

Introduction

The purpose of this research study is to describe the prevalence of Parkinson disease in patients with Fabry disease and their first-degree family members (parents, children, siblings). You are being asked to take part in this research study because you have been diagnosed with Fabry disease or because you have a first-degree family member who has been diagnosed with Fabry disease.

Being in a research study is completely voluntary. You can choose not to be in this research study. You can also say yes now, and change your mind later.

If you agree to take part in this research, you will be asked to complete a questionnaire with questions about general health information and your family history of Parkinson disease. Your participation in this study will take about 15 minutes.

You can choose not to answer any question you do not wish to answer. You can also choose to stop taking the survey at any time.

You must be a Fabry disease patient or first-degree family member of a Fabry disease patient to participate. If you do not have Fabry disease, or do not have a first-degree family member with Fabry disease, please stop now.

The possible risks to you in taking part in this research are:

- You may feel uncomfortable answering some of the questions
- Someone else might find out you were in a research study

The possible benefits to you for taking part in this research are:

- Increase the medical understanding of individuals with Fabry Disease with respect to the development of Parkinson disease.

To protect your identity as a research subject, no identifiable information will be collected and in any publication about this research, no private information will be used.

If you have any questions about this research, please contact the Principal Investigator, Dr. Amy Yang at 212-241-0915 or amy.yang@mssm.edu. You can also call the Program for the Protection of Human Subjects Office at 212-824- 8200 if you have any concerns about this research.

Demographic Information

Demographic Information:

What is your age?

What is your year of birth?

What is your sex?

- Male
 Female

Are you adopted?

- Yes
 No

Fabry Disease Information:

Fabry Disease Information:

Please select the option(s) that best describes you:

- I have been diagnosed with Fabry disease
- I am a first-degree relative (parent, sibling or child) of someone who has Fabry disease

Please select the family member(s) diagnosed with Fabry disease (please select all that apply).

- Mother
- Father
- Son
- Daughter
- Brother
- Sister
- Other

Please provide your Fabry disease genotype, if known:

(ex: G43R, g11185-delT)

PD Personal

Personal History of Parkinson Disease:

Have you been diagnosed with Parkinson disease?

- Yes
- No

What year were you diagnosed with Parkinson disease?

Have any of your family members been diagnosed with Parkinson's disease?

- Yes
- No

Maybe (please explain)

Please select all of the family members who have been diagnosed with Parkinson's disease.

Mother

Father

Sibling(s) (please enter number of siblings with Parkinson's disease)

Maternal Grandmother

Maternal Grandfather

Paternal Grandmother

Paternal Grandfather

Other (please describe)

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The remainder of this survey will ask you to answer the same series of questions related to the symptoms and diagnosis of Parkinson disease for each of your parents, children, and siblings. Please select the individual about whom you are answering this series of questions, and then proceed.

If the family member is deceased, please answer the questions based on your memory of the person, to the best of your ability (please note that all questions are written in the present-tense).

PD History: Parents

The following information pertains to my:

Mother

Father

Year of Birth

Does or did this person have the mutation in the Fabry gene or have symptoms of Fabry?

- Yes
- No
- I don't know

Is this person living?

- Yes
- No

Please indicate age at death:

What was the cause of death?

- | | |
|---|---|
| <input type="radio"/> Unknown | <input type="radio"/> Kidney disease |
| <input type="radio"/> Heart disease | <input type="radio"/> Septicemia |
| <input type="radio"/> Cancer | <input type="radio"/> Intentional self-harm (suicide) |
| <input type="radio"/> Cerebrovascular disease (stroke) | <input type="radio"/> Chronic liver disease and cirrhosis |
| <input type="radio"/> Chronic lower respiratory disease | <input type="radio"/> Essential hypertension and hypertensive renal disease |
| <input type="radio"/> Accident (unintentional injury) | <input type="radio"/> Parkinson disease |
| <input type="radio"/> Diabetes | <input type="radio"/> Homicide |
| <input type="radio"/> Alzheimer disease | <input type="radio"/> Natural causes |
| <input type="radio"/> Influenza and pneumonia | <input type="radio"/> Other |
| <input type="radio"/> | <input type="text"/> |

Was an autopsy performed?

- Yes
- No
- Unsure

Did the autopsy indicate Parkinson's disease?

- Yes
- No
- Unsure

Has he/she been diagnosed with Parkinson's disease by a physician?

- Yes
- No
- Unsure

If yes, was the physician a neurologist?

- Yes
- No
- Unsure

How old was he/she when the diagnosis was made?

Does he/she take Sinemet (Levodopa/Carbidopa)?

- Yes
- No
- Unsure

Does he/she exhibit any of the following symptoms? Please check all that apply.

- Tremor at rest or when sitting quietly
- Shuffling gait and/or steps that are short and tiny
- Stooped posture
- Arms that do not swing when he/she walks
- Muscular rigidity or slow/stiff movements

If he/she has any of the above symptoms, at what age did they begin?

If this person has any of the symptoms listed below, which were the first to develop? (Please select all that apply).

- | | |
|--|--|
| <input type="checkbox"/> rest tremor | <input type="checkbox"/> stooped posture |
| <input type="checkbox"/> action tremor | <input type="checkbox"/> small handwriting |
| <input type="checkbox"/> slowness | <input type="checkbox"/> freezing |
| <input type="checkbox"/> stiffness | <input type="checkbox"/> memory loss |
| <input type="checkbox"/> balance problem | <input type="checkbox"/> numbness |
| <input type="checkbox"/> shuffling gait | <input type="checkbox"/> hallucinations |
| <input type="checkbox"/> decreased arm swing | <input type="checkbox"/> depression |
| <input type="checkbox"/> decreased facial expression | <input type="checkbox"/> leg dragging |
| <input type="checkbox"/> decreased volume of speech | <input type="checkbox"/> other |
| <input type="checkbox"/> dystonia | <input type="text"/> |

Thank you. You will now be asked the same series of questions about your other parent. If you have already completed these questions for both parents, you will be asked to provide information about your children and siblings.

Siblings

How many siblings do you have?

PD History: Siblings

Please answer the following questions for each of your siblings.

Is this person male or female?

- Male
 Female

Year of Birth

Does or did this person have the mutation in the Fabry gene or have symptoms of Fabry?

- Yes
 No
 I don't know

Is this person living?

- Yes
 No

Please indicate age at death:

What was the cause of death?

- | | |
|---|---|
| <input type="radio"/> Unknown | <input type="radio"/> Kidney disease |
| <input type="radio"/> Heart disease | <input type="radio"/> Septicemia |
| <input type="radio"/> Cancer | <input type="radio"/> Intentional self-harm (suicide) |
| <input type="radio"/> Cerebrovascular disease (stroke) | <input type="radio"/> Chronic liver disease and cirrhosis |
| <input type="radio"/> Chronic lower respiratory disease | <input type="radio"/> Essential hypertension and hypertensive renal disease |
| <input type="radio"/> Accident (unintentional injury) | <input type="radio"/> Parkinson disease |
| <input type="radio"/> Diabetes | <input type="radio"/> Homicide |
| <input type="radio"/> Alzheimer disease | <input type="radio"/> Natural causes |
| <input type="radio"/> Influenza and pneumonia | <input type="radio"/> Other |
| <input type="radio"/> | <input type="text"/> |

Was an autopsy performed?

- Yes
 No
 Unsure

If so, did the autopsy indicate Parkinson's disease?

- Yes
 No
 Unsure

Has he/she been diagnosed with Parkinson's disease by a physician?

- Yes
 No
 Unsure

If yes, was the physician a neurologist?

- Yes
- No
- Unsure

How old was he/she when the diagnosis was made?

Does he/she take Sinemet (Levodopa/Carbidopa)?

- Yes
- No
- Unsure

Does he/she exhibit any of the following symptoms? Please check all that apply.

- Tremor at rest or when sitting quietly
- Shuffling gait and/or steps that are short and tiny
- Stooped posture
- Arms that do not swing when he/she walks
- Muscular rigidity or slow/stiff movements

If he/she has any of the above symptoms, at what age did they begin?

If this person has any of the symptoms listed below, which was the first to develop? (Please select all that apply.)

- rest tremor
- action tremor
- stooped posture
- micrographia

- slowness
- stiffness
- balance problem
- shuffling gait
- decreased arm swing
- decreased facial expression
- decreased volume of speech
-
- dystonia

- freezing
- memory loss
- numbness
- hallucinations
- depression
- leg dragging
- other
-

Thank you. You will now be asked the same series of questions about your other siblings. If you have already completed these questions for all of your siblings, you will be asked to complete this information about your children.

Children

How many children do you have?

PD History: Children

Please answer the following questions for each of your children. It may be helpful to proceed from oldest to youngest, in order to keep track of which information has already been entered.

Is this person male or female?

- Male
- Female

Year of Birth

Does or did this person have the mutation in the Fabry gene or have symptoms of Fabry?

- Yes
- No
- I don't know

Is this person living?

- Yes
- No

Please indicate age at death:

What was the cause of death?

- | | |
|---|---|
| <input type="radio"/> Unknown | <input type="radio"/> Kidney disease |
| <input type="radio"/> Heart disease | <input type="radio"/> Septicemia |
| <input type="radio"/> Cancer | <input type="radio"/> Intentional self-harm (suicide) |
| <input type="radio"/> Cerebrovascular disease (stroke) | <input type="radio"/> Chronic liver disease and cirrhosis |
| <input type="radio"/> Chronic lower respiratory disease | <input type="radio"/> Essential hypertension and hypertensive renal disease |
| <input type="radio"/> Accident (unintentional injury) | <input type="radio"/> Parkinson disease |
| <input type="radio"/> Diabetes | <input type="radio"/> Homicide |
| <input type="radio"/> Alzheimer disease | <input type="radio"/> Natural causes |
| <input type="radio"/> Influenza and pneumonia | <input type="radio"/> Other |
| <input type="radio"/> | <input type="text"/> |

Was there an autopsy performed?

- Yes
- No
- Unsure

If so, did the autopsy indicate Parkinson's disease?

- Yes
- No
- Unsure

Has he/she been diagnosed with Parkinson's disease by a physician?

- Yes
- No
- Unsure

If yes, was the physician a neurologist?

- Yes
- No
- Unsure

How old was he/she when the diagnosis was made?

Does he/she take Sinemet (Levodopa/Carbidopa)?

- Yes
- No
- Unsure

Does he/she exhibit any of the following symptoms? Please check all that apply.

- Tremor at rest or when sitting quietly
- Shuffling gait and/or steps that are short and tiny
- Stooped posture
- Arms that do not swing when he/she walks
- Muscular rigidity or slow/stiff movements

If he/she has any of the above symptoms, at what age did they begin?

If this person has any of the symptoms listed below, which was the first to develop? (Please select all that apply.)

- | | |
|---|---|
| <input type="radio"/> rest tremor | <input type="radio"/> stooped posture |
| <input type="radio"/> action tremor | <input type="radio"/> micrographia |
| <input type="radio"/> slowness | <input type="radio"/> freezing |
| <input type="radio"/> stiffness | <input type="radio"/> memory loss |
| <input type="radio"/> balance problem | <input type="radio"/> numbness |
| <input type="radio"/> shuffling gait | <input type="radio"/> hallucinations |
| <input type="radio"/> decreased arm swing | <input type="radio"/> depression |
| <input type="radio"/> decreased facial expression | <input type="radio"/> leg dragging |
| <input type="radio"/> decreased volume of speech | <input type="radio"/> other |
| <input type="radio"/> | <input style="width: 100%; height: 20px;" type="text"/> |
| <input type="radio"/> dystonia | |

Thank you. You will now be asked the same series of questions about your other children. If you have already completed these questions for all of your children, you have completed the survey.

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