

# Programme



## FRIDAY 20 MARCH

17:00-20:30 Registration

18:00-20:30 Poster session

## SATURDAY 21 MARCH

09:00-09:10 Welcome from Actelion Stefan Kolb (Actelion)

09:10-09:20 Welcome from Chairs Roberto Giugliani (Brazil)  
Thorsten Marquardt (Germany)

### INTRODUCTORY SESSION: INSIGHTS INTO THE LYSOSOME AND NP-C PATHOGENESIS

09:20-09:50 Presenter Yiannis Ioannou (USA)

### PLENARY SESSION I – IDENTIFYING NP-C: REVOLUTIONISING SCREENING AND DIFFERENTIAL DIAGNOSIS

09:50-10:10 Chairs Charles Lourenço (Brazil)  
Marc Patterson (USA)

10:10-10:30 Screening and diagnosing children with NP-C Mercedes Pineda (Spain)

10:30-10:50 Diagnostic strategies in adult metabolic practice Chris Hendriksz (UK)

10:50-11:20 The importance of networks and referral systems: the Brazilian NP-C experience Roberto Giugliani

11:20-11:40 Break

11:40-12:00 Evaluation of eye movements and motor function for early detection and monitoring of NP-C James Blundell (UK)

12:00-12:20 Differential diagnosis of NP-C and an algorithm for the diagnosis of recessive ataxias Mathieu Anheim (France)

12:20-12:30 Psychiatric manifestations in NP-C: new approaches to differential diagnosis Olivier Bonnot (France)

12:30-12:30 Q&A

12:30-14:00 Lunch & poster session

### PLENARY SESSION II – WHAT CAN WE LEARN FROM POPULATION SCREENING STUDIES? INNOVATIVE APPROACHES AND KEY FINDINGS

14:00-14:30 Chair María-Jesús Sobrido (Spain)

14:30-14:50 Q&A Amalia Bruni (Italy)  
Thomas Duning (Germany)  
Helen Michelakakis (Greece)  
Meral Topçu (Turkey)

14:30-14:50 Q&A

14:50-14:55 Changeover

14:55-15:40 Parallel workshop session I Workshops 1, 2, 3, 4 and 5

15:40-15:45 Changeover

15:45-16:30 Parallel workshop session II Workshops 1, 2, 3 and 4

16:30-17:00 Break

### PLENARY SESSION III – BREAKTHROUGHS IN DIAGNOSIS: INVALUABLE CONTRIBUTIONS FROM NOVEL BIOMARKERS

17:00-17:05 Chairs Paul Gissen (UK)  
Carlo Dionisi-Vici (Italy)

17:05-17:05 Introduction Carlo Dionisi-Vici

17:05-17:25 Improved diagnostics in NP-C by the analysis of plasma oxysterols Thorsten Marquardt

17:25-17:45 Lyso-sphingomyelin-509 is a sensitive and specific biomarker for NP-C Arndt Rolfs (Germany)

17:45-18:05 Bile acids in NP-C Peter Clayton (UK)

18:05-18:15 Q&A

18:15-18:20 Day 1 summary and announcement of poster winner Roberto Giugliani  
Thorsten Marquardt

18:20 Day 1 close

	<b>SUNDAY 22 MARCH</b>	
08:00–08:45	Breakfast session – Preclinical research in NP-C	Fran Platt (UK) Yiannis Ioannou
	<b>PLENARY SESSION IV – AFTER DIAGNOSIS: MIGLUSTAT TREATMENT AND DISEASE MANAGEMENT</b>	
	Chairs	Roberto Giugliani Marie Vanier (France)
09:00–09:20	Miglustat and the blood-brain barrier	David Begley (UK)
09:20–09:40	Structural MRI changes as biomarkers of disease status and progression in adults with NP-C	Mark Walterfang (Australia)
09:40–10:00	NP-C, miglustat and sleep disorders: a Spanish patient cohort	Jordi Gascón (Spain)
10:00–10:20	How miglustat treatment effect translates into tangible real-life benefits	Marc Patterson
10:20–10:30	Q&A	
10:30–10:55	Break	
10:55–11:40	Parallel workshop session III	Workshops 1, 2, 3 and 5
11:40–11:45	Changeover	
	<b>PLENARY SESSION V – PROGRESS IN UNDERSTANDING NP-C: MECHANISMS BEHIND THE DISEASE AND THERAPY</b>	
	Chairs	Thorsten Marquardt Peter Clayton
11:45–11:55	Poster presentation winner	
11:55–12:15	NP-C heterozygosity: the genetics of a complex disease	Miriam Stampfer (Germany)
12:15–12:35	Mechanistic approaches to treating NP-C	Fran Platt
12:35–12:55	Disorders of cholesterol metabolism	Emyr Lloyd-Evans (UK)
12:55–13:05	Q&A	
13:05–13:10	Day 2 summary and close	Roberto Giugliani Thorsten Marquardt
13:10	Day 2 close	
13:10	Lunch	



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Estoril, Portugal, 20 – 22 March 2015  
Chaired by Roberto Giugliani and Thorsten Marquardt

**INFORMATION  
FOR DELEGATES**

7<sup>th</sup> Scientific Symposium on  
Niemann-Pick Disease Type C (NP-C):  
Pioneering Approaches to  
Diagnosis and Disease  
Management



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