EUROPE

UNITED KINGDOM

SPAIN

GERMANY

FRANCE

5

European Tay-Sachs and Sandhoff Family Conference

LONDON

THE AMBA HOTEL
CHARING CROSS, LONDON
16TH-18TH JUNE 2017

5TH EUROPEAN TAY-SACHS & SANDHOFF DISEASE FAMILY CONFERENCE

THE EUROPEAN TAY-SACHS CHARITY CONSORTIUM (ETSCC) PROVIDES A UNIFIED VOICE IN THE FIGHT AGAINST TAY-SACHS AND SANDHOFF DISEASE - ETSCC.ORG
The European Tay-Sachs and Sandhoff Charity Consortium (ETSCC) was created to enable European charities to come together in the fight against Tay-Sachs and Sandhoff. Members are all recognised charities in their country of origin, and there are currently organisations from the United Kingdom, Spain, Germany and France.

The three aims of the ETSCC are to raise awareness of Tay-Sachs and Sandhoff, support the research for a potential treatment and provide a united European voice. Our goal as a collective is to help find a potential treatment for these devastating diseases.

One goal. One group. One voice.
# ETSCC2017

5th European Tay-Sachs & Sandhoff Disease Family Conference

## Table of Contents

<table>
<thead>
<tr>
<th>Page</th>
<th>Section</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>03</td>
<td>Chairman’s message</td>
<td>An introduction to this year’s conference by the Chairman of the ETSCC.</td>
</tr>
<tr>
<td>05</td>
<td>What to expect</td>
<td>The goals and objectives the consortium would like you to take away from the conference.</td>
</tr>
<tr>
<td>09</td>
<td>Day one: afternoon session</td>
<td>Detailed agenda of the afternoon session of the first day of the conference in London.</td>
</tr>
<tr>
<td>13</td>
<td>Day two: afternoon session</td>
<td>Detailed agenda of the afternoon session of the second day of the conference in London.</td>
</tr>
<tr>
<td>20</td>
<td>Our speakers</td>
<td>A list of all of the speakers at this year’s conference, including the institution they represent.</td>
</tr>
<tr>
<td>22</td>
<td>Thank you</td>
<td>A short message from the consortium to everyone who was able to join the charities in London.</td>
</tr>
<tr>
<td>04</td>
<td>Meet the charities</td>
<td>Overview of all of the charities who are members of the European Tay-Sachs and Sandhoff Charity Consortium.</td>
</tr>
<tr>
<td>07</td>
<td>Day one: morning session</td>
<td>Detailed agenda of the morning session of the first day of the conference in London.</td>
</tr>
<tr>
<td>11</td>
<td>Day two: morning session</td>
<td>Detailed agenda of the morning session of the second day of the conference in London.</td>
</tr>
<tr>
<td>14</td>
<td>Session overview</td>
<td>A short review of each of the presentations being given at this year’s conference.</td>
</tr>
<tr>
<td>21</td>
<td>Our sponsors</td>
<td>All of the sponsors of this year’s conference who have all made it possible to hold the event.</td>
</tr>
<tr>
<td>23</td>
<td>2018 conference location</td>
<td>Announcement of the conference location for the 6th European Tay-Sachs and Sandhoff Family Conference.</td>
</tr>
</tbody>
</table>
CHAIRMAN’S MESSAGE

WELCOME TO OUR 5TH EUROPEAN FAMILY CONFERENCE

I am delighted to welcome you all to this year’s conference. For the first time we are holding it in London and at the Amba Hotel.

This year, the first day of the conference will be focusing on respiratory health and we think that this is a vital topic that impacts all our children. There are some fantastic speakers who, we hope, will provide you with many useful tips. The second day will once again focus on the science and research being explored for Tay-Sachs and Sandhoff diseases.

All of the ETSCC members are really excited to share with you the latest news regarding the research into a potential treatment for Tay-Sachs and Sandhoff disease. As a group, we are committed to working hard until there is a treatment which can save our children’s lives.

Finally, I want to say a big thank you to all of our sponsors. Without their support, it would not have been possible to hold this fantastic event.

DANIEL LEWI
Chairman of the ETSCC
MEET THE CHARITIES

THE CURE & ACTION FOR TAY-SACHS (CATS) FOUNDATION
United Kingdom

The Cure & Action for Tay-Sachs (CATS) Foundation was established in 2011 to provide support and information to families affected by Tay-Sachs and Sandhoff and it’s nominated patient organisation for the clinical trial in Cambridge.

ACCIÓN Y CURA PARA TAY-SACHS (ACTAYS)
Spain

Acción y Cura para Tay-Sachs (ACTAYS) was founded in 2014 so families in Spain had access to a support network, access to information about the diseases and actively supports the research in Seville and Cambridge.

HAND IN HAND GEGEN TAY-SACHS UND SANDHOFF
Germany

Hand in Hand gegen Tay-Sachs und Sandhoff in Deutschland was set-up in 2015 to collect and distribute information, support families and the national and international research into the diseases.

VAINCRE LES MALADIES LYSOSOMALES (VML)
France

Vaincre les Maladies Lysosomales (VML) is an association based in France which was established in 1990 and provides support to parents and patients affected by lysosomal diseases.
WHAT TO EXPECT AT OUR CONFERENCE

The European Tay-Sachs and Sandhoff Family Conferences give families throughout Europe the opportunity to come together once a year.

Language
All of the presentations will be given in English. However, each speaker will answer questions after their talk.

Roundtables
Roundtable discussions will take place for fifteen minutes after each presentation according to the tables selected language.

Theme
The theme of the first day of the conference is "Respiratory Health" and all presentations will focus on this topic.

Meals
A buffet lunch will be provided during each day of the conference and there will also be a group dinner after the first day.

Remembrance
The Remembrance Corner can be visited at any time where you can leave a message to those children who have passed away.

Connecting
You will be able to connect with lots of different families and medical professionals during the two days of the conference.
Following on from a successful conference held at Disneyland Paris in 2016, we are delighted to be hosting the 5th European Tay-Sachs and Sandhoff Disease Family Conference in London. Once again we will continue with our "language round table" set-up so that everyone feels comfortable to discuss each presentation in their native language. We believe this will help everyone enjoy the talks and get the most out of them.

As we have such a busy schedule, we will be strict with our timings and running order. In order to achieve this, we will not be having coffee breaks. Instead, this will be made available at the back of the conference room and you are welcome to help yourself at any time. We will also have play facilities which will be supervised by a Portage specialist.

We invite you to join us in honoring our angel children in our Remembrance Corner which is available throughout the Conference weekend. All families are welcome to pay their respects at any time in this calm, private area. Please stop by, take a moment to remember our angels and perhaps write a message to leave on our Memory Tree.
The morning session on day one will introduce the conference theme of chest health and we will be joined by various specialists in this field.

**SESSION 1**

**Dr Brindha Dhandapani**  
Consultant Community Paediatrician  
Lewisham & Greenwich NHS Trust

The paediatrician role in respiratory health for Tay-Sachs and Sandhoff.

**SESSION 2**

**Rebecca Murphy**  
Highly Specialist Speech and Language Therapist  
Evelina Children’s Hospital

The role of the Saliva Control Clinic at the Evelina Children’s Hospital.

**SESSION 3**

**Dr Charlie Fairhurst**  
Consultant Paediatrician in Neurodisability  
Evelina Children’s Hospital

The impact Botulinum Toxin A (botox) has on managing secretions.

**INTRODUCTION**

Daniel Lewi  
Chairman & Chief Executive  
ETSCC & The CATS Foundation

Introducing the 5th European Family Conference.
REGISTRATION OPEN

09:30

The registration desk for the 5th European Tay-Sachs and Sandhoff Disease Family Conference will open at 9:30 and will be situated outside the Ballroom. Upon arrival you will be given a conference pack and name badge which must be worn at all times during the conference. Each speaker has been given a thirty minute time slot for their presentation and an extra fifteen minutes to circulate the room to answer any questions attendees may have about their talk.
DAY ONE:  
AFTERNOON SESSION

The afternoon session on day one will continue focusing on our theme and there will be presentations on how to provide practical care at home.
Lunch will be served in the Regency Room at 12:30 where everyone will be able to enjoy a three course buffet meal. The lunch break will run until 14:00 and we ask that all attendees are able to make their way back to the Ballroom so we can begin the afternoon session. There will be a short summary of the first day’s speakers and everyone will be able to spend the rest of the day relaxing before the group dinner which will be served in the Regency room from 19:30 until 22:00.

**SESSION 4**
Rebecca Lydon  
Special Needs Paediatric Nurse  
Lewisham & Greenwich NHS Trust  
Chest health management in the home setting.

**SESSION 6**
Nicki Backus  
Family Support Officer  
The CATS Foundation  
Family led discussions about respiratory health.

**SESSION 5**
Case studies  
Parents and carers  
England, Spain and Germany  
Three parents will share their experiences of being a carer.

**Summay**
Melanie Mende  
Director  
Hand in Hand Against Tay-Sachs and Sandhoff  
Overview of the presentations on the first day.
DAY TWO: MORNING SESSION

The second day of the conference will focus on the science and research being undertaken in the area of Tay-Sachs and Sandhoff disease.

10:00
INTRODUCTION
Daniel Lewi
Chairman & Chief Executive
ETSCC & The CATS Foundation
Introducing the science and research presentations.

10:15
SESSION 7
Dr Cordero & Dr Cachón-Gonzalez
Research Associate & Senior Research Associate
The University of Sevilla & University of Cambridge
Update on Inflammasone Complex treatment for GM2.

11:00
SESSION 8
Dr Eva Zaccariotto
Research Associate
The University of Cambridge
Update on the Gene Therapy Trial for GM2 at the University of Cambridge.

11:45
SESSION 9
Professor Tim Cox
Director of Research & Hon Consultant Physician
The University of Cambridge
The pre-trial process and participation in a gene therapy trial.
The Ballroom will be open from 9:30 and the presentations for the second day will begin at 10:00. We ask that attendees who are joining us for the second day only to arrive early so that they can register and collect their conference pack. Lunch will once again be served in the Regency Room and during this break there will be the opportunity to visit the charity stands which can be found in the side room of the Ballroom. During this time attendees can also take part in our Instamemory project.
DAY TWO: AFTERNOON SESSION

The afternoon of the second day of the conference will include talks highlighting the importance of charities in clinical trials.
The 5th European Tay-Sachs and Sandhoff Disease Family Conference will close at 16:30. However, there will still be an opportunity for attendees to network with one another and coffee will be made available during this time. So we are able to improve on our conferences, you will be asked to complete a short survey about your experiences over the two days. This will help the member charities of the ETSCC when we begin planning the 6th Family Conference.
SESSION OVERVIEW

SESSION 1

Dr Brindha Dhandapani
Dr Dhandapani will discuss the role of the Community Paediatrician in managing someone’s chest health. As a Consultant, the focus of the talk will be how various symptoms are identified which can have a negative impact on the respiratory system and the steps which can be taken to both improve and reduce the risk of developing a life threatening infection. These are all vital in improving someone’s quality of life.

SESSION 2

Rebecca Murphy
Rebecca will give an overview of the Saliva Control Clinic at the Evelina Children’s Hospital in London. This unique clinic provides specialist assessment, advice and treatment on issues surrounding a child’s saliva control. This may involve interventions such as injections (Botulinum Toxin A), changes to a child’s medication, oral-motor therapy (exercises designed to help children learn a more mature swallowing pattern to avoid drooling) or surgery.

SESSION 3

Dr Charlie Fairhurst
Dr Fairhurst will discuss how his role as a Paediatric Neurodisability Consultant can have a huge impact on managing the symptoms that a child with Tay-Sachs and Sandhoff disease suffer from. The talk will go into greater detail surrounding using Botulinum Toxin A to manage secretions and how identifying and treating one symptom can lead to an improvement in another.
SESSION 4

Rebecca Lydon
Rebecca will discuss how care provided by a local community team can have a dramatic impact on managing respiratory health. In the UK, these teams are the first point of contact for parents and carers.

SESSION 5

Case studies
Every family has a different experience of looking after someone’s respiratory health. This session will feature three parents and carers who will discuss the challenges they encountered and how they overcame them.
SESSION 6

Nikki Backus

The family led discussions will be facilitated by Nikki to ensure that all attendees at the conference are able to talk freely about respiratory health. After the success of these discussions last year, everyone will be encouraged to take an active part so that tips on managing respiratory health or any concerns can be addressed amongst specialists and peers.

SESSION 7

Dr Mario Cordero & Dr Begoña Cachón-Gonzalez

Dr Cordero and Dr Cachón-Gonzalez will provide an update on his work using Inflammasome Complex Treatment for Tay-Sachs. In this project, the team evaluates the role of autophagy as a cleaning mechanism and NLRP3-inflammasome as a mechanism involved in neuro-inflammation and cell death.

SESSION 8

Dr Eva Zaccariotto

Eva will give an update on the Gene Therapy Trial for GM2 at the University of Cambridge. This talk will provide a brief history of the project which began over fifteen years ago and will then focus on the huge developments we have seen in the last twelve months as the team moves towards the clinical trial starting in the very near future.

SESSION 9

Professor Tim Cox

Professor Cox will follow on from Eva’s presentation and will introduce the pre-clinical work that the team at the University of Cambridge are working on so that the clinical trial can start as soon as possible. The talk will also focus on selection and how recruitment will take place for this people who will be able to take part.
SESSION 10

Dr Heather Gray-Edwards

Dr. Heather Gray-Edwards is an Assistant Research Professor at Auburn University, AL USA. She studies Tay-Sachs disease in sheep and is currently testing AAV mediated gene therapy for Tay-Sachs disease. Dr. Gray-Edwards will be presenting her research on the efficacy of gene therapy in Tay Sachs sheep and the lessons learned that can be applied to human patients. She will provide an update on the timeline for human clinical trials for Tay-Sachs and Sandhoff disease in the USA.

SESSION 11

Toni Mathieson

Toni will give a talk which focuses on not only on her personal journey with Niemann-Pick, but also her role as the Chief Executive of Niemann-Pick UK. Toni will discuss how she became involved with the charity in 2003 after her daughter Lucy was diagnosed with Niemann-Pick Type C and how she now leads the charity which is developing treatments for the devastating disease.

SESSION 12

Beatriz Dominguez & Patricia Durão

Patricia and Beatriz will give talks about the role of families and charities and how they can inspire others. The Tay-Sachs and Sandhoff community is small and relies on a unique peer-to-peer support model to help those who need advice or support with managing a family member affected. The talk will feature families who have made significant contributions to the community.
OUR SPEAKERS

Dr Brindha Dhandapani  
Consultant Community Paediatrician  
Lewisham & Greenwich NHS Trust

Rebecca Murphy  
Specialist Speech and Language Therapist  
Evelina Children’s Hospital

Dr Chalie Fairhust  
Consultant Paediatrician in Neurodisability  
Evelina Children’s Hospital

Dr Begoña Cachón-Gonzalez  
Senior Research Associate  
The University of Cambridge

Professor Timothy Cox  
Director of Research & Consultant Physician  
The University of Cambridge

Rebecca Lydon  
Special Needs Paediatric Nurse  
Lewisham & Greenwich NHS Trust

Dr Mario Cordero  
Research Associate  
The University of Seville

Dr Eva Zaccariotto  
Research Associate  
The University of Cambridge

Toni Mathieson  
Chief Executive  
Neimann-Pick UK

Dr Heather Gray-Edwards  
Assistant Research Professor  
Auburn University, AL USA
OUR SPONSORS

SANOFI GENZYME

Sanofi Genzyme is the specialty care global business unit of Sanofi, focused on rare diseases, multiple sclerosis, immunology, and oncology.

mtn-i

mtn-i provides a global MTN news, data, analytics and communication platform for the financial sector and covers the Americas, Europe, Asia, including Canada and Japan.

NUTRICIA

Nutricia specialises in the delivery of advanced medical nutrition for the very young, the old and the sick and is the largest specialist nutrition company in Europe.

SPECIALISED ORTHOTIC SERVICES

Specialised Orthotic Services was the first dedicated special seating company in the UK and they manufacture a comprehensive range of seating solutions.

CHPK

CHPK is a leading professional practice providing property and construction consultancy services for a wide range of national and regional clients.

SBK

The Siemens-Betriebskrankenkasse (Siemens company health insurance fund - SBK) is one of the largest company health insurance funds in Germany.
THANK YOU FOR JOINING US

We hope you have enjoyed attending the 5th European Tay-Sachs and Sandhoff Disease Family Conference in London.

THE GOAL OF THE ETSCC IS TO BRING TOGETHER ALL OUR EUROPEAN FAMILIES

Receiving a diagnosis of Tay-Sachs or Sandhoff disease can be very isolating. Five years ago the ETSCC was born which not only brought together all the European based charities supporting families affected by Tay-Sachs and Sandhoff, but it also gave us the opportunity to bring all our families together once a year. Over the last five years the conference has grown from very humble beginnings. In 2013 there were only four families and in 2017 we are delighted that over twenty families have been able to join us, meet other families and hear the latest research into the diseases. We want to say thank you to everyone who is with us in London. By attending the conference you have become part of our very special family and we hope you have enjoyed it.
THE LOCATION FOR 2018

We are delighted to announce that the 6th European Tay-Sachs and Sandhoff Disease Family Conference will be held in Madrid, Spain. This vibrant city will enable us to enjoy the conference in warmer climates. ACTAYS will be leading the organising committee and we all look forward to welcoming you to Madrid.