

# CASE FOR SUPPORT

Directed by Science  
Driven by Hope



**CASK**

RESEARCH FOUNDATION

Reg. charity no. 1197434



# CASK-related disorders

CASK mutations are the most frequent cause of the serious brain disorder called PCH. Whilst being aware of and diagnosing PCH, many neurologists will never have heard of CASK.

There are around 400 known cases of CASK related disorders worldwide and over 40 UK cases.

There are no targeted treatments for CASK-related disorders.

We are the only organisation in the UK supporting CASK families and research.

We have the ability to change the outcome for children with CASK disorders.

Learning more about the CASK gene will improve our understanding of how the brain works.

Annie lost her fight with CASK in 2024. She was ten years old. Every year more children and young adults pass away as a consequence of their CASK mutation.



## Laura Hattersley, Founder

“The foundation is a mesh of my passion for science and my first hand experience of caring for a child living with the effects of a CASK gene mutation.

Knowing there are researchers out there wanting to study this disease and that there are children suffering is what drives me to find funding for research into CASK disorders.

# What are CASK related disorders?



The CASK gene produces the protein 'CASK' which is essential for brain development and function.

In affected children the CASK protein is not produced in the required quantities for healthy brain development. This leads to the brain not growing as it should, giving the child microcephaly.

Children with a CASK-related disorder can suffer from 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.

Male infants with a deletion in their CASK gene generally pass away in infancy.

Females with a CASK disorder have an unknown and variable prognosis but most if not all will require life-long care.

Children with a CASK-related disorder often cannot communicate other than by crying, moaning, smiling or laughing.

Many children with CASK cannot play independently due to the inability to control their limbs effectively.

The children who are less medically complex can display challenging behaviours due to their learning difficulties.

Caring for a child with CASK is exhausting since sleep disturbances are not uncommon and seizures often happen during the night.

# About us

CASK Research Foundation was established in 2022 and goes by the shorter name of CASK Research.

We are a Charitable Incorporated Organisation (CIO) registered with the Charity Commission in England and Wales, charity no. 1197434.

The founder and director, Laura Hattersley (MSc, BSc), is the mother of a child suffering from a CASK gene mutation. She has spent her professional life working in the field of biology.

**Our Vision:** A world where people with a CASK gene mutation have targeted treatments, better symptom management and support.

**Our Mission:** To enable medical advances to improve the lives of people with CASK gene disorders and to ensure no UK family feels alone.

In 2022, when CASK Research was established, there was no research into CASK in the UK. As a direct result of our work, at least five UK universities, including the University of Cambridge, are currently studying CASK disorders.

## *“We know Science”*

We create grants for UK researchers in order to move CASK research from the lab and towards clinical trials and the clinic.

We work closely with the University of Bristol and are working alongside NHS paediatric neurologist Dr Sam Amin to establish a Centre of Excellence in Bristol.

# The small national charity dilemma

- 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 research focused on a single rare disease.
- No government funding is available for CASK research.

We have a track record of success and garnering international support from the people that understand our predicament the most: the families, the researchers and the clinicians. We are only held back by our unique situation of being an ultra-rare disease and the lack of funding opportunities this presents.



**“We refuse to leave our children’s fate to chance.”**



# Achievements to date

## Increased awareness

- Attended and spoken at national conferences & events.
- Multiple radio appearances.
- Presented CASK disorders to over 100 doctors at the British Paediatric Neurological Association conference, 2024 & 2026.
- Created the documentary 'Life with CASK'.
- Developed personalised leaflets for families to use to spread awareness and educate their families.

## Grants provided

- Gene therapy research - University of California Davis, USA
- CASK fly model and research - University of Bristol.
- UK Patient registry and Biobank - NHS and University of Bristol

## Funds raised

- Raised over £120,000 since our creation in 2022, from individual fundraisers, supporter donations and legacy giving.

## Collaboration with Universities

- **Cambridge University** - CASK Behaviour and Emotion study - now published.
- **University of Bristol** - Professor Hodge and Dr Amin - clinical and fundamental research.
- **University of Southampton** - Neuronal work
- **University of Birmingham** - Dr Chan, metabolic aspects of CASK.
- **University of Bristol** - Dr Low, the GENROC study.
- **University of Portsmouth** - Developing a Zebra fish model, Dr Cayuso.

## CASK registries

- Over 160 global patients are registered on RARE-X, our chosen registry for CASK disorders
- We have 40+ patients registered on our UK register.

## Family support

- Annual UK CASK family meet ups.
- Virtual family socials held every other month.
- Free Counselling provision for families in the UK and Ireland.

## Genetic support

- We work with expert geneticist Prof. Kutsche, whose team first discovered CASK-related disorders, to provide families with answers and understanding.

# CASK by numbers

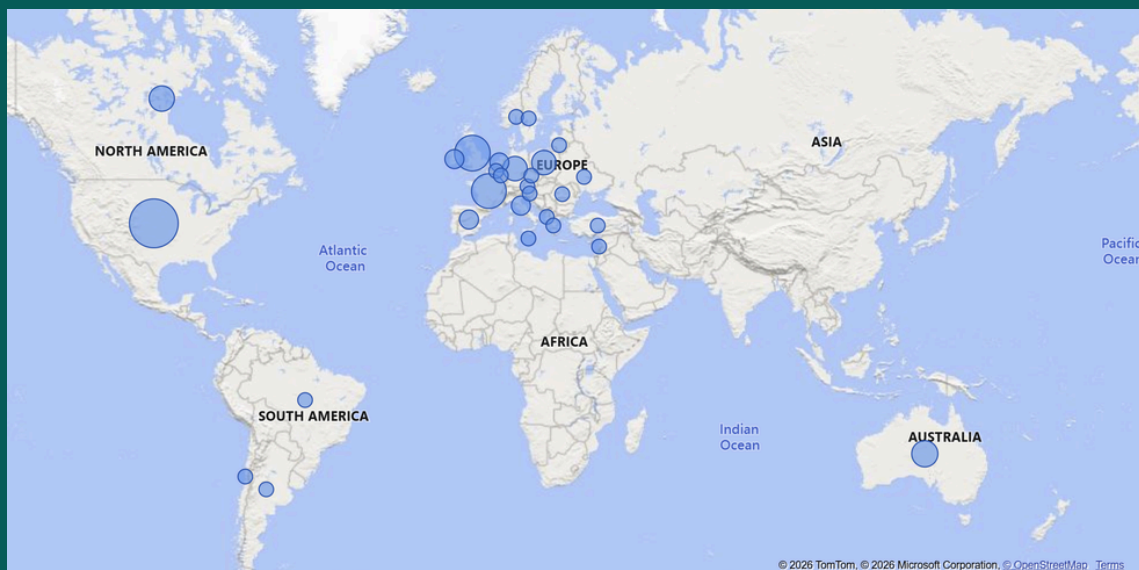
Patients registered with CASK Research Foundation , 2024 data



CASK related disorders are ultra-rare; however, there are over 3.5 million people in the United Kingdom suffering from a rare disease. If rare diseases are ignored and research into them is disregarded, that equates to 3.5 million people with no real hope of a cure or even effective treatments.

By supporting CASK Research Foundation you are doing your bit towards ending this discrepancy and providing hope where there was none.

Current patients registered with a CASK gene mutation as of May 2026 - courtesy of RARE-X registry



# Why help us?

## Efficient

100% volunteer-led means your donations will go to the research and not to capital costs or salaries.

## Personal

You will receive regular updates of how the research you helped fund is progressing.

## Dedicated

Run by CASK parents and friends, we have limitless drive and passion.

## Successful

Because there is so much to learn about CASK all research will reap benefits in terms of furthering our understanding.

## Broader Relevance

Any research into CASK will further our understanding of the brain and could enhance understanding of associated diseases such as Parkinsons, autism and cancers.

## Impactful

Before our existence there was:

- zero UK research into CASK
- no gene therapies being investigated
- no dedicated support for families
- Minimal awareness of the disease
- No research into CASK epilepsies
- no collaborative network of CASK non-profits

**AND NOW**, as a direct effect of our work:

- Multiple UK universities studying CASK.
- 2 gene therapies under development.
- Support for all UK CASK families
- Increased awareness.
- National and global registries.
- The global [CASK Coalition](#).



# How can you help?



## Sponsor an event

Offering to sponsor the hire of a hall or the cost of entertainment can mean all funds raised can go towards the important work of the charity.

## Match funding

Agreeing to match a particular fundraising initiative increases the impact and money raised from an event.

## Volunteer a specialist skill

For example, website design, social media creation or writing press releases are all valuable ways to contribute to the work of the charity.

## Fundraising

Your workplace could organise a fundraising event such as a cake sale, raffle or office challenge. This will help our charity whilst also team building.

## Payroll giving

Members of staff can make a donation directly from their salary. The donations are taken from employees pay before income tax is applied, making it a tax-effective way to give. In addition, knowing donations are coming in regularly makes it easier for our charity to plan.

## Be a charity ambassador

A great way to raise awareness and support for the work we do is by acting as an ambassador where you tell others about our work and how they can get involved to help.

# Don't take our word for it...



"I could not imagine a world and our fight without CASK Research. We have been delighted to have CASK Research help us build our charity and network connections with scientists and clinicians. We have mutual confidence in each other and share everything, which helps accelerate things for research on the CASK gene."

Thierry Kervella - President of Association Enfants CASK France

"You are doing SO much amazing work. And there will be real progress for CASK families, I'm sure. Better information is an essential starting point, as well as all the innovative experimental work."

*Dr Kate Baker, Clinical Geneticist and researcher, Cambridge University*



"Thanks to the foundation, my daughter has undergone a genetic test for scientific purposes, which helps understand why, in her case, CASK has not shown such extreme symptoms. Additionally, this test provides understanding and evidence that, in the future and with the necessary resources, CASK could be treated by modifying the bias of the X chromosome. This suggests that children can overcome regression and be capable of learning, as they are individuals with incredible abilities that will contribute to the world in significant ways due to the life experiences they have endured"

*Mother to a child with a CASK-related disorder, Chile*

# How We Work



## Governance

- We have a board of trustees, 50% of whom are parents of a child with CASK.
- The organisation is 100% volunteer led and run.
- Our Director has a scientific background.
- Volunteers are well experienced and include accountants, policy workers and branding experts.



## Costs

- Funding grants – We aim to create a least one UK research grant a year of £40,000
- The costs of the annual family meet are covered by corporate sponsor 'The Park London'.
- We are also able to support UK researchers who require small, additional funds to complete a project.



## Research Advisory Board

- Professor James Hodge - Neurobiologist, Bristol University
- Dr Mingshan Xue - Associate Professor, Dept of Neuroscience, Baylor College, USA
- Professor Silverman - neuroscientist, University California, Daivs.
- Dr Isabel Zwart (Director Regulatory CMC at AstraZeneca)
- Dr Emma Davis - Principal Investigator, Healx

## Clinical Advisory Board

- Dr Sam Amin (Consultant paediatric neurologist)
- Professor Kerstin Kutsche (deputy director of the Institute for Human Genetics, Hamburg)
- Dr Catherine Tuffrey (consultant lead for research in community paediatrics in Solent)



## Collaboration

- Our memberships: [Genetic Alliance UK](#); [Global Genes](#), [UK Rare Epilepsies Together](#), [GW4 Epilepsy Community](#), [Neurological Alliance](#), [Bristol Neurodevelopmental Hub](#).
- We are founding members of [The CASK Coalition](#)



# Inspired?

Contact us at  
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Together let's  
**#conquercask**

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