

Geneial

A stylized graphic of a DNA double helix in a light blue color, positioned to the right of the word 'Geneial'. The helix is oriented vertically, with the top of the helix pointing upwards and the bottom pointing downwards. The two strands of the helix are represented by thick blue lines, and the base pairs are represented by horizontal bars connecting the two strands.

Engineering Equitable Health Futures

Xia-Gibbs Society Webinar
September 16, 2023

Agenda – 6 “Ws”

- **History of Geneial + XGS**
- **What** is a registry?
- **Why** is a registry important?
- **“Where”** is the registry accessed?
- **Who** is involved in the XGS Registry?
- **How** do I participate?
- **When** can I join the new platform?
- **Q&A**

Announcing Partnership with HGSC



Pictured: Dr. Hansen, Dr. Gibbs, and HGSC researchers gather for Xia-Gibbs Syndrome Awareness Week.

Hi there,

Welcome to our inaugural newsletter! We're kicking off 2022 with some exciting news. You may have noticed the addition of a news section on our website, with a [press release](#) we've been eager to share.

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History of Geneial + XGS

Announcing Partnership with HGSC



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Geneial's Mission

Our mission is to help **relieve human suffering** through accelerating precision and genomic medicine.

We are a software company providing a framework advancing understanding of **rare genetic disorders** and how to treat them.

Our platform solutions enable **personal health advocacy** while **contributing to research**.

We have worked closely with BCM and XGS Society to guide our development.



What is a registry?

A collection of data about individuals and families living with a specific diagnosis, e.g. Xia-Gibbs Syndrome (XGS)

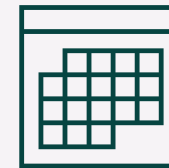


Could include...

- Medical history (symptoms, past treatments)
- Social factors that could influence lived experience
- Measurements of “quality of life”



May include follow-up information over an extended period of time



Data is structured and standardized to the fullest extent possible



Why is a registry important?

With each family that participates, we **build the community**.

Registries can help healthcare providers make treatment decisions, with the ultimate goal of improving **quality of life**.

As registries grow, they become an increasingly valuable **resource to attract external partners**.

Future features:

- Community connectivity & knowledge sharing
- Medical history organization



Why is a registry important? (continued)

Foundation of all XGS research

- How changes in *AHDC1* cause XGS
- Future therapy development as research progresses
- Potential opportunities for clinical trials

Accelerates timeline to publication

Lowers cost of research

→ Continued progress towards understanding and improving **quality of life** with XGS

ORIGINAL ARTICLE

The phenotypic spectrum of Xia-Gibbs syndrome

Yunyun Jiang, Michael F. Wangler, Amy L. McGuire, James R. Lupski, Jennifer E. Posey, Michael M. Khayat,

D RESEARCH ARTICLE

Phenotypic and protein localization heterogeneity associated with *AHDC1* pathogenic protein-truncating alleles in Xia-Gibbs syndrome

HGG
Advances

ARTICLE

AHDC1 missense mutations in Xia-Gibbs syndrome

Michael M. Khayat^{1,2,11}, Jianhong Hu^{1,11}, Yunyun Jiang^{1,11}, He Li^{1,11}, Yuesun Chander^{1,2},

Xia-Gibbs syndrome in adulthood: a case report with insight into the natural history of the condition

R. Wangler^{2,11}

Human Mutation

Variation, Informatics, and Disease



Khayat^{1,2},

:hartz⁵,

RESEARCH ARTICLE

Long read sequencing and expression studies of *AHDC1* deletions in Xia-Gibbs syndrome reveal a novel genetic regulatory mechanism

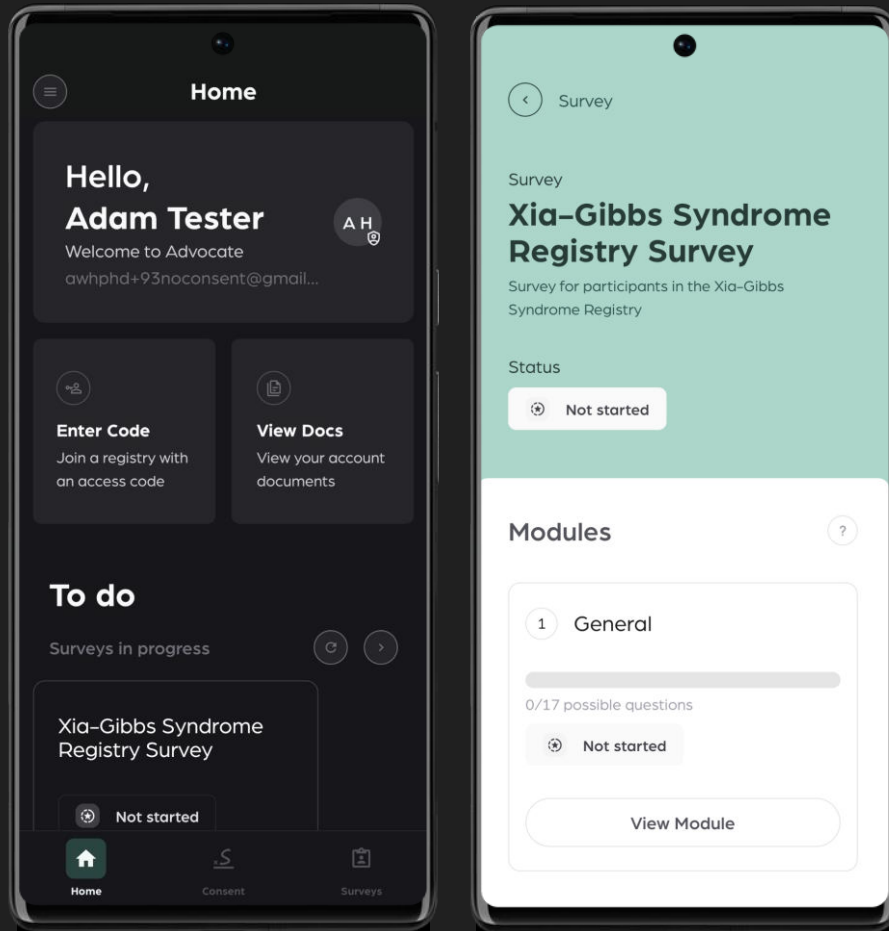
Varuna Chander, Medhat Mahmoud, Jianhong Hu, Zain Dardas, Christopher M. Grochowski, Moez Dawood, Michael M. Khayat, He Li, Shoudong Li, Shalini Jhangiani, Viktoriya Korchina, Hua Shen, George Weissenberger, Qingchang Meng, Marie-Claude Gingras, Donna M. Muzny, Harsha Doddapaneni, Jennifer E. Posey, James R. Lupski, Aniko Sabo, David R. Murdock, Fritz J. Sedlazeck, Richard A. Gibbs

... See fewer authors ^

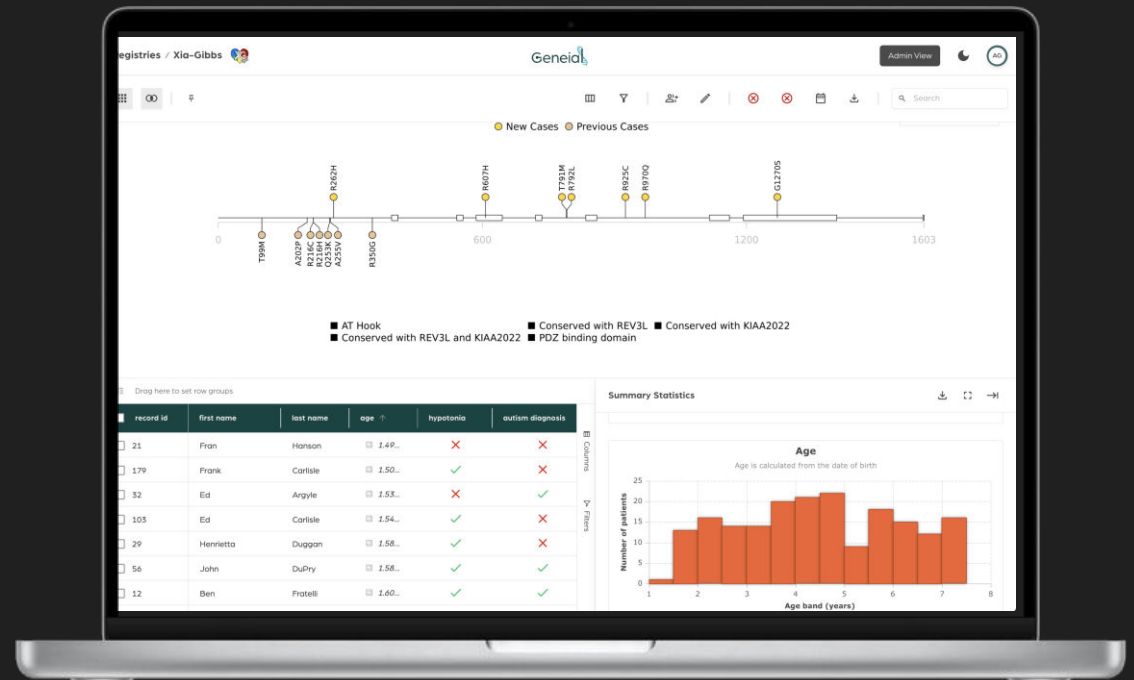
Where is the registry accessed?

Geneial's platform includes a mobile app for participants and a web app for researchers

Advocate (participants)

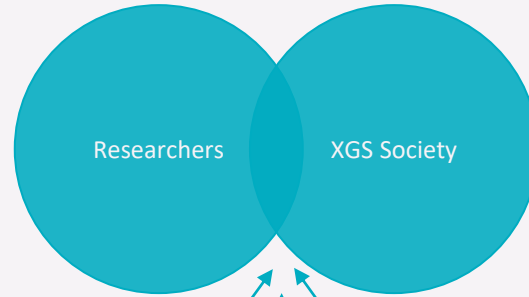


Bridge (researchers)



Who is involved in the XGS Registry?

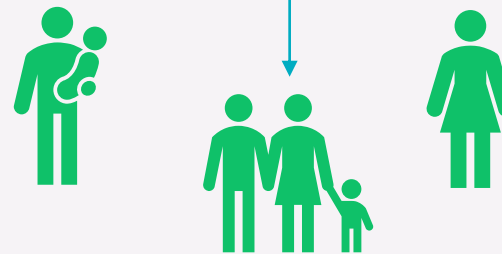
Bridge



(Public summary dashboard)

Advocate

XGS confirmed or potential diagnosis

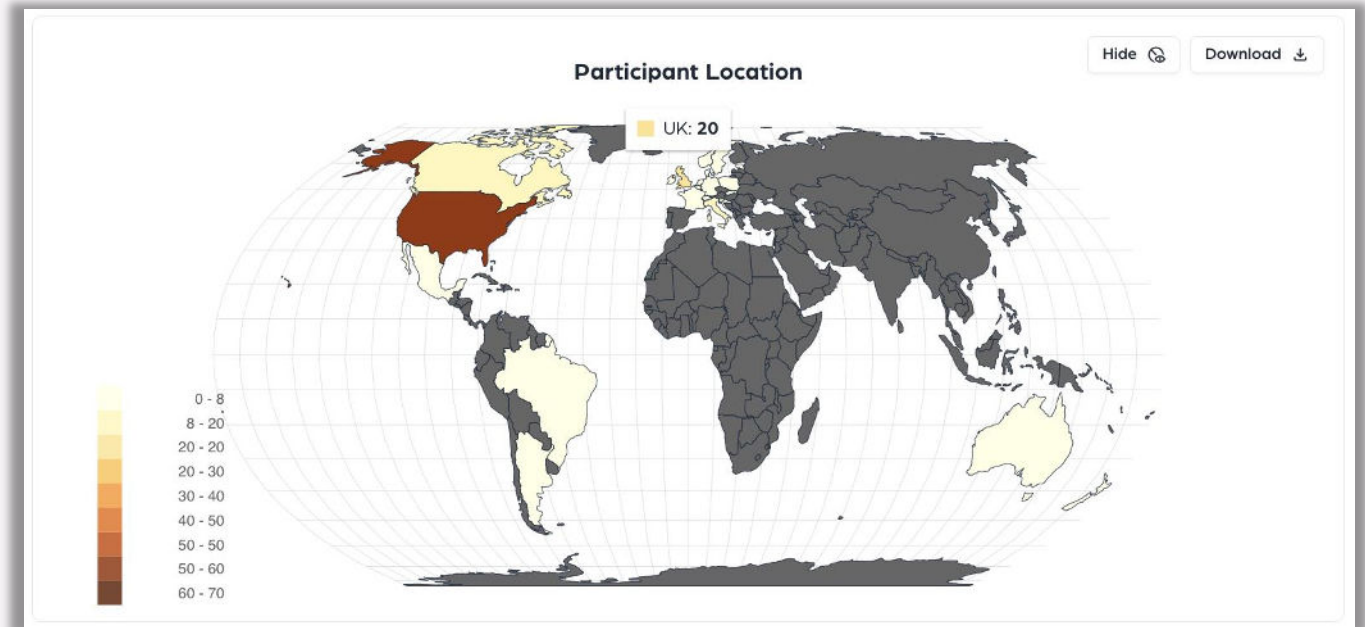
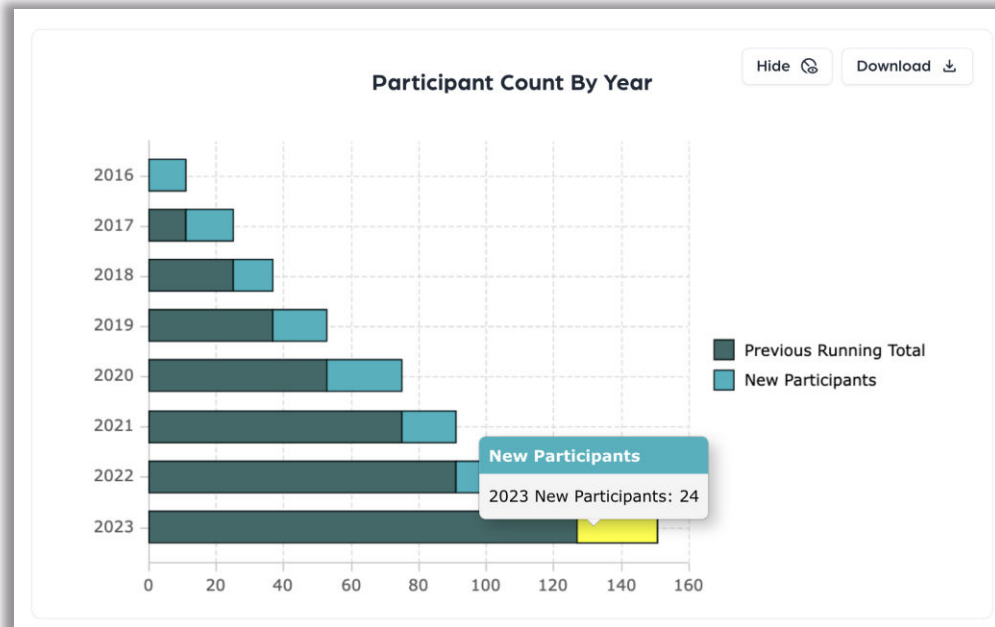


AHDC1 genetic variant

Where can I access registry summary data?

Public summary dashboard

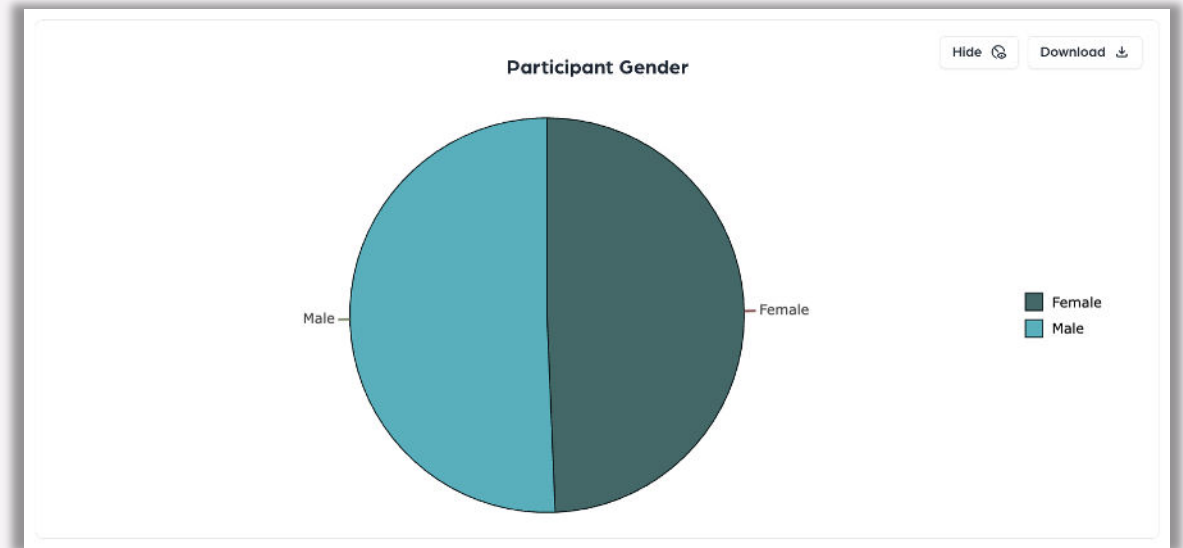
<https://app.geneial.com/stats/dashboard>



Where can I access registry summary data?

Public summary dashboard

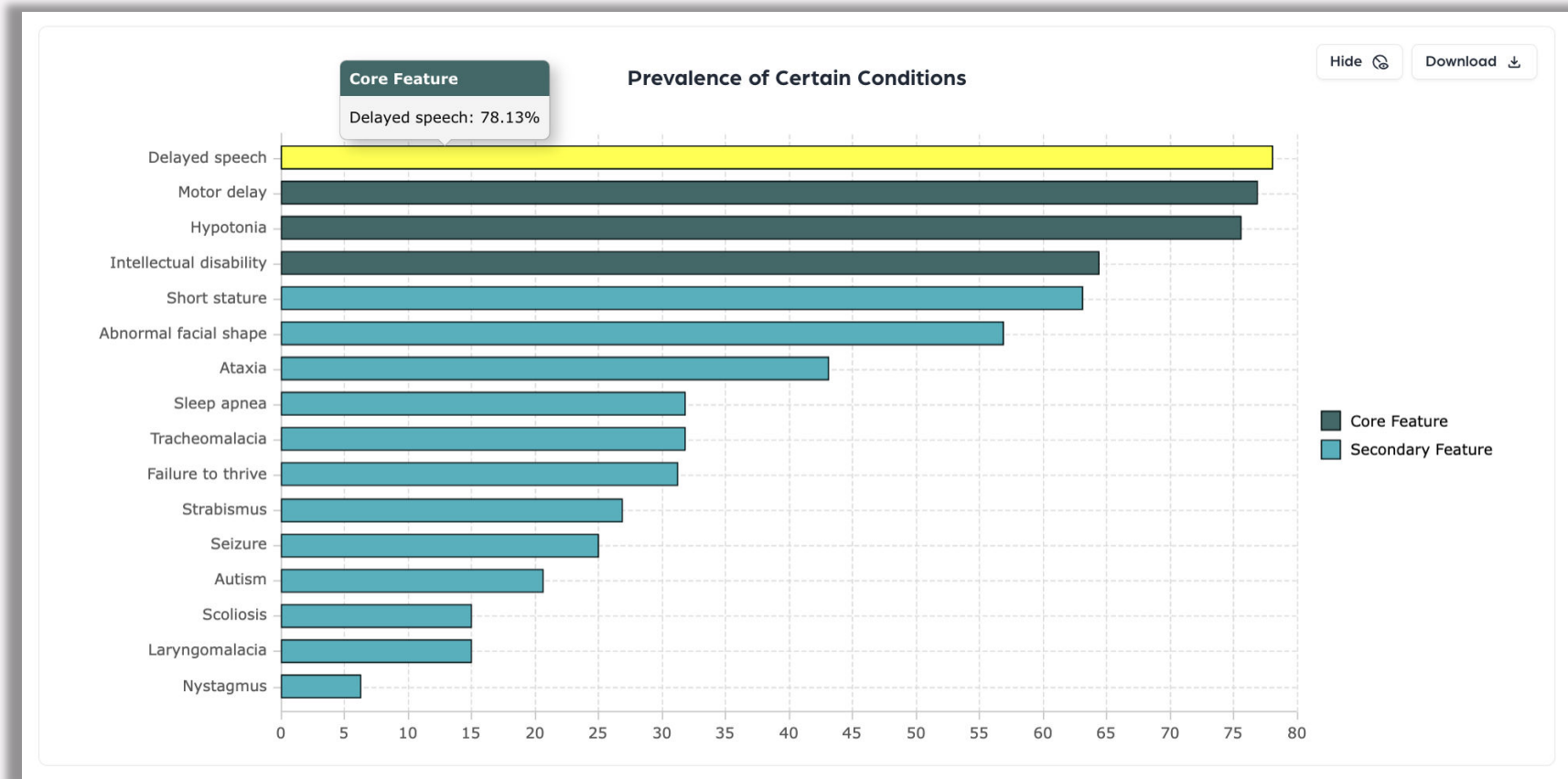
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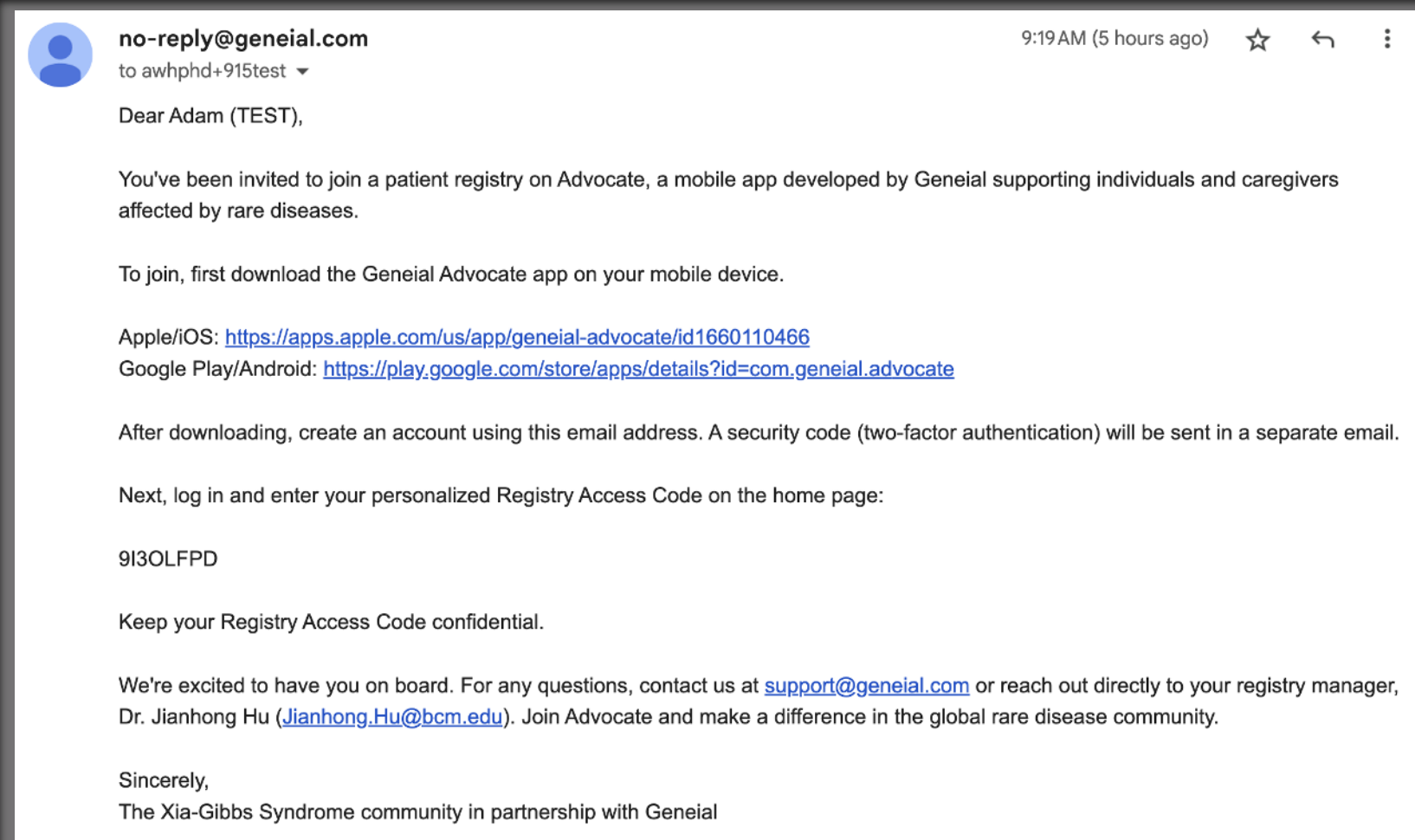
Public summary dashboard

<https://app.geneial.com/stats/dashboard>

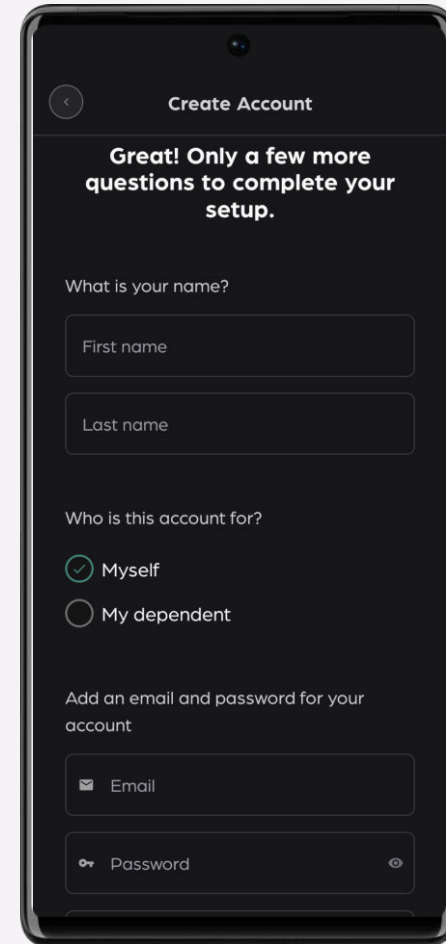
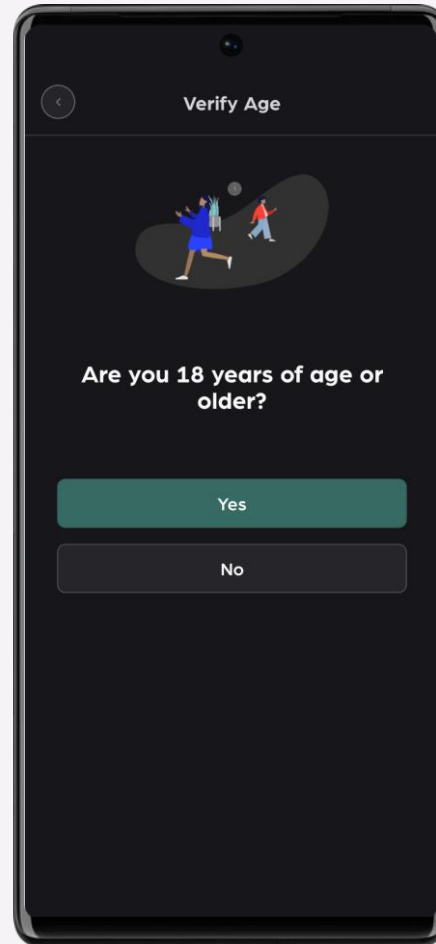
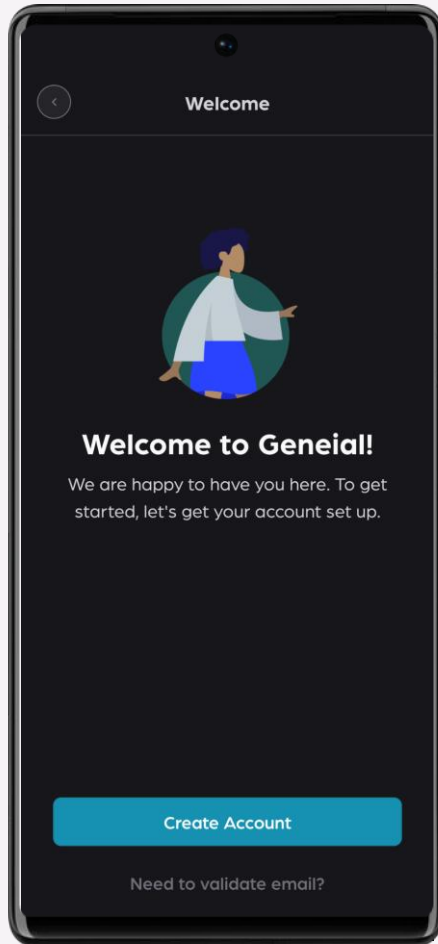


How do I participate?

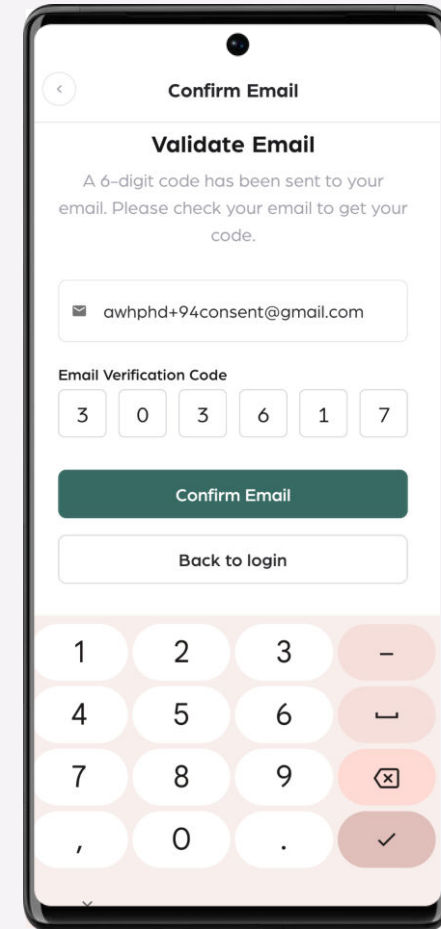
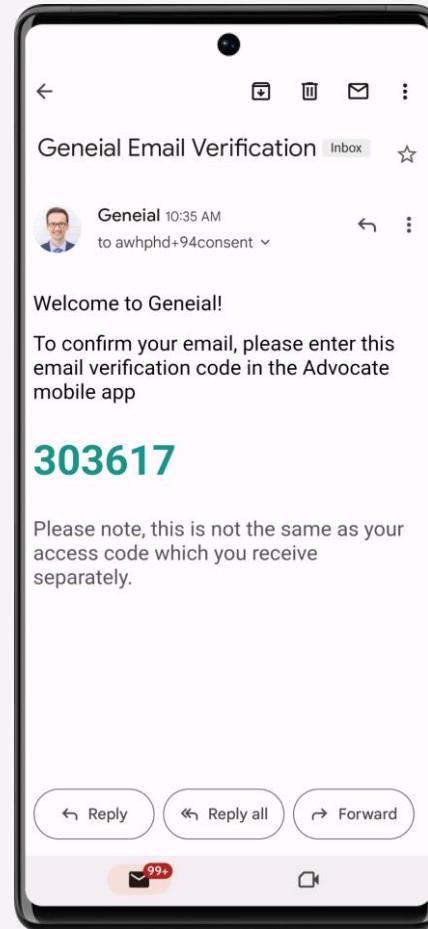
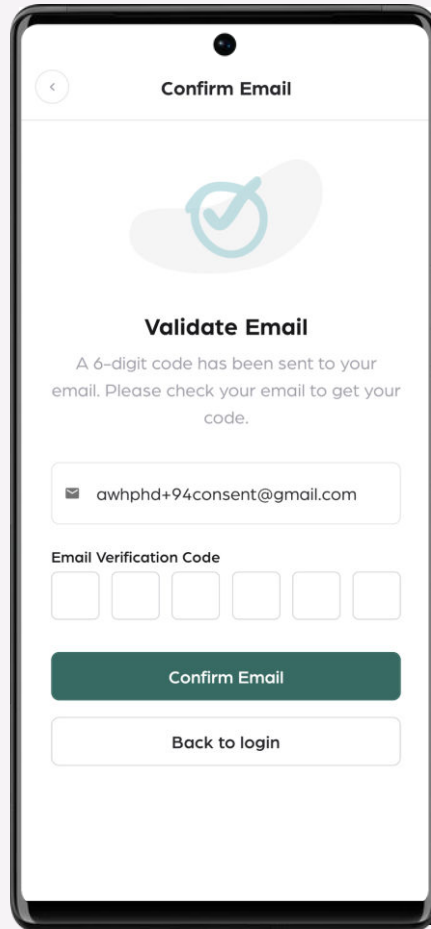
You will receive an invitation email with instructions (sent by HGSC team via Geneial app)



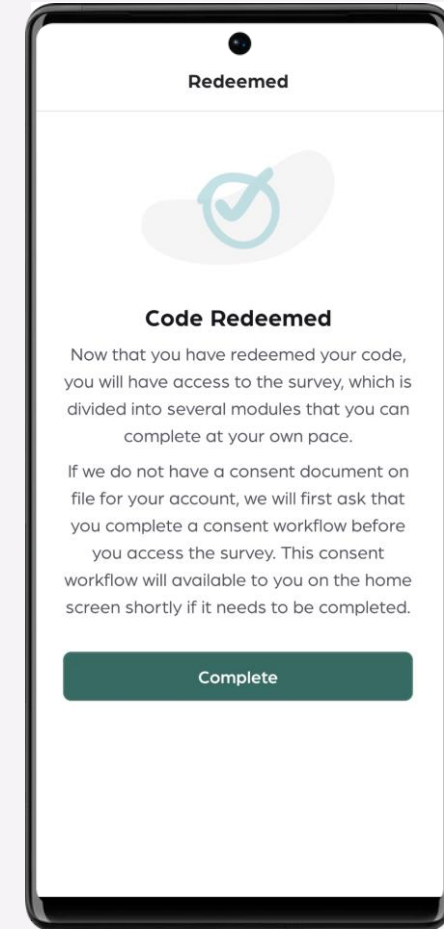
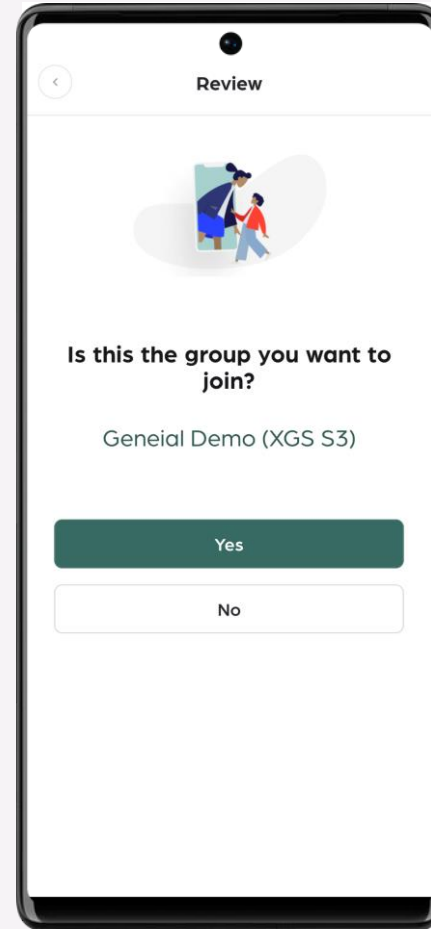
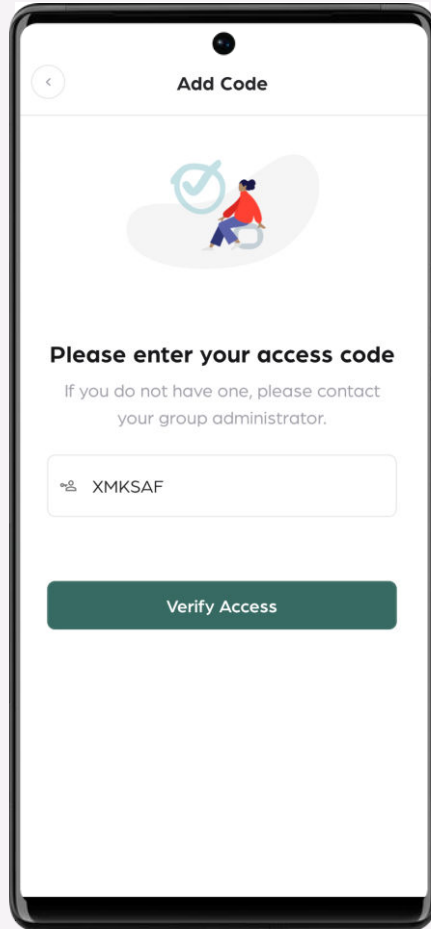
First, create an account on Advocate



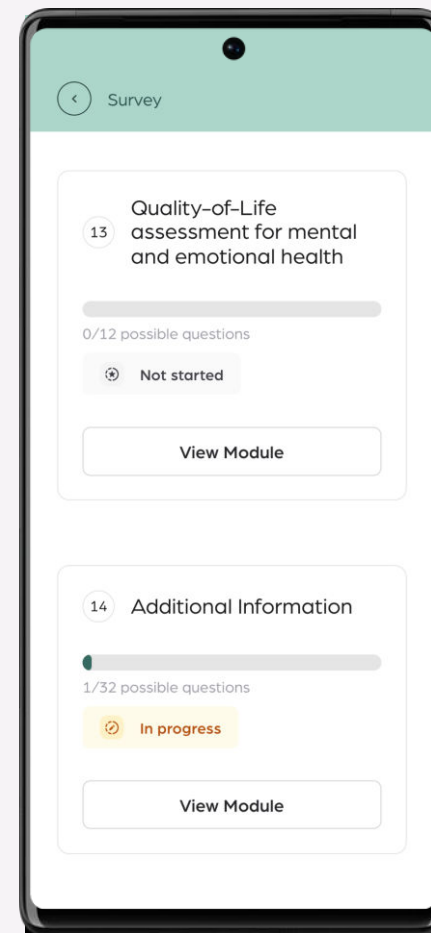
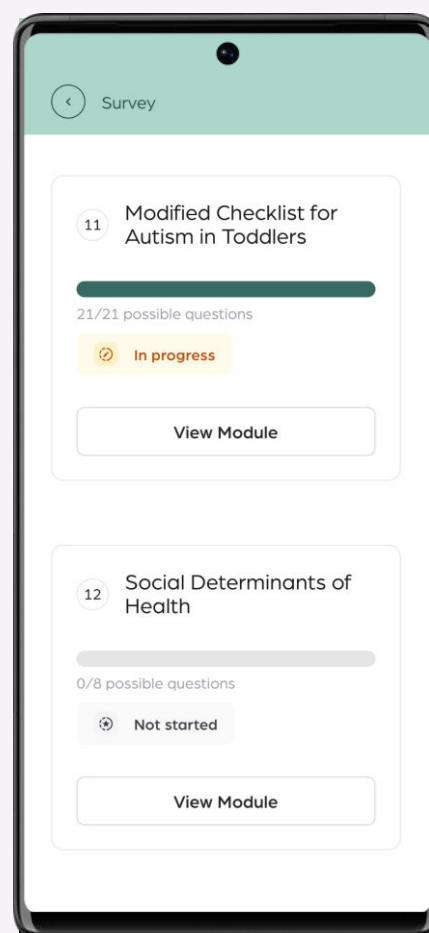
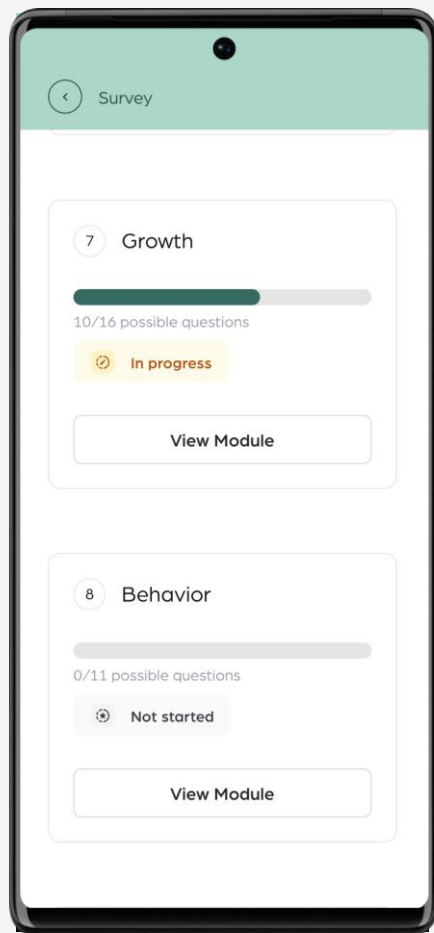
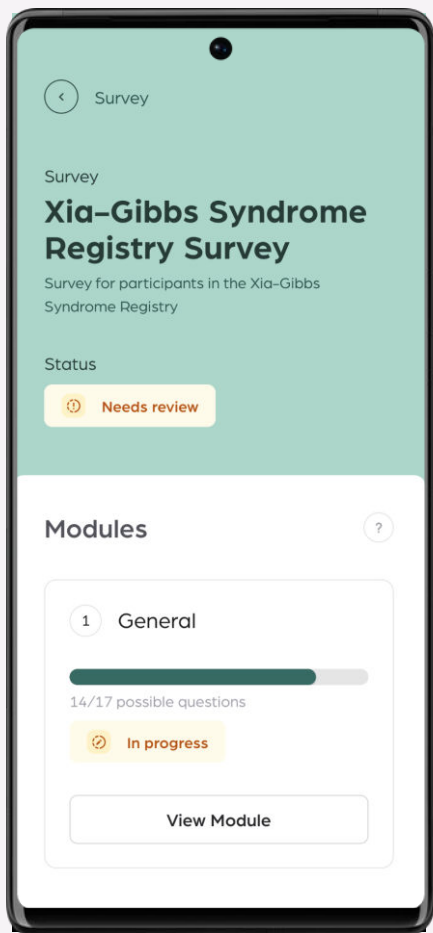
Next, verify your email and login



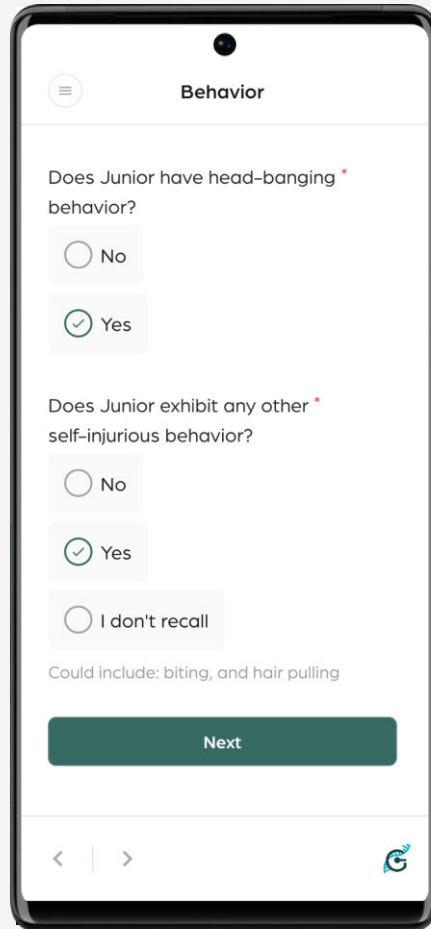
Redeem your Access Code (and complete the consent workflow if needed) to join the XGS Registry



There are currently 14 survey modules to complete at your own pace, including all previous surveys plus several new additions



...If you previously participated, your data is automatically pulled over from the old platform. (Those fields are locked, but you can Request Change after submitting if necessary.)



Behavior

Does Junior have head-banging * behavior?

No

Yes

Does Junior exhibit any other * self-injurious behavior?

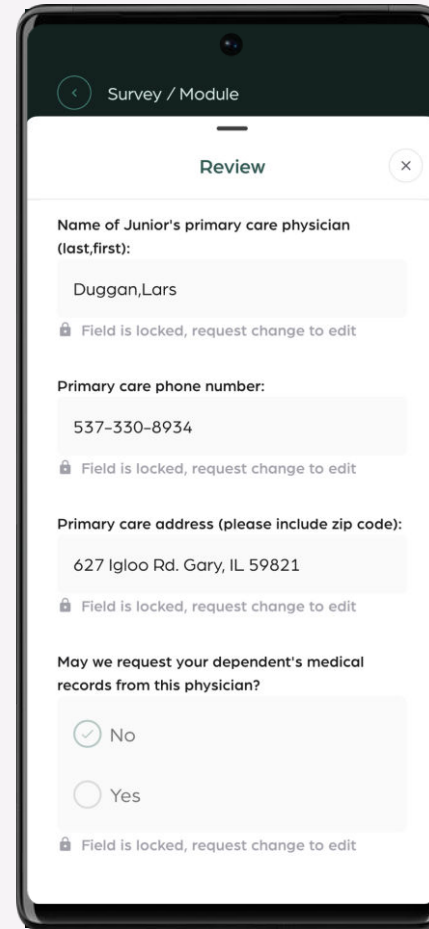
No

Yes

I don't recall

Could include: biting, and hair pulling

Next



Survey / Module

Review

Name of Junior's primary care physician (last,first):

Duggan,Lars

Field is locked, request change to edit

Primary care phone number:

537-330-8934

Field is locked, request change to edit

Primary care address (please include zip code):

627 Igloo Rd. Gary, IL 59821

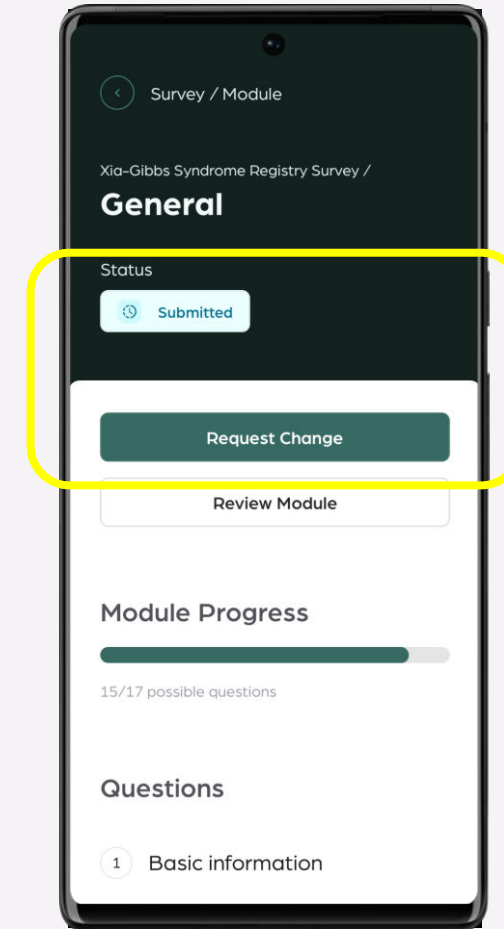
Field is locked, request change to edit

May we request your dependent's medical records from this physician?

No

Yes

Field is locked, request change to edit



Survey / Module

Xia-Gibbs Syndrome Registry Survey /

General

Status

Submitted

Request Change

Review Module

Module Progress

15/17 possible questions

Questions

1 Basic information

When can I join?

Expect an email invitation with your personalized access code **within two weeks**

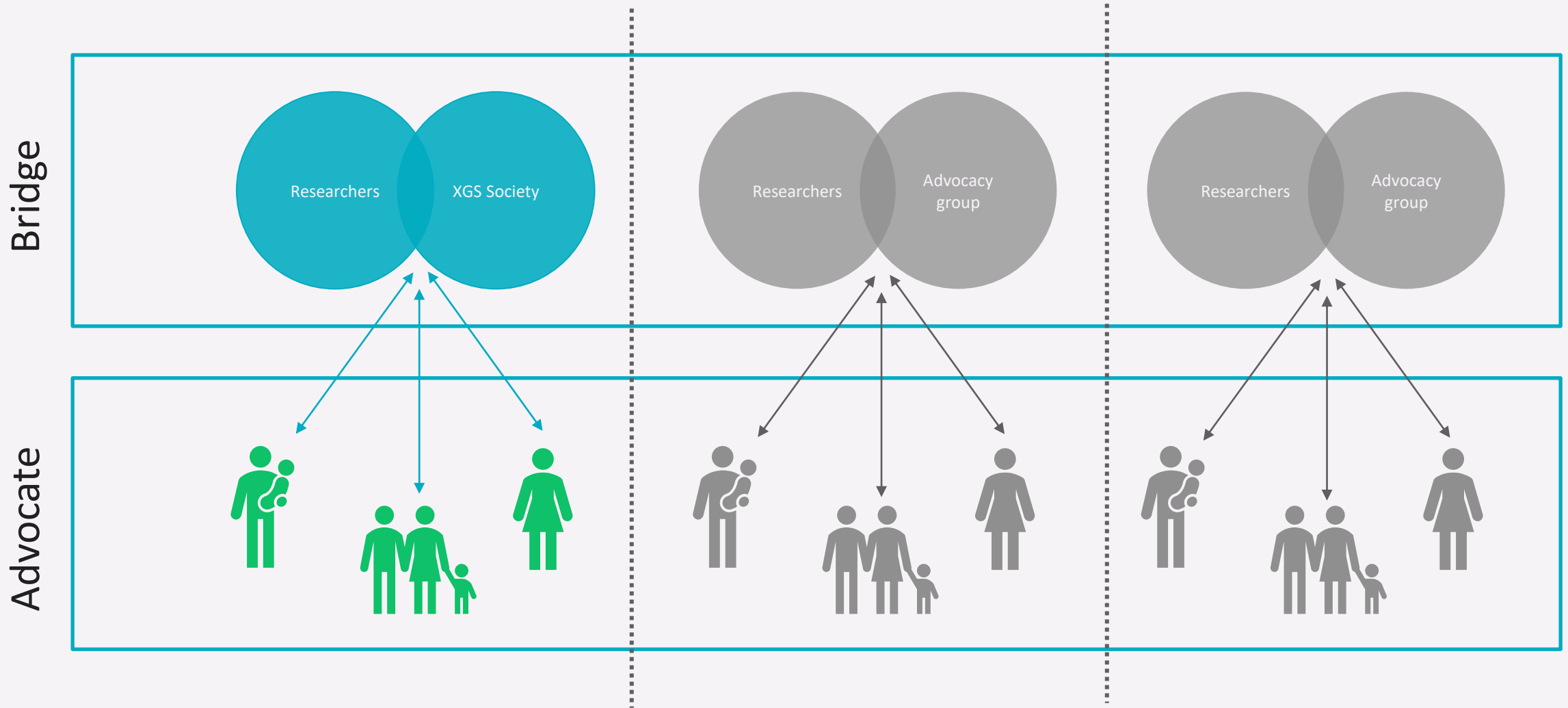
For those who previously participated in Registry:

- We have your contact info from the old platform, will send you an email

For families who have *not* previously participated in the Registry

- We have your contact info from Zoom Webinar registration, will send an email
- If you don't receive an invite after the start of October, email Dr. Jianhong Hu at jianhong.hu@bcm.edu

Strength in numbers over time -> Registry network





Acknowledgements

Alpha testing from XGS society leaders

Direct support from XGS Society in collaboration with BCM, which has led to a 2+ yrs guaranteed support from the NIH: National Human Genome Research Institute SBIR funding

All of the society members and registry participants

Geneial team



Geneial Registry Contributors



Dustin Belliston
Senior Engineer



Emily Little
Bioinformatics Engineer



Katrisa Ward
Bioinformatics Engineer



Russ Greer
Senior Engineer



Chris Hansen
COO & President

Questions?

