

Vienna, Austria, 15–17 March 2013  
Chaired by Chris Hendriksz and Frits Wijburg

# PROGRAMME

## 5<sup>th</sup> Scientific Symposium on Niemann-Pick Type C (NP-C): Next Generation of NP-C Management



### FRIDAY 15 MARCH

18:30 – 20:00 **Poster session**

### SATURDAY 16 MARCH

09:00 – 09:05 **Welcome from Actelion**

S Kolb (Actelion Pharmaceuticals Ltd)

09:05 – 09:15 **Introduction to the 5<sup>th</sup> NP-C Forum**

C Hendriksz (UK) and  
F Wijburg (Netherlands)

### OPENING SESSION

09:15 – 09:45 **An Introduction to Niemann-Pick disease type C**

C Hendriksz (UK)

### PLENARY SESSION I: GENETIC AND BIOCHEMICAL UPDATE

**Chairpersons:**

C Hendriksz (UK) and  
F Wijburg (Netherlands)

09:45 – 10:05 **Deep-sequencing to assess genetic variability in NP-C**

H Runz (Germany)

10:05 – 10:25 **A hot-spot and broad genetic carrier screening on a Greek island**

H Michelakakis (Greece)

10:25 – 10:45 **Break (including poster viewing)**

**Hilton Vienna Hotel**  
Am Stadtpark 1  
1030 Vienna, Austria

This Symposium is supported by Actelion Pharmaceuticals Ltd  
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# Programme

## 5<sup>th</sup> Scientific Symposium on Niemann-Pick Type C (NP-C): Next Generation of NP-C Management

### PLENARY SESSION II: EPIDEMIOLOGY OF NP-C

Chairpersons:

C Hendriksz (UK) and  
F Wijburg (Netherlands)

10:45 – 11:05 Final data on the ZOOM trial P Bauer (Germany)

11:05 – 11:25 Novel text mining semantic technology to identify undiagnosed  
NP-C cases: a retrospective analysis T Klopstock (Germany)

11:25 – 11:45 Newborn screening with oxysterols D Ory (USA)

11:45 – 12:15 Q&A

12:15 – 13:15 Lunch

### DEBATE: SHOULD NP-C MUTATIONS BE ACTIVELY SCREENED FOR IN THE GENERAL POPULATION?

Chairperson:

A Parker (UK)

Pro:

P Gissen (UK) and F Wijburg (Netherlands)

Con:

P McCormack (UK) and R Lachmann (UK)

### PARALLEL CASE-STUDY WORKSHOPS IN NP-C

Workshop 1: Psychiatric symptoms M Walterfang (Australia)  
H-H Klünemann (Germany)

Workshop 2: Ataxias M Anheim (France)  
C Hendriksz (UK)

Workshop 3: Developmental delays and school failure R Lachmann (UK)  
M Topçu (Turkey)

Workshop 4: Gaze palsies C Lourenço (Brazil)  
M Strupp (Germany)

Workshop 5: Infantile presentations C Dionisi-Vici (Italy)  
M Pineda (Spain)

14:55 – 15:15 Break (including poster viewing)

15:15 – 15:55 Workshops 1–5 repeated

### PLENARY SESSION III: INDICES OF DISEASE AND DIFFERENTIAL DIAGNOSTIC APPROACHES

Chairpersons:

S Jones (UK) and  
B Bembi (Italy)

16:00 – 16:20 The face of NP-C: Suspicion Index tool and dysmorphic manifestations in NP-C C Lourenço (Brazil)

16:20 – 16:40 Neonatal jaundice D Kelly (UK)

16:40 – 17:00 Differential diagnosis of NP-C: supranuclear ocular motor disorders M Strupp (Germany)

17:00 – 17:15 Monitoring disease course with Niemann-Pick type C clinical database (NPC-cdb) H Runz (Germany)

17:15 – 17:40 Q&A

17:40 – 17:50 Winning poster presentation

17:50 – 18:00 Day 1 summary and close C Hendriksz (UK)  
F Wijburg (Netherlands)

18:00 Close

### SUNDAY 17 MARCH

#### PLENARY SESSION IV: OPTIMISING DISEASE MANAGEMENT

Chairpersons:

T Marquardt (Germany) and  
A Parker (UK)

09:00 – 09:20	Treatment guidelines, NPC registry and optimised treatment with miglustat	M Patterson (USA)
09:20 – 09:40	Structural MRI changes as biomarkers of disease status and progression in adult patients with NP-C	M Walterfang (Australia)
09:40 – 09:55	Case presentation: Swallowing ability in a Taiwanese patient cohort	Y-H Chien (Taiwan)
09:55 – 10:10	Case presentation: Effect of miglustat on stabilisation of neurological disorder in NP-C in an Iranian patient cohort	P Karimzadeh (Iran)
10:10 – 10:25	Management of gastrointestinal side effects associated with miglustat therapy	M Rohrbach (Switzerland)
10:25 – 10:45	Q&A	

10:45 – 11:15 Break (including poster viewing)

#### PARALLEL CASE-STUDY WORKSHOPS IN NP-C

11:15 – 11:55 Workshops 1–5 repeated from Day 1

#### PLENARY SESSION V: FUTURE PERSPECTIVES IN NP-C

Chairpersons:

M Vanier (France) and  
C Hendriksz (UK)

12:00 – 12:20	Novel treatments for NP-C disease	D Ory (USA)
12:20 – 12:40	Genetic diagnosis in rare disorders: now and in the near future	P Gissen (UK)
12:40 – 13:00	NP-C: links to other disorders	F Platt (UK)
13:00 – 13:10	Q&A	
13:10 – 13:20	Day 2 summary and meeting close	C Hendriksz (UK) and F Wijburg (Netherlands)

13:20 Lunch