

english

7th EUROPEAN

# TAY-SACHS AND SANDHOFF FAMILY CONFERENCE

12th - 14th July 2019 Maritimhotel Würzburg



# Welcome

Welcome to our 7th European Family Conference in Würzburg. After 2015 in Günzburg our Charity Hand in Hand gegen Tay-Sachs und Sandhoff in Deutschland e.V. is happy to invite the European families to Germany for the second time. And we are glad that more than 30 families will be with us. We are hoping for some nice hours in the circle of other affected families, for an active exchange and hopefully many good news from the researchers.

Because 2019 is a very special year. It is the first time that a clinical trial will start in Europe, that includes Tay-Sachs and Sandhoff disease. We will have three experts that will tell us everything we need to know about it. And there is a good chance that with a new substrate reduction therapy there will be a second study including Tay-Sachs and Sandhoff disease soon. And last but not least we are of course hoping for good news regarding the progression of the gene therapy. We are very pleased, that we can welcome families from 11 countries. And we are a bit proud, that 17 of the attending families are part of the Hand-in-Hand-family. When we first started, back in 2015, we were the only family from Germany with this diagnosis that we knew of. At the first European Family Conference we were three families. And now the group has grown big. We are meeting regularly, help each other, laugh together, cry together. To many we have become a second family.

And now we are looking forward meeting our European extended family.

As the chairman of „Hand in Hand“ I'd like to say a special thank you to my wife Birgit Hardt



*Birgit Hardt and Melanie Mende with Ben.*



*Folker Quack and Dario.*

and to Melanie Mende. They are working hard on the organization of this conference for several weeks now. Inviting the speakers, organizing their travel, booking hotel rooms, looking for sponsors, planning the program for the conference and the kids etc.

We are looking forward seeing all of you.

***Folker Quack***

*(Chairman of Hand in Hand, Germany)*

# Greetings

We are very excited to welcome you all to the 7th European Tay-Sachs and Sandhoff Family Conference which is being held in the shadows of the Marienberg Fortress here in Wuerzburg.

For our 2019 conference, we have some very exciting speakers attending who will be giving updates on the many research projects which are investigating potential treatments for Tay-Sachs and Sandhoff disease. Since we were all last together in 2018 there has been some incredible developments in this research and we are very hopeful that everyone will learn how all our families have played an important part in moving these projects forward. Without them, we would not be hearing such great news this year.

When the European Tay-Sachs Charity Consortium (ETSCC) was established in 2012 we could only dream that one day there may be some hope that a treatment was on the horizon. We are very excited to say that there is most definitely light at the end of the tunnel and we are extremely hopeful that the breakthroughs in the research which you will hear this weekend will excite you as well.

It would be remiss to not acknowledge that these treatments have come too late for some of us. There will be families in attendance this weekend, like there always is, who have lost their children to Tay-Sachs and Sandhoff. They will be excited by the news we will hear this weekend, but it will also be tinged with the sadness that it has come too late for their loved one. However, I am sure they, like my family who has lost a child to Tay-Sachs, will be pleased to know that we are moving forward in the fight against Tay-Sachs and Sandhoff.



*Dan Lewi*



*The finish of the 6th family conference in Madrid.*

Finally, we would like to say a big thank you to Hand in Hand gegen Tay-Sachs und Sandhoff who have organised this year's conference. These events take a lot of planning and it is no small task to bring together families for one weekend from over 10 different countries! With the support of the other ETSCC charity members, this year will once again build on the work we have all achieved in bringing families together and creating our special community.

We hope you enjoy yourself!

***Dan Lewi***

*Chairman of the ETSCC*



## The city of Würzburg

Würzburg is the northernmost city in the German state Bavaria. It is known for its buildings in baroque and Rococo-style, especially for the Würzburger Residenz from the 18. Century, with magnificent halls, a large fresco from the Venetian artist Tiepolo and a fanciful stairwell. After the dome of Aachen, the Würzburger Residenz was the second German building that was added to the World Heritage List.

With its numerous wineries and wine estates as well as its idyllic location surrounded by famous vineyards, Würzburg is the heart of the wine region Franconia, that is often called the Tuscany of Germany. Even the German poet Johann Wolfgang Goethe praised the Franconian wine from the Würzburger location „Am Stein“. Until today this famous „Stein-Wein“ is planted. High above the city enthroned the fortress Marienberg. It was already built in the year 1200 as an unusually big castle, that was made even bigger in the late middle ages and Renaissance. While we will have dinner on Saturday in the panorama room of the Maritim-Hotel we will have a great view on the wine location „Am Stein“, the fortress Marienberg and the pilgrimage church „Käppele“.

Würzburg owns the oldest University of Bavaria. Wilhelm Conrad Röntgen discovered the radiance that could permeate matter here in his laboratory on 8th. November 1895 and that he would later call x-rays. For this discovery he earned the first Nobel Prize in physics that had ever been handed over. This certificate and his laboratory can be visited and are located in immediate neighborhood to the Maritim-Hotel.

A very famous contemporary son of this town is Dirk Nowitzki, that started his great career in the USA as a basketball player in the NBA, and that is still visiting his hometown every year in the summer holidays.



Rare, but not alone!



**ETSCC**  
EUROPEAN  
TAY-SACHS &  
SANDHOFF  
CHARITY  
CONSORTIUM

# Friday, 12th. July

On Friday you will have the chance to get a closer look at the city and get in touch with the other families in a relaxed atmosphere. For example while doing a city stroll, having a glass of wine on the historical bridge of Main (“Brückenshoppen”) or visiting an original bavarian fair on the Talavera. All important attractions are close enough to the hotel to walk.

**Meeting point for joint activities: 13:00 in the Lobby of the Maritim-Hotel**





**Guided city strolls in different languages**

**Friday, 12th July**

Meeting point for joint activities: 13:00 in the Lobby of the Maritim-Hotel



**A glass of wine on a historical bridge**



**Original bavarian fair**



**At 18:00 our 7th European Family Conference will start with a Welcome-Dinner in the banquet hall of the Maritim-Hotel.**

# Agenda Saturday 13th July

09:30	10:00	Registration	(Room Barbarossa)	
10:00	10:20	Welcome - Introduction	Folker Quack/ Birgit Hardt / Melanie Mende	
10:20	10:30	Presentation Charity	Hand in Hand /Germany	
10:30	11:15	Genotype/Phenotype Correlation		Prof. Konrad Sandhoff
11:15	11:40	Coffee break	(Lobby)	
11:40	11:50	Presentation charity	CATS / GB	
11:50	12:15	Psychosocial determinants in the medical care of patients with rare diseases with a particular focus on lysosomal storage disorders		Dr. Tatiana Bremova
12:15	13:00	Epilepsy in patients with LSD – treatment options		Prof. Andreas Hahn
13:00	14:30	Lunch	(Restaurant)	
14:30	14:40	Presentation charity	ACTAYS / Spain	
14:40	15:25	GM-2-Research project at Seville University		Dr. Mario Cordero
15:25	15:35	Presentation Charity	VML / France	
15:35	16:00	Coffee break		
16:00	16:50	Acetyl-DL/L-leucine for the treatment of lysosomal storage diseases: evidence, mechanisms and clinical trials		Prof. Michael Strupp Prof. Frances Platt Taylor Fields
16:50	17:00	Presentation charity	DOCE / Portugal	
17:00	17:15	Group picture		Birgit Walther-Luers
17:15	17:45	Remembrance ceremony		
18:00	19:00	Break or Aperitif		
19:00	24:00	Dinner		



**Prof. Konrad Sandhoff** discovered Sandhoff disease in 1968 while he conducted research about Sphingolipidosis. Since then the chemist is seeking for causes, correlations and treatments for Tay-Sachs, Sandhoff and allied diseases. Which influences, factors and interactions could be important for Tay-Sachs and Sandhoff disease will be part of his talk.

**Dr. Tatjana Bremova** was involved in the trials with Tanganil in patients with Tay-Sachs and Sandhoff disease. Now she is presenting a study, where she is investigating psycho-social requirements to support affected families better.



**Prof. Andreas Hahn** is treating children with epilepsy, his main emphasis are lysosomal storage diseases. He will present the special challenges and successful therapies.



He will present the special challenges and successful therapies.

## Speakers on Saturday

**Prof. Michael Strupp** is professor for Neurology at the University of Munich. **Prof. Frances Platt** is professor of biochemistry and pharmacology in Oxford. **Taylor Fields** is Senior Vice-President of the pharma company Intrabio. Together they will present the clinical trial, where the active substance Acetyl-DL/L-Leucine in patients with Tay-Sachs and Sandhoff disease is going to be researched furthermore.



**Dr. Mario Codero** is a nutritionist and molecular biologist for rare diseases at the University of Seville in Spain. He is exploring the inflammatory process in Tay-Sachs and Sandhoff disease and wants to find new treatments through his research.

# Agenda Sunday 14 th July

09:30 Registration (Room Barbarossa)

09:45 10:30 Substrate Reduction Therapy Prof. Frances Platt

10:30 11:15 Study Designs for Rare Diseases Prof. Michael Beck

11:15 11:45 Coffee break

11:45 12:30 Two Molecular Therapies Prof. Timothy Cox

12:30 14:00 Lunch

14:00 14:45 Introduction to Axovant Gene Therapies and  
AXO-AAV-GM2, a novel gene therapy for  
GM2 Gangliosidosis Dr. Gavin R. Corcoran

14:45 15:30 Distinctive features regarding phenotypes  
and response to treatment Dr. Eugen Mengel

15:30 15:45 Coffee break

15:45 16:30 Summary / Round for questions Moderation Prof. Michael Beck  
All speakers

16:30 16:45 Update on the patient registry for Tay-Sachs & Sandhoff Daniel Lewi

16:45 17:00 Close of the conference Folker Quack & Daniel Lewi

The conference language is english. We have simultan translation english-german and german-english. **Please use the haedphones.** A lot of thanks to our translators: Lucy Thomson, Christiane Spiel, Jannika Morisse and Constanca Schumm.

**Professor Timothy Cox** is professor at the University of Cambridge. His research is about the biochemical genetic of congenital metabolic disorders with focus on lysosomal storage diseases. He is searching for new active substances to treat Tay-Sachs and Sandhoff disease and is currently working on a gene therapy.



**Dr. Gavin R. Corcoran** is Chief R&D Officer at the Axovant Sciences Ltd. His firm wants to bring a gene-therapy for patients with Tay -Sachs and Sandhoff disease to a clinical trial. The aim is to restore the function of the  $\beta$ -Hexosaminidase A (Hex A) by bringing a functional copy of the HEXA- andHEXB-Gene with the help of two AAVrh8-Vectors into the brain.

## Speakers on Sunday

**Prof. Frances Platt** is dealing with lysosomal storage diseases and their possible therapies for several years now. At the moment the zoologist, bio-chemist and pharmacologist is researching into substrate reduction therapy for Tay-Sachs and Sandhoff disease.

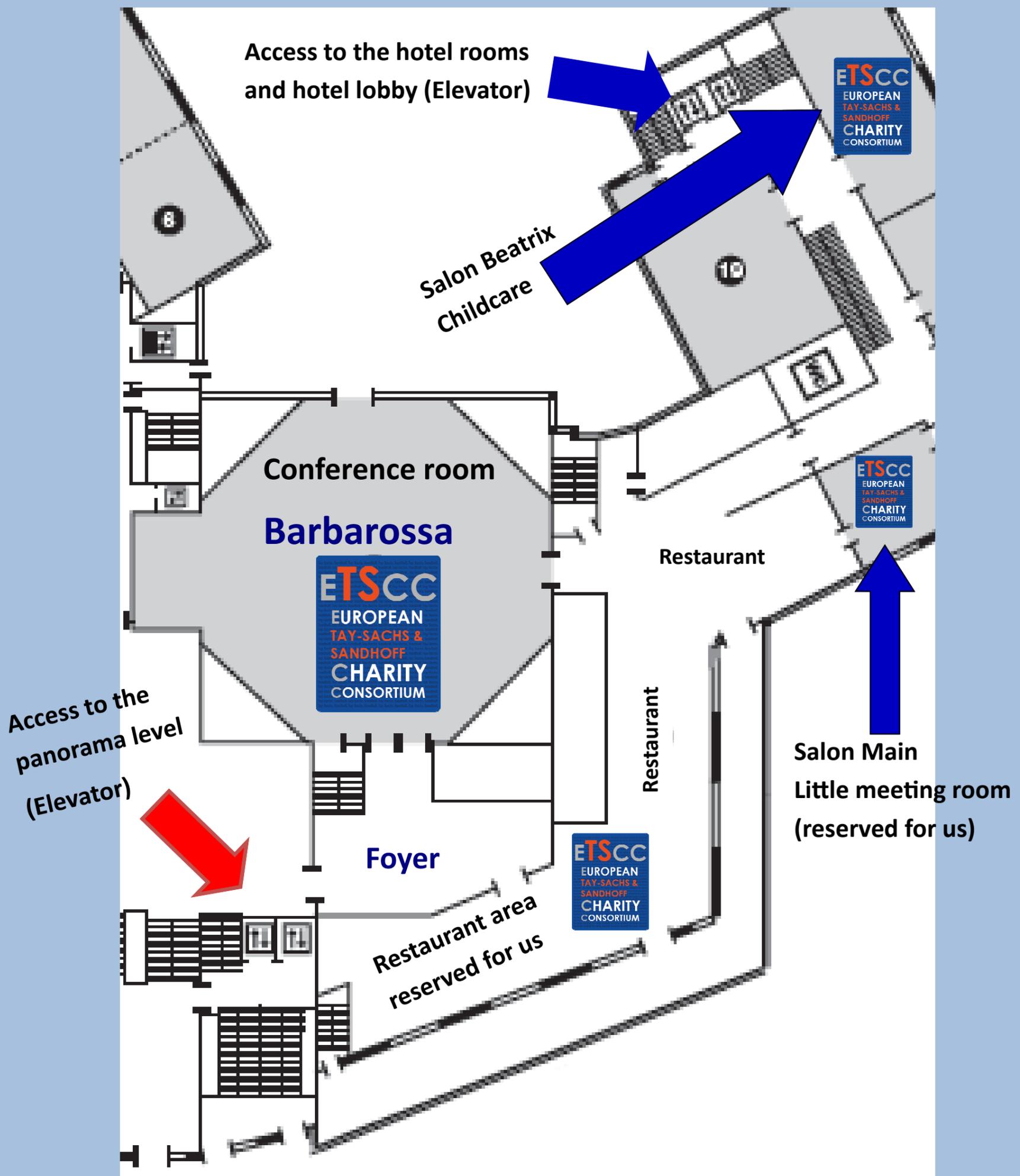


**Dr. Eugen Mengel** is clinician and investigator for lysosomal storage disease with focus on sphingolipidoses like Tay -Sachs and Sandhoff disease. He will light up the different progressive forms of the disease.

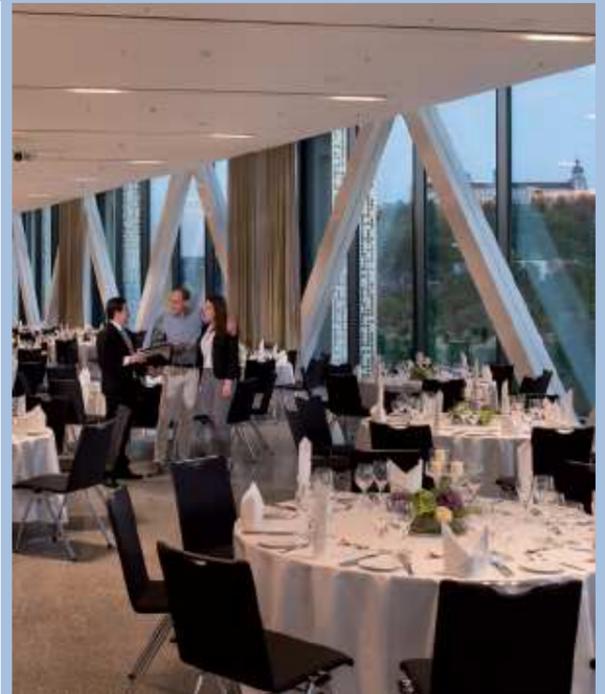
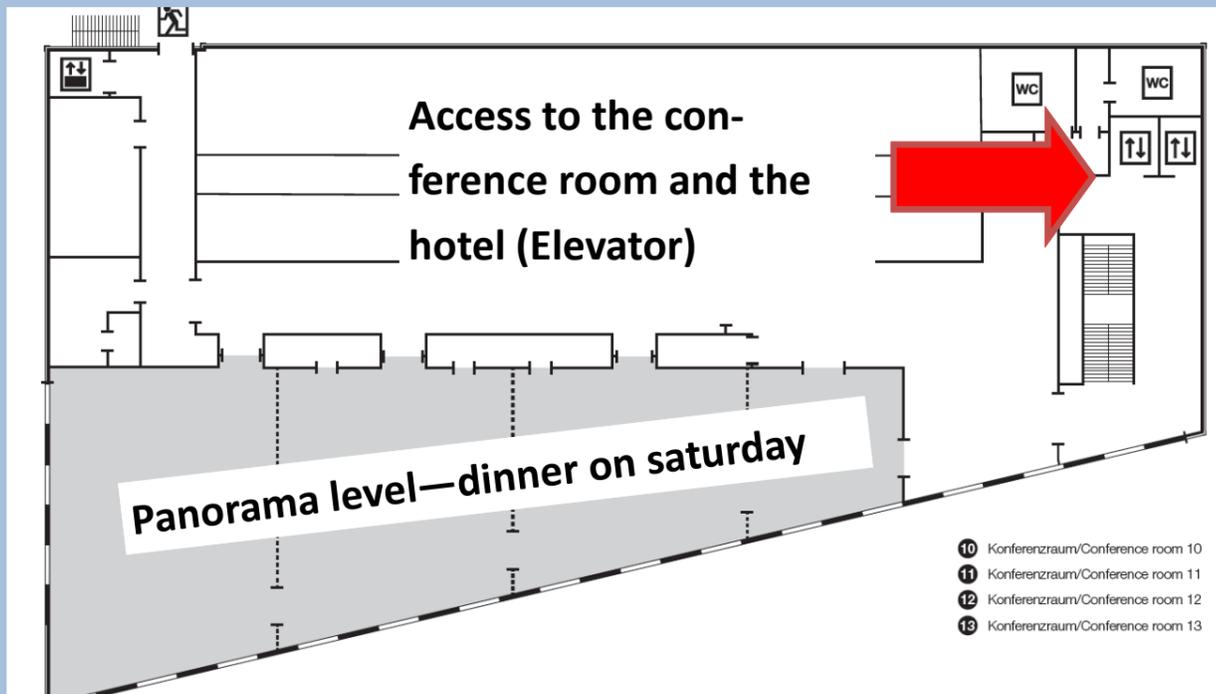


**Professor Michael Beck** dedicated his whole research life to the lysosomal storage diseases. He builded the Villa Metabolica in Mainz up to a worldwide recognized center for the research and treatment of these diseases. He will explain why the development of drugs for rare diseases take so long.

# Our conference rooms



We will meet in the room “Barbarossa” on Saturday and Sunday. Entry through the restaurant. For breakfast and the lunch on Saturday we have reserved some tables in the back area of the restaurant. All buffets, coffee breaks etc will be built up in the foyer.



On Saturday evening we invite all families and speakers to a dinner at the panorama level of the Congress Center. We will have a wonderful look at the city, fortress, the “Käppele”, the vineyard and the Killiani-Fair there. You can reach the panorama level easily with the elevators behind the foyer of the room “Barbarossa” (red arrow).

**During dinner on Saturday, you can make selfies with „Jimmy“ . Please use it— in memory of the conference and for our guestbook.**

## Hotel Maritim

**The Maritim Hotel** is part of the famous Maritim hotel chain that is leading especially conference hotels of high quality. The hotel lies on the edge of the downtown area, picturesque next to the river, with a direct view at one of the most important monuments . It is integrated into the Congress Center with numerous conference rooms and a big concert hall.

We will meet in the related conference area of the hotel. The hotel

has a wellness area with a sauna, saunarium, fitness room and a pool with 28 degrees. You will also find a hotel bar and a french wine bar. There is free wi-fi in the whole hotel.





## Childcare:

On Saturday and Sunday (from 10:00 to 17:00) we offer a childcare with a program close to the conference room (Salon Beatrix). Social workers and child nurses will take care of our children. They will do paper drafting, paint, sing and a magician will care for some magical moments. For the insurance law the supervisory duty will officially still be with the parents.

## General information

### Photo memories:

The whole conference will be accompanied by the photographer Birgit Walter-Lüers. You can make photo memories, if you want to. We ask all attendees to meet on Saturday at 17:00 in the foyer of our conference room “Barbarossa” for making a group photo of all families and speakers. We also will document the conference with pictures on our website. Due to the private policy we ask everyone that does not want pictures of them to be published to get in touch with us (Birgit Hardt, Folker Quack) or to talk to the photographer Birgit Walter-Lüers directly. We will also hand out a consent form with the participant list. Who will be part of the group photo automatically gives permission for the publication of this picture.



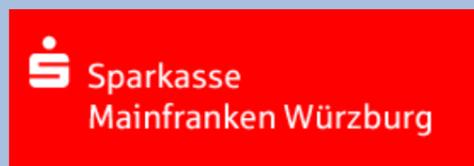


Together we are strong. Four adults affected with Tay-Sachs.

## Sponsoring

Such a large conference would not be financially viable without the support of sponsors. For years, the „Siemens Health Insurance“ has generously supported our family conferences as part of the German self-help promotion. Added to this are the foundations Kindness for Kids, the Robert Vogel Foundation and the Sparkasse Mainfranken Foundation. The „Würzburger Dolmetscherschule“ has made a school project for our conference and provides us with four interpreters for simultaneous translation. The interpreting technique is completely sponsored by the company „PCS-Konferenztechnik“. Each charity of the ETSCC supported the conference.

We thank everyone for their generous support!



Würzburger Dolmetscherschule





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



in Seville and Cambridge. ACTAYS was established by Beatriz Fernandez Dominguez. ACTAYS has extended their help to other families in Latin America providing information and counselling.

## Charities in Europe



**The CATS Foundation** was established in June 2011 by Daniel and Patricia Lewi after their daughter Amelie was diagnosed with Tay-Sachs at 15 months of age in March 2011. At the time there was no UK based charity dedicated to providing support for families affected by Tay-Sachs or Sandhoff disease and the charity's main focus is supporting families whilst also raising awareness of the diseases.



**DOCE:** On the International Rare Disease Day, February 28, 2019, a group of parents and relatives of patients with Tay-Sachs and GM1



gangliosidosis in Portugal decided to band together to make a difference! The non-profit organization supports and informs all concerned, giving everyone access to information about studies and clinical trials worldwide.



The representatives of the charities come together in the European Tay-Sachs and Sandhoff Charity Consortium (ETSCC).

# Charities in europe



**Hand in Hand gegen Tay-Sachs und Sandhoff in Deutschland e.V.** was founded in 2015 by Birgit Hardt and Folker Quack. Their son Dario suffers from juvenile Sandhoff. At the time of the diagnosis, they were told they will never meet another family, because the disease is so rare. But they didn't accept this. Today, „Hand in Hand“ represents 30 affected families



in Germany and German-speaking countries. The association organizes help and meetings for the families and collects donations for the research of Tay-Sachs and Sandhoff.



**Vaincre les Maladies Lysosomales (VML)** is a French non-profit organisation recognized as being of public utility since 2006. It has been created in 1990 by parents of children with lysosomal diseases and adults suffering a lysosomal diseases. VML gathers 52 lysosomal disorders. In 2018, VML



is gathering more than 1.000 members in France and is active in Switzerland and Belgium. Benoit Clermont represents Tay-Sachs and Sandhoff disease within the VML since 2015.

## 2013: Mondsee (Austria)



# 2013 to 2018—The Family Conferences

## 2014: Paris (France)



## 2015: Günzburg (Germany)



7th Tay-Sachs and Sandhoff Family-Conference



Hand in Hand Germany



ETSCC 2019

## 2016: Paris (France)



## 2013 to 2018 — The family conferences

### 2017: London (Great Britain)



### 2018: Madrid (Spain)



7th Tay-Sachs and Sandhoff Family-Conference



Hand in Hand Germany

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*“Life is what happens to you while you're busy making other plans”*

**John Lennon**

[www.etscc.org](http://www.etscc.org)