

How to get research done on your rare disease





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







"You probably won't meet another family with a child affected by Tay-Sachs."



4th European Tay-Sachs & Sandhoff Family Conference Disneyland Paris – 2016

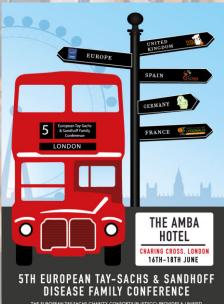






5th European Tay-Sachs & Sandhoff Family Conference

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 Gnother family with a child affected by Tay-Sachs."





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."







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

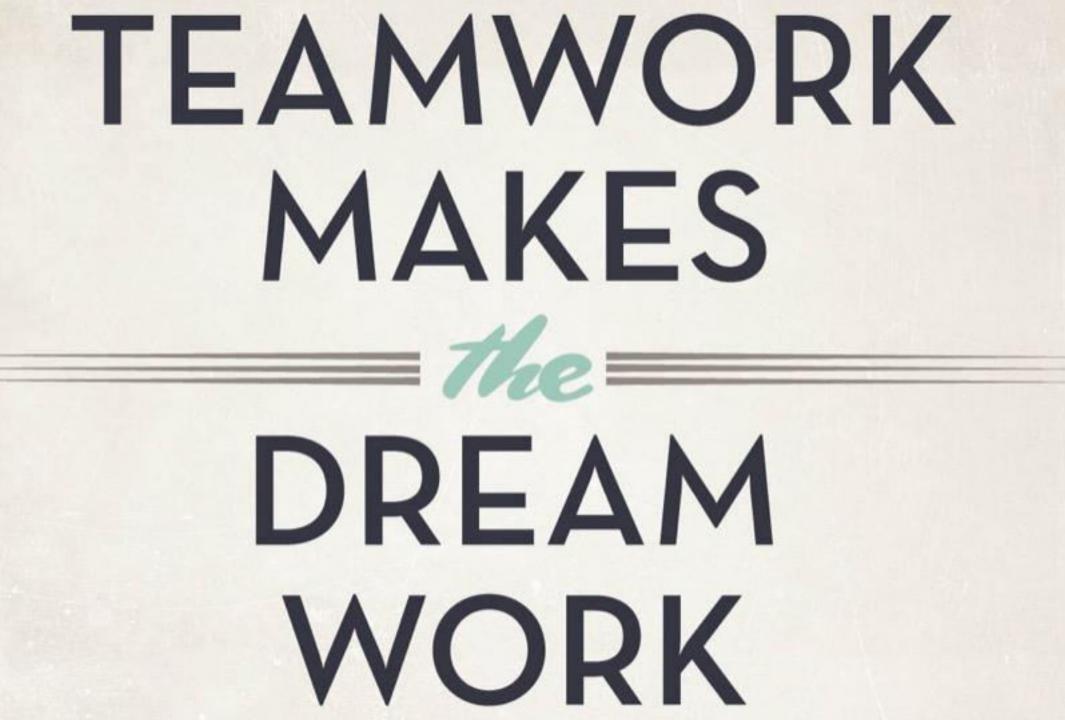
- Team at The University of Cambridge had been working with on a treatment for GM2 for over 20 years
- Needed a charity to support them to secure funding
- Needed a charity to help them identify patients
- Needed a charity to raise awareness of the Tay-Sachs and Sandhoff





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







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







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





"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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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

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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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