

What is a Patient Registry?

A patient registry simply stores an individual's personal information and their medical history. Since registries are usually disease-specific, the collective data of all patients that share a disease help clinicians and researchers to better understand causes of the disease and potentially lead to discoveries that can help the patient groups.

Why is a Patient Registry important?

A critical amount of clinical information is needed to understand what is causing any disease and the purpose of registries are to capture information from each patient that may shed light on understanding how to pursue treatments. Since SYNGAP is rare, there are so few patients; it is critical that every child is registered. We are just at the beginning of bringing together this information in order to understand the diversity of the disease.

"A patient registry will directly connect caregivers to the brain scientists working at the front lines to develop cures for SYNGAP1 disorders. Such a strong connection would ensure the work of researchers is focused on the most pressing needs of SYNGAP1 patients."

— **Gavin Rumbaugh, PhD**
Department of Neuroscience, Scripps Florida

About Us

Bridge the Gap - SYNGAP is a non-profit education and research organization whose mission is to serve, educate and fund research for families coping with the effects of SYNGAP mutations.

Contact Us

Phone: (832) 671-0010
Email: bridge.syngap@yahoo.com
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Our Partners



RARE Science is a non-profit research organization whose mission is to accelerate finding therapeutics for children with rare disease through a common computational platform that enables data sharing across all stakeholders.

E-mail: info@rarscience.org
Web: www.rarscience.org



Agility Clinical is the first orphan drug focused clinical trial services organization in the US. Due to experience and innovation in small trial design the organization has become known for successful Orphan/Rare clinical trial design and execution.

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Bridge the Gap - SYNGAP

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Education & Research Foundation



THE SYNGAP INTERNATIONAL PATIENT REGISTRY:

The First Step in Finding A Cure

Bridging the Gap of all Stakeholders
Across the Globe

What is SYNGAP1?

A mutation in the gene SYNGAP1 results in intellectual disability that ranges from moderate to severe. Attention deficits, impulsivity, and/or mood disorders can also be associated with the condition. In addition, two thirds of affected children have epilepsy. SYNGAP1 has been linked to autism in recent findings. It is currently unknown how many individuals that have SYNGAP1 have been wrongly diagnosed with autism, which could partly explain the low number of SYNGAP1 individuals.

Early developmental intervention is important to ensure that affected children reach their full potential. Most children benefit from occupational, physical and speech therapy, emphasizing the importance of early diagnosis.

Currently, there are no treatments because researchers and clinicians are still trying to understand the biology of the disease.



Each child is a step on the path to finding a cure

How do I join the SYNGAP Patient Registry?

Joining our patient community is easy and can be started with a call or email to

admin@bridgesyngap.org

The steps are simple: first, sign an informed consent so the medical record information can be collected, and second, retrieving your medical records. This can be completed independently or with our assistance.

A patient registry is critical because it provides information to clinicians about the symptoms due to SYNGAP mutations and how best to treat children with this disorder.

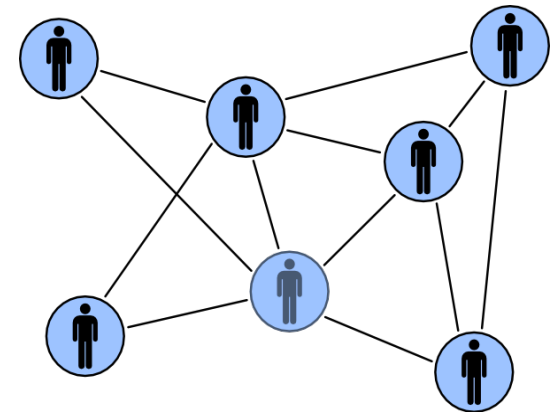
— **Jimmy Holder, MD, PhD**
Blue Bird Circle Clinic for Pediatric Neurology, Texas Children's Hospital

What will we do with the data?

The medical records will be held in a secure HIPAA compliant computer database. Data from medical records will also be put in a common format and entered into a secure database that will be anonymized to ensure privacy but enable analytical analysis to identify biological trends of the disease in one individual and across the patient community. The more patients analyzed help to understand the biology of the disease and enable creation of a "baseline" or measured understanding of the disease for clinical trials.

7 Benefits of having a Patient Registry

1. A connection to the SYNGAP1 community for support and resources
2. A resource and tool to organize and compare medical records
3. As a community, a registry can increase awareness that can drive faster diagnosis
4. Collectively with others, the information builds a foundation for clinical researchers to understand the disease and potential treatments
5. A registry is not bound to any geographical location but can unite patients across the world
6. Can be an avenue to access potential SYNGAP clinical trials
7. Can lead to improved care, even in the absence of a cure



Every child is an important piece of the puzzle