Leukoencephalopathy with Brain Calcifications & Cysts (LCC/Labrune Syndrome)

FDA Patient Listening Session

Friday, July 11, 2025

Motivation for a Patient-Listening Session

- We hope to provide the FDA with a deeper understanding of the critical challenges faced by the LCC/Labrune Syndrome community.
  - Serious impacts of disease on LCC patients
  - Effects on quality of life

- We hope to educate the FDA on the unmet medical needs of the LCC/Labrune Syndrome community.
  - No FDA-approved treatments
  - Tremendous unmet medical needs
  - Families' preferences for treatments and outcomes

The LCC Foundation
Our Mission

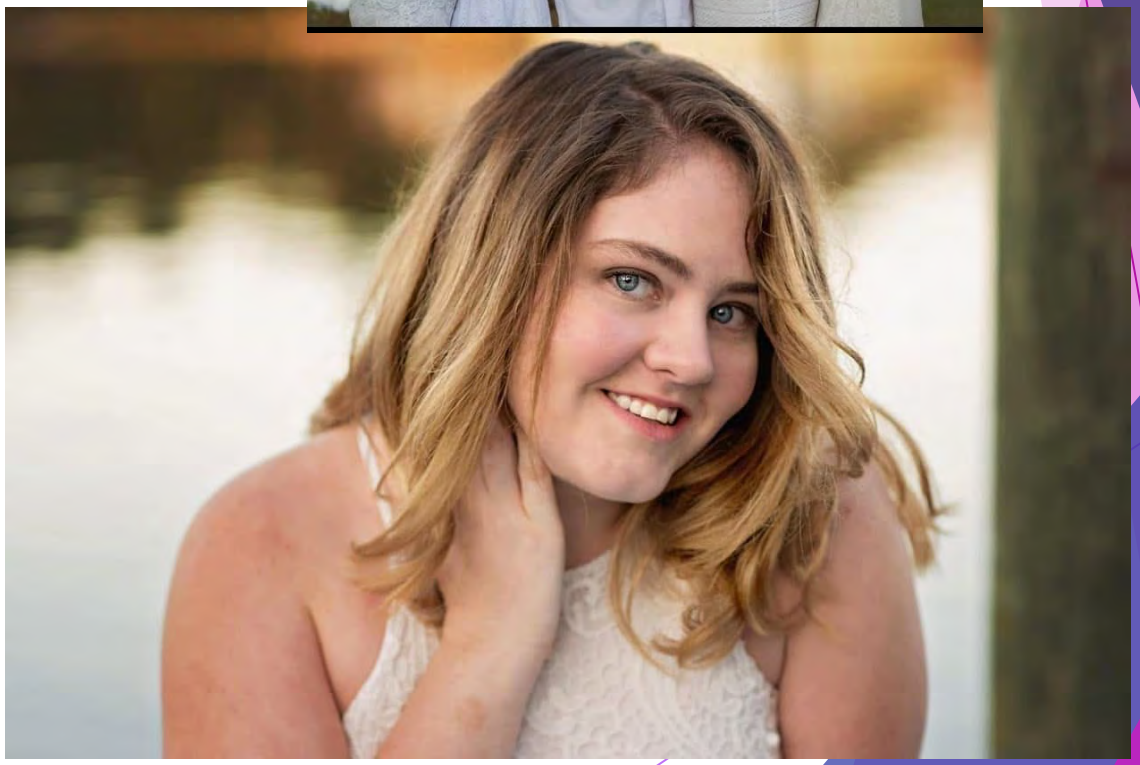
- US Based 501(c)(3) Founded in April 2022
- Proactively serve those affected by LCC by supporting programs of Education, Research, and Advocacy
- Identify sources of medical care, social services and genetic counseling; establish a communication forum among families
- Increase public awareness
- Creating information sources for health care providers
- Promote research into causes, treatment, and cure of LCC

Jamie Fraser, MD, PhD
Children's National Hospital

- Director of Leukodystrophy & Myelin Disorders Program
- Director of the Prenatal Genetics Program
- Biochemical and Medical Geneticist

Patient & Family Perspectives

Patient	Presenter	Relationship
Elisabeth	Elisabeth	Self
Zac & Ben	Danny	Dad
Ekrem	Adriana	Mom
Alice	Alice	Self
David	Lexi	Mom
Victoria	Martha	Mom



Elisabeth, USA
23 years old

Main Symptoms:

- Seizures
- Mood Disorder
- Tremors
- Dystonia

Elisabeth, USA
23 years old

Hopes for the Future:

- Any small improvement
- More normalcy/better quality of life
- More independence, stability, and dignity
- Even small improvement, I would be open to it

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Zac & Ben, USA
16 & 12 years old

- Significant Needs
- Constant Change
- Family Balance
- Mental Health
- Physical Health
- Hope

A progressive journey

Age 4

Age 6

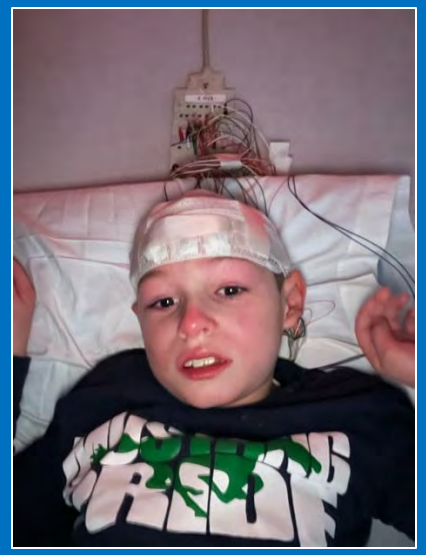
Age 7

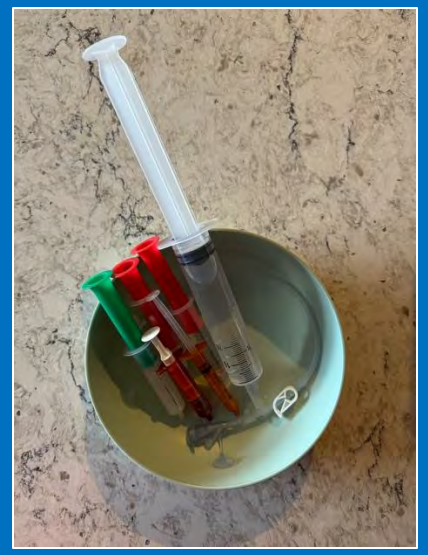
Age 8

Age 11

Just when you think you are set with equipment...

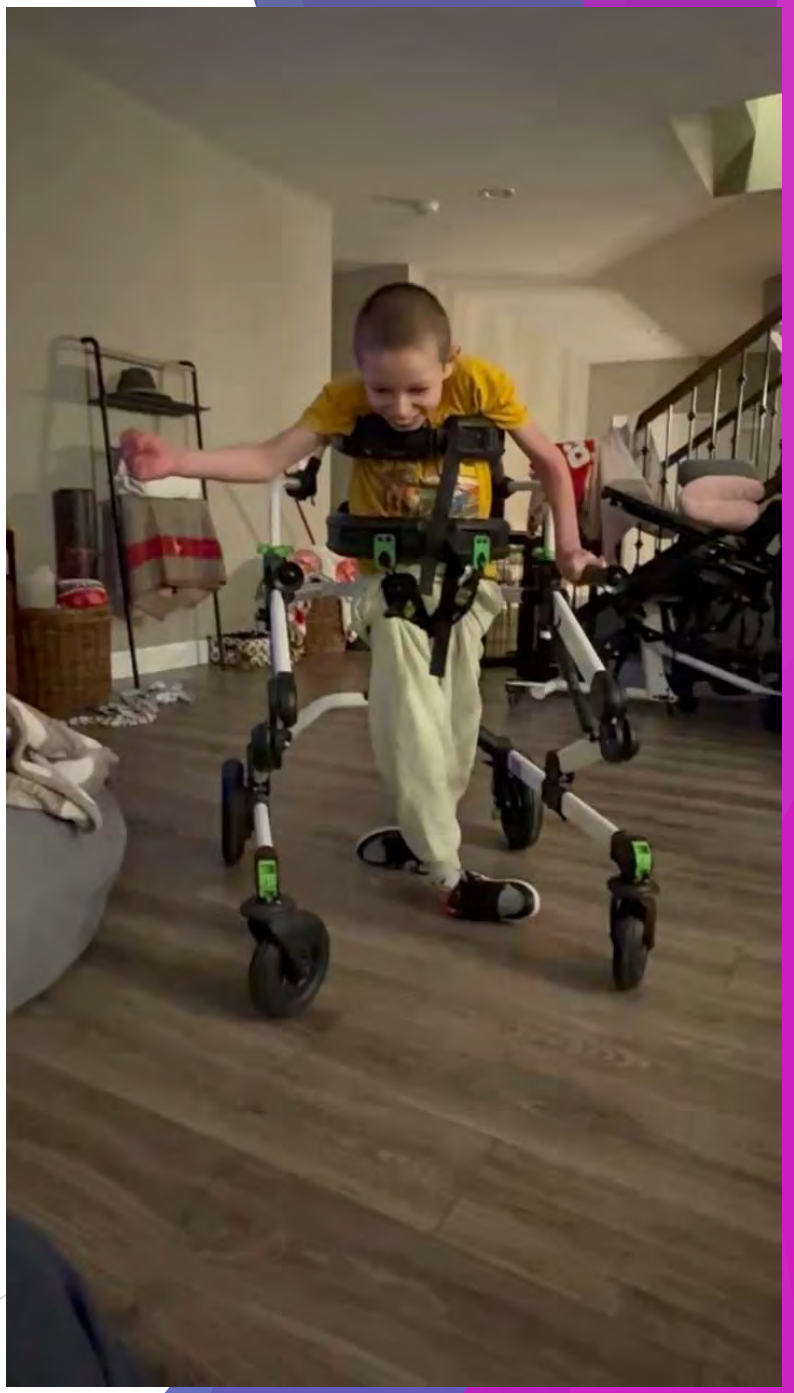
Stress doesn’t take a day off for caregivers



- Seizures
- Dystonia
- Medications
- Feeding
- Toileting
- Bathing
- Changing
- Bedtime
- Transitions

What do the physical challenges look like?



Zac & Ben, USA
16 & 12 years old

- Funding and solutions to advance research (ex. PRV)
- Continue to advance treatments that manage core patient challenges and halt or slow progression
- Partner to balance risk/reward for treatments of the future

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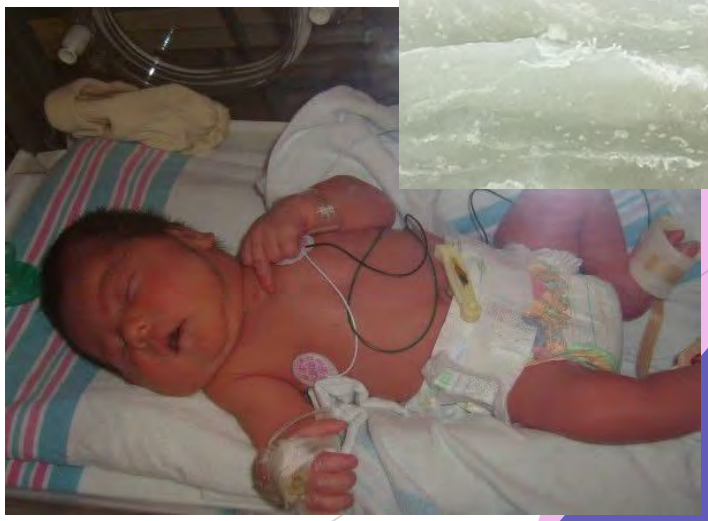
Ekrem's Journey with LCC

- Early challenges: Born prematurely; began having seizures at 4 months—misattributed to prematurity and treated with harsh side effects.
- Developmental struggles: Diagnosed with ADHD in early childhood—later revealed to be symptoms of undiagnosed brain damage.
- Life-changing diagnosis (2017): After a grand mal seizure, CT scans and genetic testing confirmed Labrune Syndrome.

Ekrem, USA
17 years old



Ekrem's Journey with LCC (continued)

- Progressive decline: From mainstream classes to life skills programs- gradual loss of speech, motor skills, mobility and social connection.
- "In-betweener" isolation: Too impaired for general education, too aware for severe-needs classrooms- left without a true place to belong.
- Rapid disease progression: One cyst has grown into ~20; major physical decline including speech loss, muscle weakness, and choking risk.

Ekrem, USA
17 years old

Ekrem's Journey with LCC (continued)

- Total dependence: Ekrem now needs round-the-clock care- cannot be left alone, tires easily, and experiences unpredictable seizures.
- A plea for hope: Ekrem still dreams of a normal life. We ask the FDA for a chance- access to clinical trials that could slow his decline.

Ekrem, USA
17 years old

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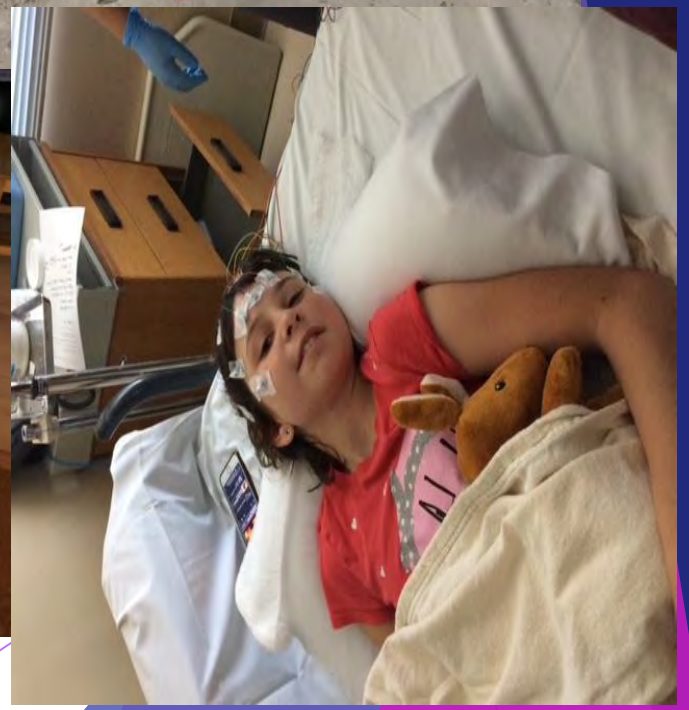
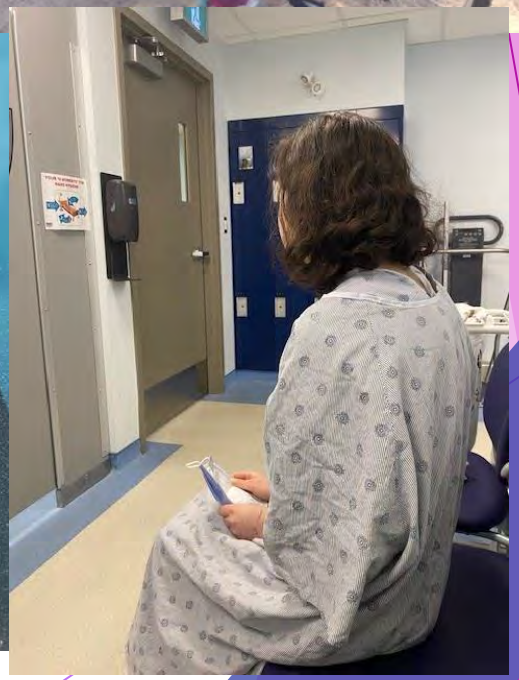


Alice, Canada
19 years old

- Seizures as baby, outgrown by 6 months
- Active early childhood years
- Seizures start to recur at 10 yrs
- Diagnosed with LCC at 11 yrs
- Effects of Calcification/Cysts
  - Seizures
  - Vision and visual processing
  - Physical activity & balance
  - Cognitive function



Alice, Canada
19 years old

- Progressive impacts as cysts grow
- Spill-over effects on mental health
- Adaptive learning strategies important
- Support from family care givers, teachers, and health professionals
- Hopes for future:
  - Effective treatment for effects of LCC
  - Recover physical/cognitive abilities
  - Increased independence

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David, USA
5 years old

Daily Challenges/Symptoms:

- Left-sided hemiplegia & spasticity
- AFO (ankle foot orthoses) for walking-wheelchair used when needed
- Developmental delays
- Affected vision - glasses and eye patch
- Scoliosis

David, USA
5 years old

- The Family Impact
  - Military Family
  - Stay-at-home mom
  - Limited time and energy
- The Emotional Landscape
  - Diagnosis
  - Advocacy
  - Education
  - Finding strength
- Hope & The Future
  - More normalcy for David
  - Priority of safety with potential treatments

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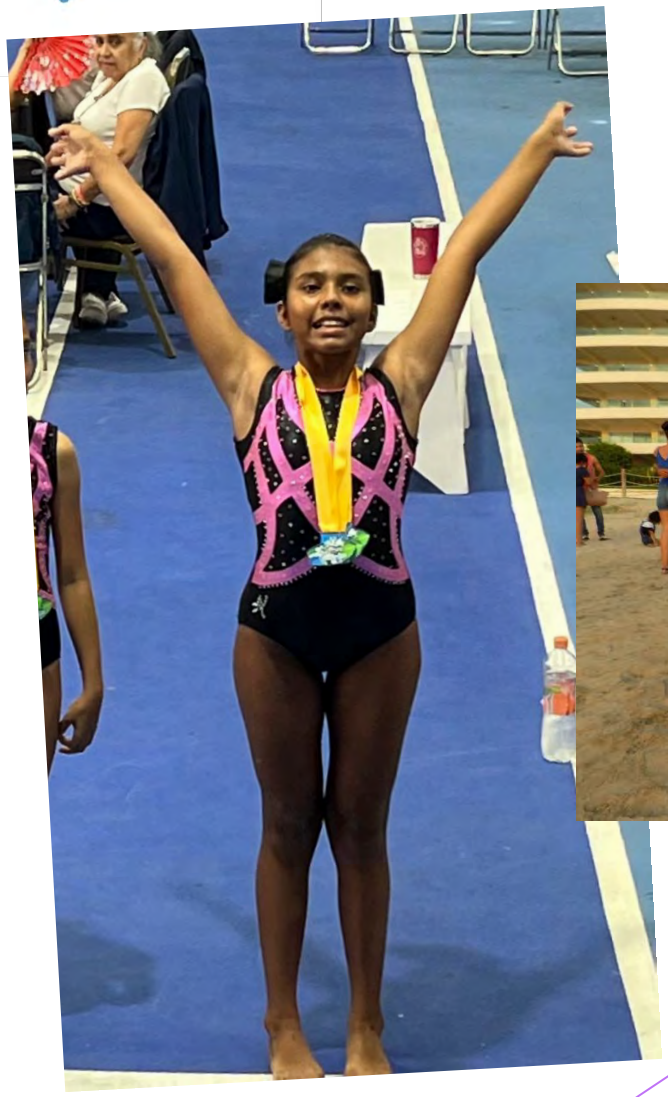

Victoria, Mexico
11 years old

Primary Symptoms:

- Seizures
- Psychomotor deficit - altered fine motor skills

Biggest Challenges:

- Intellectual Disabilities
- Psychomotor delays
- Hypothyroidism & precocious puberty
- Constipation
- Dry Skin
- Fatigue

Victoria, Mexico
11 years old

Care/Symptom Management:

- Victoria's treatment plan is symptom management and palliative care
- Neuropsychological therapy
- Academic support
- Physical therapy & gymnastics

Unmet Medical Needs:

- No disease-modifying treatment

SOME DAYS ARE FRUSTRATING FOR OUR FAMILY AND OUR SON DUE TO COMMUNICATION CHALLENGES, AND THE UNCERTAINTY OF THE FUTURE CAN FEEL HEAVY.

SEIZURES

HOPE

WE NEED MEDICINE TO HELP !

CYSTS

LCC VOICES

FATIGUE

TIME

ULTRA-RARE

"MY HOPE IS THAT THE CYSTS IN HIS BRAIN WILL STOP GROWING OR AT LEAST SLOW DOWN—SO HE CAN LIVE A LONG, HAPPY LIFE."

ENERGY

"MY CHILDREN ARE SQUARE PEGS WHICH THE HEALTHCARE SYSTEM INSIST ON TRYING TO FIT INTO ROUND HOLES"

HAVING AN LCC DIAGNOSIS HAS LEFT US FEELING ISOLATED.

CALCIFICATIONS

Question & Answer