DOSSIER INFORMATIVO

EL CAMINO DE CARLOTA

A gene therapy for Cockayne syndrome type B

Science already knows how. Now we must get there in time.

ASOCIACION COCKAYNE B ESPANA

www.elcaminodecarlota.org

Our Story: EL CAMINO DE CARLOTA

THE ORIGIN

When Carlota started walking, something caught our attention. She fell often and her movements were clumsier than usual. We thought she simply needed more time, but the doubts grew with every step.

Then began a long road of medical visits, tests and months of uncertainty, searching for an answer no one could give.

Finally, a study of her genome revealed the diagnosis: Cockayne syndrome type B, an ultra-rare neurodegenerative genetic disease.

Today Carlota is 4 years old.

She is a cheerful, loving girl full of hope. She goes to school, loves singing, playing with her friends and enjoying the little things every child should get to live. But we know that, without a treatment, the disease will keep advancing.

That is why El Camino de Carlota was born.

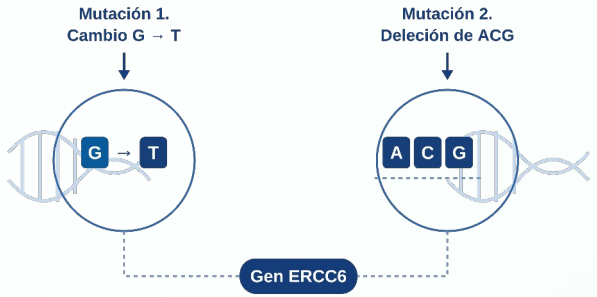
An initiative driven by her family and friends with a single goal: getting a treatment to arrive in time for Carlota and other children affected by Cockayne syndrome.

Carlota has started her path. Now we need to walk it together

CARLOTA	4 YEARS OLD	ASTURIAS

What is Cockayne syndrome?

Cockayne syndrome is an ultra-rare, neurodegenerative, multisystem disease caused by mutations in the ERCC6 (type B) or ERCC8 (type A) gene. It affects one in every 2 million births. Its symptoms cause progressive deterioration, giving patients an average life expectancy of 12 years. To date it has no curative treatment. But for the first time, research is opening a door to hope.



DNA repair dysfunction

ERCC6 produces a protein essential for repairing the DNA of cells when they are damaged. Without this protein, cells age prematurely.

What symptoms appear over time?

Neurological deficits

Progressive psychomotor decline and cognitive delay.

Ataxia

Loss of coordination, tremors and severe progressive gait impairment.

Hearing loss

Progressive bilateral hearing loss

Eye complications

Progressive retinal degeneration, optic atrophy and early cataracts.

Musculoskeletal problems

Persistent contractures, kyphosis and joint problems.

Photosensitivity

Extreme skin sensitivity to ultraviolet radiation.

Reduced life expectancy

The average life expectancy of affected children is 12 years.

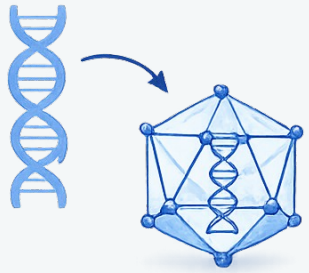
Developmental delay

Short stature and smaller head size.

The treatment: what is gene therapy?

An AAV virus carries the healthy copy of the gene to the patient's cells.

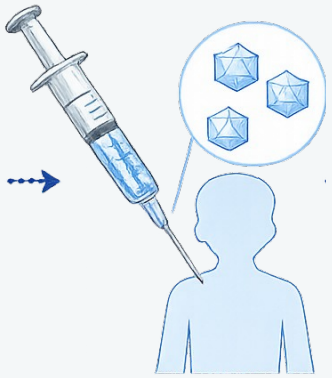
1



The healthy gene is prepared

A healthy copy of the gene is placed inside an empty, harmless AAV virus.

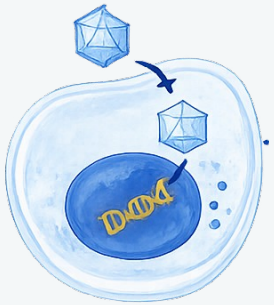
2



It is injected into the patient

A single injection introduces millions of viruses carrying the gene into the body.

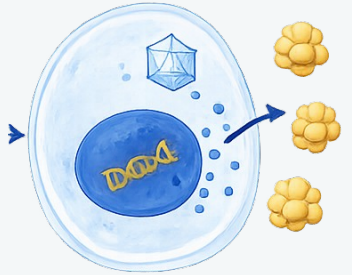
3



The virus delivers the gene

Each capsule enters a cell and deposits the healthy gene inside.

4



The cell produces normal CSB protein

With the healthy gene inside, the cell makes the protein it was missing.

The virus is only a carrier: it does not multiply and cannot spread from person to person.

Our researchers: a unique opportunity for Cockayne B

CURE FOR COCKAYNE SYNDROME TYPE B
Viljem Julijan ASSOCIATION

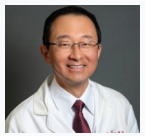
The Viljem Julijan Association in Slovenia launched a fundraising campaign in 2023 to advance gene therapy for Cockayne B and help a Slovenian girl named Karolina. This family initiative raised over €2M, which has helped fund the preclinical research of 2 working groups.

Advanced preclinical stage

University of Minnesota — Minneapolis, USA

Dra. Christina Pacak

Leader of the Pacak Laboratory research group at the University of Minnesota Medical School.



Dr. Peter B. Kang

Paediatric neurologist, professor and vice chair of research in the Dept. of Neurology, University of Minnesota Medical School.

1,8 M USD (FDA CBER)

1M € (VILJEM JULIAN)

Advanced preclinical stage

ABC-RI Portugal Algarve, Portugal

ALGARVE BIOMEDICAL CENTER

Dr. Clévio Nóbrega

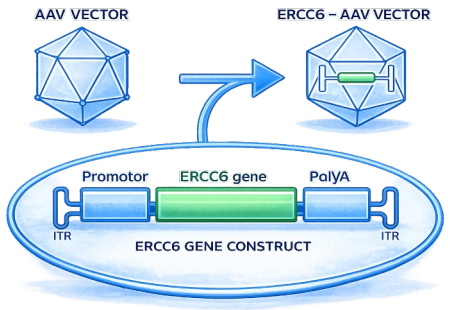
Leads the Molecular Neuroscience and Gene Therapy group at the Algarve Biomedical Research Centre, Portugal.

1M € (VILJEM JULIAN)

The treatment: current status of the therapy for Cockayne type B

Researchers have managed to introduce a functional copy of the ERCC6 gene into an AAV virus; preclinical trials have been successful in animal models, increasing life expectancy and improving symptoms. We are at an advanced preclinical stage for Cockayne type B.

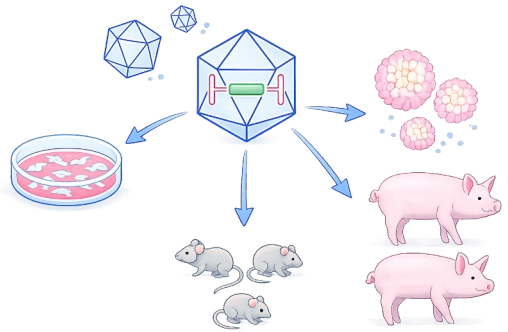
1



They have built a virus with a functional copy of the gene

The structure of an AAV virus has been optimised to carry the ERCC6 gene.

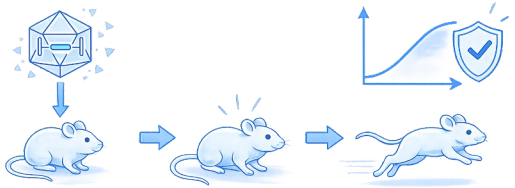
2



They have tested its efficacy in animal models and patient cells

The safety and efficacy of the ERCC6-AAV vector has been validated.

3



The results in animal models are promising

Life expectancy has multiplied, symptoms have improved and disease progression has slowed.

4

They are currently starting the regulatory studies

Talks with US and European regulatory agencies have begun with a view to starting a clinical trial.

Current status of the therapy for Cockayne type B

CIENCIA BÁSICA VALIDADA

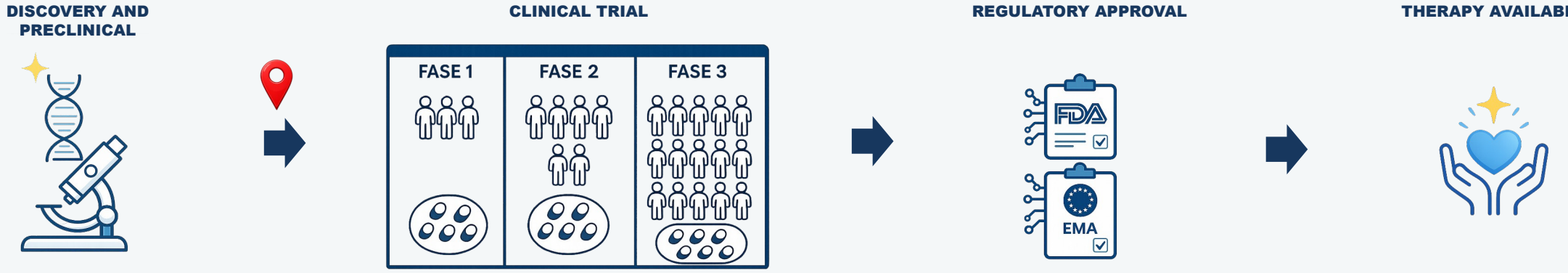
What we ALREADY have ✔

- **Gene vector design: virus ready and optimised.**
- **Proof of concept: successful functional rescue in vitro.**
- **Animal models: initial safety demonstrated in vivo.**
- **Initial funding: over €3.5M invested globally.**

CUELLOS DE BOTELLA CLÍNICOS

What is MISSING ⏳

- **GMP batch production: manufacturing the vector for human use.**
- **Regulatory phase (FDA/EMA): approval by the European/US agencies.**
- **Clinical trial design: protocol and hospitals ready in the EU.**
- **Critical translational funding: the capital to make the leap to the clinic.**



ACTION PLAN 2026-2028

El Camino de Carlota campaign goal

Our goal is to reach €2 million to fund the leap from the preclinical phase to the start of the clinical trial that will treat Carlota and other children with Cockayne B.

2 M€

FUNDING GOAL

EXECUTION PERIOD: 2 YEARS

Goal: raise the funds as soon as possible to deliver the project in 2 years.

5%	**Final preclinical studies**
65%	**Production of a GMP batch of gene therapy**
12%	**Regulatory process and FDA/EMA authorisations**
18%	**Start and execution of the clinical trial**

Every € raised lets us take one more step along this path.

Together we can turn hope into a treatment.

FOR CARLOTA

And all children with Cockayne type B

A CLEAR GOAL

€2 million to reach the first clinical trial.

A POSSIBLE PATH

Science, rigour and commitment to change lives.

REAL HOPE

We need to reach the next step together.

The greatest challenge: getting there in time

THE SCIENCE IS READY.

Now the challenge is getting there in time.

THE SCIENCE IS READY

Researchers have already developed a working gene therapy. Studies in animal models have shown very promising results, and the project is ready to take the next step.

ALL THAT REMAINS IS BRINGING IT TO CARLOTA

Before giving it to patients, the following must be completed:

- **GMP production**
- **Regulatory validation**
- **FDA and EMA authorisation**
- **Start of the clinical trial**

We are not talking about discovering a therapy. We are talking about getting it to those who need it.

TIME IS OUR GREATEST CHALLENGE

Today Carlota is four years old. She goes to school, plays, runs and sings. But Cockayne syndrome is a neurodegenerative disease. Every month that passes reduces the chance of treating her before irreversible damage appears. That is why every day counts.

THE TREATMENT IS WITHIN REACH. Now it depends on all of us.

Camina con nosotros

How can you help?

"A sum of small gestures can change the future"

Be part of “El Camino de Carlota”

Your support is essential to drive the scientific research forward

DONATE DIRECTLY

Fast and simple — 100% reaches the association

BANK TRANSFER: ASOC COCKAYNE B ESPAÑA

ES38 3059 0086 0540 8475 6628

BIZUM

Look for “Donate to an NGO” and enter the code: 14716

DONATE WITH TAX RELIEF

Through our partner foundation

If you wish to deduct your donation in your tax return, you can donate through Fundación Imp4ct, a registered public-interest entity that channels contributions to this project and issues the tax certificate.

Coming soon on our website

SPREAD THE WORD

Share Carlota's story and raise awareness

PARTNER AS A COMPANY

Sponsorships and charity initiatives. Let's join forces.

TAKE PART

Charity lottery, fundraising events and merchandising.

EL CAMINO DE CARLOTA

www.elcaminodecarlota.org

info@elcaminodecarlota.org

@el_camino_de_carlota