



Cerebrotendinous Xanthomatosis

# INTERNATIONAL SCIENTIFIC MEETING

August 27 - 28, 2023 | Jerusalem, Israel



# Thank You to Our International Scientific Meeting Organizing Committee



Dr. Andrea DeBarber  
Oregon Health and Science University  
USA  
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Prof. Tzipora C. Falik-Zaccai  
Galilee Medical Center, Nahariya & The Azrieli  
Faculty of Medicine, Bar-Ilan University, Safed  
Israel  
.....



Dr. Antonio Federico  
University of Siena  
Italy  
.....



Dr. Hidde Huidekoper  
Erasmus MC University Medical Center  
Netherlands  
.....



Dr. Robert D. Steiner  
University of Wisconsin, Prevention Genetics  
USA  
.....

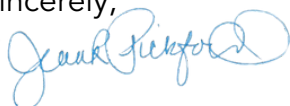
Hello everyone!

Welcome to Jerusalem and the 2023 CTX International Scientific Meeting! On behalf of the CTX Alliance and the organizing committee, it is our pleasure and privilege to host this first-ever scientific meeting exclusively dedicated to cerebrotendinous xanthomatosis (CTX).

Our goal is to catalyze much-needed international basic research and clinical collaborations for CTX and to advance our understanding for improved outcomes for CTX patients and their families.

Never has there been an international gathering of this many CTX experts at one meeting. Thank you for your attendance and commitment to CTX patients and their families around the world!

Sincerely,



Jean R. Pickford

Executive Director

[jpickford@ctxalliance.org](mailto:jpickford@ctxalliance.org) | [www.ctxalliance.org](http://www.ctxalliance.org)

# SUNDAY PM

## AUGUST 27, 2023



### Meeting Sessions Location: Queen of Sheba Hall, Ramada Hotel

Session 1: Welcome & Overview		Speakers:
1:00 - 1:05 pm	Welcome & Opening Remarks	Meeting Organizers
1:05 - 1:45 pm	Cerebrotendinous Xanthomatosis: Its History and New Perspectives	Dr. Antonio Federico
Session 2: Molecular Aspects of CTX		Speakers:
1:45 - 2:15 pm	Overview of CTX Genetics and Genotype-Phenotype Correlations	Dr. Penelope Bonnen (Remote)
Session 3: Biochemical Aspects of CTX		Speakers:
2:15 - 2:40 pm	Overview of CTX Biochemistry with Focus on Clinical Laboratory	Prof. Peter Clayton
2:40 - 3:05 pm	CTX Cases Identified with Atypical Biochemistry	Dr. Andrea DeBarber
3:05 - 3:15 pm	Questions for Session	Prof. Tzipora Falik-Zaccai
3:15 - 3:35 pm	Break	
Session 4: Clinical Experience & Disease Spectrum for International CTX Patient Cohorts		Speakers:
3:35 - 3:50 pm	CTX in Italy	Dr. Antonio Federico
3:50 - 4:05 pm	CTX in Israel	Dr. Hofit Cohen
4:05 - 4:20 pm	CTX in Netherlands	Dr. Aad Verrips
4:20 - 4:35 pm	CTX in Brazil	Dr. Jonas Saute
4:35 - 4:50 pm	CTX in Japan	Dr. Shingo Koyama
4:50 - 5:05 pm	CTX in France	Dr. Fanny Mochel
5:05 - 5:25 pm	Questions for Session	Dr. Hidde Huidekoper
Awards & Social Activities		
5:25 - 5:40 pm	Sunday Awards Presentation	Prof. Tzipora Falik-Zaccai & Dr. Robert Steiner
5:40 - 6:30 pm	Poster Presentation and Meeting Reception	All attendees

Notes:

**MONDAY**  
**AUGUST 28, 2023**



**Meeting Sessions Location: Queen of Sheba Hall, Ramada Hotel**

Session 1: Biochemical Contributions to Pathology of CTX		Speakers:
8:00 - 8:25 am	Cholesterol accumulation in the brain and eye in CTX	Dr. Irina Pikuleva
8:25 - 8:50 am	Role of bile acid pathway intermediates and bile alcohols in pathology of CTX	Dr. William Griffiths
8:50 - 9:00 am	Questions for Session	Dr. Andrea DeBarber
Session 2: Neuroimaging in CTX		Speakers:
9:00 - 9:25 am	Overview of Neuroimaging in CTX	Dr. Rosa Cortese
9:25 - 9:50 am	Novel Tools for Neuroimaging in CTX	Dr. Fanny Mochel
9:50 - 10:00 am	Questions for Session	Dr. Antonio Federico
10:30 - 10:20 am	Break	
Session 3: Epidemiology/Prevalence of CTX		Speakers:
10:20 - 10:45 am	Epidemiology/Prevalence of CTX	Dr. Tiziano Pramparo
Session 4: Treatments for CTX - Present and Future		Speakers:
10:45 - 11:05 am	Chenodeoxycholic acid treatment in patients with CTX	Dr. Antonio Federico
11:05 - 11:20 am	Long-term treatment effect in CTX depends on age at treatment - Netherlands CTX Cohort	Dr. Aad Verrips
11:20 - 11:35 am	Can we improve outcomes? Lessons taken from the Israeli CTX Cohort	Dr. Sharon Hassin-Baer
11:35 - 11:55 am	Treatment during Pregnancy in CTX	Prof. Tzipora Falik-Zaccai
11:55 - 12:15pm	Gene supplementation of CYP27A1 in the liver restores bile acid metabolism in a mouse model of CTX	Dr. Ruben Hernandez-Alcoceba
12:15-12:35 pm	Cholic acid treatment in neonates and children with CTX	Dr. Ken Setchell
12:35- 12:55pm	Cholic acid treatment for adults with CTX	Dr. Yann Nadjar (remote)
12:55 - 1:15pm	Questions for Session	Dr. Robert Steiner
1:15 - 1:45 pm	Lunch	

Notes:

Session 5: Early Diagnosis of CTX - Presentation in Infants and Children		Speakers:
1:45 - 2:10 pm	CTX Pediatric presentation	Dr. Tanyel Zubarioglu
2:10 - 2:35 pm	CTX Presenting as Juvenile Cataracts	Dr. Zvi Segal
2:35 - 3:00 pm	CTX Identification in Infants	Dr. Ken Setchell Dr. Hidde Huidekoper Prof. Tzipora Falik-Zaccai
3:00 - 3:10 pm	Questions for Session	Dr. Robert Steiner
Session 6: Newborn Screening for CTX		Speakers:
3:10 - 3:35 pm	Progress toward newborn screening for CTX	Dr. Michael Gelb
3:35 - 4:00 pm	Outcomes for CTX Affected Newborns Treated from Birth	Prof. Tzipora Falik-Zaccai
4:00 - 4:10 pm	Questions for Session	Dr. Hidde Huidekoper
4:10 - 4:30 pm	Break	
Session 7: CTX Patient, Family and Patient Advocacy Presentations		Speakers:
4:30 - 4:40 pm	AEXCT - Asociacion Española Xantomatosis Cerebrotendinosa	Dr. Ruben Hernandez-Alcoceba
4:40 - 4:50 pm	CTX Alliance	Mr. Neel Odedara
4:50 - 5:00 pm	Israeli CTX Association	Ms. Shosh Ben Yaish
5:00 - 5:10 pm	CTX Group in Netherlands	Ms. Hanka Dekker
5:10 - 5:25 pm	Questions for Session	Meeting Organizers
5:25 - 5:30 pm	Closing Remarks	Meeting Organizers

Notes:

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Providing education, support, advocacy, and promoting research  
for CTX patients, their families, and medical professionals who treat  
and study this rare disease.

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