The European STXBP1 Consortium Newsletter, March 2025

Welcome to the first edition of the European STXBP1 Consortium (ESCO) newsletter! We're excited to connect with you and share updates on research, events, and community stories. ESCO is dedicated to advancing knowledge and improving care for individuals with STXBP1 syndrome, and this quarterly newsletter will keep you informed about our latest progress, upcoming events, and ways to get involved. Thank you for being part of our journey—we look forward to working together to make a difference!

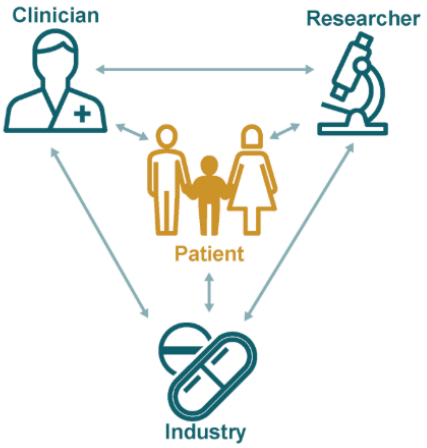
ESCO members in Antwerp, Belgium at the Kick-off meeting in February 2024.

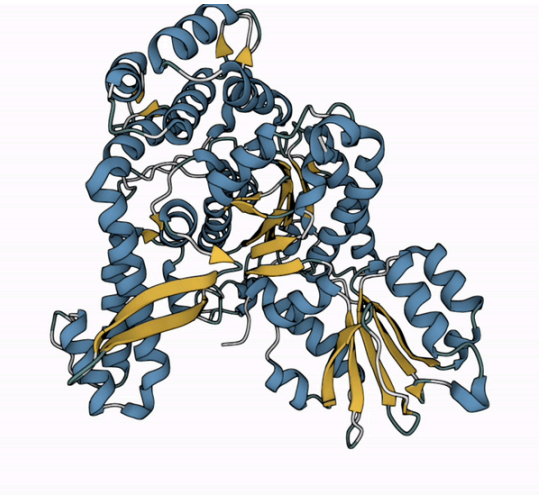
How it all began

The European STXBP1 Consortium (ESCO) was established in 2021 by Prof. Matthijs Verhage of Vrije Universiteit Amsterdam and University Medical Center Amsterdam (AUMC) and Dr. Ganna Balagura of Gaslini Hospital in Genoa. Together with Andrea Soto Padilla, business developer at IAO/IXA Neuro, and Hannah Stamberger, an adult neurologist at the University Hospital Antwerp, they form ESCO's executive board. In addition, ESCO is powered by brilliant **researchers and clinicians from across Europe and Israel**. Their vision was to create a collaborative network dedicated to advancing research and clinical trial readiness for STXBP1-related disorders (STXBP1-RD), rare neurodevelopmental conditions characterized by developmental delays, intellectual disabilities, and epilepsy.

A significant milestone for ESCO is the launch of a large-scale, pan-European natural history study focused on STXBP1-RD. Beginning with 50 patients across its eight member countries—Netherlands, Spain, Belgium, France, Germany, Denmark, Israel, and Italy—the study aims to recruit participants of various ages, providing a comprehensive representation of the STXBP1 population throughout different life stages.

Through these concerted efforts, ESCO continues to advance the understanding of STXBP1-RD, striving to improve the lives of those affected by these rare disorders.





Launching Soon: The ESCO Natural History Study & Registry

Background

In April 2025, the European STXBP1 Consortium (ESCO) will launch its Natural History Study (NHS) and patient registry across multiple sites. These initiatives are key steps toward understanding STXBP1-related disorders (STXBP1-RD) and preparing for future clinical trials. The NHS will begin with a one-year pilot phase involving roughly five patients per ESCO country, with plans to expand participation in subsequent years. Clinicians at ESCO sites will reach out to families about enrolling in the registry and NHS once the study begins.

ESCO Natural History Study (NHS)

The NHS is a structured research study designed to track the progression of STXBP1-RD over time, helping to identify clinical trial endpoints and outcome measures. After the initial clinic visit, participants will have follow-ups every three months.

ESCO Registry

The ESCO Registry is an online platform where caregivers and doctors enter medical and developmental information about individuals with STXBP1-RD. The registry collects both retrospective and prospective data, with follow-up surveys every 12 months. Once consent is given, both the caregiver and the clinician receive a link to input data.

Why Participate?

By joining the NHS or the registry, families contribute to vital research that improves understanding of STXBP1-RD and helps shape future clinical trials. The ultimate goal is to provide valuable insights into STXBP1-RD progression and establish external control data for future trials evaluating potential disease-modifying therapies.

More info on ESCO's NHS

More info on ESCO's Registry

Check out this insightful video from **STXBP1 Disorders (USA)** on Natural History Studies (English).

STXBP1 Family Across Europe Take Action for Rare Disease Month

Throughout Rare Disease Month 2025, STXBP1 family organizations across Europe came together to raise awareness and support those living with STXBP1-related disorders. From community-driven fundraisers to large-scale awareness campaigns, their efforts made a lasting impact.

🇩🇪 Germany: The German Association for STXBP1 joined the Rare Diseases Run 2025, the largest online run for rare diseases in Europe. Under the motto "Running makes you happy," the event united participants from Germany, Austria, and Switzerland to highlight the challenges faced by those with rare diseases.

🇪🇸 Spain: In Madrid, the Puerta de Las Rozas bridge was illuminated in green to mark Rare Disease Day. Additionally, before the March 2 professional football match between Leganés C.F. and Getafe C.F., two children with rare diseases—one with STXBP1 syndrome and another with Duchenne syndrome—performed the honorary kickoff. This moment of recognition was broadcast live on TV and radio, amplifying awareness across Spain.

🇮🇱 Israel: The Rare Smile Association organized a cherry tomato sale with a full greenhouse harvest donated by Seeds Technologies. Volunteers, alongside students from two local schools, sold two tons of cherry tomatoes and hundreds of kilograms of peppers, with all proceeds supporting the organization's mission. Additionally, young advocate Neomi continued her heartfelt tradition, leading a baking and cookie sale—a project she started at the age of seven—to raise funds for STXBP1 awareness and research.

🇵🇱Poland: On March 5th, Wiktor, the Polish representative for STXBP1, delivered a compelling lecture on synaptopathies, with a particular focus on STXBP1 and SYNGAP1. His audience included doctors and medical students eager to expand their knowledge. While Poland stands out in various medical fields, rare diseases remain largely unknown, even within the academic community. Many students initially believed these conditions were "ultra-rare" – something they only needed to learn about for exams, not real-world practice. This session played a crucial role in raising awareness, fostering social responsibility, and spreading vital information about STXBP1.

These incredible initiatives showcase the dedication and unity of STXBP1 families across Europe, working together to raise awareness, inspire change, and support research. ESCO is deeply grateful for these efforts and the growing strength of our community! 💜

The European STXBP1 Summit 2025 in Heidelberg, Germany

We are delighted to invite you to the **2nd European STXBP1 Summit and Research Roundtable** in **Heidelberg, Germany!**

The **Family Day** will take place on **Saturday, October 11, 2025**, and we warmly welcome all children & adults with STXBP1, as well as their parents, caregivers and siblings. This day will provide you with the opportunity to learn more about **STXBP1**, engage in discussions, and connect with other families, researchers, and clinicians.

The family registration is open on a first come, first served basis. Participation will be free of charge. To ensure that as many families as possible can participate, we kindly ask you to limit the number of attendees per family.

You can find more information about the summit and the link to the registration at **stxbp1summit2025.eu**.

We look forward to an inspiring event!

Register here!

The European STXBP1 Consortium

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