Hope on the Horizon: A Simple Guide to the ELJ-101 Gene Therapy Webinar

Hello everyone! It's Karen, Carol-Anne, Cristina, and Rodney here. We're sitting down with a big cup of tea (and, honestly, a few deep breaths) to share a warm little update about something that's been buzzing around our community lately: the ELJ-101 gene therapy webinar.

Now, we know "gene therapy" sounds like something straight out of a sci-fi movie, and the technical talk can get a bit heavy. But at its core, this is a story about hope, about our children, and about the incredible people working night and day to make their lives a little bit easier. We had Russ Addis PhD, Chief of Pipeline Strategy and Head of Genetic Medicine of the LouLou Foundation, join us to talk about a programme called ELJ-101.

Grab a cookie, get comfy, and let's break down what this all means for our CDKL5 families in a way that actually makes sense.

What is ELJ-101 anyway?

Think of our children's bodies like a giant, beautiful LEGO® set. To build it properly, you need the instruction manual. In CDKL5 deficiency disorder (CDKL5 for short), a little page of that manual is missing or smudged. ELJ-101 is essentially a way to deliver a fresh, clear copy of that missing page directly to the cells that need it.

Russ explained it so well. They use something called an AAV vector. Think of this as a tiny, harmless "delivery truck." The scientists take the shell of a virus (but they've taken out all the bits that make you poorly!), and they pop a working CDKL5 gene inside. This little truck travels into the brain cells and drops off the gene so the body can start making the CDKL5 protein it's been missing.

It's important to remember that this isn't ***"editing"*** our children's original genes, it's just adding a helpful new set of instructions.

A quick, kind pause (from us)

Before we dive deeper into the science, we want to pause for a second. Webinars like this can be an emotional rollercoaster. One minute you're thinking, "Oh wow... could this really happen?" and the next you're spiralling into timelines, eligibility, and a hundred "what ifs."

Speaking as the leadership team (Carol-Anne, Karen, Cristina, and Rodney), and as humans alongside you, please go gently with yourselves. It's completely normal to feel hopeful *and* wobbly at the same time, because you care so much.

We'll say it plainly: it's totally normal to need a minute. Sometimes the kindest thing you can do is put a hand on your chest, take one slow breath, and come back to the info when your brain is ready to take it in.

The bits we found most reassuring (and why)

One of the things that really landed for us in the webinar was the "preclinical" update (that just means the testing that happens before anything is tried in people).

Russ shared some genuinely encouraging signs from the work so far in animals (including mice and non-human primates). In plain English: the "delivery trucks" seem to be getting the gene where it needs to go, and the safety checks so far looked reassuring.

We want to be careful with language here because preclinical is not the same as "proven in humans." However, it *is* a really important step, because you only move forward if the safety picture is looking solid.

The ELJ-101 Q&A (the questions everyone asked)

This was the part of the webinar we all leaned forward for. Below is our simple, "human" summary of the detailed Q&A about ELJ-101.

How big is the first trial?

- The plan shared was **around 12 participants** in the **first** study.
- That small number is *not* because the need is small - it's because early trials are designed to be careful and safety-focused.

When might it start?

- The timing mentioned was **early 2027** for the first-in-human trial.
- As always with clinical trials, that date can move depending on regulatory reviews and the final readiness of sites.

Where will it happen (which hospitals / which countries)?

- **Trial sites weren't confirmed** in the webinar.
- Site selection usually depends on specialist experience, the ability to deliver the treatment safely, and the practical capacity to do long-term follow-up. (So, it's as much about safety and systems as it is geography.)

Who might be eligible?

- **Eligibility wasn't final** in the webinar.
- In early studies, criteria are often strict and can include things like:
    - age range

    - confirmed CDKL5 diagnosis and specific genetic details
    - overall health and medical stability
    - previous treatments/medications
    - whether someone has factors that could increase risk with a gene therapy approach
- We know that uncertainty is hard. We also heard a clear intention to build the evidence step-by-step so access can widen over time.

Will there be an age range?

- An age range is **likely**, but it wasn't confirmed in detail.
- Early trials often start with a narrower group because it makes safety monitoring clearer and more controlled.

How is ELJ-101 given (what does administration look like)?

- ELJ-101 uses an **AAV vector** (the "delivery truck").
- The webinar didn't lock down every practical detail families will care about (like exact admission length), but gene therapy delivery typically requires a specialist clinical setting so the team can monitor closely during and after dosing.

What monitoring is involved?

- Monitoring is a **big** part of the plan.
- The outline shared included:
    - a **main study period of about 1 year**
    - followed by **longer-term follow-up (around 4 additional years)**
- That's because gene therapy is intended to be long-lasting, and everyone wants a really clear picture of safety over time.

What are the main risks talked about?

No one can pretend this is "risk free." In the Q&A, the overall framing was careful and realistic. The kinds of risks that tend to be discussed with AAV-based gene therapies include:

- immune responses (the body reacting to the vector)
- inflammation-related effects that need monitoring and, sometimes, treatment
- unknowns that only become clearer once dosing begins in humans

The tone from Russ was very much: move forward **only** if the safety signals support it.

What benefits are they hoping for?

The Q&A kept circling back to quality-of-life outcomes. In early trials the first aim is safety, but the team will also be watching for signals in areas like:

- **seizure activity**
- **sleep**
- **developmental skills** (things like movement and engagement)

It's important to remember: responses can vary a lot from child to child, and early trials aren't designed to promise outcomes.

If it works, will there be longer-term access?

This came up (because of course it did - families think ahead). The webinar didn't promise a specific access pathway yet. But the long follow-up plan and the stepwise approach suggest the intent is to build evidence properly, which is what you need before anything can move toward broader availability.

If your heart is already racing at the thought of "only 12," we hear you. That number feels tiny when your whole world is your child. But those first participants (and their families) help open doors - carefully, slowly, and with as much safety as possible - or everyone behind them.

A few practical webinar highlights we're holding onto

There were a few "pin this to the fridge" moments we took away from the ELJ-101 update:

- **This first study is small on purpose** - because early trials are mainly about safety.
- **The team is planning long follow-up** - because gene therapy isn't a quick pop-in-and-out; they want to watch carefully over time.
- **The goal is careful progress, not rushing** - which is frustrating when you're living the day-to-day realities of CDKL5, but it's also how they keep it as safe as possible.
- **The science is complicated, but the idea is simple** - a working set of instructions delivered by a "truck," so cells can make the CDKL5 protein they're missing.

Also, we just want to say: it meant a lot to see real humans behind the science on the screen. Not "faceless pharma," but people who know they're speaking to families who've carried a lot for a long time.

What this means (right now) for families

From our side, our job is to keep it simple, honest, and useful.

Right now, that means:

- We **don't** yet have all the practical details families want (like confirmed sites, final eligibility rules, and the exact "what will the week look like?" pieces).
- We **do** have a clearer sense of direction: ELJ-101 is moving forward, and the intention shared in the webinar is still to begin a first-in-human trial around **early 2027**.
- We'll keep sharing plain-English highlights as and when we get them, because you shouldn't need a science degree to follow what's being said.

And emotionally? If you're excited *and* nervous *and* slightly frozen… same. We were listening and thinking of your children the whole time.

Final Thoughts: One Step at a Time

This webinar was a huge milestone. ELJ-101 is moving forward, and while 2027 feels like a long way away, it gives us time to prepare, to advocate, and to keep supporting each other.

Remember, you aren't doing this alone. Whether you're a parent, a grandparent, or a friend, we are all part of this incredible journey toward better CDKL5 treatments.

If you'd like to support this work, please keep sharing accurate information, being kind to one another in the comments, and (if you're able) supporting the organizations pushing this science forward.

Stay hopeful, stay connected, and most importantly, stay kind to yourselves.

With hope,

Karen, Carol-Anne, Cristina, and Rodney – The GCC Webinar Series Organisers

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