Name of person with Danon disease: _____

Name of person completing these questions: _____

Today's Date: _____

Demographics

1. Gender (**select one**):
   - ☐ Male
   - ☐ Female
2. Ethnic Category (**select one**):
   - ☐ Hispanic or Latino
   - ☐ Not Hispanic or Latino
   - ☐ Unknown
   - ☐ Decline to report
3. Racial Category (**select one**):
   - ☐ American Indian/Alaska Native
   - ☐ Asian
   - ☐ Native Hawaiian or Other Pacific Islander
   - ☐ Black or African American
   - ☐ White
   - ☐ More than One Race: (specify please) _____
4. Name of person with Danon Disease: _____
5. Date of birth of person with Danon Disease: _____
6. Age of person with Danon Disease (or date of death if deceased): _____
7. Name of person completing survey: _____
8. Relationship to person with Danon Disease: _____
9. Address: _____
   _____
10. Phone number: _____
11. Alternate phone number: _____
12. Email: _____

Danon Disease Symptom Questions

PART I: Diagnosis of Disease:

Question 1: **When** were you diagnosed with Danon Disease? (month/year) _____

(In these questions 'you' refers to you or your child with Danon disease)

☐ I do not remember.

☐ I am suspected to have Danon disease but have not been diagnosed yet.

Comment: _____

Question 2: How was your diagnosis of Danon Disease made?

☐ DNA test (genetic test / LAMP-2 gene sequencing).

☐ Skeletal muscle biopsy (e.g. arm or leg) showing absence of staining for LAMP2 protein

☐ Heart muscle biopsy showing absence of staining for LAMP2 protein

☐ other (specify please): _____

☐ I do not remember.

Question 3: Have you had a genetic test (of the LAMP-2 gene) to confirm a diagnosis of Danon Disease?

☐ Yes ☐ No ☐ I do not know

If yes, please list your mutation (genetic change) if you know it: _____

Question 4: **Who** made the diagnosis of Danon Disease in you?

☐ Primary Care Physician ☐ Cardiologist

☐ Pediatrician ☐ Other physician (please explain):

Question 5: **Why** were you diagnosed with Danon Disease?

☐ I was having symptoms and saw a doctor about these symptoms.

List symptoms: _____

What was your first symptom? _____

What was your second symptom? _____

How long did you have symptoms before you were diagnosed with Danon disease (days/months/years)? _____

☐ I was tested for Danon Disease because a family member already had Danon Disease.

☐ A problem was identified during a regular doctor (or other healthcare provider's) visit.

Please explain: _____

☐ Other reason (explain): _____

Question 6: Were **you** the **first person** in your family to be diagnosed with Danon Disease?

☐ Yes ☐ No ☐ I do not know/cannot remember

Question 7: Comments relating to your diagnosis of Danon Disease: _____

PART II: Symptoms and Signs of Danon Disease:

Question 1: Eye Symptoms / Disease. **Check all boxes that apply**:

- ☐ I do not have any known eye symptoms/disease(s) (**go to next question**)
- ☐ I do have (had had) eye symptoms/disease(s) (check all boxes that apply).
  - ☐ Known retinal abnormalities

    Please explain: _____
  - ☐ Visual complaints (please describe): _____

    At what age(s) did your eye disease develop? _____

    Comments: _____

Question 2: Heart Symptoms / Disease. **Check all boxes that apply**:

- ☐ I do not have any known heart symptoms/disease(s) (**go to next question**)
- ☐ I do have (have had) heart symptoms/disease(s) (check all boxes that apply).

☐ Chest pain
☐ Shortness of breath with rest
☐ Shortness of breath with exertion or exercise
☐ Shortness of breath that causes awakening from sleep
☐ Swelling in the legs or ankles
☐ Difficulty breathing when laying flat on your back
☐ Irregular heart beat
☐ Rapid heart beat
☐ High blood pressure
☐ High cholesterol

At what age were these heart symptoms first experienced? _____

Comments: _____

Question 3: Heart Procedures / Studies: **Check all boxes that apply**:

3A: Echocardiogram (heart ultrasound):

- ☐ I have never had an echocardiogram (**go to question 3B**)
- ☐ I have had an echocardiogram(s), it showed
  - ☐ Normal results
  - ☐ Hypertrophic cardiomyopathy (thickening of heart walls)
  - ☐ Dilated cardiomyopathy (enlarged heart chambers)
  - ☐ Other: _____
  - ☐ Don't know

  If your echocardiogram(s) were abnormal, at what age were the abnormalities first noted: _____

  Comments: _____

3B: Electrocardiogram (also called 'EKG' or 'ECG' that measure your heart's electrical activity):

- ☐ I have never had an electrocardiogram (**go to question 3C**)
- ☐ I have had an electrocardiogram(s), it showed
  - ☐ Normal results
  - ☐ Wolff-Parkinson White syndrome (sometimes called 'pre-excitation')
  - ☐ Heart hypertrophy (thickening of the wall(s) of the heart)
  - ☐ Atrial Fibrillation (irregular heart rhythm)
  - ☐ Other: _____
  - ☐ Don't know

  If your electrocardiogram (s) were abnormal, at what age were the abnormalities first noted: _____

  Comments: _____

3C: Invasive Heart Studies/Procedures: (heart catherization, electrophysiology study, pacemaker)

- ☐ I have never had an invasive heart study or procedure (**go to question 3D**)
- ☐ I have had an invasive heart study
  - ☐ I have had an electrophysiology study (evaluate electrical conduction in my heart). At what age(s): _____
  - ☐ I have had an ablation(s) to treat Wolff-Parkinson White or another problem with heart conduction. At what age(s): _____
  - ☐ I have a pacemaker. At what age(s): _____
  - ☐ I have a defibrillator. At what age(s): _____
  - ☐ I have had a heart biopsy. At what age(s): _____

  Comments: _____

3D: Heart Transplant:

- ☐ I have never had a heart transplant (**go to next question**)
- ☐ I have had a heart transplant. At what age(s): _____
- ☐ I am on the heart transplant list now

  Comments: _____

Question 4: Learning / cognition: **Check all boxes that apply**:

- ☐ I have no learning or cognitive symptoms/disease(s) (**go to next question**)
- ☐ I have learning or cognitive symptoms/disease(s) (check all boxes that apply)

  At what age were the learning or cognitive problems identified? _____

  Types of learning problems (please check all that apply):
  - ☐ Problems learning to talk
  - ☐ Problems learning to read
  - ☐ Problems with math
  - ☐ Other (please specify): _____
- ☐ I need/needed special education services in school

  Formal IQ test performed:

  ☐ Yes ☐ No ☐ I do not know

  If yes, what was the IQ score: _____

  Comments: _____

Question 5: Behavioral / Psychiatric Diagnoses: **Check all boxes that apply**:

☐ I have no behavioral or psychiatric diagnoses (**go to next question**)

☐ I have behavioral and/or psychiatric diagnoses (check all boxes that apply)

Types of behavioral problems or psychiatric Diagnoses (check all that apply):

- ☐ Attention Deficit / Hyperactivity disorder
- ☐ Autism
- ☐ Pervasive developmental disorder
- ☐ Depression
- ☐ Other (please specify): _____

At what age did these problems begin? _____

Comments: _____

Question 6: Muscle Disease. **Check all boxes that apply**:

☐ I do not have any known muscle symptoms/disease(s) (**go to next question**)

☐ I do have (have had) muscle symptoms/disease(s) (check all boxes that apply).

- ☐ Muscular Dystrophy (i.e. a physician has diagnosed you with muscular dystrophy)
- ☐ Muscle weakness (Which muscles? _____)
- ☐ Muscle cramping (Which muscles? _____)
- ☐ Muscle fatigue (i.e. muscles initially feel OK, but tire easily) (Which muscles? _____)
- ☐ Delay of motor milestones in childhood (rolling over, sitting, walking)
- ☐ Current difficulty walking
- ☐ Current difficulty running
- ☐ Current difficulty walking up stairs

At what age were these symptoms first experienced? _____

Comments: _____

Question 7: Other Neurological Disease **Check all boxes that apply**:

- ☐ Tingling or numbness (Where? _____)

- ☐ Seizures
- ☐ Migraines
- ☐ Stroke(s)

At what age did these problems begin? _____

Comments: _____

Question 8: Lung / breathing symptoms/disease(s) **Check all boxes that apply**:

- ☐ I do not have any known lung symptoms/disease(s) (**go to next question**)
- ☐ I do have (have had) lung symptoms/disease(s) (check all boxes that apply).
  - ☐ Difficulty breathing / shortness of breath at rest
  - ☐ Difficulty breathing / shortness of breath with activity/exertion
  - ☐ Asthma
  - ☐ Sleep apnea
  - ☐ Pneumonia(s) (at what age? How often?) _____
  - ☐ I need to wear/use oxygen (Day/Night? When started?) _____

At what age did these problems begin? _____

Comments: _____

Question 9: Digestive / Gastrointestinal symptoms/disease(s) **Check all boxes that apply**:

- ☐ I do not have any known digestive/gastrointestinal symptoms/disease(s) (**go to next question**)
- ☐ I do have digestive/gastrointestinal symptoms/disease(s) (check all boxes that apply).
  - ☐ Difficulty swallowing
  - ☐ Gastrointestinal reflux
  - ☐ Abdominal pain (chronic)
  - ☐ Liver disease (describe please): _____
  - ☐ Diarrhea
  - ☐ Constipation
  - ☐ Other gastrointestinal disease (please describe): _____

At what age were these symptoms first experienced? _____

Comments: _____

Question 10: Endocrine symptoms/disease(s) **Check all boxes that apply**:

- ☐ I do not have any known endocrine symptoms/disease(s) (**go to next question**)
- ☐ I do have (have had) endocrine symptoms/disease(s) (check all boxes that apply).

☐ High cholesterol

☐ Diabetes

☐ Type 1 ☐ Type 2 ☐ Do not know

☐ Thyroid disease

☐ Other endocrine disease (please describe):

At what age were these symptoms first experienced?

Comments:

Question 11: Other problems/symptoms **Check all boxes that apply**:

☐ Skin disease (please describe):

☐ Hearing problems (please describe):

☐ Growth delay (please describe):

☐ Arthritis (please describe):

☐ Autoimmune disease (please describe):

☐ Recurrent infections (please describe):

☐ Allergies (please describe):

Question 12: Medications

Please list the medications you are currently taking.

Please list any medications you have had allergies or reactions too (please describe).

Have you had any problems with surgery / anesthesia?

☐ No

☐ Yes (please describe):

Have you had any problems with vaccinations?

- ☐ No
- ☐ Yes (please describe): _____

Have you had any dental problems (cavities, surgeries, gingivitis)?

_____

Are there any other things you wish to tell us (medical, physical, psychological, etc)?

_____

Thank you for your participation which is instrumental in our learning more about this rare and poorly understood condition