PATRE

PATient based phenotyping and evaluation of therapy for Rare Epilepsies

IQSEC2 Natural History Study

International patient registry for IQSEC2-related disorders

Every family counts!

Together we are building the largest international collection of data on IQSEC2. Every completed registration brings us closer to better care and future therapies. Please help us get as many families as possible over the finish line!

212 families registered

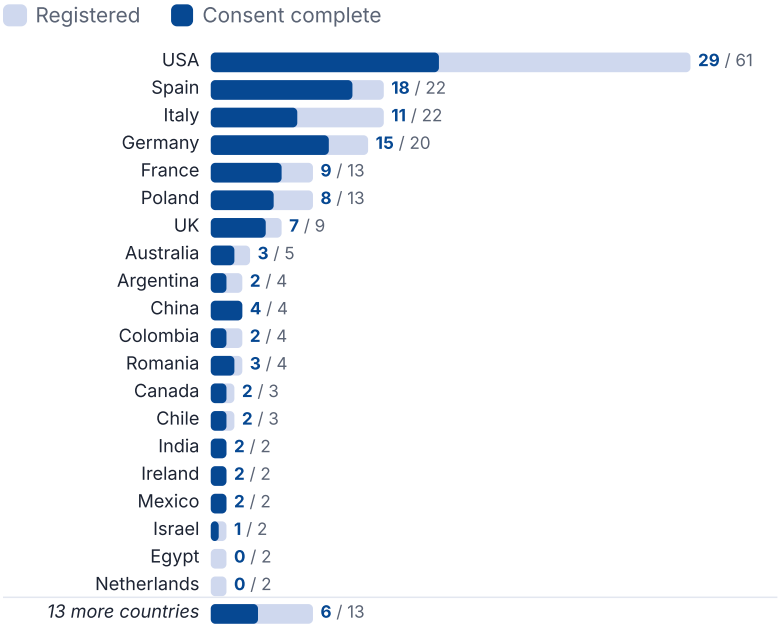
33 countries

128 consent forms complete

63 families have completed all core questionnaires

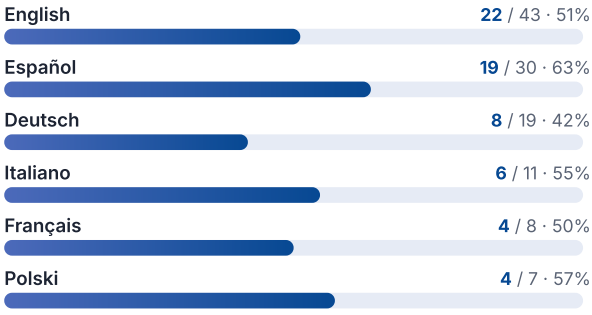
Status: 8 November 2026

Where our families come from



Progress in the app

Families in the app who have completed all 5 core questionnaires



84 registered families are still missing a complete consent form, and **55 families in the app** still have open questionnaires.

What is your next step?

1

Not registered yet?

Register at **patre.info** or scan the QR code. You will then receive the consent and data protection documents.

2

Registered, but consent still missing?

Please sign the consent form and upload it. We will send you your personal QR code for the app **within one week**.

3

Already in the app?

Please complete the **5 core questionnaires**: Genetics, Basic Information, Development, Behavior and Epilepsy, step by step on your smartphone.

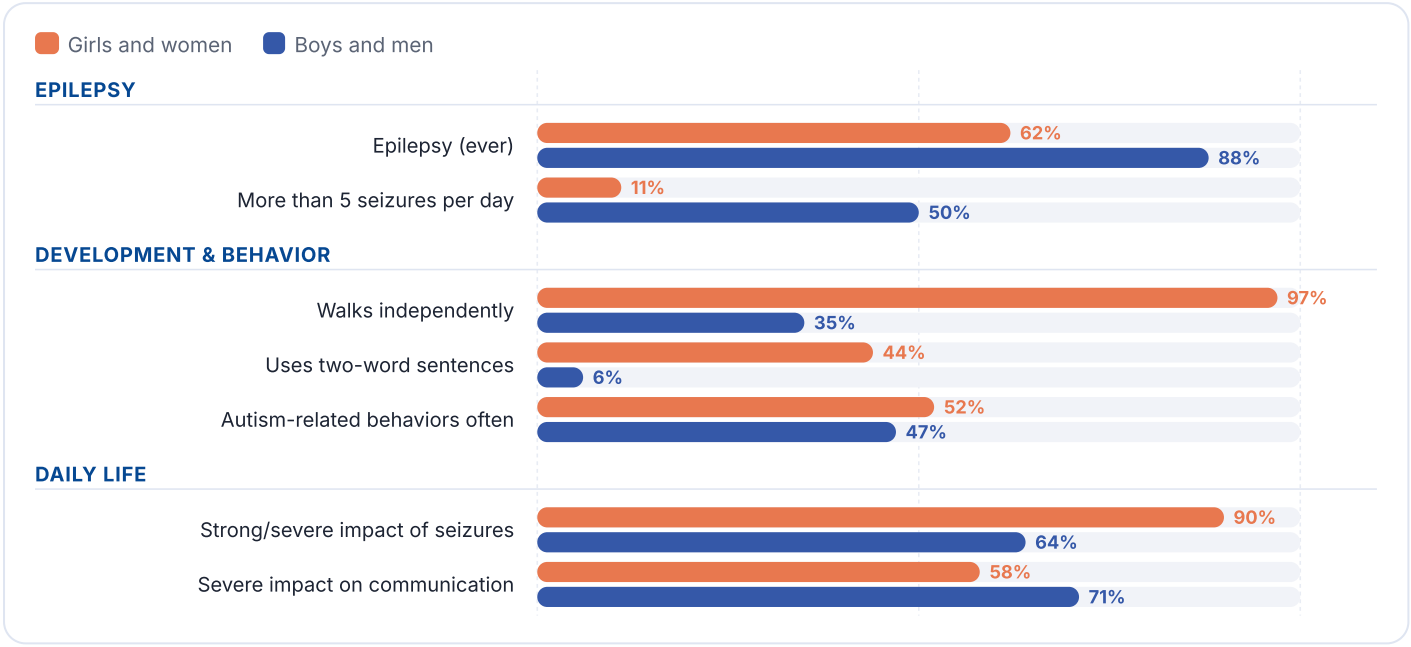
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IQSEC2 Natural History Study · PATRE

What we have already learned

First baseline results from the PATRE registry (118 registrations, data as of August 2026) will be presented at the American Epilepsy Society meeting (AES 2026) in Denver.



Caregiver-reported data; preliminary results, not age-adjusted; percentages based on available answers.

Different for boys and girls

Boys and men more often had epilepsy and very frequent seizures; girls and women more often walked independently and used two-word sentences, as expected for a gene on the X chromosome.

A high burden for all families

Caregivers of both girls and boys rated the impact of seizures and communication difficulties on daily life as substantial.

Why your complete data matter

At the time of the analysis, seizure frequency could only be evaluated for 19 girls and 28 boys. Every completed questionnaire makes the results more reliable and helps prepare future clinical trials.

What is PATRE?

PATRE (PATient-based phenotyping and evaluation of therapy for Rare Epilepsies) is a natural history study that systematically collects information on people with rare genetic epilepsies. For IQSEC2, families from all over the world take part in six languages, directly from their smartphone.

Who can take part?

People with a genetic diagnosis of IQSEC2, or their parents or legal guardians, from any country.

How are my data protected?

Data are collected via a secure app and stored on a secure server in the EU. Personal data are stored separately from medical data. The study has been approved by an ethics committee (Bavarian State Medical Association).

Study team

Kirsten Eschermann, MD · Lorenz Kiwull, MD
Paracelsus Medical University, Salzburg, Austria
team@patre.info · **patre.info**

PARACELSUS MEDICAL UNIVERSITY

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