CASE FOR SUPPORT

CASK
RESEARCH FOUNDATION

Searching for Answers
Researching for Hope
About us

We are a Charitable Incorporated Organisation (CIO) registered with the Charity Commission in England and Wales.

CASK Research Foundation was established in 2022.

Our founder, Laura Hattersley (MSc, BSc), is the mother of a child suffering from a CASK gene mutation.

Our Vision: To create the best possible outcomes for people with CASK gene disorders

Our Mission: To enable medical advances to improve the lives of people with CASK gene disorders

We are: ambitious, supportive, informative, collaborative
What are CASK related disorders?

CASK is an ultra rare genetic condition that affects the brain.

As a result, the brain doesn’t grow as it should, giving the child microcephaly (literally meaning ‘small head’).

Children with CASK 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 CASK generally pass away within the first few years of life.

Females with CASK have an unknown prognosis but many are classed as having a life-limiting condition and rely on hospice care.

Most children with CASK cannot communicate other than by crying, smiling or laughing.

Caring for a child with CASK is exhausting since most suffer from sleep disturbances.

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

The milder children with CASK can display challenging behaviours due to their low cognitive abilities.
In short

• The problems

There is no treatment for children suffering from a CASK gene mutations.

International scientists are interested in studying the disease but struggle to get funding due to the rarity of the condition.

There is little information and support for parents and caregivers.

• The solutions

Increase awareness about the condition amongst the scientific and medical community.

Create research grants to enable research to happen.

Have online resources with information for parents and guardians.

Provide meet ups for UK families.
The facts

There are just over 220 known cases of CASK related disorders worldwide.

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

We have a scientific committee dedicated to ensuring funds are allocated appropriately and effectively.

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

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

Laura Hattersley, CEO

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

Knowing there are researchers out there wanting to study this disease is what drives me in my quest to put CASK on the rare disease map.
# Achievements to date

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<tr>
<th>Increase awareness</th>
<th>Create grants</th>
<th>Provide information</th>
<th>Family support</th>
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| • Held multiple events throughout the country raising awareness  
  • Contacted scientists and clinicians  
  • Joined RARE-X, a revolutionary Data Collection Platform  
  • Taken part in the Beacon patient group pairing | • Held fundraising events  
  • Created fundraising packs  
  • Online fundraising  
  • Sponsored events | • Successful informative website  
  • Collaborated with UK charity 'Unique' to publish a pamphlet about CASK for carers and clinicians | • First annual UK CASK family meet up held in June 2022  
  • Website updated to encompass more charities that support families |
How We Work

Governance
- We have seven trustees, three of whom are parents of a child with CASK.
- The organisation is 100% volunteer led and run.
- CEO has a scientific background.
- Volunteers are well experienced and include professional fundraisers and marketing, events and branding experts.

Costs
- Funding grants – we have the aim of creating our first research grant by September 2023.
- Our online support is privately funded.
- The costs of the annual family meet are covered by corporate sponsor ‘The Park’.

Research Review Committee
- We currently have three members of the committee and are looking to expand.
- Members’ expertise include stem cell research, scientific publishing, rare disease and drug licensing.

Collaboration
- We work closely with the other CASK foundations in Australia, France and USA.
- We are members of Genetic Alliance, Global Genes and RARE-X, a data collection platform.

Advisors
- Medical advisor: Paediatric Neurologist Frances Gibbon.
- We are recruiting for further scientific advisors to grow our team.
We know Science

Our funds will go to the best science and the best scientists

In order to achieve this, we have a strict RESEARCH STRATEGY, summarised briefly below:

1. The project must be able to meet our aims as a charity: improving the lives of people suffering from CASK related disorders

2. Other questions include “Is the research underpinned by scientific excellence?” and “Has the right team been identified?”. 

3. Written peer review (for grants above £25,000)

4. Research review committee makes recommendation

5. Trustees make the final decision on whether the charity should award funding to the research application.

Our Research Review Committee

Lucy Robinson, PhD - Research Review Committee (RRC) chair
Lucy founded Insight Editing London which assists authors in publishing articles and books across diverse scientific fields including oncology, virology, immunology, genetics and embryology.

Isabel Zwart, PhD, MSc
Isabel obtained her D.Phil from Imperial College London in Clinical Medicine Research. She has worked in the pharmaceutical industry on medicinal products for neurodegenerative diseases and is the current Director Regulatory CMC at AstraZeneca.

Jessica Tamamini, PhD
Jessica obtained her D.Phil in neurobehavioural genetics and ophthalmology from the University of Oxford. She now works in scientific publishing.
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.
Our supporters

The Park, London

This award winning brand proof company supports us by providing corporate funding as well as helping us manage our branding, events and giving us business advice.

Dr Frances Gibbon

Frances is our current medical advisor. She became a consultant in Paediatric Neurology at the Children’s Hospital for Wales, Cardiff in 1997.

Frances has over 20 years experience of caring for children with epilepsy and other neurological disorders and was a member of the Cardiff ABICE (Advanced Brain Imaging in Childhood Epilepsy) group based at the Cardiff University Brain Research Imaging Centre, CUBRIC.

Frances has first hand experience caring for patients with CASK and has her expertise in epilepsy.

Evolve Psychological Services

They offer clinical psychological assessments and therapeutic support services for families, children and adults. 'Evolve' undertakes neurodevelopmental assessments and psychological therapy. Their founder, Dr Laura Powling, can offer advice regarding support, assessment and therapeutic services.
How can you help?

Sponsor an event
Putting on fundraising events costs money. Offering to sponsor the hire of a hall or the cost of entertainment can mean all funds raised become profit.

Match funding
Agreeing to match a particular fundraising initiative.

Volunteer a specialist skill
For example website design, social media creation or counselling.

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
Perhaps your members of staff would want to donate so they pay a donation directly out of their wage.

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.
The advantages of our small, unique charity

Minimal overheads
You know your money will go directly towards a grant.

No government funding
Without your support no grants will be created. You are this rare disease's hope.

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

Dedicated
Run by CASK parents, we have limitless drive and passion
Inspired?

Contact us at
admin@caskresearch.org

Together let's
#conquercask