The European STXBP1 Consortium Newsletter Winter 2026

Welcome

As we move further into 2026, we're excited to share just how much progress the ESCO community has made in a very short time. The final months of 2025 marked some truly important milestones for STXBP1 research and for our families.

Last quarter, we came together in Heidelberg for the 2nd European STXBP1 Research Roundtable and Family Summit — a powerful moment of connection between families, clinicians, and researchers. Around the same time, another major step forward was reached: the first Natural History Study (NHS) site was initiated, and shortly after, the first patient was enrolled.

Momentum continues to grow. We now have four that have completed their site initiation visits (SIV) — in Belgium, Spain, Germany, and Denmark — and will begin recruiting soon. Each new site and every participating family brings us closer to a deeper understanding of STXBP1 and to future treatments.

Looking ahead, 2026 is shaping up to be an exciting year for ESCO and for STXBP1 research. None of this progress would be possible without the dedication, trust, and involvement of families across our community — thank you for moving this work forward together with us.

💜 **If you wish to donate to STXBP1 research, you can do so through this link.** Every contribution helps us continue advancing research and supporting

our community.

Donate to STXBP1 Research

Announcements & Events

Preparation to Progress: ESCO's First Patient Enrollment

ESCO enrolled our *very first* NHS participant on the 4th of November 2025 at the University Hospital in Antwerp. Four-year-old Rosie and her family kindly devoted an entire day to help move STXBP1 research forward.

The study visit began at the Centre for Developmental Disorders where Rosie first met with the neuropsychologist, who evaluated her social and functional development. Afterwards, she saw the speech and language therapist for an assessment of her communication skills, followed by the physiotherapist who guided her through the motor evaluations.

We introduced a small break after that as the family had to change location.

In the afternoon, Rosie was seen by the clinical (pediatric) neurology team at the University Hospital in Antwerp. Together with the family, they reviewed the medical, developmental, social and family history in detail. Several scales questionnaires were further completed covering sleep, behavior, motor function, communication and also more socio-economic aspects such as financial support and quality of life. Rosie also had a clinical examination, followed by a 30-minute EEG and ECG, all handled with patience and care. The family also kindly consented to a blood draw for biosampling. As part of the study, a follow-up consultation was planned after 3 months, which will take place by phone.

Despite the long and busy day, Rosie did wonderfully and her participation marks an encouraging start for our European Natural History Study. We are grateful to Rosie and her family, and we hope that many more patients and families will follow in her footsteps. Together, we can better understand STXBP1-related disorders and continue working towards improved care for our entire community.

2nd European STXBP1 Summit: A Moment to Build On

In October, Heidelberg brought together researchers, clinicians, families, and advocates from across Europe and beyond for the 2nd European STXBP1 Summit and Family Day. Over four days, the meeting fostered scientific exchange, open dialogue, and strong connections across the community, all focused on advancing STXBP1 research and care. We were truly encouraged by the energy and collaboration throughout the summit, and planning is already underway for the next European STXBP1 Summit in 2027.

Read more in our event blog and in the summit reflections by James Goss, Scientific Director of the STXBP1 Foundation, linked below.

Click here

Our European STXBP1 Community

In this section, we highlight members of our European STXBP1 community — families, clinicians, and advocates — whose voices and efforts inspire and strengthen us all.

This is Robyne
Robyne lives in the north of Belgium, in the Noorderkempen. She was born in 1999 and received her STXBP1 diagnosis at the age of eighteen. Just two weeks after her birth, she began experiencing epilepsy. After a month in the hospital, the doctors fortunately found the right medication, which made her seizure-free.
At first, the doctors spoke of infantile epilepsy and gave us hope for a fairly normal life. But as time went on, it became clear that Robyne needed more support than other children. We saw delays in both her mental and physical development. At the same time, we discovered that living differently, adjusting expectations, and creating a supportive environment are not insurmountable. Small moments of happiness often turned out to be greater than we had ever imagined.

Robyne is usually in good spirits and absolutely loves music: listening, singing along, and everything in between. TV programs that include music are also among her favorites. She can say about fifty words, but she cannot form sentences. She also loves playing with water—every splash brings a smile to her face.

Still, there are about two moments each day when Robyne cries or becomes upset. For us as parents and caregivers, these are difficult moments. We want to help, but so far we do not know what is wrong or how to break through it. The only thing we can do is stay by her side and wait for it to pass.

Walking is also a challenge for Robyne. In familiar surroundings—such as the living room, hallway, and bedroom—she manages to walk calmly from one place

to another. Outside, however, it becomes difficult due to uneven surfaces. She can walk with support, but after about a hundred meters it becomes too much for her and she needs to rest.

At the moment, Robyne is seizure-free, and we sincerely hope it stays that way. Learn more about the STXBP1 community in Belgium by clicking the link below.

Learn more

Welcome Merel Swinnen to the Belgium team!

Dr. Merel Swinnen is a resident in adult neurology at the University Hospital of Antwerp and a PhD researcher in the Translational Epilepsy Genomics Group at VIB, University of Antwerp, in Belgium. She works in close collaboration with Dr. Hannah Stamberger in both research and clinical practice at the Neurology Department of the University Hospital.

Her PhD focuses on STXBP1-related disorders, where she helps coordinate the Belgian site for the ESCO Natural History Study and Registry. Her research further focuses on movement disorders in STXBP1, epilepsy trajectories, and genetic modifiers.

STXBP1 Science Spotlight

In this section, you'll find roundups of the latest research on STXBP1. Sometimes the updates come directly from clinicians and researchers; other times we'll share a brief summary with links for further reading.

Read here (English)

For the latest updates on all things ESCO, follow us on social media:

Let’s move together—across borders, communities, and hearts—to raise awareness and fuel progress for STXBP1.

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