The European STXBP1 Consortium Newsletter Autumn 2025

Welcome
Being STXBP1 Awareness Month, September is always a special month for the STXBP1 community — and this year has been no exception! Across Europe, families, friends, and supporters have been coming together for **Move to Cure Europe**, completing relays (“estafettes”) in their own countries to raise awareness and funds for STXBP1 research. From walks and bike rides to creative community events, each effort has brought us closer together and closer to a future with better treatments and outcomes for our children.

💜 **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

STXBP1 Awareness Month

This September, our community truly showed what it means to **move together for a cure**! Families across Europe joined forces to raise awareness, raise

funds, and celebrate the strength of the STXBP1 community.

🇳🇱 **The Netherlands**
Families and friends completed an incredible estafette relay spanning **over 300 kilometers — from one end of the country to the other!** The different legs of the estafette culminated in a 5 kilometer walk through the Amsterdamse Bos with families from across the Netheralnds. Together, they raised **more than €14,000** for STXBP1 research. Team Myrthe alone raised €7,000 — what an inspiring example of community power!

Check out the video from the Dutch Estafette!

Families across the Netherlands have also started the Oudervereniging STXBP1 (Parent Association STXBP1). Learn more by visiting their website.

🇪🇸 **Spain**
From Morón de la Frontera to Navas del Madroño — passing through Magic Island, Alhaurín de la Torre, Orcera, Isla de Arousa, Oropesa del Mar, and so many more corners of the country — Spanish families carried a symbolic baton of hope across the nation.

Spain set themselves a goal of €5,000 for STXBP1 research! Donate here to help them reach the finish line. 💙

🇩🇪 **Germany**
In Germany, many families are joining the European relay— running, cycling, rolling, and sharing videos that became part of the Europe-wide relay story. Their goals were clear: raise awareness for STXBP1, connect families and communities, and send a powerful message of hope across Europe.

💜 **And more to come…**
Families across other ESCO countries have been joining in too, and we can't

wait to share even more highlights in the coming weeks.

Together, these efforts are more than just events — they are a statement. **We are united, determined, and moving toward a better future for everyone living with STXBP1-related disorders.** We look forward to sharing the full impact of these activities at the **European STXBP1 Summit in Heidelberg this October!**

European STXBP1 Summit and Research Roundtable

The **European STXBP1 Summit** is just around the corner! This special event will bring together **researchers, clinicians, families, and community members from across Europe and beyond** — all united by a common goal: advancing research and improving the future for those living with STXBP1-related disorders.

Key highlights:

- **Research Roundtable (8-10 October):** Leading scientists and clinicians will present the latest advances in STXBP1 research. Presentations will be **recorded** and made available to our community after the event.
- **Family Summit (11 October):** Families will have the chance to connect, share experiences, and hear directly from experts. This event will be **live streamed**, making it possible for everyone to join, no matter where you are.

We are so excited for the conversations, connections, and new ideas that will emerge during these days together — and we look forward to sharing the outcomes with you all.

Heidelberg STXBP1 Summit

Supporting Families Through Knowledge

This fall, the STXBP1 Italia is launching a parent workshop series on the rights of caregivers and those living with disabilities. Developed together with ANFFAS Nazionale, a leading disability rights organization in Italy, the course will cover topics such as family support plans, school inclusion, the transition to adulthood, and caregivers' protections in the workplace.

While the sessions are held in Italian, the model itself can be adapted by associations in other countries. By starting with a simple survey of families' needs and working with legal experts, the Italian team created a program that is flexible, practical, and replicable.

We share this initiative as inspiration and hope it sparks similar conversations in other communities across Europe.

Parent Panel Meeting on Potential Future STXBP1 Therapies

On April 8, The Dutch BRAINmodel project organized a parent panel meeting on potential future gene therapies for STXBP1-related disorders (STXBP1-RD). The aim of this meeting was to explore the perspectives of European parents of children with STXBP1-RD on potential future gene therapies. Parents from seven European countries joined this online dialogue.

The meeting was organized by researchers from the Erasmus Medical Center in Rotterdam, within the context of the BRAINmodel project. At present, treatment options for children with STXBP1-RD are limited. However, several research groups worldwide are investigating potential future therapies, such as RNA and DNA therapies, that target the underlying causes of the disorder. Some of these therapies may be tested in clinical trials in the upcoming years, but most are still in very early investigational stages. At this stage, insight into the ethical perspectives of families with a child with STXBP1-RD is important to help guide research in directions that align with the needs, hopes, and concerns of these families. Read more… (English)

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.

Meet Herman, 6 years old, from Norway

Herman is an active and happy boy who lights up every room with his smile. He loves jumping on the trampoline, chasing after someone in a game, blowing soap

bubbles, splashing in water, and playing peekaboo. Silly sounds never fail to make him laugh.

Herman was diagnosed with STXBP1 just three weeks after his second birthday. Coming to terms with the diagnosis and its severity has been very difficult for his family — and coping remains an ongoing process. But finding joy in the little things, celebrating everything Herman *can* do, and connecting with other parents in the STXBP1 community has made all the difference.

From being told he might never sit, walk, or talk, Herman has gone on to prove himself a fighter. Today, he runs, jumps on the trampoline, and proudly says “mum” and “dad” — milestones that fill his family’s hearts with pride.

Message from Herman’s family to the community:

> *“It is extremely hard in the beginning emotionally, but moments of joy and laughter will come. You will be proud of your child’s milestones, knowing how hard they’ve worked for them. You’ll also realize how complicated many things we take for granted truly are — and you’ll be in awe of their determination.”*

Meet Kim Thalwitzer
University Hospital Heidelberg

Kim Thalwitzer first became fascinated by STXBP1 during her doctoral research under the supervision of Steffen Syrbe, where she studied genotype–phenotype correlations in the disorder. To deepen her knowledge, she joined Ingo Helbig’s group at CHOP for a research stay, continuing clinical research and, most importantly, meeting many families in the clinics.

Today, she is completing her pediatric residency at the University Hospital Heidelberg with the goal of specializing in pediatric neurology. Alongside her clinical training, she is part of the Heidelberg team coordinating the ESCO Natural History Study, and is excited to contribute to such a large, collaborative European project.

Thalwitzer notes that one of the biggest challenges in STXBP1 is its wide variability — while children may share some features, each child is also

remarkably different. This makes research especially complex, but also deeply meaningful.

"I hope families feel empowered, knowing that their participation truly makes a difference. Whether by sharing data, attending study visits, or raising awareness in the community, every contribution counts. I also hope families benefit from the regular exchange of information with clinicians and researchers. Joining long-term studies can be an extra challenge on top of everyday life, but every single data point helps us better understand STXBP1 and brings progress for the whole community."

Outside of the hospital, Thalwitzer has caught the cycling bug — ever since joining the Million Dollar Bike Ride in Philadelphia in 2022 with Lulu's crew, she now spends her free time exploring the Odenwald by road bike.

STXBP1 Science Spotlight

Science is always moving, and so are we! In this section, we'll share a short roundup of recent research in STXBP1. Sometimes you'll hear directly from clinicians and researchers, other times we'll provide a summary with links to learn more. Either way, our goal is to keep families connected to the latest progress.

Read the Autumn Science Spotlight (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.

The European STXBP1 Consortium

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