



GERMAN NETWORK ON  
BONE MARROW FAILURE SYNDROMES

**SECOND INTERNATIONAL WORKSHOP  
ON  
CONGENITAL BONE MARROW FAILURE SYNDROMES**

**28<sup>th</sup> February/ 1<sup>st</sup> March 2009**



---

**Kloster Wennigsen ▪ Klosteramthof 5 ▪ D-30974 Wennigsen ▪  
Germany**

# Program

## **Scientific Organization:**

Matthias Ballmaier, Hannover

Helmut Hanenberg, Düsseldorf

Hermann Heimpel, Ulm

Karl Welte, Hannover

Cornelia Zeidler, Hannover

## **Congress Secretary:**

Marina Kotz

SCNIR

Children's Hospital,

Hannover Medical School,

Telephone: +49-(0)511-557105

## **Congress Venue:**

Kloster Wennigsen

Klosteramthof 5

30974 Wennigsen

Telephone: +49-(0)5103-9398-0

Fax: +49-(0)5103-9398-55

## **Information:**

Dr. med. Cornelia Zeidler

SCNIR

Department of Molecular Hematopoiesis,

Children's Hospital,

Hannover Medical School,

Carl-Neuberg-Straße 1

30625 Hannover

Telephone: +49-(0)511-557105

Fax: +49-(0)511-557106

## 2. INTERNATIONAL WORKSHOP ON CONGENITAL BONE MARROW FAILURE SYNDROMES

28<sup>th</sup>February/ 1<sup>st</sup> March 2009

Kloster Wennigsen ▪ Klosteramthof 5 ▪ D-30974 Wennigsen ▪ Germany

---

### **Saturday, 28<sup>th</sup> March 2009, 9 a.m. – 6 p.m.**

#### **9:00 Welcome and Introduction**

Karl Welte, Hannover, Germany

#### **9:10 The Israeli Bone Marrow Failure Registry**

Hannah Tamary, Petah Tikva, Israel

#### **9:30 Congenital Anemias** (chair: Hermann Heimpel)

- **Extra-Medullary Hematopoiesis in CDA and Other Rare Anemias**

Hermann Heimpel, Ulm, Germany

- **Ribosomal Protein Mutations and Biogenesis in DBA**

Lydie Da Costa, Paris, France

- **Stem Cell Transplantation in Diamond Blackfan Anemia**

Jörg Meerpohl, Freiburg, Germany

- **Diamond Blackfan Anemia: Data from the Czech DBA Registry**

Dagmar Pospisilova, Olomouc, Czech Republic

#### **10:50**

#### **COFFEE BREAK**

#### **11:15 Fanconi Anaemia** (chair: Helmut Hanenberg)

- **Preclinical Studies for Gene Therapy in Fanconi Anemia**

Juan A Bueren, Madrid, Spain

- **Mapping the Fanconi Anemia Interactome**

Miguel A. Pujana, Barcelona, Spain

- **Translational Approaches in Fanconi Anemia**

Helmut Hanenberg, Düsseldorf, Germany

- **Relevance of Acquired Clonal Chromosome Aberrations in Bone Marrow Cells of Fanconi Anemia Patients**

Heidemarie Neitzel, Berlin, Germany

- **Splice Mutations in Fanconi Anemia**

Heiner Schaal, Düsseldorf, Germany

- **Reasons for Fanconi Anemia Patients to Reach Adulthood**

Detlev Schindler, Würzburg, Germany

## **2. INTERNATIONAL WORKSHOP ON CONGENITAL BONE MARROW FAILURE SYNDROMES**

28<sup>th</sup> February/ 1<sup>st</sup> March 2009

Kloster Wennigsen ▪ Klosteramthof 5 ▪ D-30974 Wennigsen ▪ Germany

---

**13:15**

**LUNCH**

**14:40 Congenital Neutropenia Part I** (chair: Karl Welte)

- **Update on the Original Kostmann Families**  
Jan Palmblad, Stockholm, Sweden
- **New Gene Defects in Congenital Neutropenias**  
Christoph Klein, Hannover, Germany
- **Glycogen Storage Disease Type Ia and Ib: Molecular Insights**  
Rene Santer, Hamburg, Germany
- **WAS Mutations in Congenital Neutropenias**  
Karolien Beel and Peter Vandenberghe, Leuven, Belgium

**16:00**

**COFFEE BREAK**

**16:30 Congenital Neutropenia Part II** (chair: Jan Palmblad)

- **Insights in Pathomechanisms of CN: An Overview**  
Karl Welte, Hannover, Germany
- **G-CSF Receptor Mutations in Congenital Neutropenia**  
Ivo Touw, Rotterdam, Netherlands
- **Leukemia in Congenital Neutropenia**  
Cornelia Zeidler, Hannover, Germany
- **Risk Factors for Bone Marrow Failure and Leukemia in Shwachman-Diamond Syndrome**  
Jean Donadieu, Paris, France

**17:50 Congenital Thrombocytopenia** (chair: Matthias Ballmaier)

- **Congenital Amegakaryocytic Thrombocytopenia (CAMT)**  
Matthias Ballmaier, Hannover, Germany
- **Biology of Thrombopoietin in the Central Nervous System**  
Christof Dame, Berlin, Germany

## **2. INTERNATIONAL WORKSHOP ON CONGENITAL BONE MARROW FAILURE SYNDROMES**

28<sup>th</sup>February/ 1<sup>st</sup> March 2009

Kloster Wennigsen ▪ Klosteramthof 5 ▪ D-30974 Wennigsen ▪ Germany

---

### **18:30 Initial Survey on Conditions and Quality of Life in Patients with Inborn Bone Marrow Failure Syndromes**

Gabriele Calaminus, Münster, Germany

**20:00**

**CONFERENCE DINNER**

## **2. INTERNATIONAL WORKSHOP ON CONGENITAL BONE MARROW FAILURE SYNDROMES**

28<sup>th</sup>February/ 1<sup>st</sup> March 2009

Kloster Wennigsen ▪ Klosteramthof 5 ▪ D-30974 Wennigsen ▪ Germany

---

**Sunday, 1<sup>st</sup> March 2009, 9.00 a.m. – 12:30 p.m.**

### **9:00 Special Case Reports** (chair: Matthias Ballmaier)

**- Case Report on a Patient with ICF-1 Syndrome and Pancytopenia**

Georg Ebetsberger, Linz, Austria

**- An unusual Patient with CAMT**

Joanne Yacobovich, Petah Tikva, Israel

**- Case Reports of Two CDA - Like Diseases**

Martina Suková and Ester Pindurova, Prague, Czech Republic

**10:00**

**COFFEE BREAK**

**10:30**

### **Networking and Common Projects:**

Anaemia Working Group

Fanconi-Anaemia Working Group

Neutropenia Working Group

Thrombocytopenia Working Group

**12:00**

### **Closing Remarks**

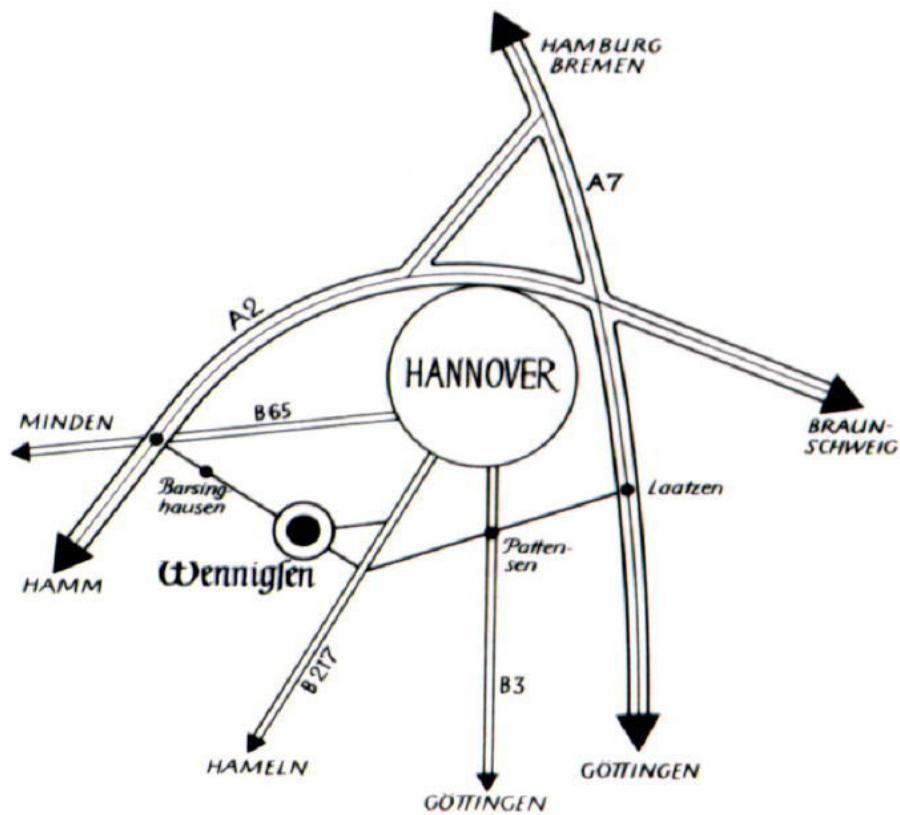
**LUNCH AND ADJOURNMENT**

## How to get to Kloster Wennigsen

Klosteramthof 5

30974 Wennigsen

**By car:** Take the B 217 towards Hameln/Springe, take the exit Sorsum. Pass through Sorsum and follow the road to Wennigsen. Turn right at the church and immediately left on the parking place of the “Johanniter Haus”.



**By train from Hannover airport:** take the subway **S5** at Terminal C (departure every hour at .06 and .36 minutes) to Hannover central station (arrival at platform 2 at .23 or .53), than continue as from Hannover central station (below)

**By train from Hannover central station:** take the train S1 or S2 to Haste (departure S1 on platform 1 every hour at .33, S2 on platform 1 every hour at .03). After approximately 25 min. **exit the train in Wennigsen.** Use the underground crossing and walk down Bahnhofstraße to the convent (approximately 10 minutes walk)

