

**International Workshop    March 14-16th, 2012, University of  
Konstanz**

**“New Developments of Lysosomal storage disease diagnostics &  
therapeutics  
LSD Protein – aggregation & interactions”**

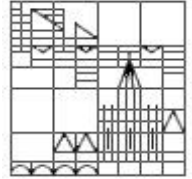
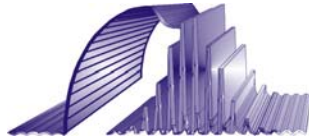
Lysosomal storage disorders (LSDs) are a group of approximately 50 rare inherited metabolic disorders (classified as orphan disorders) caused by deficiency of single lysosomal enzymes in metabolizing biopolymer substrates such as glycolipids or mucopolysaccharides, or by a defect in a lysosomal membrane protein. The accumulation of lysosomal substrates causes a broad spectrum of clinical symptoms that differ by disease, age of onset and severity. The clinical presentation can vary from seriously debilitating and life-threatening early in life, to late-onset, mild symptoms not requiring specific treatment.

In recent years, therapies have become available for a few LSDs, including hematopoietic stem cell transplantation, organ transplantation, enzyme replacement therapy and administration of small molecules. However, for most LSDs symptomatic treatment is still the only option. Early diagnosis of treatable LSDs can be life saving and allows timely treatment before irreversible damage occurs. Therefore, it is of utmost importance to develop new and improve existing diagnostic techniques for LSDs. However, timely diagnosis of lysosomal storage disorders is still a significant challenge in clinical practice.

In this workshop, new developments in therapeutics for LSDs, new tools of mass spectrometry for characterization of structure and interactions, and quantitative determination of LSD proteins, and diagnostic of LSDs, will be presented and discussed by biopolymer scientists, clinical chemists, and mass spectrometry experts. In addition, the programme will comprise course practicals on affinity-mass spectrometry, and diagnostics of lysosomal storage diseases by mass spectrometric and fluorimetric determinations.

We look forward to welcome you in Konstanz!

Stefan Maeser, Centogene, Freiburg/Rostock  
Michael Przybylski, University of Konstanz



Fachgruppe Affinity- Mass Spectrometry

International Workshop:

**Affinity Mass Spectrometry: New Methods and Applications  
New Developments of Lysosomal Storage Disease  
Diagnostics & Therapeutics**

14 - 16<sup>th</sup> March, 2012

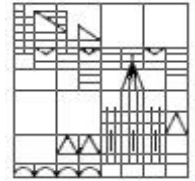
Hörsaal/Lecture Rooms A 704 / L 829 / L 846

## Programme

Wednesday, 14<sup>th</sup> March 2012

A 704

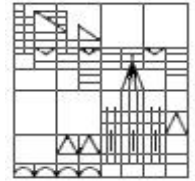
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| 14:00 - | <b>Registration</b>  |
| 16:30   | <b>Michael Przybylski, Konstanz<br/>Stefan Maeser, Freiburg/Rostock<br/>Welcome &amp; Introduction</b>                           |
| 17:00   | <b>Michael Przybylski, Konstanz<br/>New Methods for Diagnostics of Lysosomal Storage Diseases</b>                                |
| 17:30   | <b>Michael Preiss, Basel<br/>(Introduction by Stefan Maeser)<br/>Overview and State of Lysosomal Storage Disease Diagnostics</b> |
| 18:15   | <b>Welcome &amp; get-together</b>  |



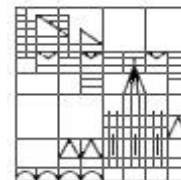
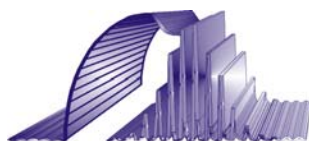
Thursday, 15<sup>th</sup> March 2012

A 704

- 09:00            **Therapeutic Developments of Lysosomal Storage Diseases**
- 09:00            **Stefan Maeser, Freiburg & Rostock**  
**State and Development of Lysosomal Storage Diseases**
- 09.30            **Hanna Mandel, Haifa**  
**Pompe's Disease in the ERA of Enzyme Replacement**  
**Therapy: Pros and Cons"**
- 10:00            **Stefan Kolb, Basel**  
**The Niemann-Pick C Suspicion Index: A novel diagnostic**  
**approach for a rare disease**
- 10:30            **Eugen Mengel, Mainz**  
**Enzyme Replacement as Therapeutic Principle for Lysosomal**  
**Storage Diseases - Most Recent Developments**
- 11:10            *Coffee break*
- 11:30            **Diagnostics of Lysosomal Storage Diseases**
- Eser Sozmen, Izmir**  
**Enzymatic Diagnosis of Lysosomal Storage Diseases by**  
**Fluorimetric assay: Effectiveness and Reliability**
- 12:00            **Adolf Mühl, Rostock**  
**Tandem Mass Spectrometry Assays of Lysosomal Enzyme**  
**Activities in Dried Blood: Results from Large Scale Screenings**
- 12.50            **Zoltan Lukacs, Hamburg**  
**Dried Blood Spots for the Diagnosis of Lysosomal Storage**  
**Disorders - Ten Years Experience**



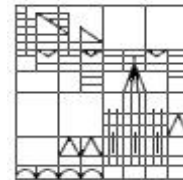
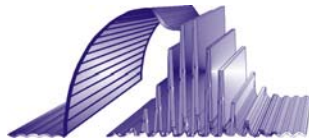
- 13:15 *Lunch Break*
- 14:00 **Michael Przybylski, Konstanz**  
**Affinity – Mass Spectrometric Determination and Aggregation of Lysosomal Proteins**
- 14:30 **Stephanie Fischmann, Ludwigshafen**  
**Mass Spectrometric Determination of Antibodies**
- 15:00 **Mark MacDowell, Manchester**  
**Application of T-Wave Ion Guides for ETD and Ion Mobility- Mass Spectrometry**
- 15:30 *Coffee Break*
- 16:00 **N.N., Bremen**
- 16:30 **Silva Hecimovic, Zagreb**  
**The molecular links between lysosomal storage diseases and Alzheimer’s disease**
- 17:00 **Michael Glocker, Rostock**  
**Proteome Profiling by Affinity - Mass spectrometry: Application to Gestation-related Diseases**
- 17:30 **Introduction of Course Practicals (S. Maeser; Moderation)**  
**Claudia Cozma: Mass spectrometric and Fluorimetric Diagnostics of Lysosomal Storage Diseases**
- 17:45 **Adrian Moise: Mass Spectrometric Identification of Protein-carbohydrate recognition epitopes**
- 18:00 **Stefan Slamnoiu : Online Bioaffinity- Mass Spectrometry**
- 19:30 *Workshop Dinner*



Friday, 16<sup>th</sup> March 2012

### Workshop Practical Courses

8:30	<b>Introduction</b> (M. Przybylski/S. Maeser/C. Cozma/ S. Slamnoiu/A. Moise)	<b>L 829</b>
9:00 – 11:30	<b><u>Course Practical I:</u></b> “Affinity-Mass Spectrometry: “Identification of Protein- carbohydrate recognition epitopes”	<b>L 846 / 844</b>
12:00 – 13:30	<b><u>Course Practical II:</u></b> “Affinity-Mass Spectrometry: Online- Biosensor- MS”	<b>L 846 / 844</b>
	<i>Lunch Break</i>	
14:00 –	<b><u>Course Practical III / IV:</u></b> Fluorimetric diagnostics of Lysosomal Storage Diseases	<b>L 846 / 844</b>
17:30	<b>Mass spectrometric diagnostics of Lysosomal Storage Diseases</b>	
18:00	<b>Final Discussion</b>	
18:30	<b>End of Workshop Refreshments</b>	



## General information

### Venue

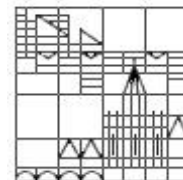
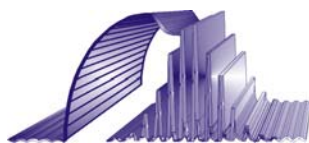
The international workshop "New Developments of Lysosomal Storage Disease Diagnostics & Therapeutics" will be held at the University of Konstanz. All lectures will be in Building A, lecture hall A 704. The Workshop will start with an opening lecture on Wednesday, 14<sup>th</sup> March 2012, 16.30 pm, Workshop Registration starts earlier from 14.00 p.m. Course on mass spectrometric and fluorimetric determination of lysosomal storage proteins will be held for interested participants on Friday, March 16<sup>th</sup>. Please see separate announcement.

### Accommodation

Since Konstanz is a major tourist attraction, accommodation limitations are always possible, therefore we recommend booking in advance. For extensive information on accommodation in Konstanz please visit: <http://www.konstanztourismus.de/>. Major hotels located downtown in walking distance from the train station are listed below.

Viva Sky Hotel KN  
Sigismundstr. 19, 78462 KN  
Tel. 07531 - 692362 - 0  
[welcome@hotel-viva-sky.de](mailto:welcome@hotel-viva-sky.de)  
<http://www.hotel-viva-sky.de/>  
ab 80.- EUR

Hotel Arcadia Halm KN  
Bahnhofplatz 6  
78462 KN  
Tel. 00 800 - 97 33 42 26  
[http://arcadia-halm-konstanz.hotelrez.com/index\\_de.html](http://arcadia-halm-konstanz.hotelrez.com/index_de.html)  
ab 69 EUR



City Hotel KN  
Bahnhofstr. 10  
78462 KN  
Tel. 07531 - 1331-0  
[info@city-hotel-konstanz.de](mailto:info@city-hotel-konstanz.de)  
<http://www.city-hotel-konstanz.de/>  
EZ ab EUR 65.- inkl. Frühstück

Gästehaus Centro KN  
Bahnhofstr. 4  
78462 Konstanz  
07531 - 457 181 8  
[info@gaestehauscentro.de](mailto:info@gaestehauscentro.de)  
<http://www.gaestehauscentro.de/>  
EZ ab EUR 55.- inkl. Frühstücksbuffet

Hotel Bayrischer Hof KN  
Rosgartenstr. 30  
78462 KN  
Tel. 07531 - 1304-0  
[info@bayrischer-hof-konstanz.de](mailto:info@bayrischer-hof-konstanz.de)  
<http://www.bayrischer-hofkonstanz.de/>  
EZ ab EUR 68.- inkl. Frühstück

### **Parking**

There are four parking lots at the University Campus area which can be recognized by the given signs.

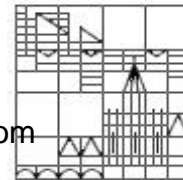
### **Transportation**

You can reach the University with bus lines 9A and 9B from the main train station Konstanz. From Wollmatingen you can use the bus line 11 to get to the University, and from Litzelstetten and Dingeldorf the bus line 4 is available, with the bus stop "Egg" convenient to reach the University.

### **Wardrobe**

On level B4 you can find a supervised wardrobe. However, we recommend you not to leave any valued articles unattended.

### **Conference office/Registration**



...k will be located adjacent to A704, and will ... starting from  
Wednesday, 14 March 2012, 14.00 p.m.

### **Speaker's PC**

In the lecture hall A704 a laptop will be available for presentations. We ask you to bring your PowerPoint presentation on an USB flash drive, for easy loading on the presentation computer.

### **Registration**

The registration form can be found as a Word document together with the announcement on the websites:

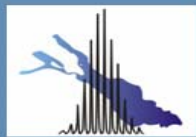
[www.afinity-ms.de](http://www.afinity-ms.de) and

[www.uni-konstanz.de/agprzybylski/chemie](http://www.uni-konstanz.de/agprzybylski/chemie)

Please send the registration to [adrian.2.moise@uni-konstanz.de](mailto:adrian.2.moise@uni-konstanz.de).

A workshop dinner is planned, details of which will soon be announced.





## **Fachgruppe Affinity – Mass Spectrometry International Workshop**

**14 - 16<sup>th</sup> March 2012  
University of Konstanz**

# **Affinity-Mass Spectrometry: New Methods and Applications New Developments of Lysosomal Storage Disease Diagnostics & Therapeutics**

- **Lysosomal proteins**
- **Clinical LSD diagnostics**
- **LSD Therapeutics**
- **Protein-carbohydrate interactions**
- **Biomarker identification**
- **Quantitative Affinity-MS**
- **MS & hyphenated methods**

**Introductory course:  
LSD diagnostics by MRM-MS and fluorometry**

Organizers:

**Michael O. Glocker, Rostock  
Michael Przybylski, Konstanz  
Joan Keutzer, Cambridge  
Stefan Maeser, Freiburg  
Stefaan Sansen, Amsterdam  
David Andreu, Barcelona**

Further information:  
[www.affinityms.de](http://www.affinityms.de)  
[www.uni-konstanz.de/agprzybylski/chemie/](http://www.uni-konstanz.de/agprzybylski/chemie/)