

## Online Resource 1 Survey Questions

This supplemental file includes the participant survey.

### Screening Questions

Q1 - Are you 18 years or older?

- Yes
- No

Q2 - What is your sex assigned at birth?

- Female
- Male

Q3 - Are you currently living in the United States?

- Yes
- No

Q4 - Are you comfortable taking this survey in English?

- Yes
- No

### Carrier Status/Experience

Q5 - Which X-linked disease do you carry/are you affected with?

- Aarskog syndrome
- Adrenoleukodystrophy (or Adrenomyeloneuropathy)
- Alport syndrome
- Barth syndrome
- Chronic granulomatous disease
- Coffin-Lowry syndrome
- Duchenne muscular dystrophy/Becker muscular dystrophy
- Fabry disease
- Fragile X syndrome (full carrier with >200 CGG repeats)
- Fragile X syndrome (premutation carrier with 55-200 repeats)
- Hemophilia A/B
- Hyper IgM syndrome
- Kennedy's disease (or Spinal-bulbar muscular atrophy)
- L1 syndrome (or LICAM)
- Lesch-Nyhan syndrome
- Myotubular myopathy
- Pelizaeus-Merzbacher disease
- X-linked ichthyosis
- Other \_\_\_\_\_

Q6 - What age did you learn you were a carrier of your X-linked disease? If you are unsure, around what age were you?

- Use age input as a number

Q7 - At what age do you believe individuals should be told they are carriers?

- Use age input as a number

Q8 - Which statement best describes how/why you initially learned you were an X-linked disease carrier?

- I gave birth to a son who was diagnosed with the X-linked disease
- I gave birth to a daughter who was found to be a carrier
- I have a father who was diagnosed with the X-linked disease
- I have a brother or brothers who were diagnosed with the X-linked disease
- I have other male relatives who were diagnosed with the X-linked disease
- I have female relatives who are known carriers
- Routine carrier screening (no family history)
- Genetic testing due to personal symptoms of the X-linked disease

### Symptoms

Q9 - When you first learned of your carrier status, were you aware that you could be at risk for symptoms?

- Yes
- No

Q10 (Skip logic - pertains to only those who answered “Yes” to Q10) - Who was the first to inform you that you could be at risk for symptoms as a carrier?

- Parent(s)
- Other family member
- Doctor
- Genetic counselor
- I found out myself through research or social media

Q11 - Do you feel that you have any symptoms attributable to your X-linked disease?

- Yes
- No
- Maybe

Q12 (Likert scale) - Are you aware of any medications/treatments/therapies for symptoms experienced by females that are carriers of your X-linked disease? Would you say that you are extremely aware, moderately aware, somewhat aware, slightly aware or not at all aware?

- Extremely aware
- Moderately aware
- Somewhat aware
- Slightly aware
- Not at all aware

Q13 (Skip logic) - If you are aware of medications/treatments/therapies for carriers of your X-linked disease, please list them. \_\_\_\_\_

Q14 (Likert scale) - Do you feel that females who carry your X-linked disease have sufficient access to knowledgeable healthcare providers and/or medical information concerning possible symptoms and/or risks associated with being a carrier?

- Strongly agree
- Agree
- Neither agree nor disagree

- Disagree
- Strongly disagree

### Reproductive Options

Q15 - Which of the following options have been discussed with you by a healthcare provider as reproductive options? Select all that apply.

- Prenatal diagnostic testing, i.e. chorionic villus sampling (CVS) or amniocentesis to determine if an ongoing pregnancy is affected
- In vitro fertilization with preimplantation genetic testing (IVF with PGT)
- Using an egg donor
- Adoption
- Natural conception (with option of postnatal genetic testing and/or postnatal genetics evaluation)
- None of the above

Q16 - If cost or insurance were not a barrier, which, if any, of the following reproductive options would you be most likely to utilize?

- Prenatal diagnostic testing, i.e. chorionic villus sampling (CVS) or amniocentesis to determine if an ongoing pregnancy is affected
- In vitro fertilization with preimplantation genetic testing (IVF with PGT)
- Using an egg donor
- Adoption
- Natural conception (with option of postnatal genetic testing and/or postnatal genetics evaluation)
- None of the above

### Needs/Priorities

Q17 - How important are the following to you, with respect to carriers of your X-linked disease? 1 - Very important, 2 - Important, 3 - Moderately important, 4 - Slightly important, 5 - Not important.

- Raising community awareness of carrier issues (physical/emotional symptoms, reproductive options, etc.)
- Ensuring that all potential carriers have access to affordable genetic testing to determine or confirm carrier status
- Ensuring that females carriers have insurance coverage for or can afford alternative reproductive options
- Improving carrier access to medical information, research studies, and new treatments and reproductive methods
- Mobilizing the medical community to perform studies and develop clinical trials for female carriers to help identify treatments/cure
- Uniting female carriers of X-linked disease to support one another
- Advocating for laws and policies beneficial to carriers

Q18 - If you had found out or known about your carrier status earlier, how would you have used this information? Select all that apply.

- I would have known that there were associated symptoms with my carrier status and sought out medical evaluation
- I would have looked for more information/support groups
- I would have been able to advocate for myself more
- I would have considered an alternative reproductive option (i.e. prenatal diagnostic testing, IVF with PGT, using an egg donor, or adoption)
- I would have had my child(ren) undergo genetic testing at birth

- I would have chosen not to have children
- I would have not had to endure the stress of genetic testing and waiting for results
- Nothing would have changed

Q19 - How has learning information about your carrier status impacted you? Select all that apply.

- I know that the symptoms I am experiencing are due to my carrier status
- I sought medical evaluation for my symptoms
- I looked for more information/support groups
- I chose to advocate for myself
- I chose or plan to choose an alternative reproductive option
- I had or will have my child(ren) undergo genetic testing at birth
- I chose not to have children
- I was able to or plan to inform my other family members about their risks
- Nothing has changed

Q20 - If you would like to elaborate on any of the questions listed above and provide insight on your experiences as a female X-linked carrier, please do so below.

- Fill in response

### **Demographic Questions**

Q21 - What is your age?

- Use age input as a number

Q22 - What is your gender identity?

- Female
- Male
- Transgender
- Non-binary
- Prefer not to disclose
- Prefer to self-describe (please specify)

Q23 - What state do you currently live in?

- (Choose state)

Q24 - Which of the following best describes where you live the majority of the year?

- City
- Suburban
- Rural

Q25 - What race/ethnicity do you identify as? Select all that apply.

- South Asian
- East Asian
- Southeast Asian
- Black/African American
- Hispanic/Latinx
- Native American or American Indian/Alaskan Native
- White
- Native Hawaiian/Pacific Islander

- Other \_\_\_\_\_
- Prefer not to answer

Q26 - If applicable, please specify your religious beliefs. Select all that apply.

- Christianity
- Judaism
- Islam
- Buddhism
- Hinduism
- Atheist
- Agnostic
- Spiritual but not affiliated/religious
- Prefer not to answer

Q27 - What is the highest degree or level of school you have completed? (If you're currently enrolled in school, please indicate the highest degree you have completed.)

- Less than high school degree
- High school degree or equivalent (e.g. GED)
- Some college
- Associate degree
- Bachelor degree
- Graduate degree (Master's degree, professional degree, doctorate)
- Prefer not to answer

Q28 - What is your current employment status?

- Employed full-time
- Employed part-time
- Unemployed (currently looking for work)
- Unemployed (not currently looking for work)
- Student
- Retired
- Self-employed
- Unable to work

Q29 - What is your marital status?

- Single (never married)
- Married
- In a domestic partnership
- Separated/divorced
- Widowed
- Prefer not to answer

Q30 - What is your annual household income?

- Less than \$10,000
- \$10,000 to \$24,999
- \$25,000 to \$49,999
- \$50,000 to \$74,999
- \$75,000 to \$99,999
- \$100,000 to \$149,999

- \$150,000 and greater

Q31 - Which of the following describes you?

- Have child(ren) and plan to have more biological children
- Have child(ren) and am done with family planning
- Have child(ren) and am unsure of having more biological children
- Do not have children but plan to have biological children one day
- Do not have children and do not plan to have biological children because of my carrier status
- Do not have children and do not plan to have biological children for other reasons
- Do not have children and am unsure of having biological children