

# Monthly Newsletter

May 2026

CTNNB1 FOUNDATION

Issue #11

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## Dear CTNNB1 community,

This month's newsletter brings several important milestones and updates from across the CTNNB1 community. We share exciting progress from the GAIN clinical trial, including the successful treatment of the second patient in the world with our investigational gene therapy URBAGEN intended to treat CTNNB1 syndrome. 🧬

We also reflect on recent community activities, from the ESGCT Summer School in Coimbra to the first round of Dragonfly NHS clinical examinations in Ljubljana.

Inside, you will also find important clarifications regarding the difference between the Dragonfly Natural History Study and the GAIN Clinical Trial, updates about conference childcare services, and ways to support our mission through fundraising and partnerships. 🙌

Finally, we are honored to feature a deeply moving story written by Robbin Verwest, whose words capture the realities, emotions, strength, and love that shape life within the CTNNB1 community. 💙

*The CTNNB1 Team*

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## Community News

### ☀️ ESGCT Spring School in Coimbra

Špela recently visited the beautiful city of Coimbra to attend the [ESGCT Spring School](#), where she gave a presentation on the journey of developing gene therapy for CTNNB1 syndrome. The event brought together young researchers, clinicians, and emerging leaders in gene and cell therapy from across Europe, creating valuable opportunities for collaboration, discussion, and knowledge sharing.



## NHS Clinical Examinations in Ljubljana

We are happy to share that we have successfully completed the first round of NHS clinical examinations in Ljubljana. A heartfelt thank you to all the families who traveled to participate. It was truly wonderful to reconnect with familiar faces and welcome many new ones to our growing community.

We would also like to express our deepest gratitude to our incredible study team — **Nina, Maša, Damjan, Alenka, and Lavra** — as well as our dedicated translators. Their commitment, professionalism, and teamwork made these examinations possible, and we are immensely grateful for all their hard work and support. 🙌

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## Research & Clinical Trial Updates

### 2nd patient treated with URBAGEN gene therapy

In the first week of May, another important milestone was reached. The **second patient in the world** has now been treated with URBAGEN, gene replacement therapy intended for the treatment of CTNNB1 syndrome.

The second patient, Mafalda from Portugal, underwent the procedure successfully and handled it very well. 🙌 She is currently recovering in the hospital and will remain in Ljubljana for the coming months for close monitoring and follow-up observations as part of the clinical trial.



This marks an extraordinary achievement for both the research field and the CTNNB1 community. Each step forward brings us closer to the possibility of future approvals from international regulatory agencies, including the U.S. Food and Drug Administration (FDA) and regulatory authorities in Europe. 📋

We are deeply grateful to Mafalda and her family for their trust and courage in joining this journey. We look forward to supporting her recovery and hope this treatment will contribute to meaningful improvements in her quality of life and future. 🤍

Follow Mafalda's journey on social media:

[Facebook](#)

[Instagram](#)

With the clinical trial now approved in Slovenia, our team is working intensively to prepare the documentation required for regulatory submissions in Spain and the United States. We will continue to keep you updated, but our current goal is to submit all required applications by the end of summer 2026 and open additional clinical sites in late 2026.

At the same time, we are actively exploring funding opportunities to support the manufacturing of an additional batch and the expansion of clinical sites across Europe. If the treatment proves successful, our commitment is to make it accessible to all eligible patients as quickly as possible.

## **Important Clarification About CTNNB1 Studies**

Please read this carefully.

We have noticed that within the CTNNB1 community there is some confusion about our ongoing projects. To ensure full understanding, we would like to clearly explain the difference between the two studies currently led by the CTNNB1 Foundation:

- **Dragonfly Natural History Study (NHS)**
- **GAIN Clinical Trial**

These are two **completely separate** studies, and each **requires a separate application**.

### **1. Dragonfly Natural History Study (NHS)**


The Dragonfly NHS is a **multi-year (2024-2029) observational study** designed to track how CTNNB1 syndrome progresses over time.

It includes standardized assessments such as:

- Medical examinations
- Psychological evaluations
- Physiotherapy assessments

#### **Key points:**

- This study is for **research purposes only**.
- Participation **does NOT include treatment**.
- Children enrolled in the NHS are **not automatically included** in the clinical trial.
- Even if your child is enrolled, you must **register each year for clinical examinations** via our [website](#) for logistical planning.

In simple terms:  The Dragonfly NHS helps us **understand the disease**, not treat it.

### **2. GAIN Clinical Trial**

The GAIN study is an **interventional clinical trial**, which means that a gene therapy product is administered to children as part of a clinical research study.

In this study:

- Selected participants receive a gene therapy that is currently being **evaluated for its safety and efficacy**.
- The treatment involves a **neurosurgical procedure**.
- Patients are asked to come live for at least 4-months close to the clinical site and then return for 5-years to follow-ups.

#### **! Important considerations:**

- This is the first time this therapy is being tested in humans with CTNNB1 syndrome
- As with any first-in-human study, there are **potential risks**, including serious ones
- Although the likelihood of severe side effects is considered **low**, it is essential that families **fully understand** the experimental nature and risks before applying

 **Current status:**

Applications for the clinical trial are **currently closed** in Slovenia. The CTNNB1 Foundation is working on preparing regulatory documents to apply to other regulatory agencies and open new sites soon.

### **How Are These Two Studies Connected?**

Participation in the Dragonfly NHS can be beneficial **if you later apply** to the GAIN clinical trial.

**Why?** Children in the NHS:

- Have **well-documented medical data**
- Are **regularly assessed** over time
- Provide the clinical team with a **clear understanding of disease progression**

This means they may have an advantage during the selection process but participation in the NHS **does NOT guarantee** selection into the clinical trial.



### **Study Communication Channels**

Each study has its own dedicated communication channel. Please use the correct email for your inquiries:

**Dragonfly NHS:** [nhs@ctnnb1-foundation.org](mailto:nhs@ctnnb1-foundation.org)

**GAIN Clinical Trial:** [gain@ctnnb1-foundation.org](mailto:gain@ctnnb1-foundation.org)

### **Need More Information?**

If anything remains unclear, please visit our [website](#) where both studies are explained in detail.



### **Conference Update: Childcare Services**

Childcare services will be available during the conference on **June 18–19** to help support families attending the event.

**Please note:** Childcare is organized directly by the CTNNB1 Foundation and **not by the hotel.**

If you have any questions regarding childcare arrangements, please contact us directly at [nhs@ctnnb1-foundation.org](mailto:nhs@ctnnb1-foundation.org)

Families interested in using the childcare service are kindly asked to complete the following application form:

[Apply for childcare](#)

## Donate to the Cause

Donations to the CTNNB1 Foundation have significantly decreased in recent months. It is now more than anything critical that we step together and fundraise for this cause.


We would like this program to stay within non-profit organization and have control over the cost and accessibility.

Please consider donating or organizing a fundraiser to help cover hospital and operational expenses and manufacture another batch.

Your support directly helps us continue this important work and move closer toward making treatment available to families around the world.

[Donate through Donorbox](#)

[Donate through Givebutter](#)

 If you know of any organizations, companies, foundations, or individuals who may be interested in supporting a meaningful cause, we would be incredibly grateful if you shared our contact information with them or invited them to reach out to us directly at [contact@ctnnb1-foundation.org](mailto:contact@ctnnb1-foundation.org)

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## Stories of Impact: Kai

*This month, we are honored to share an incredible story written by Robbin Verwest, mother to Kai, a little boy with CTNNB1 syndrome from the Netherlands. Her writing beautifully captures the whirlwind of emotions, challenges, and experiences that come with caring for a child with a rare neurodevelopmental disorder.*

In the past I was asked a very blunt question about how Kai's level compares to other children with CTNNB1. I was asked to put my child on a scale. To weigh him against others. To sum up what he can do and what he cannot. This happens in the outside world because Kai differs from what is considered "normal," and therefore he needs to be explained.

Kai is part of the natural history study for the CTNNB1 Foundation. Every year Kai and I partake in the research and attend the CTNNB1 Conference. For me one of the highlights of the year. Suddenly I am among a group were I don't

have to explain Kai. It almost feels like I'm with his long lost brothers and sisters. In this group, they follow, test, and examine a large group of children with CTNNB1. It's about gaining more knowledge about the diagnosis and seeing how children develop. There's also a gene therapy being developed. We have hope that one day Kai will be treated and that it will improve his quality of life, bring him more joy, even more joy. Maybe he'll take a few steps or say a few words.

I gathered my thoughts, trying to put this impossible answer into words that the "normal world" could understand. I found myself explaining that there are children who can walk and talk but will most likely need support and guidance for the rest of their lives. There are also older children who are confined to wheelchairs. The spectrum is wide because the diagnosis is relatively new. And we are aware there is a group "out there" who never received an official diagnosis. "Oh, so Kai is one of the worst cases," she replied. For her, it was about understanding the diagnosis. Learning about the spectrum. Showing interest in our life. But for me? My son had just become a case. A number. An object to be placed somewhere on a shelf of qualified abilities.

In 2023, I attended the first CTNNB1 conference in Madrid. Heavily pregnant with my third son, nervous, and full of determination to learn more. I met my first Dutch CTNNB1 mom friend. We were the only two Dutch moms attending. We stood right next to each other when the conference started, perfectly on time, how Dutch. I held up my Dutch passport, and from behind me I heard, "Hey, jij komt uit Nederland!" A friendship rooted in understanding and compassion was born in that moment. She has the most adorable little girl, who was with her in Spain. When I met her, my heart ached. It was like meeting Kai's sister. A strange familiar feeling I always have when I see other CTNNB1 children. Looking into her eyes, into the eyes of so many CTNNB1 children, I see it. The love. The dedication. Their ability to bring people to tears of joy and love without speaking a single word. She has it all. Just like Kai.

Over the years, I watched her grow. I saw her achieve milestones, followed by intense setbacks. I saw her battle those setbacks with an unknown inner strength, only to conquer them again. And then came the news: She is talking! It is a whole chapter on its own to define the gravity of not being able to fully understand your child. You see them in pain, frustrated or confused. You run through your mental checklist. You replay the past days, the events, the routines, the small details. You assess their current needs and try to piece the puzzle together. And sometimes, even after doing everything you can, you are still left in the dark. Unsure if all your puzzling helped and their needs were truly met.

So do I hold a grudge? Am I jealous of those who walk and talk? Let me dig deeper into jealousy. It's Wednesday evening. I just bribed my seven-year-old neurotypical child with three pancakes with Nutella to go to his rugby practice. After that, I wrestled his rugby shoes onto his feet and dragged him into the cargo bike. With a nervous system already on edge, I pedalled to the rugby field. I chat with other parents. They tell me about their son's dedication to rugby. "He even watches competition matches on TV," they say proudly. And there I stand, internally exhausted, watching my child chase butterflies at the edge of the field instead of the ball. That is where I feel jealousy. But when it comes to my friend and all the devoted parents within the CTNNB1 community, it is a totally different ballgame.

Because our life is about showing up where nobody else is going and declining to the life that everybody else lives. Because when you have limited energy, you choose carefully where it goes and we choose to give it all to our children. For us, it's not about buying the fancy Miffy milestone cards that say, I was X months old when I crawled. When I took my first step. When I said my first word. For us, it's about 685 hours of therapy just to make sure your child can safely swallow a piece of bread or climb in and out of their floorbed without getting injured. It's about continuing when progress is measured in millimetres or doesn't show up at all. It's about not giving up when everything seems to stand still. It's about believing. Believing in your child when doctors say she can't. Knowing, deep in your bones, that she will. When the world says, "You shouldn't," you quietly go out and do it anyway.

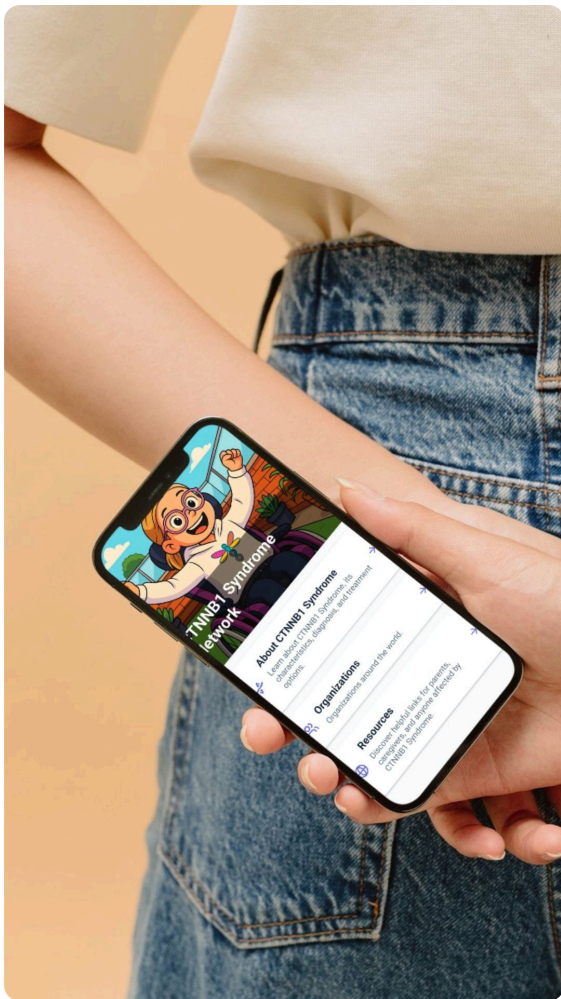
When I am low, I think of her and all the children I got to meet. Of their resilience. Of their relentless drive to push through limitations. I recall the arguments we have with the system. A system meant to help our children, yet constrained by rules, regulations, and ceilings that don't fit them. She stands there fighting for her child. Jealousy doesn't live there.

Yes, I get jealous. I get jealous of ease. Of simplicity. Of children who chase balls instead of butterflies. But I am not jealous of this path. Because this path builds warriors. It builds advocates. It builds a love that is disciplined and fierce and unshakeable.

When I heard she is talking, my eyes filled with tears, not because I wished it was us, but because I know the price that was paid for every word. Jealousy cannot survive in a place built on that kind of devotion.

- by *Robbin Verwest*

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## 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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