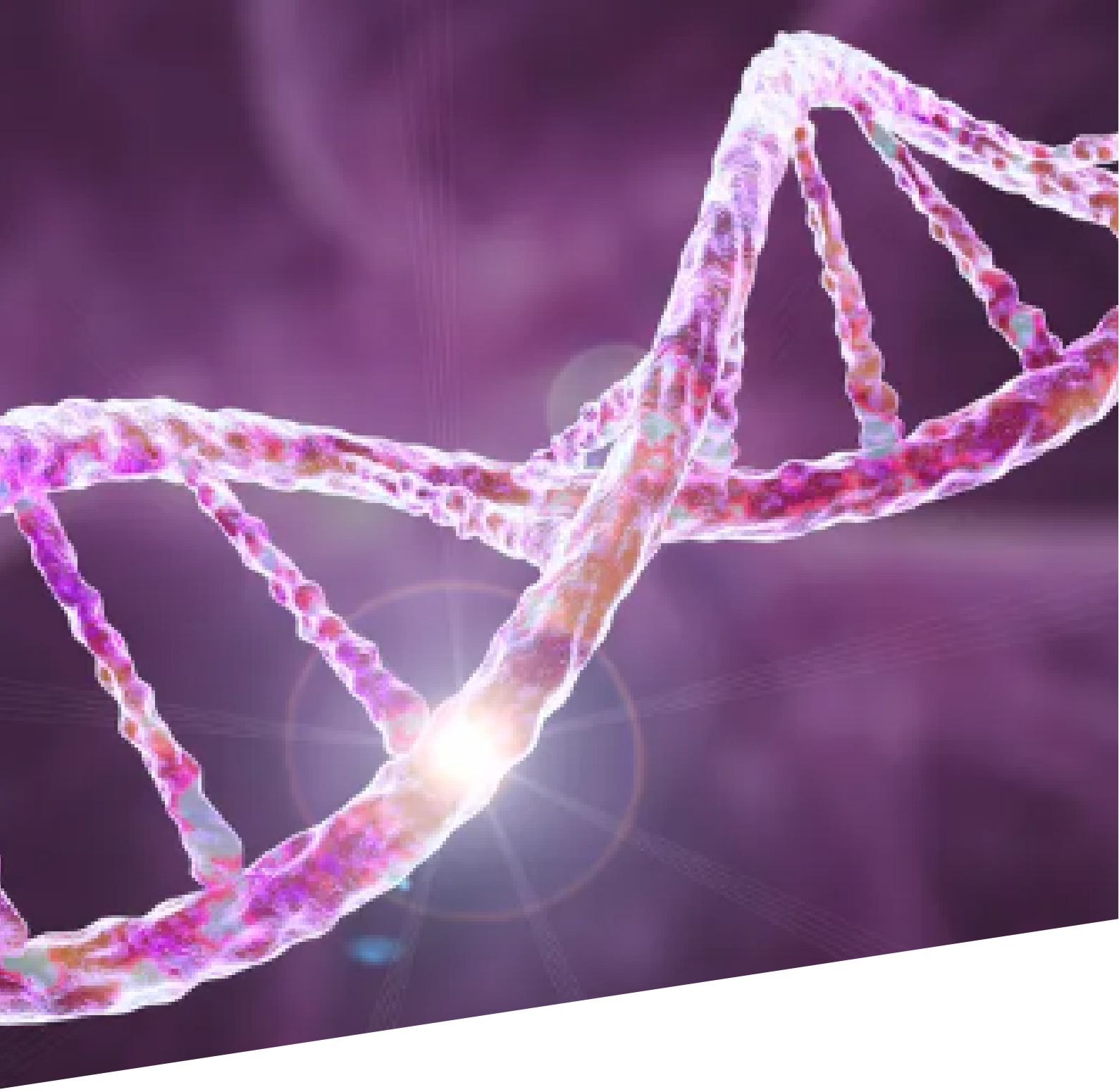

AECF

ASSOCIATION ENFANTS CASK FRANCE

N°RNA: W782009952 - N° SIRET: 918 157 991 00012

JULY 2026

ASSOCIATION ENFANTS CASK FRANCE

AECF



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THE ASSOCIATION

WHO ARE WE?

The association was created by parents of French children suffering from genetic mutations of the CASK gene (pathogenic variants).
N° RNA: W782009952 / N° SIRET: 918 157 991 00012
President : Thierry KERVELLA
Secretary: Claire PORTENSEIGNE
Treasurer : Magali Dalle-Palle
Members: 39 member families in France and 1 in Luxembourg, each with a family member affected by genetic mutations of CASK gene (children or young adults).
Scientific commitee: constituted of scientists (doctors and researchers)
Secondary facilities : 10 regional branches

SPONSORS

Bruno GUILLON (animateur radio et TV)



Marche des Maladies Rares 2021 à Paris

WHEN ?

Created July 2022

HISTORY

AECF is a **non-lucrative general interest caritative association**. Prior to its creation, several « family » associations have been created in order to finance care and for children in France or other countries; the material costs materiel were taken care of by the families, but no provision was made for financing medical research. These associations continue to exist.
AECF is an association **for ALL French children suffering from CASK-related pathogenic variants.**
Our missions are described below.

OUR MISSIONS

AECF is a non-lucrative caritative general interest association whose missions are:

1. **Provide financial aid for innovations** in the area of world-wide medical research related to diagnostics, prevention, and treatment of CASK gene mutations.

2. Use public and private sector input to establish a **scientific committee** to validate and select the best medical research projects in a professional, legal, and fair manner.

3. Maintain and make available **clear and precise information** to help patients diagnosed with CASK and/or their families.

4. **Establish and facilitate relationships** between families impacted by this rare disease.

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5. Ensure that the general public as well as the medical sector are made **aware of this rare genetic disease**, both in France and other countries.

6. To actively participate and help the **development of medical research methods as well as the treatment for CASK sufferer**s in order to improve their support, development, and quality of life.

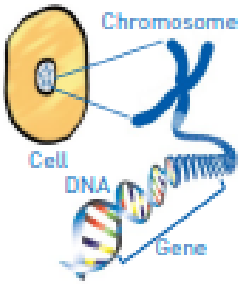
7. Establish **partnerships** with other caritative organisations in France and other countries.

8. Be an association that values diversity, honesty, and integrity

WHAT IS CASK?

The CASK gene is situated on the X chromosome and codes for the protein calcium/calmodulin-dependent serine protein kinase. The CASK protein plays a vital role in brain development, allowing brain cells (neurones) to function correctly. It also interacts with a large number of other proteins thus making its role widely important.







Origin

Genetic (pathogenic variants).
In most cases, random "de novo" mutations, more rarely mosaicism in parental gametes.

Consequences

- Cerebral malformation (microcephaly and ponto-cerebellar hypoplasia)
- intellectual deficiency
- Speech/language difficulty
- Motor function problems (axial hypotonia and hypertonia of the extremities)
- Troubles with vision and audition
- High risk of pharmacoresistant epilepsy, digestive problems, scoliosis...
- Sleep problems

Social consequences :

- Multiple handicaps
- Frequent medical visists & follow-ups
- Heavy impact on family and social life for the parents and close helpers

Rare orphan disease

Currently only **350 world-wide cases of CASK have been diagnosed** since its discovery in 2008. In France, about 50 families are affected.

No treatment exists at the moment, and CASK diagnosis can more or less be detected depending on the severity of the disease.

* Different types of mutations responsible of various phenotypes depending on cases (serious *de novo* mutations or resulting from mosaicism in parental gametes, less severe mutations for the gene and mainly observed on boys suffuring from remote intellectual deficience)

CASK COALITION

What is CASK Coalition ?

The CASK Coalition is a group of like-minded and dynamic CASK gene collaborative research associations.

The aim of the CASK Coalition is to harmonize and mobilize all the vital forces of the various foundations dealing with the CASK gene mutation.

Coalition members pool their resources and skills to help each other, exchange ideas, share contacts and work together to find a treatment or cure for CASK disorders.

AECF France and its partners are acting together and calling on all CASK-related organizations to give priority to collaboration rather than competition, in the interests of those who suffer: patients and carers alike.

Why a coalition ?

With around 350 confirmed diagnoses, the CASK community is small and scattered across different countries. United by a Facebook group that provides an essential platform for sharing information and supporting families affected by the disease, the community is currently represented by several foundations around the world.

Given the size of the community and the enormous obstacles to the advancement of treatments and cures for rare diseases, the CASK community would benefit from a collaboration to define a community-wide research roadmap, promote unified data collection and collectively undertake awareness-raising activities.

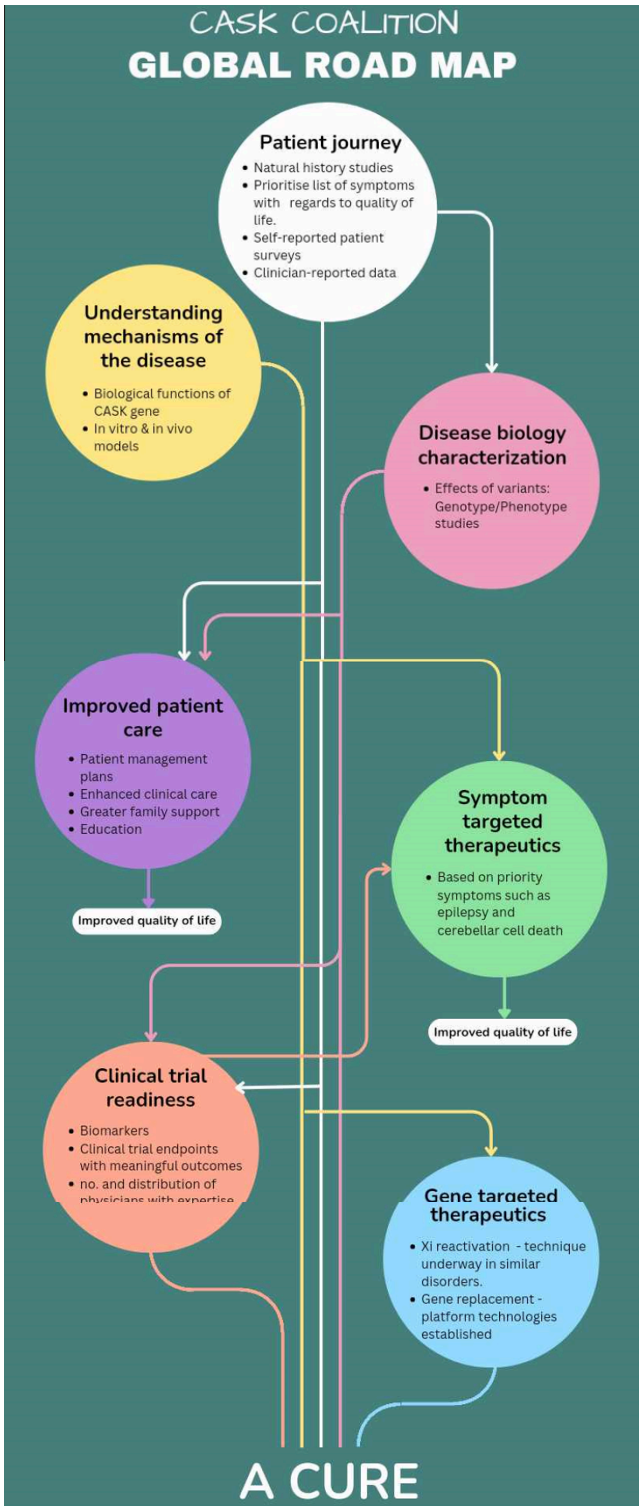
CASK COALITION
United for our CASK warriors

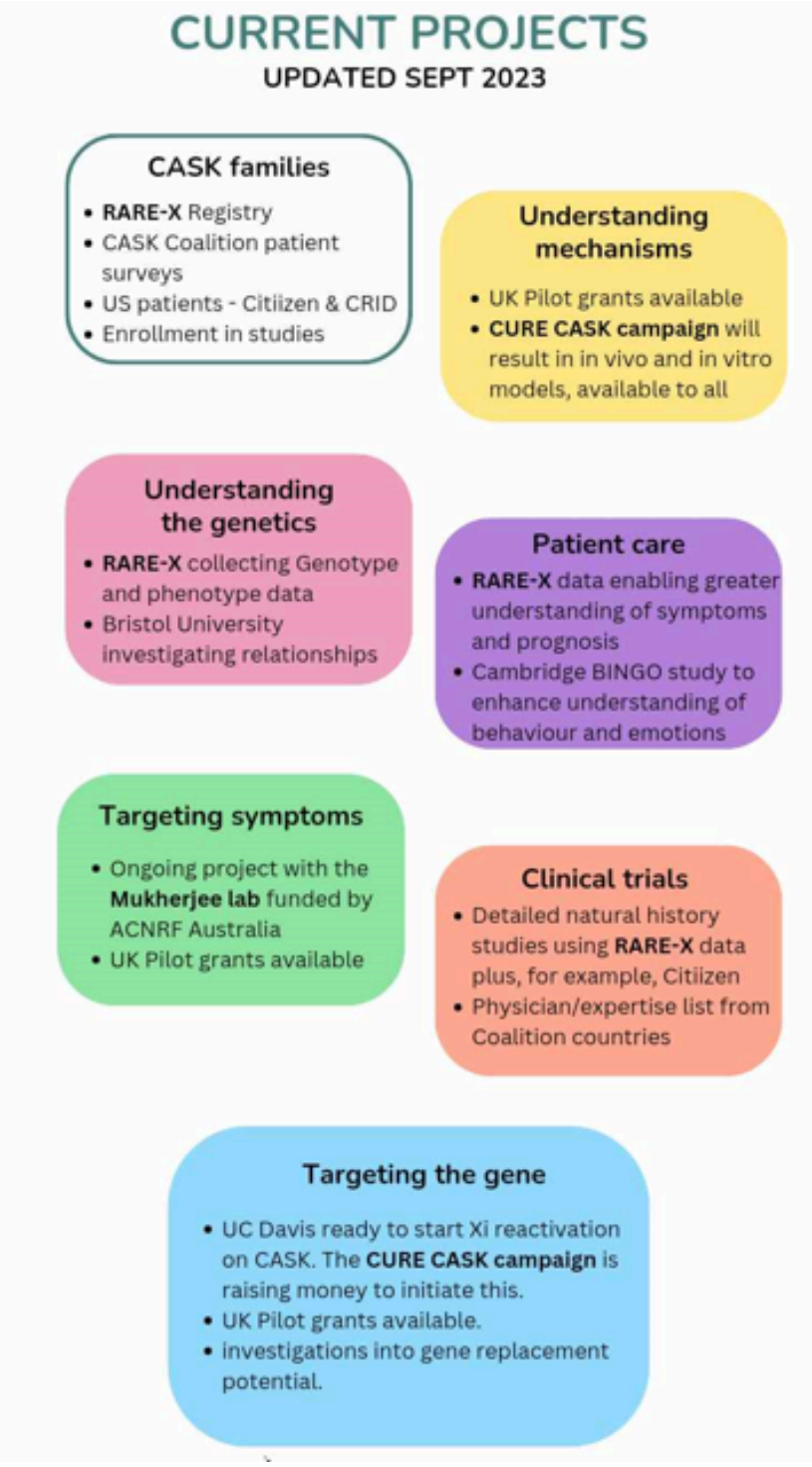
CURE CASK
CASK RESEARCH
PROCASK ITALIA
CASK Luxembourg asbl

What is its role ?

The CASK Coalition provides a platform to streamline collaboration between CASK foundations that become members, reduce redundancy between foundations for activities that can be managed globally, and promote greater engagement with the global CASK community to advance research. We advocate best practices and transparency within the non-profit CASK arena.

CASK COALITION ROADMAP



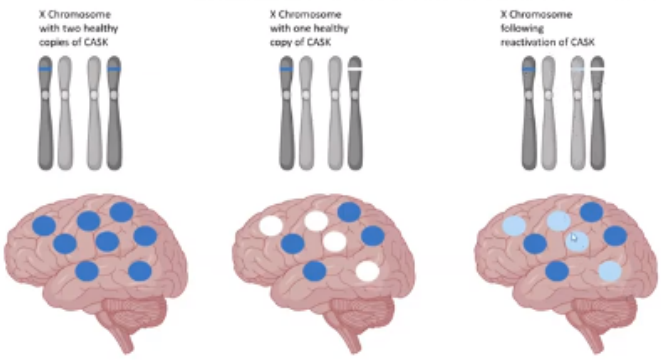


CASK COALITION
United for our CASK warriors

The CASK coalition prioritizes neglected aspects of the roadmap. These include a detailed long-term patient pathway, improved quality of life, clinical trial readiness and gene-targeted therapies. The CASK gene, its mechanisms and its role in the disease are the subject of global, growing and diversified research. What is lacking is translational research with the ultimate goal of a cure.

CURE CASK: A HOPE FOR ALL OF US

The AECF is associated with the CASK Research Foundation and the Angelina CASK Neurological Research Foundation in order to raise funds and finance a genetic therapy research project with the aim of reactivating the X chromosome in girls. The project is named CURE CASK.



The Research

2 lines of study :

- Work "in vitro" on pluripotent human stem cells and transform them into different types of brain cells in order to evaluate this approach
- Examine and study CASK murine models; evaluate the ability of therapeutic treatment to reactivate the healthy CASK copy in the brains of mice and test their recovery

The theory

All girls possess two X chromosomes and thus two copies of the CASK gene. Early in the development of the foetus, one chromosome is inactivated due to some biochemical mechanisms (such as XIST genes, DNA methylation, etc) . As a result of the gene mutation, only about 50% of brain cells possess the healthy CASK protein.

The aim of the CURE CASK project is to be able to reactivate, in the brain cells, on the inactive chromosome specifically the CASK gene. Thus, a synthesis of a working, healthy, CASK protein would become possible.

Expected effects

Successful activation of silent CASK genes in a sufficient number of cells would have a major impact on the lives of CASK sufferers, possibly even reversing certain symptoms.

CURE CASK: A HOPE FOR ALL OF US

CURE CASK

History

The reactivation of the X chromosome has been successfully accomplished on murine models for the CDML5 and MeCP2 genes (Rett syndrome). Research teams claim that the CASK gene is a good candidate for this approach.

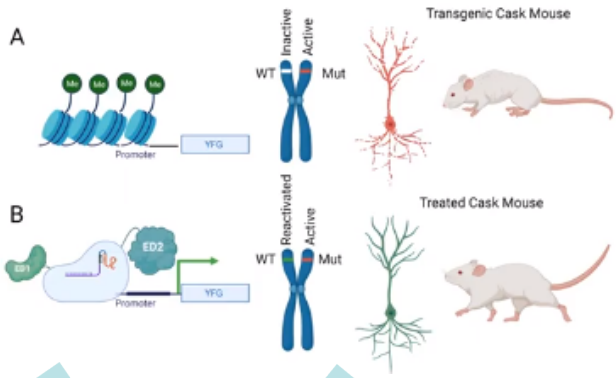
UC Davis

'The UC Davis MIND institute, located in California, is at the origin of the project. It is a collaborative international research centre engaged in providing awareness, understanding, prevention and treatment for neurodevelopmental handicaps and rare disorders relating to the X chromosome.

Project cost

The cost of the whole CURE CASK project is **estimated at 1,5 million US dollars**. We splitted it into small part in order to start with validation of the concept on induced pluripotent stem cells.

This is clearly a very high cost for the different associations, but we are convinced that we can raise this sum. Furthermore, if this revolutionary technique is successful, it can greatly improve other research projects related to the X chromosome, including research to find a cure for boys affected with CASK gene mutation.



Parents say:

"Given that the disease is so rare, the creation of this research project is a real once-in-a-lifetime godsend".

HOW CAN YOU HELP?

« Small streams become great rivers »

COMMUNICATE !

Share information about the association with family and friends; propose partnerships with other associations, organize events ...

MAKE A DONATION

You can donate via the associations's web site. Your donations help to finance international CASK-related medical research

1 € donated = 1 € for research

VOLUNTEERING OR PROPOSING ACTIONS BY THE MUNICIPALITY

Festive or sports events, patronage, volunteering, or a word of encouragement (always welcome!). You can join us on our Facebook page
Association Enfants CASK France | Facebook

CLICK HERE TO DONATE

All donations are 66% tax-deductible for individuals, and 60% tax-deductible for enterprises; a tax receipt is automatically provided.

Example for an individual:

100€ donated → 66% tax deduction → real cost is **34€**

SUPPORTING OUR ASSOCIATION

RARE ≠ DESPAIR

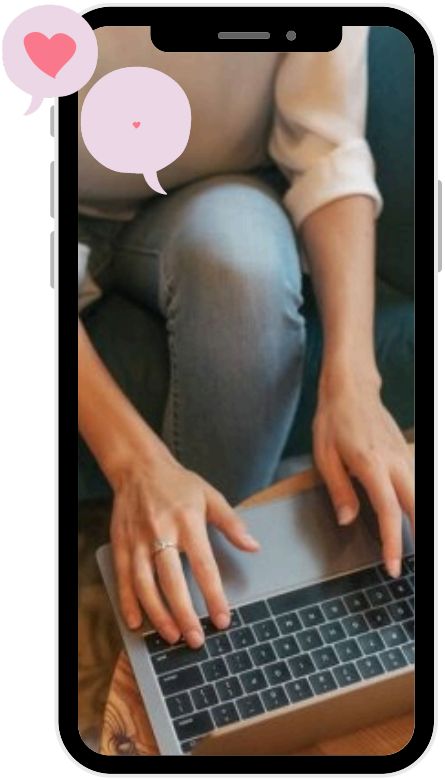
We are the only association that finances and promotes research into CASK-related problems in France. We do not receive government funding. SO, PLEASE, help us to save lives.

BECOME A MEMBER OF THE ASSOCIATION

The membership fee is 25€ per year

EVERY ACTION COUNTS!
GET TOGETHER TO HELP US!

CONTACTS & USEFUL REFERENCES



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AECF ASSOCIATION ENFANTS CASK FRANCE

www.aecf-france.fr

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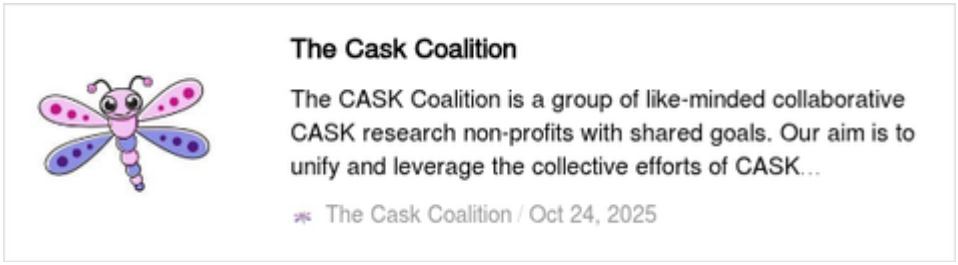
Other useful links :

https://thecaskcoalition.com



Alliance maladies rares
UNIS POUR VAINCRE

MEMBER OF EURORDIS RARE DISEASES EUROPE

THANK YOU FOR YOUR SUPPORT

CASK warriors in France



ALL DIFFERENT BUT ALL TOGETHER !