



CASK
RESEARCH UK



Accelerating a path to a cure

CASK is a gene essential for healthy brain development. Mutations in this gene cause a range of disorders including MICPCH. Children with MICPCH can have a multitude of problems, including intractable epilepsy, inability to feed or drink, inability to walk, low tone, poor balance, global developmental delay, vision and/or hearing loss. Males with MICPCH pass away in early infancy. Females have a variable and unpredictable prognosis.

WHY SUPPORT US?

We are the only UK charity funding research into CASK and supporting families.

WHAT WE DO

Fund Research

99% of the money raised through donations and fundraisers goes directly to research.

Genetic support

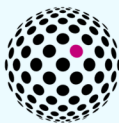
Our geneticist in Germany, Professor Kutsche, investigates the more complex cases to give families answers.

Family support

We have monthly online calls where all registered families can meet and chat and we provide every new family with a welcome goody bag.

Family meet ups

We organise an annual meet up for families in the UK. These provide much needed peer-peer support and reduce isolation.



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What have we achieved?

- **Funded academic research** at Bristol University, Aston University and UC Davis (USA).
- **Increased awareness** by attending conferences, creating a documentary, contacting researchers and being part of larger umbrella groups.
- **Provided information** via our website, pamphlets, a clinical diagnosis webpage and an accessible animation.
- **Facilitated studies** with Cambridge University, Southampton University and Cornell (USA).
- **Created** the global CASK registry - essential for clinical trials.
- **Founded the CASK Coalition** - A group of collaborative CASK non-profits that work together to find a treatment for children with CASK-related disorders. Current members are in USA, France, Australia and UK.



Why support such a rare disease?

Being national we cannot apply for 'local' or 'community' grants that are abundantly available.

National funding pots have extensive competition, often from large well-funded charities. It is difficult for us to compete.

There are very few grants available for scientific or clinical research.

We don't even know why or how our children die. This is unacceptable.

We have a track record of success and garnering international support from the families, researchers and clinicians. We are only held back by our unique situation of being an ultra-rare disease.

"WE REFUSE TO LEAVE OUR CHILDREN'S FATE TO CHANCE."



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