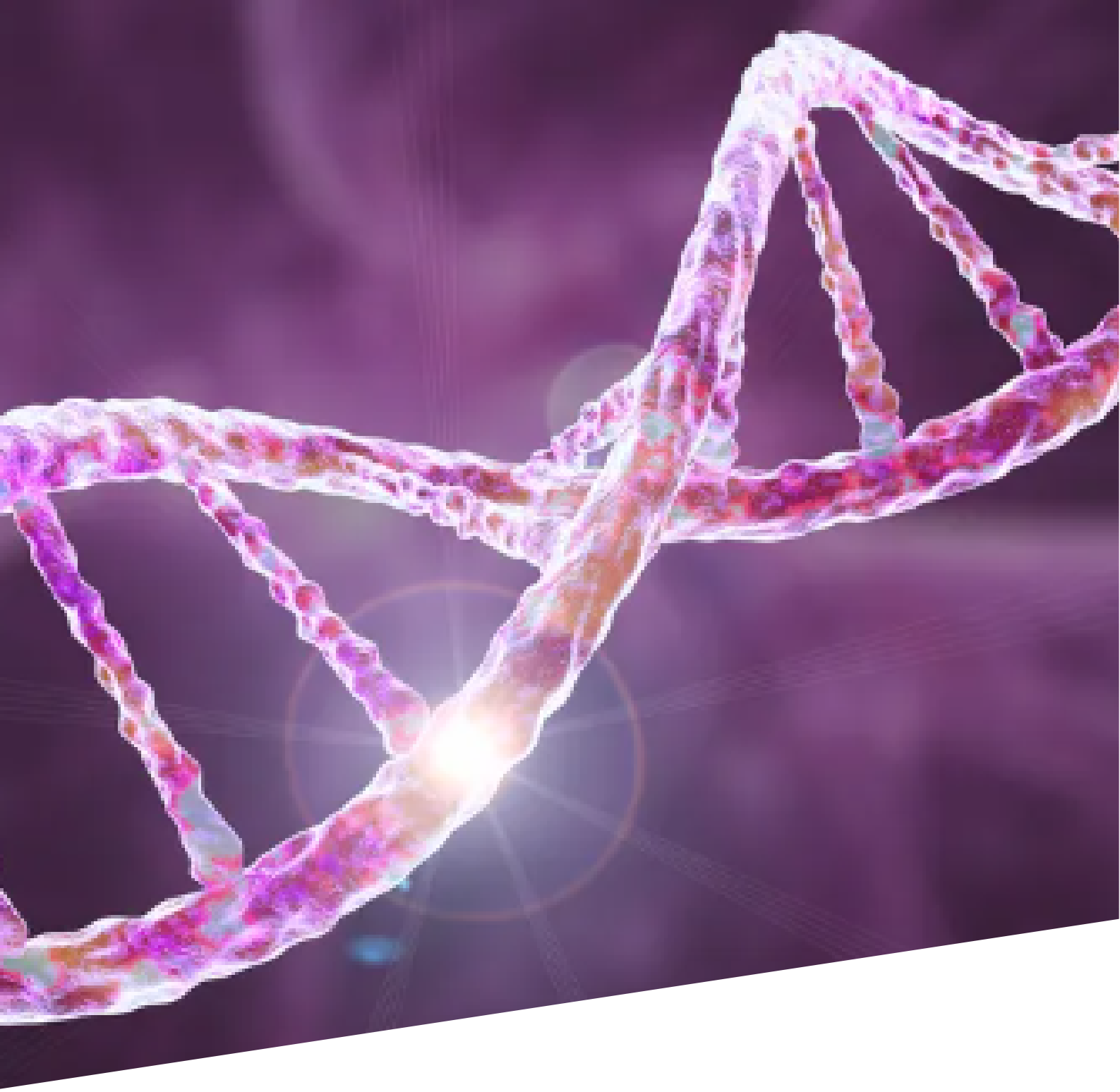

AECF

ASSOCIATION ENFANTS CASK FRANCE

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

JMAY 2023

AECF

MEMBER OF EURORDIS RARE DISEASES EUROPE

AECF

Summary

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
President : Thierry KERVELLA
Treasurer : Jennifer MARTIN / Vice-Trésorier: Mathieu BEHLOULI
Secretary: Claire PORTENSEIGNE
Members: Luisa GUERGUET, Ghyslaine MARTIN, Géraldine DE NORONHA DUPRAT, Marilys REICHELL, Charlotte SCHOTSMANS, Nathalie TOURE, Elodie TEXEIRA, Sarah COSTANTINI, Livia NONORGUES-DAHAN, Charlotte DEBRUYNE, Amal MOUCHARIK, Marie-Andéole HALLE, Anne BECOURT, Dominique CARCEL, Nicolas DEJEAN, Jennifer & Nicolas BOUTRON, Nathalie RICHARD, Evelyne CUBELLS.
Scientific commitee: constituted of scientists (doctors and researchers)

SPONSORS

To be identified - on-going

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.

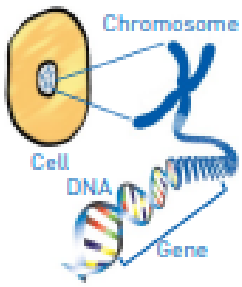
6. To actively participate and help the **development of medical research methods as well as the treatment for CASK sufferer**s in order to improve their support, development, and quality of life.

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

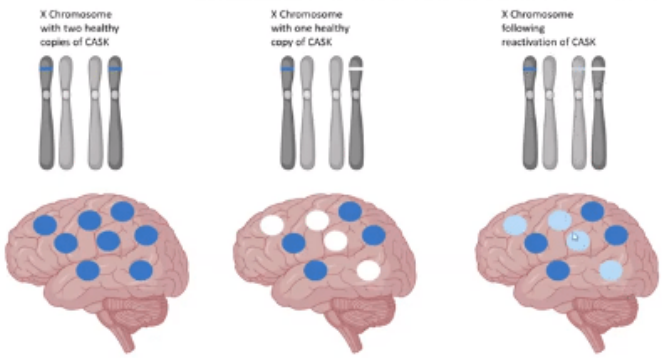
Currently only **200 world-wide cases of CASK have been diagnosed** since its discovery in 2008. In France, about 15 families are affected.

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)

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

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.

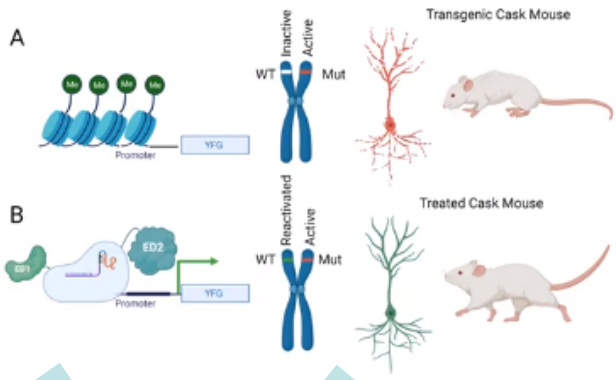
UC Davis

'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 CURE CASK project is **estimated at 1,5 million US dollars**.

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

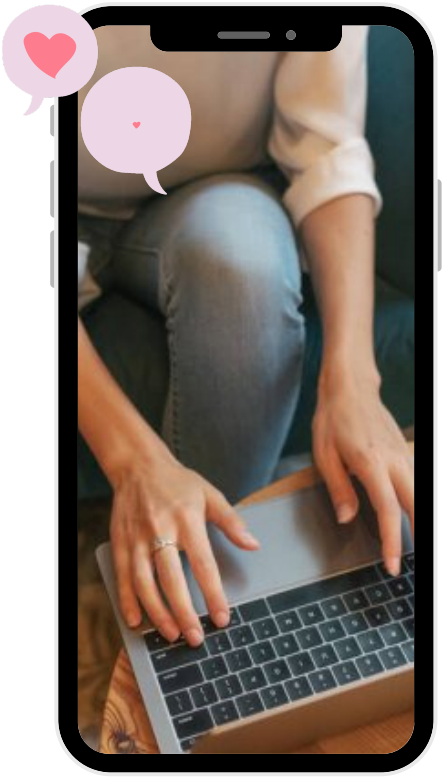
We are the only association that finances and promotes research into CASK-related problems in France. We do not receive government funding. SO, PLEASE, help us to save lives.

BECOME A MEMBER OF THE ASSOCIATION

The membership fee is 20€ per year

EVERY ACTION COUNTS!
GET TOGETHER TO HELP US!

CONTACTS & USEFUL REFERENCES



Association Enfants CASK France

3, allée des Peupliers
29290 MILIZAC-GUIPRONVEL

06 82 97 45 93

associationenfantscaskfrance@gmail.com

Association Enfants CASK France | Facebook

www.aecf-france.fr

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Other useful links :

Websites :

- CASK Research Foundation
- CASK gene Foundation
- ACNRF

Facebook:

- The CASK gene Parent Support Group

THANK YOU FOR YOUR SUPPORT

CASK warriors in France

ASSOCIATION ENFANTS CASK FRANCE

Clément Cassy Olivia Emma Tom Chloé Inaya Tara Elyse Shérazade
Enora Mia Kylian Hortense Maude Romane Rose Elisa Ali Lucie Diane

ALL DIFFERENT BUT ALL TOGETHER !