Why am I being invited to take part in a registry? We invite you to take part in this Registry if you are 18 or over and are a carrier of a fragile X premutation or are a member of a family affected by fragile X. If you agree to participate you will be asked to provide contact information demographic information and basic medical information so that you can be invited to participate in future research projects focused on understanding and treating fragile X premutation-associated conditions. Premutation carriers who have been diagnosed with or have symptoms of fragile X-associated tremor/ataxia syndrome (FXTAS) or fragile X-associated primary ovarian insufficiency (FXPOI) as well as carriers without these problems are encouraged to participate.

Why is this registry being created? The primary goal of the Registry is to build up a large and diverse group of people interested in participating in research to better understand the effects of the premutation on human health and find effective treatments for premutation-related conditions. Registrants may be contacted by the Registry team about research studies for which they may be eligible. Registrants will not be directly contacted by the researchers. To date the majority of premutation studies have not adequately represented people from different backgrounds. The Registry team will share research opportunities via multiple methods to help investigators recruit a diverse and inclusive group of participants for their studies. People who join the Registry will be connected to an international community interested in premutation research and will periodically be sent updates via a newsletter about new research findings pertaining to the premutation.

FOR MUCH MORE DETAIL ABOUT THE REGISTRY AND YOUR RIGHTS AND PROTECTIONS AS A PARTICIPANT PLEASE BE SURE TO READ THE INFORMED CONSENT PDF DOCUMENT BELOW.

Consent form
[Attachment: "Premutation Registry Consent.pdf"]

Do you wish to join the registry? *must provide value (note)
- Yes
- No

Please tell us the primary reason that you have chosen not to join the registry.
- I do not qualify.
- I have concerns about sharing personal identifying information.
- I have concerns about sharing health information.
- There was a technical problem.
- I changed my mind about wanting to participate in future research.
- It appeared that it will take too much time/I am too busy.
- Other reason

Language
- English
- Español

Your signature documents that you have read the informed consent and have given permission to be part of this registry. *must provide value

(Note)
Legal Authorized Representative Signature  A legally authorized representative (LAR) may provide consent on behalf of a registrant. You acknowledge that you have read the informed consent form, and by signing you give permission for the person to register. Please respond to all questions according to registrants responses.

(note)

Legal Authorized Representative Name

(note)

Today’s Date *must provide value

(note)

First Name (as it appears on your birth certificate) *must provide value

Last name (current) *must provide value

Email address *must provide value

PERSONAL REGISTRATION DETAILS

** NOTE: Your internet browser may be set to “autofill” or “autocomplete” form fields, such as an address field. This can cause difficulty in completion of the registry information. We recommend turning off this function in your browser’s settings. See the following website for instructions for each browser type: autofill (Link opens in new window)

** NOTA: Su navegador de internet puede estar configurado para "auto llenar" o "autocompletar" campos del formato como el de dirección. Esto puede causar dificultades para completar la información del registro. Recomendamos desactivar esta función en las herramientas de su navegador. Vea el siguiente sitio web para instrucciones para cada tipo de navegador: autocompletar (abre en una ventana nueva.)

Nickname

Middle name
Confidential

Last name (as it appears on your birth certificate)

__________________________________

City or municipality of birth (as it appears on your birth certificate)

__________________________________

Sex at birth *must provide value

☐ Female
☐ Male

Birth year *must provide value

__________________________________

((Use el formato AAAA.))

Date of birth *must provide value

__________________________________

(note)

Street address of residence *must provide value

__________________________________

City of residence *must provide value

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<thead>
<tr>
<th>State or territory of residence (U.S. ONLY)</th>
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Zip code or mail code of residence *must provide value

30/03/2022 11:33am

Country of residence

*must provide value

- Afghanistan
- Albania
- Algeria
- Andorra
- Angola
- Antigua and Barbuda
- Argentina
- Armenia
- Australia
- Austria
- Azerbaijan
- Bahamas
- Bahrain
- Bangladesh
- Barbados
- Belarus
- Belgium
- Belize
- Benin
- Bhutan
- Bolivia
- Bosnia and Herzegovina
- Botswana
- Brazil
- Brunei
- Bulgaria
- Burkina Faso
- Burundi
- Cabo Verde
- Cambodia
- Cameroon
- Canada
- Central African Republic (CAR)
- Chad
- Chile
- China
- Colombia
- Comoros
- Democratic Republic of the Congo
- Costa Rica
- Cote d'Ivoire
- Croatia
- Cuba
- Cyprus
- Czechia
- Denmark
- Djibouti
- Dominica
- Dominican Republic
- Ecuador
- Egypt
- El Salvador
- Equatorial Guinea
- Eritrea
- Estonia
- Eswatini (formerly Swaziland)
- Ethiopia
- Fiji
- Finland
- France
- Gabon
- Gambia
- Georgia
- Germany
- Ghana
- Greece
- Grenada
- Guadeloupe
- Guatemala
- Guinea
- Guinea-Bissau
- Guyana
- Haiti
- Heard Island and McDonald Islands
- Honduras
- Hungary
- Iceland
- India
- Indonesia
- Iran
- Iraq
- Ireland
- Israel
- Italy
- Jamaica
- Japan
- Jordan
- Kazakhstan
- Kenya
- Kiribati
- Kuwait
- Kyrgyzstan
- Laos
- Latvia
- Lebanon
- Lesotho
- Liberia
- Libya
- Liechtenstein
- Lithuania
- Luxembourg
- Macau
- Madagascar
- Malawi
- Malaysia
- Maldives
- Mali
- Malta
- Marshall Islands
- Martinique
- Mauritania
- Mauritius
- Mayotte
- Mexico
- Micronesia
- Moldova
- Monaco
- Mongolia
- Montenegro
- Montserrat
- Morocco
- Mozambique
- Namibia
- Nauru
- Nepal
- Netherlands
- New Caledonia
- New Zealand
- Nicaragua
- Niger
- Nigeria
- Niue
- Norway
- Oman
- Pakistan
- Palau
- Panama
- Papua New Guinea
- Paraguay
- Peru
- Philippines
- Poland
- Portugal
- Qatar
- Romania
- Russia
- Rwanda
- Samoa
- San Marino
- São Tomé and Príncipe
- Saudi Arabia
- Senegal
- Serbia
- Seychelles
- Sierra Leone
- Singapore
- Slovakia
- Slovenia
- Solomon Islands
- Somalia
- South Africa
- South Korea
- South Sudan
- Spain
- Sri Lanka
- Strauss
- Sudan
- Suriname
- Sweden
- Switzerland
- Syria
- Taiwan
- Tajikistan
- Tanzania
- Thailand
- Timor-Leste
- Togo
- Tokelau
- Tonga
- Trinidad and Tobago
- Tunisia
- Turkey
- Turkmenistan
- Tuvalu
- Uganda
- Ukraine
- United Arab Emirates
- United Kingdom
- United States
- United States Virgin Islands
- Uruguay
- Uzbekistan
- Vanuatu
- Venezuela
- Vietnam
- Virgin Islands
- Yemen
- Zambia
- Zimbabwe
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Lithuania  
Luxembourg  
Madagascar  
Malawi  
Malaysia  
Maldives  
Mali  
Malta  
Marshall Islands  
Mauritania  
Mauritius  
Mexico  
Micronesia  
Moldova  
Monaco  
Mongolia  
Montenegro  
Morocco  
Mozambique  
Myanmar (formerly Burma)  
Namibia  
Nauru  
Nepal  
Netherlands  
New Zealand  
Nicaragua  
Niger  
Nigeria  
North Korea  
North Macedonia (formerly Macedonia)  
Norway  
Oman  
Pakistan  
Palau  
Palestine  
Panama  
Papua New Guinea  
Paraguay  

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○ San Marino
○ Sao Tome and Principe
○ Saudi Arabia
○ Senegal
○ Serbia
○ Seychelles
○ Sierra Leone
○ Singapore
○ Slovakia
○ Slovenia
○ Solomon Islands
○ Somalia
○ South Africa
○ South Korea
○ South Sudan
○ Spain
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○ Suriname
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○ Trinidad and Tobago
○ Tunisia
○ Turkey
○ Turkmenistan
○ Tuvalu
○ Uganda
○ Ukraine
○ United Arab Emirates (UAE)
○ United Kingdom (UK)
○ United States of America (USA)
○ Uruguay
○ Uzbekistan
○ Vanuatu
○ Vatican City (Holy See)
○ Venezuela
○ Vietnam
○ Yemen
○ Zambia
○ Zimbabwe

Phone number (including country code if outside the U.S.) *must provide value
Password *must provide value

(note)

Which language do you usually speak and read? *must provide value

- Mandarin Chinese
- Spanish
- English
- Hindi
- Bengali
- Portuguese
- Russian
- Japanese
- Western Punjabi
- Marathi
- Telugu
- Wu Chinese
- Turkish
- Korean
- French
- German
- Vietnamese
- Tamil
- Yue Chinese
- Urdu
- Italian
- Other - list below

Other Language
Other Languages

☐ Mandarin Chinese
☐ Spanish
☐ English
☐ Hindi
☐ Bengali
☐ Portuguese
☐ Russian
☐ Japanese
☐ Western Punjabi
☐ Marathi
☐ Telugu
☐ Wu Chinese
☐ Turkish
☐ Korean
☐ French
☐ German
☐ Vietnamese
☐ Tamil
☐ Yue Chinese
☐ Urdu
☐ Italian
☐ Other - list below

Other additional languages

FRAGILE X-RELATED INFORMATION

fmr1 DNA Test?

☐ Yes
☐ No
☐ I don't know

What is the main reason that you had fragile X testing?

☐ Prefer not to answer
☐ I had a clinical problem that was thought to be due to fragile X
☐ Research participation
☐ A family member tested positive for fragile X
☐ Other (please list below)

Other reason for fragile X testing:
What is your Fragile X (FMR1) Status  *must provide value
(If mosaic (more than one CGG category), please choose the largest CGG category.)

- Prefer not to answer
- Unknown
- Normal (up to 44 CGG repeats)
- Grey or intermediate (45-54 CGG repeats)
- Premutation (55-200 CGG repeats)
- Full mutation (>200 CGG repeats)
- Not tested, assumed to be a fragile X premutation carrier by family history
- Not tested, assumed NOT to be a fragile X premutation carrier by family history

CGG repeat number

__________________________________

Are you the first person in your extended family to be identified with a fragile X mutation?

- Yes
- No
- I don't know

Please upload a copy of your most recent FMR1 DNA test result. If you cannot locate it or never had the testing, please skip this item. If you do not have a copy available, you may be able to obtain a copy from your primary physician or the lab that provided the testing, which can be uploaded the next time you update your registry information.

How many biological children do you have?

__________________________________

How many of your biological children have fragile X syndrome (full mutation) confirmed by fragile X DNA testing?

__________________________________

How many of your biological children have the fragile X premutation?

__________________________________

How many of your biological children do not have a fragile X mutation (normal result by DNA testing)?

__________________________________

How many of your biological children have not had fragile X DNA testing (their status is unknown)?

__________________________________
Confidential

Have you been diagnosed with fragile X-associated tremor/ataxia syndrome (FXTAS) by a medical professional? *must provide value

- Prefer not to answer
- Yes
- No
- I don't know

Have you been diagnosed by a medical professional with fragile X-associated primary ovarian insufficiency (FXPOI), premature ovarian failure (POF), or abnormally early menopause? *must provide value

- Prefer not to answer
- Yes
- No
- I don't know

FUTURE SHARING OF BIOLOGICAL SAMPLES

Are you interested in providing biological samples (e.g., blood, saliva) in future research studies? *must provide value

- Yes
- No
- Maybe/depends

Are you interested in the possibility of being a tissue donor after death (e.g. brain or other body tissues)? *must provide value

- Yes
- No
- Maybe/depends

SECONDARY CONTACT

Name and address of the secondary contact.

Email address for the secondary contact (if you don't have an email address, please write "NA")

Phone number for the secondary contact (including country code if outside the U.S.). If you do not have a phone number for this person, please write "NA".
How did you hear about this registry? (Check all that apply.)

☐ Prefer not to answer  
☐ National Fragile X Foundation  
☐ Local fragile X support organization  
☐ Family member  
☐ Medical or other health provider  
☐ Social media  
☐ Fragile X clinic  
☐ Researcher  
☐ Friend  
☐ Other - please describe below

Other way you heard about the registry

__________________________________

YOU HAVE COMPLETED THE ESSENTIAL REGISTRY INFORMATION. Thank you!

__________________________________

languages  
☐ English  
☐ Español

DEMOGRAPHICS  The questions in this section will help the registry team and researchers better determine your eligibility for future studies. By answering "yes", the registry survey will give you the opportunity to provide this information (e.g., gender identity, education, marital status, race and ethnicity, and languages you speak). You can choose to answer or not answer any question. Answering "no" will take you to the next section. Do you choose to provide demographic information? *must provide value

☐ Yes  
☐ No

Gender  
☐ Prefer not to answer  
☐ Female  
☐ Male  
☐ Transgender female  
☐ Transgender male  
☐ Gender non-binary  
☐ Not listed
Confidential

Education level

- Unknown or prefer not to answer
- No schooling completed
- 8th grade or less
- 9th-12th grade-no diploma
- High school diploma
- GED or alternative credential
- Partial college
- Associates degree (for example: AA, AS)
- Bachelor's degree (for example: BA, BS)
- Master's degree (for example: MA, MS, MEng, MEd, MSW, MBA)
- Professional degree beyond bachelor's degree (for example: MD, DDS, DVM, LLB, JD)
- Doctorate degree (for example, PhD, EdD)

Marital status

- Prefer not to answer
- Married
- Living with a partner
- Divorced
- Separated
- Never been married
- Widowed

Most recent occupation

- Unknown or prefer not to answer
- Management Occupations
- Business and Financial Operations Occupations
- Computer and Mathematical Occupations
- Architecture and Engineering Occupations
- Life, Physical, and Social Science Occupations
- Community and Social Service Occupations
- Legal Occupations
- Education, Training, and Library Occupations
- Arts, Design, Entertainment, Sports, and Media Occupations
- Healthcare Practitioners and Technical Occupations
- Healthcare Support Occupations
- Protective Service Occupations
- Food Preparation and Serving Related Occupations
- Building and Grounds Cleaning and Maintenance Occupations
- Personal Care and Service Occupations
- Sales and Related Occupations
- Office and Administrative Support Occupations
- Farming, Fishing, and Forestry Occupations
- Construction and Extraction Occupations
- Installation, Maintenance, and Repair Occupations
- Production Occupations
- Transportation and Materials Moving
- Homemaker
- Other - list below

Other occupation

__________________________________
Employment status

- Prefer not to answer
- Full time (more than 35 hours/week)
- Part time (less than or equal to 35 hours/week)
- Unemployed
- Student
- Not working, disabled
- Retired

Income

- Unknown or prefer not to answer
- Less than $10,000
- $10,000 to $19,999
- $20,000 to $34,999
- $35,000 to $49,999
- $50,000 to $74,999
- $75,000 to $99,999
- $100,000 to $149,999
- $150,000 to $199,999
- $200,000 to $299,999
- $300,000 or more

Race (please choose ALL that apply, and then specify below in primary origin questions)

- Unknown or prefer not to answer
- African or Black
- Native, Indigenous, First Nations
- Asian
- White
- Hispanic or Latino/Latina

Describe white origin (e.g., English, Italian, German)

______________________________________________

Describe Black origin (e.g., Nigerian, Jamaican, Somali)

______________________________________________

Describe Asian origin (e.g., Vietnamese, Chinese, Japanese, Korean, Hmong, Pakistani)

______________________________________________

Describe native or indigenous origin (e.g., Australian Aboriginal, Aztec, Navajo)

______________________________________________

Describe Hispanic or Latino origin (e.g., Mexican, Spanish, Cuban, Salvadoran)

______________________________________________
Religion

- Prefer not to answer
- Protestant
- Catholic
- Mormon
- Orthodox such as Greek or Russian Orthodox
- Jewish
- Muslim
- Buddhist
- Hindu
- Atheist
- Agnostic
- Nothing in particular
- Other (describe below)

Other religion:

__________________________________

YOU HAVE COMPLETED THE DEMOGRAPHICS REGISTRY INFORMATION. Thank you!

HEALTH INFORMATION

The questions in this section will help the registry team and researchers better determine your eligibility for future studies. By answering "yes", the registry survey will give you the opportunity to provide this information (e.g., height and weight, neurological difficulties, psychological difficulties, reproductive health for females, general health conditions). You can choose to answer or not answer any question. Answering "no" will take you to the next section.

Do you choose to provide health information?

*must provide value

- Yes
- No

Metric Standard

- Metric (cm/kg)
- Standard/Imperial (ft/lbs)

Height (centimeters)

__________________________________

Weight (kilograms)

__________________________________

Height (feet) - enter additional inches below

__________________________________
Height (additional inches)

__________________________________

Weight (pounds)

__________________________________

Neurological symptoms

☐ Prefer not to answer
☐ Resting tremor (tremor when the muscle is relaxed and the body is at rest)
☐ Intention tremor (tremor during purposeful movement to a target, or to perform an action, such as reaching or a cup or while handwriting)
☐ Postural tremor (tremor when holding a position, such as when arms are outstretched)
☐ Head tremor
☐ Slowness of movements
☐ Balance problems
☐ Walking problems
☐ Repeated falls
☐ Use of cane or walker
☐ Use of a wheelchair
☐ Numbness or tingling sensations in legs, feet, arms or hands
☐ Swallowing or choking problems
☐ Migraine headache
☐ Parkinson's disease or parkinsonism
☐ Dementia or Alzheimer's disease
☐ Vertigo or dizziness
☐ Hearing loss
☐ Chronic pain
☐ Fibromyalgia
☐ None of the above
☐ Other

Other Neurological Problems
Autoimmune

☐ Prefer not to answer
☐ Rheumatoid arthritis
☐ Psoriasis/psoriatic arthritis
☐ Multiple Sclerosis
☐ Systemic lupus erythematosus
☐ Inflammatory bowel disease
☐ Addison's disease
☐ Grave's disease
☐ Hyper-thyroidism
☐ Sjögren's syndrome
☐ Hashimoto's thyroiditis
☐ Hypo-thyroidism
☐ Myasthenia gravis
☐ Celiac disease
☐ Pernicious anemia
☐ Autoimmune vasculitis
☐ None of the above
☐ Other

Other Autoimmune

POI Symptoms

☐ Prefer not to answer
☐ Irregular or skipped periods
☐ Hot flashes
☐ Mood swings
☐ Problems with fertility
☐ Osteopenia or osteoporosis
☐ None of the above

Menopause status

☐ Prefer not to answer
☐ Pre-menopause (before menopause; having regular periods)
☐ Peri-menopause/menopause transition (changes in periods, but have not gone 12 months in a row without a period)
☐ Post-menopause (after menopause)
☐ I am not sure

Menopause Age

__________________________________
Confidential

Other Health Conditions

☐ Prefer not to answer
☐ Cardiac/heart disease
☐ Hypertension (high blood pressure)
☐ Hypo-tension (abnormally low blood pressure)
☐ Kidney disease
☐ Liver disease
☐ Sleep apnea
☐ Respiratory disease
☐ Type I Diabetes
☐ Type II Diabetes
☐ Alcohol or substance use problems
☐ Sleep disorder
☐ Any type of cancer
☐ Sexual dysfunction
☐ Bladder or bowel incontinence
☐ Other (please describe below)
☐ None of the above

Other medical problems

Psychiatric

☐ Prefer not to answer
☐ Anxiety (e.g., phobias/fears, generalized anxiety, social anxiety, panic)
☐ Depression (e.g., major depression, dysthymia, postpartum depression)
☐ Bipolar disorder or mania
☐ Stress-related disorder [post-traumatic stress disorder (PTSD), acute stress disorder, adjustment disorder]
☐ Eating disorder (anorexia, bulimia)
☐ Sleep disorder (insomnia, restless legs syndrome)
☐ Alcohol or substance use problem or diagnosis
☐ Psychotic disorder such as schizophrenia
☐ Obsessive-compulsive type disorder (OCD, hoarding disorder, skin picking, hair pulling)
☐ Personality disorder
☐ Attention deficit hyperactivity disorder (ADHD)
☐ Autism spectrum disorder/Asperger's disorder
☐ Intellectual disability or developmental delay
☐ Specific learning disorder (dyslexia, math disorder)
☐ Language or communication disorder
☐ Tourette's disorder or other tic disorder
☐ Other (Please describe below)
☐ None of the above

Other Psychiatric Problems

YOU HAVE COMPLETED THE REGISTRY HEALTH INFORMATION. Thank you!

YOU HAVE COMPLETED ALL THE SECTIONS OF THE REGISTRY. Thank you! In order for your responses to be entered into the Registry, you must click "Submit". You will be provided with a copy of your responses in PDF format.