LUCAS & ISLA

Believing for a cure for NKH

LUCAS

MEET LUCAS:

Lucas was born on March 6, 2018. He arrived after a healthy pregnancy and delivery. In the early hours of his life, he was very sleepy and did not cry. Lucas had been cleared to go home, but his nurse sensed something was not right and kept insisting on running more tests. Within hours we went from going home to a horror show that would last weeks.

All the cards were stacked against Lucas as we sat around a board room table filled with his family members and a team of NICU physicians. The head doctor, a geneticist, spoke calmly as she began to describe the findings of the lab reports, MRI, and additional tests completed on Lucas. She was soft-spoken and talked in circles with medical jargon way above the family's heads. As her voice turned into a low and far away murmur, she explains to the family that their new baby has a very rare genetic disease called NKH (Non-ketotic hyperglycinemia). She added she was confident he would not be going home. In fact, she said she was 100% sure of it.

After the devastating news, there was an odd silence of expectation. They expected us to breakdown and crumble to the floor. They even brought in tissues. Instead, Chancee, Lucas's mom, slammed her fist on the table, shocking them all, and said, "We are not going to just let him die!" And by the grace of God, he didn't.

ISLA

MEET ISLA:

After a wonderful pregnancy, Isla was born on September 30, 2018. When Isla was five days old, Sara, Isla's mom noticed her new daughter's hands twitch, and her eyes rolled up slightly! She told family and friends, but everyone told her it was typical newborn tremors. Sara still felt something was wrong, so she took her back to the hospital. Isla stayed there for four days, but the doctors couldn't find anything wrong with her. They planned an EEG in late November and sent the new family home! A few days later, Sara noticed the movements again, and this time they brought Isla to Alder Hey Children's Hospital. Isla ended up staying there for six weeks while the staff did multiple tests, including MRI, MRS, IVS, three lumbar punctures, sedation, and EEGs.

After the tests were completed, the hospital staff gave the family the devastating news: Isla has Nonketotic Hyperglycinemia (NKH). Isla's mom described the feeling when she heard the news, "It was as if I was shot, and I couldn't breathe."

GET INVOLVED TO HELP TODAY.

Website: lucasjohnfoundation.com

Email: info@lucasjohnfoundation.com

SEE OTHER SIDE TO LEARN ABOUT THIS CONDITION

WHAT IS NKH?

Non-ketotic hyperglycinemia (NKH) is a rare, genetic, metabolic disorder caused by a defect in the GLDC or AMT gene code, which affects the enzyme system that breaks down the amino acid glycine, resulting in an accumulation of glycine in the body's tissues and fluids. NKH typically presents itself in the first week of life with low muscle tone, lethargy, seizures, coma, and apnea requiring ventilator support.

RACE FOR THE CURE:

Lucas' family established the Lucas John Foundation, Inc. earlier this year to raise funds for research. Together we are learning, growing, and moving forward to make an impact and find the cure of this rare disease. Our goal is to secure funding and finance the gene therapy program, completely paving the way for all the babies suffering from this terrible genetic condition.

Non-ketotic hyperglycinemia (NKH)

ABOUT NKH

NKH is caused by genetic variants (mutations) in the genes that encode the components of the glycine cleavage enzyme system.

The incidence of NKH is predicted to be approximately 1:76,000.

NKH can occur in individuals of any ancestry.

There are around 500 children with NKH worldwide

MEDICATIONS

NKH requires multiple anti-epileptic medications to attempt to control seizures. Sodium Benzoate is taking every four hours around the clock to keep glycine levels lowered. The biggest battle is in the blood brain barrier.

NKH requires approximately 20 syringes of medications—all given throughout day and night.

Daily therapy and specialist are required

Twenty syringes of medications given daily

THERAPIES

NKH has minimal options and no known medical cure.

The multiple sedative medications make development a difficult journey. Almost **every single day,** the babies are either seeing a specialist or a therapist such as PT, OT, Vision, Speech, Feeding, Movement, and other alternative therapies.

GOAL

There is no current medical curative treatment for NKH at this time but we are going to change that with your help!

We are on a mission to raise three million dollars for gene therapy.

HELP US FUND THE CURE!

help us

FUND THE CURE

#WhateveritTakes

LUCAS JOHN FOUNDATION

TAKE ACTION AND GET INVOLVED TO HELP TODAY.

Website: lucasjohnfoundation.com | Email: info@lucasjohnfoundation.com