

## MDCR Survey

*Thanks for taking this survey! Here's why the MDCR Advisory Board approved it.*

*The goal of MDCR is to gather the patient community perspective on mitochondrial disease in an environment that is credible and secure. This foundational survey serves as an introduction to how MDCR works for new registrants while establishing a baseline of information of potential value in future surveys.*

## Introduction

*Welcome to the Mitochondrial Disease Community Registry!*

*Depending on how much detail you include in your answers, this survey will take approximately 20 minutes to complete.*

*If you get tired, you can take a break and when you return, continue from where you left off. However, it is very important that you ultimately complete the entire survey so that everyone in the community has answered the same questions.*

*Thank you for your willingness to take part in this important work.*

*Select "Continue" to begin.*

*(About this profile.) First of all...*

*We will begin by asking a few questions to establish your relationship to mitochondrial disease.*

I am affected by mitochondrial disease... (one response can be selected)

Yes

No

Have you demonstrated symptoms of mitochondrial disease?

If yes, we would like to ask a few additional questions about your experience. If no, we will collect general health information. Even if you have no symptoms of mitochondrial disease, medical researchers often search for medical information of blood relatives of those affected by mitochondrial disease when they are designing studies.

I am (or was) a caregiver for individual(s) affected by mitochondrial disease... (one response can be selected)

Yes

No

I have relative(s) who are (or were) affected by mitochondrial disease... (one response can be selected)

Yes

No

You indicated that you have a relative with mitochondrial disease. How is this person related to you? (Please select all that apply)

- Aunt(s)
- Brother(s)
- Daughter(s)
- Father
- Granddaughter(s)
- Grandson(s)
- Maternal grandfather
- Maternal grandmother
- Mother
- Paternal grandfather
- Paternal grandmother
- Sister(s)
- Son(s)
- Uncle(s)

Generally speaking, I would consider participating in a clinical trial if I were confident that this might improve the diagnosis of, or lead to better treatments for individuals affected by mitochondrial disease... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
Strongly Agree Strongly Disagree

## Mitochondrial Disease Specific Questions

*To start out with...*

*We will begin by asking a few mitochondrial disease-specific questions. We will start with a few questions about your diagnosis, and then collect general information as well as opinions on a few topics, including your willingness to help in advancing the development of treatments and cures for mitochondrial disease.*

My mitochondrial disease has been classified by my medical doctors as...  
(one response can be selected)

- Definite
- Possible
- Probable
- Unlikely
- Not classified

My last response indicates that my doctor concluded it is unlikely the symptoms I am experiencing are due to mitochondrial disease. The statement below that most closely explains why I responded in this way is... (one response can be selected)

- Oops....that's not what I meant. There has been a specific mitochondrial disease mentioned as a possible explanation.
- Although the doctor has said it is unlikely, I still think mitochondrial disease represents a possible explanation for the symptoms experienced.
- While there are a large number of mitochondrial disorders, and while some of these have been ruled out, testing is still undergoing to identify what is responsible for the problems experienced.

My last response indicates that my diagnosis is not yet classified. The statement below that most closely explains why I responded in this way is... (one response can be selected)

Other

While the doctors are convinced that a mitochondrial disorder of some sort is responsible, they have not yet been able to identify a specific mitochondrial disease, but are considering several possibilities.

The doctors have mentioned mitochondrial disease as a potential explanation, but this diagnosis has not yet been confirmed.

While there are a large number of mitochondrial disorders, and while some have been ruled out, testing is still undergoing to identify what is responsible for the symptoms.

Other

The Other reason is....

\_\_\_\_\_

As I have gone through testing for a diagnosis... (one response can be selected)

The doctors have said they are looking carefully at several mitochondrial diseases, but I don't know which ones.

The doctors have said they are looking carefully at several mitochondrial diseases; and I/he/she may be able to indicate which ones (show a list please)

The doctors have said that they have ruled out several mitochondrial diseases, but I don't know which ones

More specifically, the possible explanations being considered are...  
(multiple responses can be selected)

The doctors have said that they have ruled out several mitochondrial diseases; and I/he/she may be able to indicate which ones (show a list please)

A biochemical disorder

A mitochondrial DNA deletion syndrome

A mitochondrial DNA mutation syndrome

A nuclear gene disorder

A system-based disorder (no genetic or biochemical answer known)

Other, not classified, or uncertain which of the above categories apply to diagnosis

More specifically, my doctors have ruled out the following diagnosis or diagnoses... (multiple responses can be selected)

A biochemical disorder

A mitochondrial DNA deletion syndrome

A mitochondrial DNA mutation syndrome

A nuclear gene disorder

A system-based disorder (no genetic or biochemical answer known)

Other, not classified, or uncertain which of the above categories apply to diagnosis

I was diagnosed with (or may have) the following mitochondrial DNA mutation syndrome(s)... (one response can be selected)

- AID (Aminoglycoside-Induced Deafness)
- LHON (Leber Hereditary Optic Neuropathy)
- LHON-Plus (Leber Heredity Optic Neuropathy Plus)
- MELAS (Mitochondrial Encephalopathy Lactic Acidosis with Stroke-like Episodes)
- MERRF (Myoclonus Epilepsy Ragged-red Fibers)
- MIDD (Maternally Inherited Diabetes and Deafness)
- MILS (Maternally Inherited Leigh Syndrome)
- NARP (Neuropathy Ataxia and Retinitis Pigmentosa)
- Other form(s) of mitochondrial DNA mutation syndrome (provide list)

The specific mitochondrial DNA mutation syndrome(s) with which I was diagnosed (or may have) is (are)...

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I was diagnosed with (or may have) the following Biochemical Disorder(s)... (multiple responses can be selected)

- CoQ Deficiency
- Complex I Deficiency
- Complex II (SDH) Deficiency
- Complex III Deficiency
- Complex IV Deficiency
- Complex V Deficiency
- Multiple Respiratory Chain Enzyme Deficiencies

The specific biochemical disorder(s) with which I was diagnosed (or may have) is (are)...

- Other form(s) of biochemical disorder(s) (provide list)
- SANDO (Sensory Ataxia, Neuropathy, Dysarthria, Ophthalmoplegia)
- Sensory Ataxia Neuropathy

\_\_\_\_\_

I was diagnosed with (or may have) the following mitochondrial DNA deletion syndrome(s)... (multiple responses can be selected)

- CPEO (Chronic Progressive External Ophthalmoplegia)
- KSS (Kearns-Sayre Syndrome)
- Other deletion(s) of mitochondrial DNA (provide list)
- Pearson syndrome

\_\_\_\_\_

The specific mitochondrial DNA deletion syndrome(s) with which I was diagnosed (or may have) is (are)...

I was diagnosed with (or may have) one of the following Nuclear Gene based Clinical disorder(s)... (multiple responses can be selected)

- Alpers syndrome
- Barth syndrome
- MNGIE (Mitochondrial Neurogastrointestinal Encephalomyopathy)
- Mitochondrial DNA Depletion Syndrome

- None of the above... Please continue to the next group of diagnoses
- SANDO (Sensory Ataxia, Neuropathy, Dysarthria, Ophthalmoplegia)
- Sensory Ataxia Neuropathy

The specific Nuclear Gene Disorder(s) with which I was diagnosed (or may have) is (are)...

\_\_\_\_\_

I was diagnosed with (or may have) the following system-based disorder(s)... (multiple responses can be selected)

- Dysarthria Ophthalmoplegia
- Encephalomyopathy
- Encephalopathy
- FBSN (Familial Bilateral Striatal Necrosis)
- Hepatocerebral disease
- Leukoencephalopathy
- Other, or multiple forms of system-based disorder(s) (provide list)

The specific system-based disorder(s) with which I was diagnosed (or may have) is (are)...

\_\_\_\_\_

The set of questions below were only presented to survey-takers who could not classify their mitochondrial disease in the categories of Nuclear Gene Disorders, Biochemical Disorders, mitochondrial DNA mutation syndrome, mitochondrial DNA Deletion syndrome, and system-based disorders.

*That's not a problem. The next few questions will present each of these categories one at a time. (one response can be selected)*

*For each category, select any diagnosis that applies (or if there has been no diagnosis, indicate any explanations that are being considered at this time). If none of the listed options apply, simply select "None of the above." Continue to the next group of possible diagnoses."*

In order to get started, please indicate which of the following applies:

I was diagnosed with (or may have) the following syndrome(s)... (one response can be selected)

- There has been a diagnosis, but not sure which category the diagnosis falls into.
- Several possible explanations are being considered at this time

- AID (Aminoglycoside-Induced Deafness)
- LHON (Leber Hereditary Optic Neuropathy)
- LHON-Plus (Leber Heredity Optic Neuropathy Plus)
- MELAS (Mitochondrial Encephalopathy Lactic Acidosis with Stroke-like Episodes)
- MERRF (Myoclonus Epilepsy Ragged-red Fibers)
- MIDD (Maternally Inherited Diabetes and Deafness)
- MILS (Maternally Inherited Leigh Syndrome)
- NARP (Neuropathy, Ataxia and Retinitis Pigmentosa)
- None of the above... Please continue to the next group of diagnoses

I was diagnosed with (or may have) the following syndrome(s)... (multiple responses can be selected)

- CPEO (Chronic Progressive External Ophthalmoplegia)
- KSS (Kearns-Sayre Syndrome)
- Pearson syndrome
- None of the above...Please continue to the next group of diagnoses

I was diagnosed with (or may have) one of the following disorder(s)...  
(multiple responses can be selected)

- Alpers syndrome
- Barth syndrome
- MNGIE (Mitochondrial Neurogastrointestinal Encephalomyopathy)
- Mitochondrial DNA Depletion Syndrome
- SANDO (Sensory Ataxia, Neuropathy, Dysarthria, Ophthalmoplegia)
- Sensory Ataxia Neuropathy
- None of the above... Please continue to the next group of diagnoses

I was diagnosed with (or may have) the following disorder(s)... (multiple responses can be selected)

- CoQ Deficiency
- Complex I Deficiency
- Complex II (SDH) Deficiency
- Complex III Deficiency
- Complex IV Deficiency
- Complex V Deficiency

I was diagnosed with (or may have) the following disorder(s)... (multiple responses can be selected)

- Multiple Respiratory Chain Enzyme Deficiencies
- Dysarthria Ophthalmoplegia

- Encephalomyopathy
- Encephalopathy
- FBSN (Familial Bilateral Striatal Necrosis)
- Hepatocerebral disease
- Leukoencephalopathy
- None of the above.... Please continue

Any other diagnosis (or diagnoses) I received (or may have) is (are) described below...

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Any other possible explanation(s) that is (are) being considered at this time is (are)...

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The reason I continue to think that mitochondrial disease may be responsible for my symptoms is...

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The reason I selected "other" is...

I have access to the results of genetic testing... (one response can be selected)

- Genetic testing was performed and I/he/she have/has a copy of a report from the doctor or testing lab that lists the genetic variations that are a possible cause of the medical condition.

- While I/he/she do/does not have a copy of the report, I/he/she recall(s) that genetic testing was performed. I/he/she would be willing to request a copy of the report if this would be useful to gathering information concerning the mitochondrial disorder.
- Although I/he/she recall(s) that genetic testing was performed, I/he/she do/does not have a copy of the report and would not be inclined to request a copy for this purpose.
- Genetic testing was never done.
- Other (please specify)

The reason I selected "other" is...

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## Health Insurance

The type of health insurance that I use is... (multiple responses can be selected)

- Health Maintenance Organization (HMO)
- High-Deductible Health Plan (HDHP)
- Indian Health Service (IHS)
- Medicaid
- Medicare
- Military Health Benefits (DoD)
- No health insurance

- Other
- Preferred Provider Organization (PPO)
- State Children's Health Insurance Program (SCHIP)
- State Risk Pool
- Veterans Health Administration (VA)

The other type of health insurance that I use is...

\_\_\_\_\_

The insurer, if any, that I receive my health insurance from is... (one response can be selected)

Response options: Aetna Group, Blue Cross Blue Shield, California Physicians' Service, Cambia Health Solutions Inc., Carefirst Inc. Group, Centene Corp. Group, Cigna Health Group, Coventry Corp. Group, HCSC Group, Health Net of California, Inc., Highmark Group, HIP Insurance Group, Humana Group, Independence Blue Cross Group, Kaiser Foundation Group, Lifetime Healthcare Group, Metropolitan Group, Molina Healthcare Inc. Group, UHC of California, United Healthgroup, Wellcare Group, Wellpoint Inc. Group, Other

My company is Blue Cross Blue Shield of... (one response can be selected)

Response options: Alabama, Alaska, Arizona, Arkansas, California, Colorado, Connecticut, Delaware, Florida, Georgia, Hawaii, Idaho, Illinois, Indiana, Iowa, Kansas, Kentucky, Louisiana, Maine, Maryland, Massachusetts, Michigan, Minnesota, Mississippi, Missouri, Montana, Nebraska, Nevada, New Hampshire, New Jersey, New Mexico, New York, North Carolina, North Dakota, Ohio, Oklahoma, Oregon, Pennsylvania, Rhode Island, South Carolina, South Dakota, Tennessee, Texas, Utah, Vermont, Virginia, Washington, West Virginia, Wisconsin, Wyoming, District of Columbia

The insurer, if any, I receive insurance from is called...

\_\_\_\_\_

My health insurance is provided by... (one response can be selected)

Response options: Work or school pays for it, Spouse or guardian's work pays for it, Purchase through work or school, Purchased through a health exchange, Privately purchased from a insurance company, Other

The other place I get my insurance from is...

\_\_\_\_\_

I have received health insurance from my current company for the last... (one response can be selected)

Response options: Month, 3 months, 6 months, 9 months, 1 year, 2 years, 3 to 5 years, More than 5 years

In general, my medical expenses are... (one response can be selected)

Not covered

My health has impacted my ability to get insurance... (multiple responses can be selected)

- Covered a little
- Mostly covered
- Completely covered
  
- Disability insurance
- Don't know
- Health insurance
- Life insurance
- Long-term care insurance
- Not at all

## Opinions on Mitochondrial Disease Research and State of Clinical Trials

*The next few questions focus on your perception about the state of mitochondrial disease research and consideration of clinical trials.*

*What is a clinical trial?*

*A major step in developing any new drug or medical device is called 'clinical trials'. These are special medical research studies that determine if a new therapy is helpful and safe for affected individuals to use. Volunteers from the targeted patient population and the general public participate in these trials so that everyone involved can learn important information. Some examples of information learned in a clinical trial are the effectiveness of a proposed new treatment, side effects, and effective dosages.*

Regarding myself personally... Thinking about my own condition, my stage in the disease process, and perspectives about the state of science in the field, I feel... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
 Pessimistic anything can be done in time to address my needs      Optimistic a treatment will be available in time to address my needs

Regarding individuals who may be affected by mitochondrial disease in the future... Given what I know about the disease, the activities of UMDF and others, and my perspectives about the state of science in the field, I feel... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
 Pessimistic anything learned from my experiences could be meaningful to helping others in the foreseeable future      Optimistic my participation in research may make a meaningful contribution to helping others in the foreseeable future

Generally speaking, I would consider participating in a clinical trial of a treatment for mitochondrial disease even if it was uncertain I would gain any direct health benefit from it... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
 Strongly Agree      Strongly Disagree

Generally speaking, I would consider participating in a clinical trial for a treatment for mitochondrial disease even if it meant I would have to stop my current treatment(s)... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
 Strongly Agree      Strongly Disagree

## General Health

Please select system of measurement for weight and height...  
(one response can be selected)

U.S. Customary System or the inch-pound system in lbs or ft-in (example: 150 lbs and 5 ft, 6 in)

International System of Units or the metric system in kg or m-cm (example: 60 kg and 1m70cm)

My current weight is approximately... (one response can be selected)

Response options: [pull-down menu with values in the unit of measurement specified]

My current height is approximately... (one response can be selected)

Response options: [pull-down menu with values in the unit of measurement specified]

Please select a system of measurement for birth weight: (one response can be selected)

U.S. Customary System or the inch-pound system in lbs (example: 7 lbs, 6 oz)

International System of Units or the metric system in kg or m-cm (example: 3 kg 010 g)

My birth weight was... (one response can be selected)

Response options: [pull-down menu with values in the unit of measurement specified]

In general, my health is... (one response can be selected)

Excellent

Very good

Good

I am able to function physically... (one response can be selected)

Fair

Poor

Without any difficulty

With a little difficulty

With some difficulty

With much difficulty

Cannot do physical activities

Pain interferes with the enjoyment of my life... (one response can be selected)

Not at all

A little bit

Somewhat

Quite a bit

Very much

I feel fatigued... (one response can be selected)

Never

Rarely

Sometimes

Often

Always

I feel depressed... (one response can be selected)

Never

- Rarely
- Sometimes
- Often
- Always

## Demographic Questions

*About these questions...*

*This section of the survey will collect basic demographic information such as gender, race and from where the survey is being taken.*

My gender (self-identified) is... (one response can be selected)

- Female
- Male
- Other (specify on next question)

I describe my gender as "Other" because I am... (one response can be selected)

- Intersex (ambiguous at birth)
- Intersex (XXY, XXYY or other chromosomal variation from XX-female or XY-male)
- Male-to-female transsexual
- Female-to-male transsexual
- Other

My gender is...

\_\_\_\_\_

My biological sex (genotype) is... (one response can be selected)

- Female (XX)
- Male (XY)
- Neither XX or XY (specify on next question)

I describe my genotype as "Neither" because I have... (one response can be selected)

- Turner syndrome (45, XO)
- XX male syndrome (also, de la Chapelle syndrome)
- Trisomy X (47, XXX)
- Klinefelter's syndrome (47, XXY)
- Jacob's syndrome (47, XYY)
- 48, XXXY, 48, XXXX, XYY syndrome (48, XYY)
- 49, XXXXY
- Penta X syndrome (49, XXXXX)

My race or origin is... (multiple responses can be selected)

- American Indian or Alaskan Native (i.e. Navajo, Mayan, Tlingit, etc.)

- Asian (i.e. Asian Indian, Chinese, Filipino, Japanese, Korean, Vietnamese, Hmong, Laotian, Thai, Pakistani, Cambodian, etc.)
- Black or African American (i.e. African American, Haitian, Nigerian, etc.)
- Hispanic, Latino, or Spanish origin (i.e. Mexican, Mexican American, Puerto Rican, Cuban, Argentinian, Colombian, Dominican, Nicaraguan, Salvadorian, Spaniard, etc.)
- Native Hawaiian or Other Pacific Islander (i.e. Native Hawaiian, Guamanian or Chamorro, Samoan, Fijian, Tongan, etc.)
- Some other race or origin
- White (i.e., European, Middle Eastern, Northern Africa, etc.)

The name of my tribe is...

\_\_\_\_\_

More specifically my Asian heritage is... (multiple responses can be selected)

- Asian Indian
- Bangladeshi
- Cambodian
- Chinese
- Filipino
- Hmong

- Indonesian
- Japanese
- Korean
- Laotian
- Malaysian
- Other
- Pakistani
- Sri Lankan
- Thai
- Vietnamese

More specifically my Pacific Islander heritage is... (multiple responses can be selected)

- Chamorro
- Fijian
- Guamanian
- Native Hawaiian
- Other
- Samoan
- Tongan

More specifically my Hispanic or Latino heritage is... (multiple responses can be selected)

- Central American
- Cuban

- Mexican
- Other Spanish, Hispanic or Latino
- Puerto Rican
- South American

More specifically my "Other" background is...

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Right now I am taking this survey from... (one response can be selected)

- Home
- Work
- School
- A hospital or clinic
- Another location

The location where I am taking this survey is...

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## Background

*About these questions...*

*This section of the survey will collect background information about birthplace, language, education level, family income and travel.*

I was born in... (one response can be selected)

- United States
- Canada
- Caribbean
- Mexico
- United Kingdom
- Other European Countries
- Africa
- Asia
- Central America
- Middle East
- South America
- South Pacific
- Other

The specific Caribbean country in which I was born is... (one response can be selected)

- Aruba
- Bahama Islands
- Dominican Republic
- Jamaica
- Other

The specific European country in which I was born is... (one response can be selected)

- France
- Germany
- Ireland
- Italy
- Netherlands
- Spain
- Switzerland
- Other

The specific African country in which I was born is... (one response can be selected)

- Egypt
- Morocco
- Senegal
- South Africa
- Other

The specific Asian country in which I was born is... (one response can be selected)

- China / Mainland
- China / Taiwan
- Hong Kong
- India
- Japan
- Korea

The specific Central American country in which I was born is... (one response can be selected)

- Philippines
- Other

- Costa Rica
- El Salvador
- Guatemala
- Panama
- Other

The specific Middle Eastern country in which I was born is... (one response can be selected)

- Yemen
- Oman
- Saudi Arabia
- Jordan
- Egypt
- Israel
- United Arab Emirates
- Kuwait
- Qatar
- Lebanon
- Bahrain
- Other

The specific South American country in which I was born is... (one response can be selected)

- Argentina
- Brazil
- Columbia
- Peru
- Venezuela
- Other

The specific South Pacific country in which I was born is... (one response can be selected)

- Australia
- New Zealand
- Polynesia
- Other

The specific country in which I was born is...

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The state in which I was born is... (one response can be selected)

Response options: Alabama, Alaska, Arizona, Arkansas, California, Colorado, Connecticut, Delaware, Florida, Georgia, Hawaii, Idaho, Illinois, Indiana, Iowa, Kansas, Kentucky, Louisiana, Maine, Maryland, Massachusetts, Michigan, Minnesota, Mississippi, Missouri, Montana, Nebraska, Nevada, New Hampshire, New Jersey, New Mexico, New York, North Carolina, North Dakota, Ohio, Oklahoma, Oregon, Pennsylvania, Rhode Island, South Carolina, South Dakota, Tennessee, Texas, Utah, Vermont, Virginia, Washington, West Virginia, Wisconsin, Wyoming, Puerto Rico, District of Columbia, Northern Mariana Islands, United States Virgin Islands, American Samoa, Guam

The city, town or village in which I was born is...

\_\_\_\_\_

The language I most frequently speak at home is... (one response can be selected)

- English
- Spanish
- Creole
- Chinese
- Tagalog
- French
- Vietnamese
- German
- Korean

Russian

Arabic

Other

The language I most frequently speak at home is...

\_\_\_\_\_

My preferred method(s) of communication is (are)... (multiple responses can be selected)

Non-Verbal

Sign Language

Spoken Language

Written Language

I am currently in school... (one response can be selected)

Yes

No

The highest grade level I completed... (one response can be selected)

Response options: Nursery school, Kindergarten, Grade 1, Grade 2, Grade 3, Grade 4, Grade 5, Grade 6, Grade 7, Grade 8, Grade 9, Grade 10, Grade 11, Grade 12 - No Diploma

My highest level of education is... (one response can be selected)

Response options: No schooling completed, Some primary or high school (no diploma), Regular high school diploma, Special Education Certificate, GED or alternative credential, Some college, but less than 1 year, 1 or more years of college, no degree, Vocational, trade school or associate's degree, Bachelor's degree, Master's degree, Professional degree beyond a bachelor's degree, Doctorate degree

The highest grade level my mother completed... (one response can be selected)

Response options: Nursery school, Kindergarten, Grade 1, Grade 2, Grade 3, Grade 4, Grade 5, Grade 6, Grade 7, Grade 8, Grade 9, Grade 10, Grade 11, Grade 12 - No Diploma

My mother's highest level of education completed is... (one response can be selected)

Response options: No schooling completed, Some primary or high school (no diploma), Regular high school diploma, Special Education Certificate, GED or alternative credential, Some college, but less than 1 year, 1 or more years of college, no degree, Vocational, trade school or associate's degree, Bachelor's degree, Master's degree, Professional degree beyond a bachelor's degree, Doctorate degree

The highest grade level my father completed... (one response can be selected)

Response options: Nursery school, Kindergarten, Grade 1, Grade 2, Grade 3, Grade 4, Grade 5, Grade 6, Grade 7, Grade 8, Grade 9, Grade 10, Grade 11, Grade 12 - No Diploma

My father's highest level of education completed is... (one response can be selected)

Response options: No schooling completed, Some primary or high school (no diploma), Regular high school diploma, Special Education Certificate, GED or alternative credential, Some college, but less than 1 year, 1 or more years of college, no degree, Vocational, trade school or associate's degree, Bachelor's degree, Master's degree, Professional degree beyond a bachelor's degree, Doctorate degree

My total family income is... (one response can be selected)

Response options: Less than \$10,000 (USD), \$10,000 - \$14,999, \$15,000 - \$19,999, \$20,000 - \$24,999, \$25,000 - \$29,999, \$30,000 - \$34,999, \$35,000 - \$39,999, \$40,000 - \$44,999, \$45,000 - \$49,999, \$50,000 - \$54,999, \$55,000 - \$59,999, \$60,000 - \$74,999, \$75,000 - \$84,999, \$85,000 - \$99,999, \$100,000 - \$149,999, \$150,000 - \$199,999, \$200,000 - \$249,999, \$250,000 and above, Decline to provide

Including me, the number of persons that live in my household is... (one response can be selected)

Response options: 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, More than 12

I have traveled outside my own country... (one response can be selected)

Yes

No

I have traveled to the following world regions... (multiple responses can be selected)

- Africa
- Asia
- Canada
- Caribbean
- Central America
- Mexico
- Middle East
- Other European Countries
- South America
- South Pacific
- United Kingdom
- United States

The specific regions of the Caribbean I visited were... (multiple responses can be selected)

- Aruba
- Bahama Islands
- Dominican Republic
- Jamaica
- Other

The specific European countries I visited were... (multiple responses can be selected)

- France
- Germany
- Ireland
- Italy
- Netherlands
- Other
- Spain
- Switzerland

The specific regions in Africa that I visited were... (multiple responses can be selected)

- Egypt
- Morocco
- Other
- Senegal
- South Africa

The specific regions in Asia that I visited were... (multiple responses can be selected)

- China / Mainland
- China / Taiwan
- Hong Kong
- India
- Japan
- Korea

The specific regions in Central America that I visited were... (multiple responses can be selected)

- Other
- Philippines

- Costa Rica
- El Salvador
- Guatemala
- Other
- Panama

The specific regions in the Middle East that I visited were... (multiple responses can be selected)

- Israel
- Jordan
- Other
- Saudi Arabia
- Turkey

The specific regions in the South Pacific that I visited were... (multiple responses can be selected)

- Australia
- New Zealand
- Other
- Polynesia

The specific regions in South America that I visited were... (multiple responses can be selected)

- Argentina
- Brazil
- Columbia
- Other
- Peru
- Venezuela

## Relationship with Research

*About these questions...*

*This section of the survey will collect information about biospecimens you may have provided as well as opinions about participating in research.*

Within the last 5 years, I have had a major surgery that required general anesthesia...

- Yes
- No

I have previously donated a biospecimen... (one response can be selected)

- Yes
- No

The type of biospecimen (blood, tissue or cell line) I previously donated was...

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I donated the biospecimen to... (multiple responses can be selected)

- Mayo Clinic Mitochondrial Disease Biobank
- NAMDC (North American Mitochondrial Disease Consortium)
- Other (Research Lab, Clinic, etc.)

To the best of my knowledge, the biospecimen I donated is located at...

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If it was quick and easy to do, I would be willing to update my health profile on a regular basis... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

My preferred methods to receive questions and respond with periodic updates would be... (multiple responses can be selected)

- Email
- From a computer browser

I would prefer the total number of questions asked in each update be limited to... (one response can be selected)

- Mail
- Phone call
- Text message
- 1 question
- 3 questions
- 5 questions
- 10 questions
- More than 10 questions would be fine

In these future updates, the sorts of topics I think would be most important to explore are... (multiple responses can be selected)

- Challenges of diagnosis
- Clinical trial participation
- Disease or condition's natural history
- Other
- Quality of life issues
- Symptoms experienced
- Usage of dietary supplements

Please specify the other topic(s)...

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If I could control who saw it, I would be willing to share information collected by a device or monitor I wore... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

If I could control who saw my information, I would be willing to ask my health providers to update my profile and/or link all or some of my health records to it... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

If I could control who had access to it, or who saw the information from it, I would be willing to donate a biospecimen (tissue or body fluid) for research... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely

I am interested in taking part in research in general... (one response can be selected)

- Very unlikely
- Definitely not

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am interested in testing a treatment where I would need to go to a clinic one or more times... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am interested in testing a treatment given by phone or over the internet (like getting advice about my health)... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am interested in testing a treatment where I have to take a medicine or other treatment, and come for clinic visits... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am interested in testing a treatment where I have to take a medicine or other treatment, come for clinic visits, AND give blood samples... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely

I am interested in taking part in a project that involves meeting at a local community center or school... (one response can be selected)

- Very unlikely
- Definitely not

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am interested in taking part in a project in which I would stay in the hospital for one or more days... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am interested in taking part in a project that involves a procedure such as a special x-ray or new type of surgery... (one response can be selected)

- Definitely would want to do this
- Very likely

I am interested in taking part in a project that requires me to wear a device or monitor that collects information about my activities, such as a heart rate or blood pressure monitor... (one response can be selected)

- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

- Definitely would want to do this
- Very likely

- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am interested in taking part in a project where I would have to respond to phone calls or text messages to provide information about what I am doing with diet, exercise or other issues about my daily health... (one response can be selected)

- Definitely would want to do this
- Very likely

- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I would be willing to donate a blood sample that would be used to study my DNA... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

I am not interested in participating in research because... (multiple responses can be selected)

- Do not want to be a "guinea pig"
- Don't know
- No obvious benefit to self or family
- Not paid for taking part
- Results may not be kept private or confidential
- The project involves medical tests, like drawing blood or having x-rays
- The project was not recommended by a doctor
- The project will take a lot of time
- The research might reveal something bad
- Worried about privacy
- Would rather not say

I would be more likely to take part in research if it were run by... (multiple responses can be selected)

- A drug company
- A medical school or hospital
- A non-profit foundation (such as the American Diabetes Association)
- A tobacco company
- An insurance company
- Personal doctor
- Private research institute
- The US government (like the National Institutes of Health)

Aside from this survey, I have taken part in research projects related to my health or to the health of a family member... (one response can be selected)

- Yes
- No
- Don't Know

Assuming a limited number of questions was asked each time, I would prefer to respond to such updates on a... (one response can be selected)

- Daily basis
- Weekly basis
- Monthly basis
- After each medical appointment
- Never

I am interested in taking part in a project that involves me and other people in my family... (one response can be selected)

- Definitely would want to do this
- Very likely
- Somewhat likely
- Uncertain
- Somewhat unlikely
- Very unlikely
- Definitely not

The most important thing I want scientists and physicians engaged in research for this disease, as well as drug approval agencies such as the FDA, to know is...

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Please share any other thoughts about this disease registry and this survey...

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Reflecting on my experience of setting up an account and responding to the questionnaire...

I found signing up for a Private Access account (choosing username, password, security questions and site key)... (one response can be selected)

- Very easy
- Fairly easy
- Some parts easy
- Some parts difficult
- Somewhat difficult

I found setting privacy and sharing preferences (choosing a guide, and setting search, usage and contact preferences)... (one response can be selected)

- Very easy
- Fairly easy
- Some parts easy
- Some parts difficult
- Somewhat difficult

I found responding to the health questionnaire... (one response can be selected)

- Very easy
- Fairly easy
- Some parts easy
- Some parts difficult
- Somewhat difficult

I felt the process of creating privacy directives pertaining to who could access and use my information and for what purposes was... (multiple responses can be selected)

- About the same as other health websites
- Cold and off putting
- Confusing
- Conveys a sense of security
- Easy to understand and follow
- Friendly and welcoming
- Helped by the use of guides
- Helpful in building trust
- More difficult due to the use of guides
- Not professional
- Professional
- Suspicious

Now that I have registered for the Private Access service and expressed my wishes for how my personal health and contact information should be handled, I feel... (one response can be selected)

- Complete trust wishes will be followed exactly as directed
- A general sense of trust that wishes will be followed essentially as directed
- Only some trust that wishes will be followed as directed
- Uncertain whether wishes will be followed
- A sense of concern that wishes will not be followed
- Certain that wishes will not be followed

I felt that being permitted to declare who could access and use my information, and for what purposes is... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
 Much less assuring than relying on a standard Privacy Policy      Much more assuring than relying on a standard Privacy Policy

I found the ability to see how others responded to the same questions I answered to be... (one response can be selected)

Helpful in keeping interest high  
 Tiresome or distracting

From start to finish my impression of the length of time it took...

To sign up for an account (one response can be selected)

It took about the right amount of time  
 It took less time than I thought it would  
 It took too long, but it was worth it  
 It took too long

To set privacy preferences (one response can be selected)

It took about the right amount of time  
 It took less time than I thought it would  
 It took too long, but it was worth it  
 It took too long

To answer the questionnaire (one response can be selected)

It took about the right amount of time  
 It took less time than I thought it would  
 It took too long, but it was worth it

It took too long

## Caregiver

*You indicated that you're a caregiver for someone with mitochondrial disease.*

*We'd like to ask you a few questions about that experience. Please click "Continue" to be redirected to our caregiver survey.*

As a caregiver working with individual(s) who are (or who were) affected by mitochondrial disease, I have experience working with: (one response can be selected)

- 1 individual
- 2-5 individuals
- 6-10 individuals
- 11-15 individuals
- More than 15 individuals

The individual(s) diagnosed with mitochondrial disease for whom I am a caregiver... (one response can be selected)

- Include at least one family member or personal friend
- Are a patient(s) or client(s) with whom I have a professional relationship (such as a doctor, nurse or other health provider)
- Are a member(s) of a support group or organization for which I serve as a paid staff member
- Are a member(s) of a support group or organization for which I serve as a volunteer

The individual who is a friend or family member, and for whom I am a caregiver is... (one response can be selected)

- Father
- Son
- A male sibling
- A male spouse or domestic partner
- A male friend or other family member
- Mother
- Daughter
- A female sibling
- A female spouse or domestic partner
- A female friend or family member
- Less than 6 months
- 6 months to less than 1 year
- 1 year to less than 2 years
- 2 years to less than 5 years
- 5 years to less than 10 years
- 10 years or more

I have been a caregiver for... (one response can be selected)

I most often provide care for this individual (or these individuals) in... (one response can be selected)

- A clinic
- A hospital
- A doctor's office

- Home (one on one in person)
- Home or office (Contact with them through email and/or by telephone)
- The individual's home
- Other

Regarding the individual(s) with mitochondrial disease for whom I serve as a caregiver... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
 Pessimistic anything can be done in time to address their needs      Optimistic a treatment will be available in time to address their needs

Regarding the other member(s) of my family who are affected by a disease or condition... (select number on a scale)

0 *(Place a mark on the scale below)* 100  
 Pessimistic anything can be done in time to address their needs      Optimistic a treatment will be available in time to address their needs

The other place I most often provide care for this individual (or these individuals) is...

\_\_\_\_\_