## 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.