

## WE KNOW WHAT CAUSES CASK

CASK is a single-gene disorder caused by a mutation on the CASK gene which is on the X chromosome. This makes it an X-linked disorder. Females have two X chromosomes whilst males only have one. As a result of the science of X-linked disorders, approximately half of the cells in a female's brain won't produce any CASK protein, whilst half will. In some males with the disease, none of their brain cells are able to produce the CASK protein, resulting in a fatal prognosis.

Patients with the form of disease called MICPCH have brain abnormalities that can be seen on an MRI. They may also exhibit abnormal EEGs.

Future possibilities for treatments include replacing the mutated gene, turning on the normally silenced copy of the gene in females or replacing the CASK protein.



ASK Research Foundation has joined forces with ACNRF Australia and USA, Association Enfants CASK France and the award winning research team at the University California, Davis, to activate a backup copy of CASK in females.

CASK aims to follow in the footsteps of other X-linked genetic disorders: CDKL5 deficiency disorder and Rett syndrome. X-reactivation has successfully been done for the CDKL5 gene (in vitro) and MECP2 gene (in mouse models). The team at UC Davis are eager to have the opportunity to try their technique on the CASK gene – a gene that displays all the markers of being a successful candidate for this novel therapeutic.

For more information, go to [www.caskresearch.org/help-to-cure-cask/](http://www.caskresearch.org/help-to-cure-cask/)



## FAST FACTS CASK GENE DISORDERS

CASK is a gene that, if mutated, causes disorders such as 'MICPCH' or 'XL-ID with or without nystagmus'. The patient community uses the term 'CASK' to encompass any disease resulting from a mutation in the CASK gene. CASK causes neurological problems with wide-ranging and complex symptoms.

# CASK OVERVIEW

CASK gene disorders (CASK) are rare neuro-genetic disorders that likely affect 1 in 2 million people worldwide.

Individuals with CASK typically can't walk, are non-verbal and have severe intellectual disability. Forty percent have seizures, may require feeding tubes and suffer vision and/or hearing loss. Disturbed sleep and behavioural problems can be serious challenges for families.

Individuals affected usually require continuous care and are unable to live independently.

Some affected males have mutations resulting in intellectual disability whilst those males with severe mutations often pass away in infancy. Females with CASK have an unknown prognosis but some are classed as having a life-limiting condition.

Typical characteristics of CASK are usually not evident at birth. Individuals with the disorder may

have feeding difficulties, poor head control and noticeable delayed development between four-twelve months of age.

Many children with CASK need intensive therapies to help develop or retain skills. In most cases, CASK isn't inherited. CASK affects every race and both genders, although females make up the majority of the living population. CASK may be misdiagnosed as cerebral palsy.

CASK has some distinct characteristics, namely great beauty, a gentle demeanor and a love of lights and certain sounds.



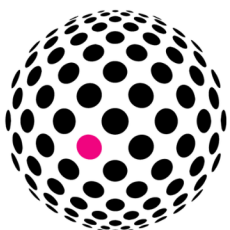
**Website**  
[www.caskresearch.org](http://www.caskresearch.org)

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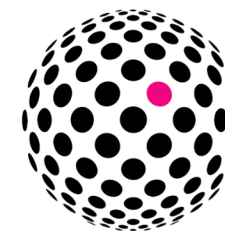
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**CASK**  
RESEARCH FOUNDATION

CASK Research Foundation is a registered charity in England and Wales, no. 1197434. Laura Hattersley, the mother of a young girl with CASK, founded the charity in 2022. We are registered with the Fundraising Regulator and are part of a collaborative group called the CASK Coalition, working together to further research into this rare disease.

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## Website

[www.caskresearch.org](http://www.caskresearch.org)

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