



How to get research done on your rare disease

9th European Conference on Rare Diseases & Orphan Products (ECRD) 2018



The CATS Foundation

- Founded in 2011
- Supports families affected by Tay-Sachs and Sandhoff
- Provides equipment, respite trips and a support network
- Support and involved in the the research for a potential treatment for Tay-Sachs and Sandhoff



What is Tay-Sachs & Sandhoff

- Terminal illness in children
- Inability to produce a vital enzyme in the brain
- Genetic condition – autosomal recessive inheritance
- Affects 1 in 320,000
- Carrier rate of 1 in 300
- Average life expectancy of around 5 years of age





Amélie

- Diagnosed with Tay-Sachs in 2011
- Told she will die in early childhood
- No dedicated charity providing support
- Very isolating & very scary





The future

“You probably won’t meet another family with a child affected by Tay-Sachs.”



4th European Tay-Sachs & Sandhoff Family Conference

Disneyland Paris – 2016



ETSCC
EUROPEAN
TAY-SACHS &
SANDHOFF
CHARITY
CONSORTIUM



ETSCC
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SANDHOFF
CHARITY
CONSORTIUM

5th European Tay-Sachs & Sandhoff Family Conference

London – 2017





What we were told

“You probably won’t meet another family with a child affected by Tay-Sachs.”



What we were told

~~*“You probably won’t meet another family with a child affected by Tay-Sachs.”*~~

WRONG



Quick history

- Set-up the charity to support families in 2011
- Looked into the research for a treatment
- Raised our profile with the research community
- Set a clear goal



Our goal

“To save children from Tay-Sachs and Sandhoff disease. We want to give them an opportunity to have a future.”

SALES DE UNION - MEETING ROOMS
PORTES D'EXHIBITION
TOILETTES



ETSCC
EUROPEAN
TAY-SACHS
CHARITY
CONSORTIUM

10-13TH
OCTOBER 2014
DISNEYLAND
PARIS, FRANCE

2nd European Tay-Sachs
& Sandhoff Disease
Family Conference

One goal. One group. One voice.

Organized by
CATS
CENTRE FOR
TAY-SACHS
AND
SANDHOFF
DISEASE

HAND-TO-HAND
EUROPEAN
TAY-SACHS
AND
SANDHOFF
DISEASE
CONSORTIUM

ACTAVS
ACTIVITIES
AND
THERAPY
VAINCRE le Tay-Sachs

The European Tay-Sachs Charity Consortium (ETSCC) provides a unified voice to the Tay-Sachs and Sandhoff Disease community.



The power of a community

- Although rare there were lots of families in Europe
- Research into Tay-Sachs and Sandhoff was gathering pace
- Put in contact with Professor Timothy Cox





What the research team needed

- Team at The University of Cambridge had been working with on a treatment for GM2 for over 20 years
- Needed a charity to support them to secure funding
- Needed a charity to help them identify patients
- Needed a charity to raise awareness of the Tay-Sachs and Sandhoff



What we needed

- Needed more families to know a support network existed
- Needed a research team to find a treatment
- Needed someone to communicate with families

TEAMWORK
MAKES

the

DREAM
WORK



Collaboration is key

- We work closely with the research team
- Together we have a planned Gene Therapy Clinical Trial due to start by 2019
- The CATS Foundation is the nominated patient organisation supporting this work



Our role

- Advisors to the research team
- Attend vital regulatory meetings with the team
- Met with the Medicines and Healthcare Products Regulatory Agency (MHRA), Medical Research Council (MRC) and European Medicines Agency (EMA) to get approval for the trial
- Keep patients at the forefront of the conversation



Emma Murphy
United Kingdom

RESENTER



Putting a face to the disease

- We are able to share a real life experience
- We act in the best interests of the families
- “False hope” is the worst hope
- Realistic with the information and message we share



The trial

- It will be a Phase 1/2 clinical trial
- Using AAV2/1-HEXA and AAV2/1-HEXB vectors (successful in the animal models)
- We will have a dosing rationale based on each dose having a therapeutic impact on an individual
- We will treating younger children before those up to 18 years-old
- The first six participants admitted to the trial are most likely to be from the younger cohort



Created a European Consortium





Working together

- There is a clear and consistent message across Europe
- We all work together and fund research projects as a group
- We have the only patient registry for Tay-Sachs and Sandhoff
- No point in us working as individual groups
- Together we are stronger



ETSCC message

“One goal. One group. One voice.”



How far we have travelled

- In 2011 there were no charities support Tay-Sachs and Sandhoff
- In 2018 there are charities in the UK, Spain, France and Germany
- A European Charity Consortium working together
- A trial due to start in 2019 for all European patients



The future

*“There’s no treatment that will help a child like her.
We’re so very sorry but there is nothing we can do
to save Amélie.”*



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WRONG
CORRECT





Our commitment

- We are committed to finding a treatment which will save children from these cruel diseases
- Amélie's life was not in vain and is inspiring people and their work so we can give children a future



Our goal

“To save children from Tay-Sachs and Sandhoff disease. We want to give them an opportunity to have a future.”



Our goal

*“To save children from Tay-Sachs and Sandhoff disease. We want to give them an **opportunity** to have a future.”*



Thank you

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