Thank you! Together we are moving SYNGAP1 research forward

Every questionnaire you complete and every genetic report you upload makes research possible. Thank you for your time and trust!

586	47	2
families registered	countries	new studies in 2026

NEW WEBSITE · 7 LANGUAGES

Our SYNGAP1 page now speaks your language

EN DE ES FR IT PL NL

patre.info/syngap1

NEW BROCHURES

Brochure in 25 languages

English · Deutsch · Español · Français · Italiano · Polski · Nederlands · Dansk · Norsk · Svenska · Suomi · Ελληνικά · Čeština · Slovenčina · Slovenščina · Hrvatski · Magyar · Română · Български · Latviešu · Lietuvių · Eesti · Português · Türkçe · Русский

RESEARCH FROM THE REGISTRY

What your data have already shown

Two studies based on PATRE SYNGAP1 data have been published as preprints in 2026. They show what becomes possible when many families share their experience, and why every additional participant matters.

PREPRINT · MEDRXIV · APRIL 2026

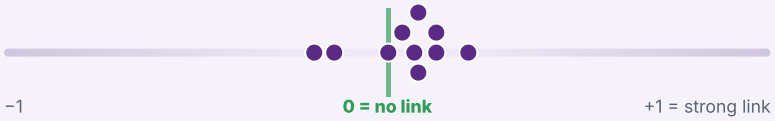
1

Beyond Seizure Burden: Seizure Semiology, but not Frequency, is Associated with Caregiver-Reported Autistic Behaviors in SYNGAP1-DEE

In 337 participants, certain seizure types, above all **atypical absences** and other seizures of the thalamocortical network, were linked to more autism-related behaviors. How *often* seizures occurred was not.

Seizure frequency made no difference to autism

0 / 10 seizure types showed a link between how often seizures occurred and autism-related behaviors



Each dot = one of 10 seizure types (Spearman correlation, none significant)

The seizure type, however, did:



- Thalamocortical seizure types (n = 232)
- No thalamocortical seizure types (n = 105)

Median · 1 = never, 5 = all the time

Why so many participants matter

Only families who had completed **both** the epilepsy and the behavior questionnaire could be included: 337 of 522. Some seizure types were reported by only 16 to 30 people, too few for firm conclusions. Every completed questionnaire makes the next analysis stronger.

Why this matters for clinical trials

If seizure counts alone do not reflect the burden on development and behavior, future trials need additional outcome measures. Registry data help choose the outcomes that really matter to families.

Read the preprint (English) → Preprints, not yet peer-reviewed

Funded by the European Union
Grant No. 101080580

PATRE SYNGAP1 · Paracelsus Medical University, Salzburg · EURAS
Questions about registration: **verena@patre.info** · Medical questions: **team@patre.info**

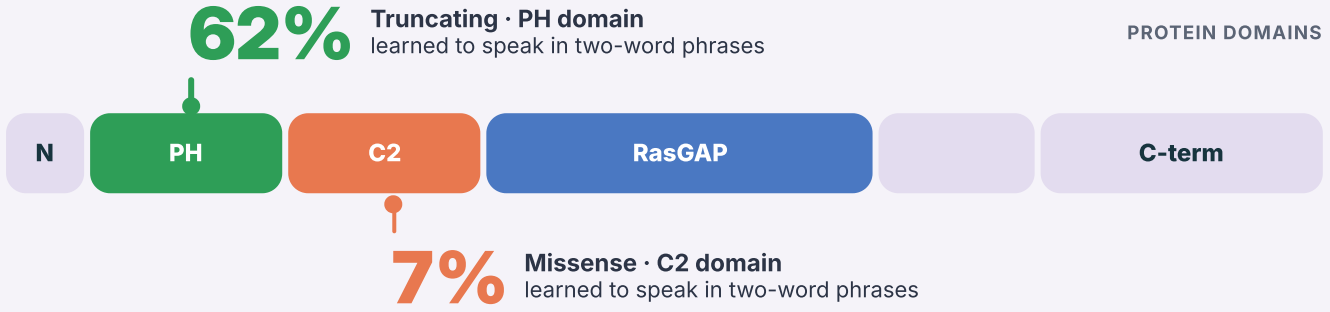
PREPRINT · MEDRXIV · NOVEMBER 2026

2

Beyond Haploinsufficiency: Protein Domain and Variant Class Shape Language Outcomes in SYNGAP1-DEE

Where in the SynGAP protein a variant lies, and what kind of variant it is, is linked to speech development. People with **missense variants in the C2 domain** rarely learned two-word phrases (7%), while people with **truncating variants in the PH domain** did so more often (62%).

Where the variant lies in the SynGAP protein



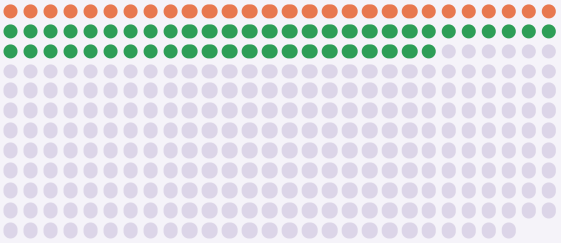
Learning to speak in two-word phrases



Share who have learned to speak two-word phrases

Rare subgroups need many families

28 of 334 participants had a C2 missense variant



Why so many participants matter

Each genetic subgroup is small: only 28 people had a C2 missense variant, and some protein regions were represented by just 3 to 6 people. Only with many families, and an uploaded genetic report, can such subgroups be studied reliably.

Why this matters for clinical trials

Antisense oligonucleotides (ASOs) for SYNGAP1 are approaching clinical trials. Knowing how language develops in each genetic subgroup helps to compose balanced study groups, choose suitable outcome measures and interpret treatment effects correctly.

Read the preprint (English) →

Preprints have not yet been peer-reviewed and should not be used to guide clinical practice. The full articles are in English.

Do you know other families with SYNGAP1?

Please tell them about the registry. Every family counts!

Register for PATRE SYNGAP1 →

Scan to register

Funded by the European Union
Grant No. 101080580

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