What is PATRE - SYNGAP1?

PATRE - SYNGAP1 is a natural history study for systematic collection of data on individuals affected by SYNGAP1. It is used to collect, analyze, and utilize health information in order to improve research, diagnosis, therapy development, and care, as well as to promote understanding of the disease.

Why should I take part?

- The data from PATRE - SYNGAP1 is used directly by the EURAS research consortium for research on therapies
- As a cross-border patient registry it enables meaningful results
- Every single data record is important in rare diseases
- Help to improve diagnosis, care, and therapy development
- Preparation for and easier access to clinical trials
- The project is supported by SYNGAP Global Network

About us

The study is medically led by:
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Patient recruitment is led by the EURAS Patient Board Officer, Verena Schmeder, and is supported by members of the EURAS Patient Board and patient organizations for SYNGAP1.

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Funded by the European Union
Grant No. 101080580

PATRE

PATient-based phenotyping and evaluation of therapy for Rare Epilepsies

PARACELSUS MEDICAL UNIVERSITY

SYNGAP ELTERNHILFE Gemeinsam sind wir stark

EURAS

Patient Research Partnership – Towards New Treatments for Neurodevelopmental RASopathies

SYNGAP1

euras-project.eu

What is a RASopathy?

RASopathies are a group of genetic disorders that can affect physical, neurological, and psychological development to different degrees. They include Noonan syndrome, CFC syndrome, Costello syndrome and also SYNGAP1.
Since these diseases share a common signaling pathway, the RAS pathway, they are being researched jointly in our project EURAS. The various genetic mutations have a major influence on the severity of the clinical picture. The exact biological mechanisms underlying the symptoms of the disease are still poorly understood, and there are currently few targeted treatment approaches. This is particularly true for neurological and psychological developmental disorders.

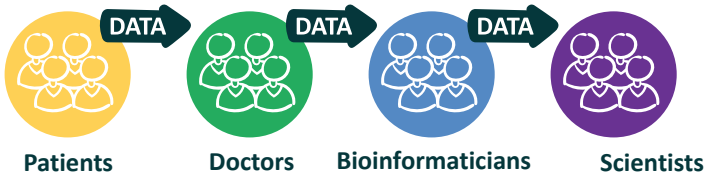
Who can take part in PATRE - SYNGAP1?

All affected individuals or parents or legal guardians of affected individuals can register individuals with SYNGAP1.
Have you received a genetic diagnosis for SYNGAP1? If you have, we invite you to participate in PATRE - SYNGAP1.

How will my data be used?

The data from PATRE - SYNGAP1 are used directly by the EURAS research consortium. This EU-funded project aims to gain a better understanding of the diseases and find new treatment options to improve the quality of life of patients and their families.

The patient data collected is anonymized, evaluated, and made available to researchers. Anonymized data may also be used for other research projects.



How are my data protected?

Your data is collected via a secure digital app (myCap) and stored on a secure server within the EU. Personal data is stored separately from your medical data. The handling and protection of your data has been reviewed and approved by an ethics committee and the data protection officer.

How do I take part in the study?

1.

Register online

Visit the website **www.syngap1.eu** or scan the **QR code**. After completing the registration form, you will receive consent and data protection documents to sign and a link for uploading.

2.

Your personal invitation

After receiving your consent, we will send you your personal invitation with a link and a QR code for the myCap app.

3.

Using the myCap app

The myCap app makes it very easy to participate in our registry. At various intervals, you will receive short tasks/questionnaires that you can answer on your smartphone while enjoying a cup of coffee or tea.