger

[ ]  Uses a cecostomy to induce bowel movements

[ ]  Requires polyethylene glycol solution by mouth or tube

[ ]  Other, specify:

[ ]  I don't know

1. What is the consistency of bowel movements? (Bristol stool scale classification)

[ ]  Watery

[ ]  Mushy

[ ]  Soft

[ ]  Formed

[ ]  Hard

[ ]  Hard and large

[ ]  Variable

[ ]  Greasy/oil droplets

[ ]  Type 1: Separate hard lumps, like nuts (hard to pass)

[ ]  Type 2: Sausage-shaped, but lumpy

[ ]  Type 3: Like a sausage but with cracks on its surface

[ ]  Type 4: Like a sausage or snake, smooth and soft

[ ]  Type 5: Soft blobs with clear cut edges (passed easily)

[ ]  Type 6: Fluffy pieces with ragged edges, a mushy stool

[ ]  Type 7: Watery, no solid pieces. Entirely liquid

[ ]  I don't know

1. Does the participant use a toilet or a diaper?

[ ]  Toilet

[ ]  Diaper

## Bowel Movement Problems

1. What was the participant's age when the bowel movement problems began?

[ ]  On the day of birth

[ ]  Less than 2 mo old (not on day of birth)

[ ]  2 mo through 5.9 mo

[ ]  6 mo through 12.9 mo

[ ]  13 mo through18.9 mo

[ ]  19 mo through 3.9 y

[ ]  4 y through 6.9 y

[ ]  7 y through 13.9 y

[ ]  14 y through 18.9 y

[ ]  Greater than 18 y

[ ]  I don’t know

1. Does the participant have to strain to pass bowel movements?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Does the participant ever have blood in the stool?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. What color is the blood in the stool?

[ ]  Bright red

[ ]  Dark red (maroon)

[ ]  Black

[ ]  I don't know

1. What is the largest amount of blood that has been in the stool?

[ ]  Small streaks (i.e., not three dimensional)

[ ]  Small clumps of clots

[ ]  Big clots of blood (larger than a dime)

1. Does the participant use medicine or other treatment to help with bowel movements?

[ ]  Yes

[ ]  No

1. What treatment does the participant use to help with bowel movements?

[ ]  Glycolax, PEG, Polyethylene Glycol

[ ]  Mineral oil

[ ]  Kondremul

[ ]  Milk of Magnesia

[ ]  Senokot or other Senna

[ ]  Colace

[ ]  Dulcolax suppository

[ ]  Prune juice, apple juice, Karo syrup

[ ]  Fleets enema

[ ]  Saline enema

[ ]  Milk and molasses enema

[ ]  Imodium

[ ]  Lomotil

[ ]  Pepto-Bismol

[ ]  Linaclotide, Plecanatide, Lubiprostone, Tenapanor

[ ]  Prucalopride

[ ]  Other, specify:

1. Has the participant used any other treatments in the past to help with bowel movements?
2. Did these treatments help or worsen the bowel movement problem?
3. Please provide additional details about bowel movement problems.

## Poor Appetite Affecting Growth

1. Please provide additional details about poor appetite affecting growth.

## General Medication/ Treatment/Surgery Questions

1. What other medicines is the participant taking now?

[ ]  Proton pump inhibitor: Prilosec (omeprazole), Nexium, Prevacid (lansoprazole), Protonix (pantoprazole) or Aciphex (rabeprazole)

[ ]  H-2 receptor antagonist: Pepcid (famotidine), Zantac (ranitidine), Tagamet (cimetidine)

[ ]  Carafate

[ ]  Ondansetron (Zofran)

[ ]  Tricyclic antidepressant: Amitriptyline, desipramine, imipramine, nortriptyline

[ ]  SSRI: Citalopram (Celexa), escitalopram (Lexapro), fluoxetine (Prozac), paroxetine (Paxil, Pexeva), sertraline (Zoloft)

[ ]  Erythromycin

[ ]  Cisapride (Propulsid)

[ ]  Domperidone (Motilium)

[ ]  Reglan (metoclopramide)

[ ]  Tegaserod (Zelnorm, Zelmac)

[ ]  Linaclotide (Linzess)

[ ]  Prucalopride (Motegrity)

[ ]  CoQ10 (Ubiquinol, Ubiquinone)

[ ]  Riboflavin (vitamin B2)

[ ]  L-Creatine

[ ]  L-Arginine

[ ]  L-Carnitine

[ ]  B vitamins (other)

[ ]  Vitamin E

[ ]  Vitamin C

[ ]  Alpha lipoic acid

[ ]  Folinic acid

[ ]  Carbamazepine (Tegretol, Carbatrol)

[ ]  Ethosuximide (Zarontin)

[ ]  Felbatol (Felbamate)

[ ]  Tiagabine (Gabitril)

[ ]  Levetiracetam (Keppra)

[ ]  Lamotrigine (Lamictal)

[ ]  Pregabalin (Lyrica)

[ ]  Phenytoin (Dilantin)

[ ]  Topamax (Topiramate)

[ ]  Oxcarbazepine (Trileptal)

[ ]  Gabapentin (Neurontin)

[ ]  Pancreatic enzyme supplements (Creon)

[ ]  Levbid/Levsin/Bentyl

[ ]  Antibiotics

[ ]  Other, specify:

1. Has the participant taken any medications in the past that helped any of their GI problems?
2. Has the participant taken any medications in the past that have worsened their GI problems?
3. Has the participant tried any "alternative medicine approaches" to help with any of the problems we discussed?

[ ]  Yes

[ ]  No

1. What types of alternative medicine has the participant tried?

[ ]  Meditation

[ ]  Yoga

[ ]  Acupuncture

[ ]  Crystal

[ ]  Faith healer

[ ]  Prayer

[ ]  Massage

[ ]  Exercise

[ ]  Herbal supplements or medicines

[ ]  Homeopathic medicine

[ ]  Chiropractic care

[ ]  Colon cleansing

[ ]  Aromatherapy

[ ]  Antioxidants

[ ]  Supplemental vitamins (not prescribed)

[ ]  Marijuana

[ ]  Ginger

[ ]  Over the counter medicine

[ ]  Items bought over the internet

[ ]  Energy therapies

[ ]  Traditional Chinese medicine

[ ]  Native American medicine

[ ]  Other traditional forms of medicine

[ ]  Other, specify:

1. Did any of these alternative medicine treatments help with the participant’s GI issues?
2. Did any of these alternative medicine treatments worsen the participant’s GI problems?
3. Has the participant ever had surgery?

[ ]  Yes

[ ]  No

1. What type of surgery or procedures has the participant had?

[ ]  G-tube

[ ]  J-tube

[ ]  G J - tube (e.g., for venting and feeding)

[ ]  Colostomy

[ ]  Ileostomy

[ ]  Jejunostomy

[ ]  Cecostomy (to treat refractory constipation)

[ ]  Hirschsprung (Swenson, Duhamel, Soave)

[ ]  Fundoplication (Nissen, Toupet, Thal, Dor)

[ ]  Bowel resection

[ ]  Bowel dilation

[ ]  Botox injection

[ ]  Upper endoscopy

[ ]  Colonoscopy

[ ]  Esophageal manometry

[ ]  Antroduodenal manometry

[ ]  Colon manometry

[ ]  Anorectal manometry

[ ]  Gastric emptying studies

[ ]  Esophageal pH monitoring

[ ]  Esophageal impedance monitoring

[ ]  Bladder surgery

[ ]  Brain surgery

[ ]  Heart surgery

[ ]  Lung surgery

[ ]  Airway surgery (tracheostomy, laryngomalacia)

[ ]  Kidney surgery

[ ]  Liver surgery

[ ]  Spleen surgery

[ ]  Orthopedic surgery (bone or joint)

[ ]  Organ transplant

[ ]  Muscle biopsy

1. Please provide additional details about the type of surgery.

## GI and Social Issues

1. How much do the symptoms we discussed affect the participant’s life? (1 being not at all, 10 being severe interruption)

[ ]  1

[ ]  2

[ ]  3

[ ]  4

[ ]  5

[ ]  6

[ ]  7

[ ]  8

[ ]  9

[ ]  10

1. If there was a clinical trial regarding GI issues, would the participant enroll?

[ ]  Yes

[ ]  No

[ ]  I don’t know

1. How long did the participant have GI symptoms before they were diagnosed with mitochondrial disease?
2. How many GI doctors has the participant seen?
3. Were these GI symptoms what led to the participant’s mitochondrial disease diagnosis?
4. Were the participant’s GI symptoms recognized as significant before diagnosis of mitochondrial disease?
5. Has the participant’s treatment for their GI symptoms ever been delayed for fear of not being believed about these symptoms?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Has there ever been a referral to a psychiatric professional related to the participant's GI symptoms?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Has there ever been concern about a Munchausen by proxy accusation?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. Has a health care professional ever had concern about Munchausen by proxy syndrome?

[ ]  Yes

[ ]  No

[ ]  I don't know

1. What setting was this in?

[ ]  ER

[ ]  Outpatient

[ ]  Clinic

[ ]  Hospital stay

[ ]  Other, specify:

## Pancreas

1. Has the participant been told they have pancreatic disease?

[ ]  Yes

[ ]  No

1. If they were told they had pancreatitis, have they had?

[ ]  One episode

[ ]  Multiple episodes

1. How long ago did the episode(s) occur?
2. Over what period of time did the episodes occur?
3. Age at first episode:
4. Age at last episode:
5. Total number of episodes:
6. Has the participant been told they have acute pancreatitis?

[ ]  Yes

[ ]  No

1. Has the participant been told they have acute recurrent pancreatitis?

[ ]  Yes

[ ]  No

1. Has the participant been told they have chronic pancreatitis?

[ ]  Yes

[ ]  No

1. Has the participant been told they have pancreatic insufficiency?

[ ]  Yes

[ ]  No

1. What symptoms did the participant have that prompted the medical team to check for pancreatitis?
2. Was the participant hospitalized for pancreatitis?

[ ]  Yes

[ ]  No

1. Has the participant had an imaging study of their pancreas?

[ ]  Yes

[ ]  No

1. If YES, what study did the participant have?

[ ]  MRCP

[ ]  ERCP

[ ]  US

[ ]  CAT Scan abdomen

[ ]  MRI

[ ]  Endoscopic ultrasound

1. Has the participant had genetic testing for pancreatitis?

[ ]  Yes

[ ]  No

1. What did the genetic tests show?
2. Has the participant had a fecal elastase test done?

[ ]  Yes

[ ]  No

1. If YES, was the fecal elastase test result normal or abnormal?

[ ]  Normal

[ ]  Abnormal

1. Is the participant on pancreatic enzyme replacement therapy?

[ ]  Yes

[ ]  No

1. Is the participant on chronic pain medication for the pancreatitis?

[ ]  Yes

[ ]  No

1. Please provide any additional comments.

Recorder Signature: Date:

## General Instructions

Important note: None of the data elements included on this CRF Module are classified as Core (i.e., strongly recommended for all mitochondrial disease clinical studies to collect). All of the data elements are classified as Supplemental 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.