ABOUT CASK

The CASK gene is found on the X chromosome and provides instructions for making a protein called calcium/calmodulin-dependent serine protein kinase (CASK). This protein is found in nerve cells in the brain (neurons) and helps control the activity of other genes involved in brain development. It also helps regulate neurotransmitters that carry signals from neurons to other cells.

WHAT IS A CASK RELATED DISORDER?

A CASK related disorder occurs when there is a pathogenic (disease-causing) mutation on the CASK gene. There are two types of CASK related disorders both of which affect brain development:

- Microcephaly (a small head) with pontocerebellar hypoplasia (MICPCH)
- X-linked intellectual disability (XL-ID), with or without nystagmus (uncontrolled eye movments)

VISION

Project CASK believes in a world free of the life limitations and debilitating effects of CASK gene disorders.

MISSION

To accelerate breakthroughs in research to develop treatments and a cure for CASK gene disorders.

Values

Collaboration. Transparency. Urgency.

project cask

TAKE ACTION

CONNECT + SHOP + GIVE

An innovative rare disease non-profit, driving breakthroughs for treatments and a cure for CASK gene disorders.

@PROJECTCASK | HELLO@PROJECTCASK.ORG

WWW.PROJECTCASK.ORG

HELLO!

Today, there are no treatments available for CASK gene disorders. With limited ongoing research and even less funding available for ultra-rare pediatric disorders, the parents behind Project CASK feel a strong sense of urgency to expand and accelerate therapeutic development.

Our strategy is characterized by continuous collaboration across scientific disciplines for maximum efficiency and learning. We are exploring all available therapeutic avenues that are viable for CASK and engaging in partnerships that can speed up our route to effective treatments.

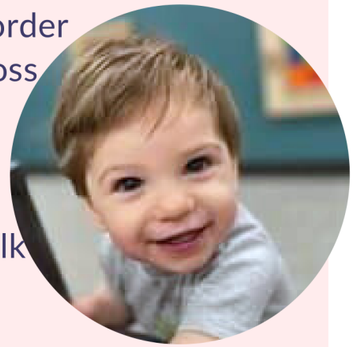
At the heart of our strategy are the children and families affected by this debilitating disorder, and our commitment to help them live their fullest, healthiest and happiest lives possible!

HOW ARE MALES AND FEMALS AFFECTED?

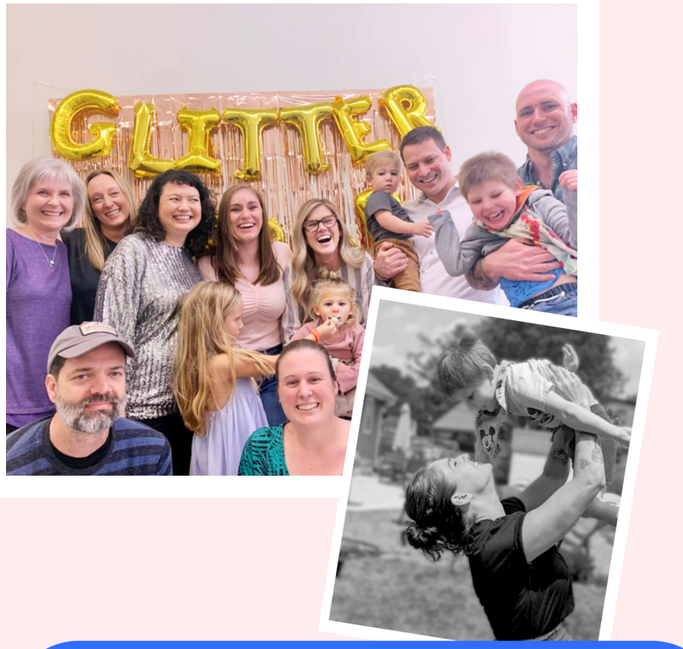
There is considerable variability in people with CASK gene disorders. Since it is x-linked, females have two copies of the gene and males only have one, which can lead to different symptoms and severity. Also, there are different types of mutations that can occur - for example, deletions where part of the gene is missing or substitutions where one or more “letters” of the DNA are switched. Depending on where these occur on the gene, there can be different outcomes. For females, x-inactivation skewing can also lead to diverse levels of severity regardless of the mutation.

COMMON SYMPTOMS

- microcephaly with or without pontocerebellar hypoplasia (MICPCH)
- seizures (diverse types, including infantile spasms, myoclonic and subcortical myoclonus, among others)
- sensory processing disorder
- sensorineural hearing loss
- hypotonia
- hypertonia
- nystagmus and CVI
- ataxia or inability to walk
- feeding issues
- sleep disturbance



For further information please see our website www.ProjectCASK.org/about-cask



CASK LIOCORNS

RARE AS UNICORNS. STRONG AS LIONS.™

Some say our kids had a better chance of winning the Powerball than being born with a CASK gene mutation. Some in the medical community call our kids "unicorns."

Unicorns are rare, magical, pure and innocent. That sounds a lot like our kids.

But there is so much more within our children, our families, and our community. We possess the tenacity, strength and courage of lions. A fierce determination to move mountains to help our kids reach their true potential. And from this, our spirit animal, the Liocorn was born.