



13TH INTERNATIONAL SYMPOSIUM ON MUCOPOLYSACCHARIDOSES AND RELATED DISEASES

COSTA DO SAUIPE | BAHIA | BRAZIL | 13TH - 17TH AUGUST 2014

Beszámoló a 13. Nemzetközi MPS Szimpóziumról

News from the 13th International Symposium on MPS and Related Diseases

Dr. Komlósi Katalin

Orvosi Genetikai Intézet, PTE KK

20. Magyar Mukopoliszaccharidózis és társult betegségek konferenciára,
2014. október 3-5., Mater Salvatoris Lelkigyakorlatos Házban, Gödöllő





Total attendees: 1036

Families: 363

Scientists: 472

Volunteers: 51



Abstracts: 219

Speakers: 81

Scientific sessions: 6

Family sessions: 6

Satelite Symposia: 5

Countries : 43







MPS International Network Meeting



Reorganisation and sustainability of the Network

- **three major leaders “retiring”**

- Christine Lavery, UK MPS Society
- Barbara Wedehase, National MPS Society, USA
- Kirsten Harkins, Canadian MPS Society

- **new working group created**

- Hanka Meutgeert, Holland
- Fer Pidden, UK
- Carmen Kunkel, Germany
- Mary Boushel, Ireland





Research updates in the MPS field the industry's perspective

- Lysogene: The future of Gene Therapy
 - MPSIIIA gene therapy: Phase I/II study: safety trial (open label mono-centric study, 4 patients, from 2-6 years)
 - open-label, single arm, multi-centric, clinical study for MPS IIIA: 2015 Q3
 - Natural History Study
 - Discussion:
 - Identification of patients for clinical trials: 3-4 years of age (Diagnostic problems encountered worldwide)
 - Clinical trial: patient access

Research updates in the MPS field the industry's perspective

- Genzyme: MPSI research
 - use of ERT in the post-transplant setting
 - intrathecal Laronidase administration
 - benefit of early treatment: sibling studies
 - Genzyme Humanitarian Programme
 - Global Programme for Patient Organisations:
 - PAL Awards: grants for Societies
 - „Expression of Hope” Programme

Research updates in the MPS field the industry's perspective

- Biomarine: MPSIII, MPSIV and MPSVI research
 - natural history and galsulfase in MPSVI study
 - growth charts for MPSVI patients available!
 - growth charts under ERT!
 - MORCAP study (2008-): MPSIVA
(elosulfase alfa: Apr 28, 2014)
 - MPSIVA registry: start in September 2014
 - MPSIIIB study: NAGLU conjugated to IGF-2 tag:
direct brain delivery
 - clinical study for Batten disease (NCL, Hamburg)

Research updates in the MPS field the industry's perspective

- Shire: MPS II, MPS III
 - Phase I/II: intrathecal ERT in MPSII completed
 - Phase III/IV: AIM-IT: intrathecal Idursulfase administration
 - MPS IIIA development programme:
 - natural history study (U of Minnesota)
 - Phase II/III: intrathecal ERT , currently enrolling (12-48 months)
 - MPS IIIB natural history study: soon to be published

Research updates in the MPS field the industry's perspective

- Ultragenyx (Dr. Emil Kakkis)
 - MPS VII: ultra-rare, underdiagnosed (NIHF?!)
 - MPS VII clinical trials:
 - MPS VII survey: completed
 - Phase II study: ongoing
 - Phase III and „Under 5 yrs” study: planned for 2014/2015

Toward Treatment of MPS VII (Sly's Syndrome)

40 years from bedside to lab and back again



First patient with MPS VII
(1970)

- 1973: First patient report of β -glucuronidase deficiency
- 1993: First ERT research in murine MPS VII
- 2013: First ERT treatment of an MPS VII patient under eIND

**Dr. Emil Kakkis, Ultragenyx CEO, Int. MPS Network Meeting,
Aug 13, 2014**



Summary and Plan for MPS VII Program

- Great progress in development of ERT
- Four patients currently treated
- Phase 1/2 Data is encouraging
- One expanded access patient doing well
- Possible 4 month old patient requesting expanded access, may start in Aug
- Phase 3 by end 2014

First infusion of
rhGUS for MPS VII
Oct 16, 2013



Research updates in the MPS field the industry's perspective

- PTC-Bio:

- RNA biology for novel therapeutics of rare diseases
- nonsense readthrough mechanism
 - Ataluren in Duchenne Muscular Dystrophy: Phase II
 - Phase III study in CF
 - Phase II in MPSI: >60% have nonsense mutations
- alternate splicing approach (Phase I SMA study)

Research updates in the MPS field the industry's perspective

- Green Cross, Korea:
 - Idursulfase beta (Hunterase®) in MPSII:
 - Phase III trial in Korea
 - no FDA approval yet
 - 70 MPSII patients in South-Korea

Research updates in the MPS field investigator initiated studies

- Univ. of Manchester (Brian Bigger)
 - Phase III trial for high dose Genistein in MPSIIIA
 - HSCT gene therapy for MPS IIIA
- Nationwide Children's Hospital:
 - MPS IIIA and IIIB gene therapy research









13TH INTERNATIONAL SYMPOSIUM ON MUCOPOLYSACCHARIDOSES AND RELATED DISEASES

BIOMARIN

genzyme
A SANOFI COMPANY

Shire

AMGEN

AMGEN CROSS

AMGEN

ultragen

Symposium and Related Diseases

ran do
da Bahia

ultragenyx



Upcoming Events

- **MPS and Adulthood Meeting: October 2-4, Dublin: medical and social aspects**
- **MPS Network Meeting 2015**
 - Mid-July/August, Salzburg region,
 - 30th Anniversary of the Austrian MPS Society
- **14th International MPS Symposium**
 - 14.-17. July, 2016: Bonn
- **15th Int. MPS Symposium: 2018**
 - USA West-coast

MPS 2016
14th INTERNATIONAL SYMPOSIUM
ON MPS AND RELATED DISEASES



14 – 17 JULY 2016
BONN, GERMANY



Destination Bonn



International Scientific Committee

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Mainz, Germany

Thomas Bräulke
Hamburg, Germany

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Kurt Ullrich
Hamburg, Germany

Frits Wijburg
Amsterdam, Netherlands

Destination Bonn

Impressions



Old Town Hall



Cathedral of Bonn



Art Museum Bonn



Beethoven's monument



„Rhineau“



Castle of Bonn | University

Children & Youngsters Program

Theme: Saga of the Nibelungs



- Outdoor playing area with different activity stations such as:
 - face painting
 - Design / Crafting costumes like a knight costume etc.
 - Treasure hunt
 - Crafting swords, the Loreley rock etc.
 - Swordplay



- Indoor: Relaxing area



Program – Friday

In the morning...

	SCIENTIFIC PROGRAM	FAMILY PROGRAM	
7:00-8:45	Reserved for breakfast symposium		
9:00-10:30	Recent advances in therapy <ul style="list-style-type: none">• Intrathecal ERT• Gene therapy		
10:30-11:00	Coffee/Tea break		
11:00-12:30	New diagnostic approaches <ul style="list-style-type: none">• Mass-spectrometry• Metabolics	Palliative care	Meeting of adult patients

Website

To much information for now?

**Don't mind –
just get all necessary information on our website**

www.mps2016.com

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- Hungarian MPS Society
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