Monthly Newsletter

March 2026 **CTNNB1 FOUNDATION** **Issue #10**

Dear CTNNB1 community,

We took a short break at the beginning of the year due to an exceptionally busy period, but we're excited to be back with a new edition of our monthly newsletter. 🗞️

This issue brings both hope and honesty. 💙

We're pleased to share key advancements in our gene therapy program, including early post-treatment observations from the first patient and the initiation of screening for the next one. This issue also covers progress in our Natural History Study, upcoming conference developments, newly awarded grants, and continued efforts to expand research. At the same time, we highlight the critical importance of sustained funding to ensure successful clinical translation. 🚀

The CTNNB1 Team

Community News

👩‍🏫 CTNNB1 Foundation at SPARK NS event

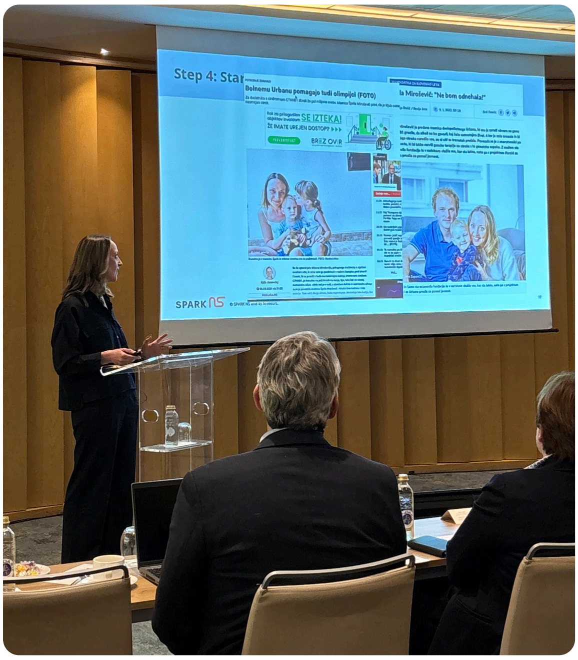
Last week, our President Špela Mirošević and our Research & Regulatory Associate, Lavra Debeljak, participated in the international short course "Positioning Neuroscience Discoveries for Successful Drug Development" (March 25-27, 2026, Madrid, Spain), organized by SPARK NS. 🇪🇸

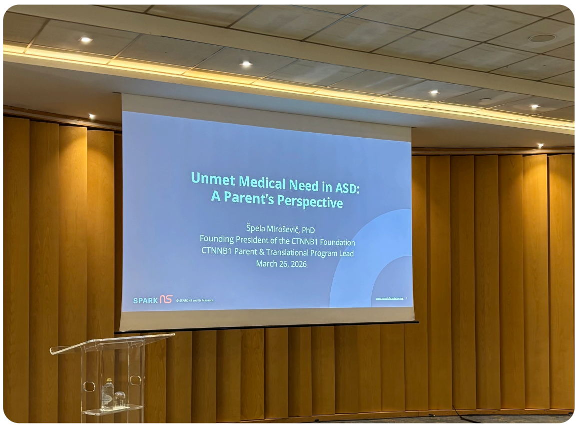
The program brings together selected academic innovators and translational leaders to accelerate the development of therapeutics in neuroscience, particularly in autism spectrum disorder (ASD) and Parkinson's disease (PD). It provides not only education but also access to a highly competitive funding opportunity of up to $2 million, along with mentorship and industry partnerships. 🤝

⚠️ The grant will not be funded directly to the CTNNB1 Foundation but toward predefined activities to support clinical trial and evaluate biomarkers.

The CTNNB1 Foundation was selected as one of the grantees, recognizing the strong translational potential of our AAV9-based gene therapy program for CTNNB1 syndrome. As part of the official agenda, Špela delivered a featured presentation titled "**Unmet Medical Need in ASD: A Parent's Perspective,**" where she highlighted the urgent need for disease-modifying therapies and presented the foundation’s work in advancing gene therapy from research toward clinical application.

Participation in this program further strengthens our positioning within the international translational research ecosystem and opens opportunities for strategic partnerships, funding, and accelerated clinical development.





Unmet Medical Need in ASD:
A Parent’s Perspective

Špela Miroševič, PhD
Founding President of the CTNNB1 Foundation
CTNNB1 Parent & Translational Program Lead
March 26, 2026

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🔍 Opportunity to participate in additional CTNNB1 research

As you know, most current research is focused on individuals with CTNNB1 syndrome themselves - which is essential, given how under-researched and underrepresented this condition remains.

However, another equally important aspect is often overlooked**: the impact of CTNNB1 syndrome on primary caregivers, especially parents.** 👨‍👩‍👧‍👦

For this reason, we invite you to participate in a master’s thesis study aiming to better understand the burden experienced by caregivers of children with CTNNB1 syndrome.

The study consists of a short (~20-minute) survey, which you can complete either **online**, or **in person during clinical examinations (at any site).** 📑

If you are also participating in the NHS, you will have the option to link your responses with your child's data, allowing for a more accurate and meaningful assessment of caregiver burden.

Participation is entirely voluntary. You will be invited to take part once ethical approval for this study has been obtained.

Research & Clinical Trial Updates

Clinical trial

We are very happy to share that the first patient is doing well. Špela has already shared updates in the CTNNB1 Facebook group, but for those who are not part of the group, we would also like to share this progress here:

We are now 3 months post-dosing. Urban is consistently walking with support—either holding one hand or using furniture—and is regularly pulling himself up to stand. With assistance, he can now walk for 30–40 minutes.

We visited a nearby shop without a stroller for the first time, which was a very special milestone. He even helped scan items and pay. During a recent trip to the zoo, he walked for most of the visit—even on what we would consider a "bad walking day." Recently, he even made **a few independent steps**, all on his own.

We are also seeing early improvements in speech. Urban is making more sounds, attempting new words (like "Ifff" for Iva and "Zaa" for Zara), and even tried to say "television." Most importantly, he has started initiating speech on his own—something he had not done before.

With gene therapies, progress is typically gradual. Improvements often begin after 2–3 months, with continued development over time.

Urban will start tapering off of corticosteroids soon and we are looking forward to seeing how he feels once the medication is fully tapered.

Important note: Before we can confidently attribute any observed progress to the gene therapy, the 6-month assessment must be completed.

A more comprehensive and reliable update on the first patient's progress will therefore be presented at the conference in June (you can still register for the conference here: https://ctnnb1-foundation.org/conference-2026/)

Urban enjoying his day exploring the zoo 🐑 🐅 🐘 👇

What’s next?

We are also pleased to share that the next patient is already in Slovenia and is currently being prepared to begin the screening process for treatment. 🇸🇮 🩺

📑 Natural History Study (NHS) update

Registration for clinical examinations is now closed for **new participant**s across all sites.

For those who were unable to register as new participants this year, please contact nhs@ctnnb1-foundation.org to be added to the waitlist (if you have not already done so). We will reach out to everyone on the waitlist ahead of next year's registration.

For anyone interested in participating in the gene replacement therapy clinical trial, **participating in the NHS is essential.**

📅 NHS Exam schedules

Participants attending examinations in Ljubljana have already received their preliminary schedules.

For those attending in **Boston** or **Barcelona**, thank you for your patience—our team is currently finalizing schedules and will share them as soon as possible.

🎤 Conference update

We will soon begin announcing speakers and presentations across our social media platforms. Stay tuned for more details about the program and what each session will cover—we're excited to share what we've been preparing. 📲

Fundraising Updates

We are encouraged to share several important milestones for the CTNNB1 Foundation:

- We have been awarded a highly competitive grant from SPARK NS, enabling us to expand our clinical trial efforts into the United States.
- Another grant awarded to us from ERDERA will help fund this year's CTNNB1 conference in Barcelona.
- A Slovenian national grant will allow us to develop the **first-ever clinical practice guidelines for CTNNB1 syndrome,** something we have worked toward for years—to clearly define what truly helps our children.

These are major steps forward. 🏆

At the same time, we want to remain fully transparent with our community.

In recent months, donations have slowed significantly. As the sponsor of the clinical trial, the foundation carries a substantial financial responsibility. Advancing a gene therapy from research to patients requires not only innovation—but sustained, reliable funding. 🤝

This means we must move forward carefully to ensure we can support the entire trial, from the first patient to the last.

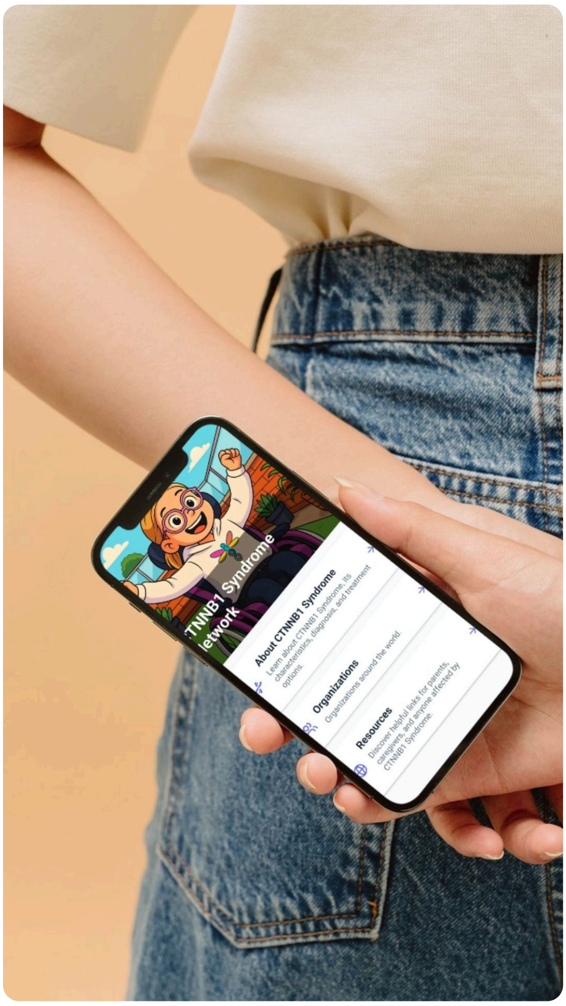
This is where our community becomes essential.

If you would like to help, one of the most powerful ways is by sharing our mission and organizing fundraising efforts within your own networks. Every conversation, every post, every initiative makes a difference. 💙

You can support the project directly here: https://givebutter.com/ctnnb1genetherapy

Every contribution—no matter the size—brings us one step closer to delivering a treatment for all children with CTNNB1 syndrome.

Thank you for standing with us and continuing this journey together.



Don't forget to download the CTNNB1 app!

This app is your central hub for **connection, support, and information-sharing** within the CTNNB1 community.

Available in **English, Deutsch, Español, Français, Italiano, Português, Slovenščina,** and **Polski**.

Through the app, you can:

- Stay up to date with the latest news and research
- Discover community events and meetups
- Join support group discussions
- Access educational resources
- Get informational support through a CTNNB1-focused AI assistant

Download for Android

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