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 body will have the mutated gene, whilst half will have a healthy copy.

MICPCH is a disease caused by a mutation that results in the inability to produce CASK protein. Patients have brain abnormalities that can be seen on an MRI. They may also exhibit abnormal EEGs and have microcephaly. This is the most common disease found in females.

XL-ID with or without nystagmus is a disease caused by a mutation that creates partially functioning or less CASK protein. This is the most common form found in males. It results in intellectual disability.

ABOUT CASK RESEARCH UK

We fund and assist research to help accelerate the path to a treatment or cure. We also advocate for more awareness and increased diagnosis.

We support families via meet ups, peer-peer support, webinars and assistance with the science and diagnosis.

The CASK Coalition

Members work together to expedite research and funding goals.

Current members are: CASK Research UK, ACNRF Australia and USA & Association Enfants CASK France

The group have teamed up with the University of California, Davis, to investigate a novel gene therapy for CASK disorders.

Visit www.thecaskcoalition.com

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

ICASK mutations are the most frequent cause of a brain condition called PCH.

The prevalence of CASK disorders is unknown but fewer than forty people in the UK have been identified.

Individuals with CASK typically can't walk, are non-verbal and have severe intellectual disability. Approximately 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.

Females with CASK have an unknown prognosis. Although there are cases of death in childhood some children can live relatively healthy lives. Males with MICPCH will likely pass away in infancy.

Typical characteristics of CASK are usually not evident at birth. Individuals with the disorder may have 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 or Dandy-Walker syndrome .

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

CASK RESEARCH UK

CASK Research UK is a registered charity in England and Wales, no. 1197434. Laura Hattersley, the mother of a young girl with MICPCH, founded the charity in 2022. We are registered with the Fundraising Regulator.

We work with leading UK Universities and NHS Clinicians.

Website

www.caskresearch.org

Our website includes support for families, FAQs, common conditions associated with CASK disorders and information for researchers and clinicians.

Subscribe

You can subscribe to our monthly newsletter on our website

Email info@caskresearch.org

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