

# PROGRAM

[www.IDMC-7.com](http://www.IDMC-7.com)

September 9 – 12, 2009 • Würzburg, Germany



# IDMC-7

100 YEARS MYOTONIC DYSTROPHY AND STEINERT DISEASE



7<sup>th</sup> International  
Myotonic Dystrophy  
Consortium Meeting

IDMC-7

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Hans Steinert

## Welcome to the 7<sup>th</sup> International Myotonic Dystrophy Consortium Meeting (IDMC-7) in Würzburg, Germany

September, 9-12, 2009

The **International Myotonic Dystrophy Consortium (IDMC)** is a group of scientists and clinicians with a shared interest in the understanding of myotonic dystrophy (DM), both type 1 (DM1) and type 2 (DM2), and the principal aim of providing improved understanding and treatment for patients. 2009 marks the 100<sup>th</sup> anniversary of the first description of *dystrophica myotonica* (DM) in the clinical literature by Hans Steinert (1875-1911, Leipzig, Germany). In Würzburg Kenneth Ricker (1934–2004) was one of the first clinicians to recognize DM2 as a disease distinct from DM1. In memory of both scientists, it is therefore with great pleasure that we welcome you all to Würzburg for this special anniversary meeting.

Since the first IDMC meeting in Paris in 1997, remarkable progress has been made in DM research. Still much remains to be done to provide patients and their care providers with effective therapies for the many symptoms of this disease. The bi-annual IDMC meeting provides a unique forum to discuss the latest findings in the DM field. In keeping with previous IDMC meetings, IDMC-7 will cover a broad range of topics, including historic perspectives on DM1 and DM2, mechanisms of disease pathogenesis and repeat instability of the underlying DNA mutations, model systems for DM, and advances in treatment of these multisystem disorders. We hope IDMC-7 will provide a setting and atmosphere for open and stimulating exchange of the newest findings and ideas between basic researchers, clinicians (including neurologists, geneticists; pediatricians, internists, physical therapists, ophthalmologists, anesthesiologists, gynecologists), and translational scientists, as well as patients and their care providers.

Finally, we would like to thank the IDMC membership for electing us to organize IDMC-7 and all those who have helped with the organization of this meeting: our generous sponsors, who have made this congress possible, the organizing agency Carlo Praetorius (Munich), and last but not least all of you--scientists, patients and caregivers--who have come from 16 countries and four continents to Würzburg to make this another successful meeting. We hope the meeting will be memorable and you will enjoy the scientific and patient sessions and the social programs.

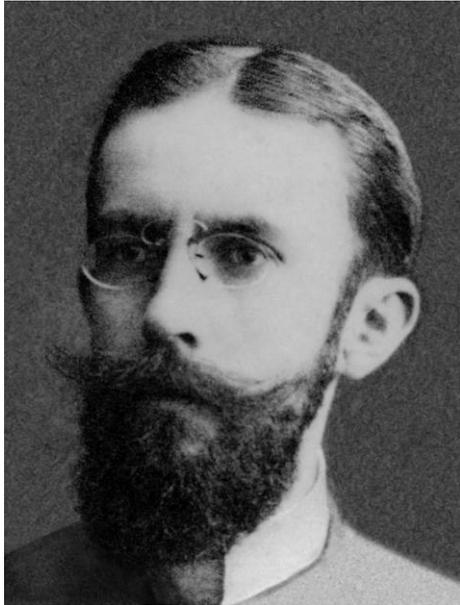
Thank you – Danke!

Ralf Krahe  
Houston, USA

Tiemo Grimm  
Würzburg, Germany

Benedikt Schoser  
Munich, Germany

The IDMC-7 Chairmen



## **Hans Gustav Wilhelm Steinert**

German Internist, 1875-1911

After attending the Gymnasium in Freiberg, Hans Gustav Wilhelm Steinert attended the Universities of Leipzig, Freiberg, Berlin and Kiel. He graduated in 1898 and subsequently held junior appointments in Halle an der Saale and Berlin. He settled in Leipzig, where he worked in the pathological institute and the medical clinic before being appointed professor in 1910. Most of his written works are in the field of neurological and muscular conditions.

*Steinert H. Myopathologische Beitrage 1. Ueber das klinische und anatomische Bild des Muskelschwunds der Myotoniker. Dtsch Z Nervenheilkd, 1909,37:58-104*

Der Bayerische Ministerpräsident  
Horst Seehofer



## Welcoming Address

### 100 Years of Myotonic Dystrophy and Steinert Disease

### 7<sup>th</sup> International Myotonic Dystrophy Consortium Meeting in Würzburg, September 9-12, 2009

I cordially welcome all participants of the 7<sup>th</sup> International Myotonic Dystrophy Consortium Meeting to Bavaria.

Fortunately, in both of its forms Myotonic Dystrophy is a relatively rare disease. But whoever is affected with it – especially type 1 – has a heavy burden to carry. Modern genetics research has been able to identify the underlying cause of the disease. Thus, there is hope that one day it will be possible to sufficiently understand the disease mechanisms and to substantially help the patients. So far, medical practice has been restricted to treating the varied symptoms.

The neurologist Hans Steinert provided the first description of the classic type of the disease in Leipzig 100 years ago. This anniversary is a good opportunity to take stock where research has gotten us and to look ahead into the future. The patient day provides a unique opportunity for all participants to closely interact, including the patients, their care-givers and advocacy groups. I dearly thank the participating patient support-groups.

This is the first time the IDMC congress convenes in Germany, and Würzburg provides an excellent setting for it. With its expertise, the University has acquired an outstanding reputation in the field of medicine. Case in point is the work by the late neurologist Kenneth Ricker who here in 1994 described Myotonic Dystrophy type 2.

Over the last decade Bavaria has developed itself into a “health-land.” In terms of quality of our healthcare institutions, our basic and translational biomedical research, and biotechnology, Bavaria has taken a leading position in the international comparison. The close ties between research and clinical practice guarantee outstanding healthcare to our people. In addition, the healthcare sector has become an important industry for the future and increasingly impacts our economy. The Bavarian State Government will continue to invest into it.

I sincerely wish all of the participants of the congress an interesting and stimulating discussions as well as a pleasant stay in Würzburg.

A handwritten signature in blue ink, appearing to read 'H. Seehofer'.

Horst Seehofer  
Prime Minister of Bavaria

# **Scientific Committee**

## ***Congress Chairmen***

Ralf Krahe, University of Texas M. D. Anderson Cancer Center, Houston, USA  
Tiemo Grimm, University of Würzburg, Germany  
Benedikt Schoser, University of Munich, Germany

## ***Local Chairmen***

Wolfram Kress, University of Würzburg, Germany  
Clemens R. Müller-Reible, University of Würzburg, Germany  
Karlheinz Reiners, University of Würzburg, Germany  
Klaus Victor Toyka, University of Würzburg, Germany

## ***International Committee***

Tetsuo Ashizawa, University of Florida, Gainesville, USA  
Giovanni Meola, University of Milan, Italy  
Darren G. Monckton, University of Glasgow, Glasgow, UK  
Charles Thornton, University of Rochester, New York, USA  
Nakaaki Ohsawa, Aino Institute For Aging Research, Osaka, Japan  
Geneviève Gourdon, Inserm U383, Hôpital Necker-Enfants Malades, Paris, France  
Jack Puymirat, CHU Laval Research Center, Quebec, Canada  
Shannon M. Lord, Hunter Lord Funds, Atlanta, Georgia, USA  
Lisa Vittek, Myotonic Dystrophy Foundation, Rocklin, California, USA  
Margaret Bowler, Myotonic Dystrophy Support Group, Nottingham, UK  
Claude Bourlier, French Myotonic Dystrophy Support Group, Evry, France

## ***Congress Secretary***

Brigitte Wolf  
Institut für Humangenetik  
Julius-Maximilians-Universität Würzburg  
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## ***Congress Organisation***

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## ***Congress Venue:***

Julius-Maximilians-Universität Würzburg  
Neue Universität, Lecture halls  
Sanderring 2  
97070 Würzburg, Germany

## ***General information***

### **Badges**

Participants should collect name badges from the conference registration desks.

**Coffee Breaks and Lunch**

During the session breaks refreshments (coffee, tea, water) and lunch will be served free of charge to participants wearing name badges.

**Currency**

The official currency of Germany is the Euro (€)

**Electricity**

220V – 50Hz AC. Connectors can be obtained from your hotel reception or electronic shops.

**Language**

The official language of the congress will be English. On Saturday, September 12, will be simultaneous translation to German

**Smoking Policy**

The IDMC-7 is a non-smoking conference. Please note that smoking is banned from all public buildings and restaurants.

***Social Events***

**Wednesday, September 9, 2009, 19:00**

**Welcome Reception**

Foyer of the Lecture Hall

**[Bassiona Amorosa](#) - an international contrabass sextet**

**Friday, September 11, 2009, 17:00 hrs**

**Visiting the Residence**

Admission by ticket only, you should pre-book at moment of your registration.

The former residence of the Würzburg prince-bishops is one of the most important baroque palaces in Europe and today it is on UNESCO's World Cultural Heritage list. Originally designed for Prince-Bishop Johann Philipp Franz von Schönborn by the then young and unknown architect Balthasar Neumann, it took sixty years to complete; the shell of the palace was built from 1720 to 1744 and the interior finished in 1780.

Neumann's world-famous staircase, roofed by an unsupported vault, was decorated in 1752/53 by the Venetian Giovanni Battista Tiepolo with a ceiling fresco representing the four continents. The painting, measuring 18 x 30 meters, is one of the largest frescos ever created. The magnificent sequence of rooms begins with the Vestibule and Garden Hall and continues via the staircase and White Hall to the Emperor's Hall, also with frescos by Tiepolo. The vaulting of these rooms even withstood the devastating fire of 1945, while the ceilings and floors of the Imperial Apartments flanking the Emperor's Hall were destroyed. The furnishings and wall paneling had been removed beforehand enabling the rooms to be reconstructed. Restoration was completed in 1987 with the reopening of the Mirror Cabinet. There are a total of over 40 palace rooms to visit, with a rich array of furniture, tapestries, paintings and other 18th century treasures.

**Friday, September 11, 2009, 18:45 hrs**

**Winetasting Reception at the Cellar of the Residence**

Meeting point at the fountain (*Frankonia-Brunnen*) in front of the Residence.



**Entrance to  
the wine cellar**

**Meeting point  
“Frankonia-Brunnen”  
(fountain before the residence)**

You will spend the evening in a historical Wine Cellar, the temperature will be max. 20° C  
Please consider adequate clothing.

Admission by ticket only, you should pre-book at moment of your registration.

**Contribution towards expenses:**

**Participants, accompanying persons Euro 10,00**

**Saturday, September 12, 2009, 19:00 hrs**

**Farewell-Dinner**

**Riverboat trip at the river Main**

Meeting point: Alter Kranen - the Old Crane at the river Main.

Admission by ticket only, you should pre-book at moment of your registration

**Contribution towards expenses:**

**Participants, accompanying persons Euro 50,00**

# **IDMC-7 Scientific Program**

## **Wednesday, September 9, 2009**

Lecture Hall 1 (HS216, Audimax)

- S1 Opening Ceremony 17.00 - 19.00**  
**Welcoming addresses by:**  
**Krahe, Ralf (on behalf of the Congress Chairmen)**  
**President of the University of Würzburg**  
**Vice president Prof. Dr. Heidrun Moll**  
**Dean of the Medical Faculty of the University Würzburg**  
Vice dean Prof. Dr. med. Manfred Gessler
- Special Lectures**  
Chairs: Grimm, Tiemo (Würzburg, Germany)  
Schoser, Benedikt (Munich, Germany)
- S1-01 A Hundred Years of Myotonic Dystrophy 17:30 – 18:15**  
Harper, Peter (Cardiff, UK)
- S1-02 Kenneth Ricker and PROMM/DM2 18:15 – 19:00**  
Moxley III, Richard (Rochester, USA)
- Welcome Reception 19:00 – 21:00**  
*Foyer of the Lecture Hall*

## **Thursday, September 10, 2009**

Lecture Hall 1 (HS216, Audimax)

- S2 Pathomechanisms in DM—Part 1 08.00 - 12.30**  
Chairs: Timchenko, Lubov (Houston, USA)  
Ranum, Laura (Minneapolis, USA)
- S2-01 Current understanding of pathomechanisms in DM 08:00 – 08:30**  
Thornton, Charles (Rochester, USA)
- S2-02 Multi-step deregulation of MBNL1 complex stoichiometry results in progressive RNA splice defects in myotonic dystrophy 08:30 – 08:45**  
Reddy, Sita Reddy (Los Angeles, USA)
- S2-03 Tau mis-splicing recovery in myotonic dystrophy type 1 08:45 – 09:00**  
Tran, H el ene (Lille, France)
- S2-04 A (CCUG)<sub>n</sub>-binding protein that may explain the lesser severity of DM2 09:00 – 09:15**  
Hammer, Caroline (Illkirch, France)
- S2-05 Identification of CUGBP1 mRNA Targets by CUGBP1/RNA Immuno-capture 09:15 – 09:30**  
Bachinski, Linda (Houston, USA)
- S2-06 Protein and micro-RNA binding behavior of DMPK messenger RNA 09:30 – 09:45**

Wieringa, Be (Nijmegen, The Netherlands)

<b>S2-07</b>	<b>Identification of novel candidates that intervene in Myotonic Dystrophy Type 1 (DM1)</b> Francois-Xavier Laurent (Gif sur Yvette, France)	<b>09:45 – 10:00</b>
	<b><i>Coffee break / Poster Viewing / Exhibition</i></b>	<b>10:00 – 10:30</b>
<b>S2-08</b>	<b>The DMSXL transgenic mice carrying very large expansions exhibit molecular and physiological defects: an animal model for gene therapy experiments?</b> Gourdon, Geneviève (Paris, France)	<b>10:30 – 10:45</b>
<b>S2-09</b>	<b>A second example of RNA-gain-of-function disease: Sam68 and MBNL1 sequestration by CGG repeats in Fragile X Tremor Ataxia Syndrome</b> Sellier, Chantal (Illkirch, France)	<b>10:45 – 11:00</b>
<b>S2-10</b>	<b>Development of a muscleblind-like 1 (Mbnl1) overexpression model to assess the use of MBNL1 as a potential therapeutic for myotonic dystrophy</b> Chamberlain, Christopher (Minneapolis, USA)	<b>11:00 – 11:15</b>
<b>S2-11</b>	<b>Misregulation of diacylglycerol kinase eta (DGK<math>\eta</math>) splicing as a potential cause of neuropsychiatric symptoms in myotonic dystrophy type 1</b> Matsuura, Tohru (Okayama, Japan)	<b>11:15 – 11:30</b>
<b>S2-12</b>	<b>Study on the mechanisms of regulation and deregulation of hcTNT exon 5 inclusion</b> Behm-Ansmant, Isabelle (Vandoeuvre-les-Nancy, France)	<b>11:30 – 11:45</b>
<b>S2-13</b>	<b>MBNL1 associates with YB-1 in cytoplasmic stress granules</b> Ishiura, Shoichi (Tokyo, Japan)	<b>11:45 – 12:00</b>
<b>S2-14</b>	<b>Abnormal secretion of prostaglandin E2 inhibits differentiation of congenital myotonic dystrophy muscle cells</b> Furling, Denis (Paris, France)	<b>12:00 – 12:15</b>
<b>S2-15</b>	<b>Quantitative semi-automated expanded CUG repeats assessment: application to the DM-SXL mouse.</b> Bassez, Guillaume (Paris, France)	<b>12:15 – 12:30</b>
	<b><i>Lunch / Poster Viewing with Presenters (odd numbers)/ Exhibition</i></b>	<b>12:30 – 14:00</b>
<b>P1</b>	<b>Poster Session 1: Basic Research—Part 1</b> Chairs: Müller-Reible, Clemens R. (Würzburg, Germany) Pearson, Christopher (Toronto, Canada)	<b>13:00 – 14:00</b>
<b>P2</b>	<b>Poster Session 2: Basic Research—Part 2</b> Chairs: Timchenko, Lubov (Houston, USA) Thornton, Charles (Rochester, USA)	<b>13:00 – 14:00</b>
<b>P3</b>	<b>Poster Session 3: Clinical Issues—Part 1</b> Chairs: Van Engelen, Baziel (Nijmegen, The Netherlands) Eymard, Bruno (Paris, France)	<b>13:00 – 14:00</b>
<b>P4</b>	<b>Poster Session 3: Clinical Issues—Part 2</b>	<b>13:00 – 14:00</b>

Chairs: Udd, Bjarne (Helsinki; Finland)  
Reiners, Karlheinz (Würzburg, Germany)

<b>P5</b>	<b>Poster Session 4: Translation and Therapy</b>	<b>13:00 – 14:00</b>
	Chairs: Ishiura, Shoichi (Tokyo, Japan) Schneider-Gold, Christiane (Bochum, Germany)	
<b>S3</b>	<b>Pathomechanisms in DM—Part 2</b>	<b>14:00 – 16:15</b>
	Chairs: Gourdon, Genevieve (Paris, France) Wieringa, Be (Nijmegen, The Netherlands)	
<b>S3-01</b>	<b>Investigating cellular and molecular abnormalities in the central nervous system of mice carrying large CTG repeat expansions</b>	<b>14:00 – 14:15</b>
	Gomes-Pereira, Mario (Paris, France)	
<b>S3-02</b>	<b>An unusual family co-segregating myotonic dystrophy type 1 and Charcot-Marie-Tooth disease present with an imperfect CTG repeat allele at the DM1 locus</b>	<b>14:15 – 14:30</b>
	Braida, Claudia (Glasgow, UK)	
<b>S3-03</b>	<b>De novo base substitution mutations at the myotonic dystrophy type 1 locus</b>	<b>14:30 – 14:45</b>
	Couto, Jillian (Glasgow, UK)	
<b>S3-04</b>	<b>Molecular pathophysiology and CNS effects in mouse models of myotonic dystrophy types 1 &amp; 2</b>	<b>14:45 – 15:00</b>
	Kang, Yuan-Lin (Minneapolis, USA)	
<b>S3-05</b>	<b>A novel role for the protein RFDM in restoring fusion of fetal DM1 muscle satellite cells</b>	<b>15:00 – 15:15</b>
	Pelletier, Richard (Quebec, Canada)	
<b>S3-06</b>	<b>The role of microRNAs in the regulation of gene expression in Myotonic Dystrophy</b>	<b>15:15 – 15:30</b>
	Timchenko, Lubov (Houston, USA)	
<b>S3-07</b>	<b>Study on human MBNL1 RNA binding properties</b>	<b>15:30 – 15:45</b>
	Vautrin, Audrey (Vandoeuvre-Les-Nancy, France)	
<b>S3-08</b>	<b>Testing the role of CUGBP1 in skeletal muscle wasting in DM1</b>	<b>15:45 – 16:00</b>
	Ward, Amanda (Houston, USA)	
<b>S3-09</b>	<b>Accumulation of Mutant DM2 (CCUG)<sub>n</sub> Transcripts Triggers Activation of PKR and ER Stress Response</b>	<b>16:00 – 16:15</b>
	Wojciechowska, Marzena (Houston, USA)	
	<b><i>Coffee break / Poster Viewing / Exhibition</i></b>	<b>16:15 – 16:45</b>
<b>S4</b>	<b>Models for DM</b>	<b>16:45 – 19:15</b>
	Chairs: Maurice Swanson, Maurice (Gainesville, USA) Mahadevan, Mani (Charlottesville, USA)	
<b>S4-01</b>	<b>Animal models for DM</b>	<b>16:45 – 17:15</b>
	Cooper, Tom (Houston, USA)	
<b>S4-02</b>	<b>Muscleblind Loss-of-Function Models for Myotonic Dystrophy</b>	<b>17:15 – 17:30</b>
	Swanson, Maurice (Gainesville, USA)	
<b>S4-03</b>	<b>Insights from mouse models of RNA toxicity</b>	<b>17:30 – 17:45</b>

Mahadevan, Mani (Charlottesville, USA)

- S4-04 Mouse Models for Myotonic Dystrophy Type 2 (DM2) Lacking Aberrant Splicing Implicate Novel Cytoplasmic Pathomechanisms** 17:45 – 18:00  
Krahe, Ralf (Houston, USA)
- S4-05 CCUG RNA gain-of-function effects in a conditional mouse model of myotonic dystrophy type 2 (DM2)** 18:00 – 18:15  
Margolis, Jamie M. (Minneapolis, USA)
- S4-06 Mouse model of congenital myotonic dystrophy type 1** 18:15 – 18:30  
Srinivasan, Varadamurthy (Charlottesville, USA)
- S4-07 A Drosophila model for Myotonic Dystrophy Type 2 (DM2)** 18:30 – 18:45  
Bergmann, Andreas (Houston, USA)
- S4-08 Towards a Zebrafish Model of Myotonic Dystrophy** 18:45 – 19:00  
Todd, Peter (Michigan, USA)
- S4-09 Toxicity of noncoding CUG and CCUG repeats** 19:00 – 19:15  
Timchenko, Lubov (Houston, USA)

## Friday, September 11, 2009

### Lecture Hall 1 (HS216, Audimax)

- S5 Clinical Issues in DM** 08:00 – 12:00  
Chairs: Eymard, Bruno (Paris, France)  
Meola, Giovanni (Milan, Italy)
- S5-01 Unusual presentations and a large proportion of mild phenotypes expand the spectrum and epidemiology of DM2** 08:00 – 08:20  
Udd, Bjarne, (Tampere, Finland)
- S5-02 Brain imaging in DM** 08:20 – 08:40  
Day, John (Minneapolis, USA)
- S5-03 Congenital myotonic dystrophy: Canadian surveillance and cohort study** 08:40 – 08:55  
Campbell, Craig (London, Canada)
- S5-04 Spectrum of disease manifestations of juvenile myotonic dystrophy type 1 (JDM) patients** 08:55 – 09:10  
Luebbe, Elizabeth (Rochester, USA)
- S5-05 Visual function in Congenital and Childhood Myotonic Dystrophy Type 1** 09:10 – 09:25  
Ekström, Anne-Berit (Gothenburg, Sweden)
- S5-06 Sleep-Disordered Breathing in a cohort of 40 Italian Steinert's Dystrophy patients. A clinical and polisomnographic study** 09:25 – 09:40  
Morandi, Lucia (Milan, Italy)
- S5-07 Cerebral white matter affection in myotonic dystrophy type 1 and 2 - A Diffusion-Tensor-Imaging Study at 3T** 09:40 – 09:55  
Minnerop, Martina (Juelich, Germany)
- Coffee break / Poster Viewing / Exhibition** 09:55 – 10:30

<b>S5-08</b>	<b>White Matter Microstructural Abnormalities in DM Observed with Diffusion Tensor Imaging</b> Franc, Daniel (Minneapolis, USA)	<b>10:30 – 10:45</b>
<b>S5-09</b>	<b>Depression in Myotonic dystrophy type 1: clinical and neuronal correlates</b> Lindberg, Christopher (Göteborg, Sweden)	<b>10:45 – 11:00</b>
<b>S5-10</b>	<b>French registry for myotonic dystrophies (DM1-DM2): toward the characterization of a large DM population</b> Guiraud-Dogan, Céline (Créteil, France)	<b>11:00 – 11:15</b>
<b>S5-11</b>	<b>Efficacy And Limitation Of Group Exercise For Swallowing Training In Myotonic Dystrophy</b> Saito, Akiko (Aomori, Japan)	<b>11:15 – 11:30</b>
<b>S5-12</b>	<b>General anesthesia in myotonic dystrophy type 2</b> Kirzinger, Lukas (Munich, Germany)	<b>11:30 – 11:45</b>
<b>S5-13</b>	<b>Double genetic trouble (DM2/FSHD) in a Sardinian DM2/PROMM family</b> Meola, Giovanni (Milan, Italy)	<b>11:45 – 12:00</b>
	<b><i>Lunch / Poster Viewing with Presenters (even numbers)/ Exhibition</i></b>	<b>12:00 – 14:00</b>
<b>P1</b>	<b>Poster Session 1: Basic Research—Part 1</b> Chairs: Müller-Reible, Clemens R. (Würzburg, Germany) Pearson, Christopher (Toronto, Canada)	<b>13:00 – 14:00</b>
<b>P2</b>	<b>Poster Session 2: Basic Research—Part 2</b> Chairs: Timchenko, Lubov (Houston, USA) Thornton, Charles (Rochester, USA)	<b>13:00 – 14:00</b>
<b>P3</b>	<b>Poster Session 3: Clinical Issues—Part 1</b> Chairs: Van Engelen, Baziel (Nijmegen) Eymard, Bruno (Paris, France)	<b>13:00 – 14:00</b>
<b>P4</b>	<b>Poster Session 3: Clinical issues—Part 2</b> Chairs: Udd, Bjarne (Helsinki; Finland) Reiners, Karlheinz (Würzburg, Germany)	<b>13:00 – 14:00</b>
<b>P5</b>	<b>Poster Session 4: Translation and Therapy</b> Chairs: Ishiura, Shoichi (Tokyo, Japan) Schneider-Gold, Christiane (Bochum, Germany)	<b>13:00 – 14:00</b>
<b>S6</b>	<b>TREAT-NMD / Marigold Foundation Working group Summaries</b>	<b>14:00 – 15:00</b>
<b>S7</b>	<b>Keynote Lecture</b> Chairs: Ashizawa, Tetsuo (Gainesville, USA) Krahe, Ralf (Houston, USA)	
<b>S7-1</b>	<b>Developing treatment for hereditary neuromuscular disease</b> Fischbeck, Kenneth H. (Bethesda, USA)	<b>15:00 – 16:00</b>
	<b><i>Coffee break / Poster Viewing / Exhibition</i></b>	<b>16:00 – 16:30</b>
	<b><i>Walk to the Residence</i></b>	<b>16:30 – 16:50</b>

**Visiting the Residence** 17:00 – 18:45

*Lift available; admission by ticket only.  
Pre-booking at registration requested*

**Winetasting Reception at the Cellar of the Residence** 18:45 – 23:00

*Meeting point at the fountain before the Residence.  
Lift available; admission by ticket only.  
Pre-booking at registration requested*

## Saturday, September 12, 2009

Lecture Hall 1 (HS216, Audimax) (deutsche Simultanübersetzung)

**S8 Molecular and symptomatic therapy** 08:30 – 10:30  
Chairs: Puymirat, Jack (Quebec, Canada)  
Toyka, Klaus (Würzburg, Germany)

**S8-01 Molecular therapy for myotonic dystrophy: current status and future strategies** 08:30 – 09:00  
Wansink, Derrick G. (Nijmegen, The Netherlands)

**S8-02 IPLEX (rhIGF-I/rhIGFBP-3) Treatment of Myotonic Dystrophy Type-1 (DM1): A Safety and Tolerability Trial** 09:00 – 09:15  
Heatwole, Chad (Rochester, USA)

**S8-03 Reversal of myotonia and splicing defects by antisense oligomers in a transgenic mouse model of myotonic dystrophy type 1 (DM1)** 09:15 – 09:30  
Wheeler, Thurman (Rochester, USA)

**S8-04 Development of therapies against RNA toxicity in DM1** 09:30 – 09:45  
Yadava, Rahmesh (Charlottesville, USA)

**S8-05 Pentamidine reverses splicing defects associated with Myotonic Dystrophy Type 1 (DM1)** 09:45 – 10:00  
Warf, M. Bryan (Eugene, USA)

**S8-06 Chemically modified (CAG)<sub>n</sub> antisense oligonucleotides as molecular tools to silence toxic, expanded DMPK transcripts** 10:00 – 10:15  
Mulders, Susan (Nijmegen, The Netherlands)

**S8-07 Assays to screen for drugs to treat myotonic dystrophy** 10:15 – 10:30  
Brook, J David (Nottingham, UK)

**Coffee break / Poster Viewing / Exhibition** 10:30 – 11:00

**S9 Heart and DM** 11:00 – 12:30  
Chairs: Ertl, Georg (Würzburg, Germany)  
Duboc, Denis (Paris, France)

**S9-01 Clinical Aspects of Cardiac Involvement in Myotonic Dystrophy: Current Knowledge and Future Directions** 11:00 – 11:15  
Groh, William J. (Indianapolis, USA)

**S9-02 Is Cardiac Involvement in Adult Survivors of Congenital- or Childhood-Onset DM1 Different than in Classical Adult-Onset DM1?** 11:15 – 11:30

Groh, William J. (Indianapolis, USA)

- S9-03 Cardiac abnormalities in Congenital Childhood Myotonic muscular dystrophy(DM1)** 11:30 – 11:45  
Mishra , Shri (Los Angeles, USA)
- S9-04 High Prevalence of Brugada Syndrome in Patients with Steinert's Disease. A New Insight in the Pathophysiology of Arrhythmias in Steinert's Disease** 11:45 – 12:00  
Wahbi, Karim (Paris, France)
- S9-05 Protein Kinase C inhibition ameliorates the cardiac phenotype of a mouse model for myotonic dystrophy type 1** 12:00 – 12:15  
Kuyumcu-Martinez, Muge N. (Houston, USA)
- S9-06 The progression of Muscular Impairment Rating Scale (MIRS) and the development of cardiac conduction abnormalities in DM1** 12:15 – 12:30  
Antonini, Giovanni (Rome, Italy)

### Lecture Hall 2 (HS224)

- M1 Patient Session in German / Vorträge für Patienten und Angehörige auf Deutsch** 08:30– 12:30
- M1-1 Einführung** 08:30 – 09:00  
Schoser, Benedikt (München)
- M1-2 Klinische Symptome der DM1 und DM2** 09:00 – 09:45  
(klinischer Verlauf, CDM, Herz, Auge, Hirn, Muskel, endokrine Organe, Immunsystem)  
Schneider-Gold, Christiane (Bochum)
- M1-3 Grundlagen und Diagnose der DM1 und DM2** (Ursache, Genetik, Beratung, Pränataldiagnostik) 09:45 – 10:30  
Kress, Wolfram (Würzburg), Grimm, Tiemo (Würzburg)
- Kaffeepause** 10:30 – 11:00
- M1-4 Komplikationen und Behandlungsmöglichkeiten der DM1 und DM2** (Herz, Müdigkeit, Schmerzen, Operationen, Darm) 11:00 – 11:45  
Schoser, Benedikt (München)
- M1-5 Berichte von Patienten und Beantwortung von Fragen. Wie geht es weiter?** 11.45 – 12.30

### Lecture Hall 3 (HS210)

- M2 Patient Session in English** 08:30 – 12:30  
**Organized by Moxley III, Richard (Rochester, USA)**
- Lunch / Poster Viewing / Exhibition** 12:30 – 13:30

### Lecture Hall 1 (HS216, Audimax) (deutsche Simultanübersetzung)

- S10 Genetic counseling** 13:30 – 14:50  
Chairs: Grimm, Tiemo (Würzburg, Germany)  
Schoser, Benedikt (Munich, Germany)

<b>S10-1</b>	<b>Genetic counseling in DM1</b> Rogers, Mark (Cardiff, UK)	<b>13:30 – 14:10</b>
<b>S10-2</b>	<b>Genetic counseling in DM2</b> Kress, Wolfram (Würzburg, Germany)	<b>14:10 – 14:45</b>
	<b>Coffee break / Poster Viewing / Exhibition</b>	<b>14:45 – 15:30</b>
<b>S11</b>	<b>Messages from the patient organizations</b> Chairs: Moxley III, Richard (Rochester, USA) Monckton, Darren G (Glasgow, UK)	<b>15:30 – 16:45</b>
<b>S11-1</b>	<b>Awards (award committee)</b>	<b>15:30 – 16:00</b>
<b>S11-2</b>	<b>Presentation of the patients organizations</b>	<b>16:00 – 16:45</b>
<b>S12</b>	<b>Conference Highlights</b> Chairs: Moxley III, Richard (Rochester, USA) Monckton, Darren G (Glasgow, UK)	<b>16:45 – 18:00</b>
<b>S12-1</b>	<b>Late-breaking Session</b>	<b>16:45 – 17:15</b>
<b>S12-2</b>	<b>Conference Highlights and Concluding Remarks</b> Wierenga, Be (Nijmegen, The Netherlands) Ashizawa, Tetsuo (Gainesville, USA) Brook, David (Nottingham, UK) Thornton, Charles (Rochester, USA)  Krahe, Ralf (Houston, USA) Grimm, Tiemo (Würzburg, Germany) Schooser, Benedikt (München)	<b>17:15 – 18:00</b>
	<b>Farewell-Dinner Riverboat Cruise on the Main River</b>	<b>19:00 – 23:00</b>
	<i>Meeting point: "Alter Kranen - The Old Crane" at the river Main; admission by ticket only. Pre-booking requested</i>	

## Poster Sessions

**Thursday, September 10, 2009 (odd numbers) and  
Friday, September 11, 2009 (even numbers)  
13.00 - 14.00 h, Foyer of the lecture hall**

### **P1 Poster Session 1: Basic Research—Part 1**

Chairs: Müller-Reible, Clemens R. (Würzburg, Germany)  
Pearson, Christopher (Toronto, Canada)

**P1-01 Searching for trans-acting genetic modifiers of somatic mosaicism and disease severity in myotonic dystrophy type 1**

Fernando Morales (San José)

**P1-02 CTCF Induces Replication Fork Pausing Around DM1 Repeats**

Katharine A. Hagerman (Ontario)

**P1-03 Study of DM1 associated RNAs using atomic force microscopy (AFM)**

Francois Meullenet (Nottingham)

**P1-04 Disease-associated trinucleotide repeats form transcription-induced RNA:DNA hybrids**

K. Reddy (Ontario)

- P1-05 CpG methylation proximal to the CTG/CAG tract of the DM1 locus in patients and transgenic mice**  
A. López Castel (Ontario)
- P1-06 Tissue-specificity of trinucleotide repeat instability and DNA replication in myotonic dystrophy type 1**  
JD Cleary (Ontario)
- P1-07 Studies of the distribution of Muscleblind-like proteins in myotonic dystrophy.**  
J David Brook (Nottingham)
- P1-08 A bi-chromatic fluorescent assay to measure splicing efficiency in Myotonic Dystrophy.**  
Javier T Granados-Riveron (Nottingham)
- P1-09 Validation of sensitivity and specificity of Tetraplet-Primed PCR (TP-PCR) in the molecular diagnosis of for Myotonic Dystrophy type 2**  
Claudio Catalli (Rome)
- P1-10 Effect of RNAi directed against CUG repeats in a mouse model of DM1**  
Krzysztof Sobczak (Poznan)
- P1-11 DNA methylation at the DM1 Locus**  
John Day (Minneapolis)
- P1-12 DMSXL mice carrying over 1000 CTG: characterization of the muscle function**  
Fadia Medja (Paris)
- P1-13 The splicing of MBNL1 is altered in DM1 muscles**  
Camille Lemercier (Paris)
- P1-14 Premature activation of the p16 stress pathway in congenital DM1 myoblasts**  
Erwan Gasnier (Paris)
- P1-15 The effort to obtain longer CTG triplet repeat DNA by using *Saccharomyces cerevisiae***  
Noboru Sasagawa (Tokyo)
- P1-16 Altered splicing of CAMKIID in brain from patients with myotonic dystrophy type 1**  
Koichi Suenaga (Nishinomiya)
- P1-17 Normal myogenesis and increased apoptosis in myotonic dystrophy type 1 muscle cells.**  
Emanuele Loro (Padova)
- P1-18 Altered mRNA splicing of the MYH14 gene in the skeletal muscle of myotonic dystrophy type 1 patients**  
Fabrizio Rinaldi (Rome)
- P1-19 The role of Twist in DM1**  
Andrie Koutsoulidou (Nicosia)
- P1-20 Cytoplasmic export of DM1 transcripts benefits muscle cell differentiation**  
Nikolaos Mastrogiannopoulos (Nicosia)
- P1-21 Cell model for repeat instability and senescence in DM1**  
Masayuki Nakamori (Rochester)
- P1-22 Mis-splicing of microtubules-associated tau exon 10 is associated to a CELF proteins gain of function but not to a MBNL1 loss of function**  
Claire-Marie Dhaenens (Lille)
- P1-23 Specific micro-RNA processing alteration in DM**  
Frédérique Rau (Illkirch)

## **P2 Poster Session 2: Basic Research—Part 2**

Chairs: Timchenko, Lubov (Houston, USA)  
Thornton, Charles (Rochester, USA)

**P2-01 The role of microRNAs in the regulation of gene expression in Myotonic Dystrophy**  
Lubov Timchenko (Houston)

**P2-02 Abnormal expression of ZNF9 in myotonic dystrophy type 2 (DM2)**  
Olayinka Raheem (Tampere)

**P2-03 Proteomic analysis of DM2 human myotubes reveals alterations in mitochondrial components, in the unfolded protein response and in the ubiquitin proteasome system**  
Francesco S. Rusconi (Milan)

**P2-04 Progressions of (CTG) n expansions, muscular disability rating scale (MDRS), and abnormal glucose metabolism are age dependent in myotonic dystrophy type 1 (DM1)**  
Masanobu Kinoshita (Tokyo)

**P2-05 High-throughput screening to identify modulators of aberrant splicing in DM1**  
Debra O'Leary (San Diego)

**P2-06 Subcellular localization of Drosophila Muscleblind changes from preferentially nuclear to cytoplasmic during muscle development**  
M. Beatriz Jiamusi Trios (Burgesses-Valencia)

**P2-07 DMPK-interacting proteins**  
Sergio Salvatore (Padova)

**P2-08 Transcriptional defects in DM1 result from nuclear exclusion of SHARP**  
Sita Reddy (Los Angeles)

**P2-09 Autoregulation of MBNL1: coupling of splicing regulation and intracellular localization**  
Yoshihiro Kino (Saitama)

**P2-10 Muscleblind-like proteins in normal and myotonic dystrophy muscle and their role in rapid diagnostic testing.**  
Ian Holt (Shropshire)

**P2-11 The mechanisms of MBNL1 regulated splicing**  
Andy Berglund (Eugene)

**P2-12 What are RNA foci? Interactions of the mutant DMPK mRNA.**  
Shagufta Rehman (Charlottesville)

**P2-13 Abnormal splicing of myomesin in DM muscle**  
Michinori Koebis (Tokyo)

**P2-14 Mathematical models of dynamic DNA in myotonic dystrophy**  
Catherine Higham (Glasgow)

**P2-15 Aberrant expression of microRNA in myotonic dystrophies**  
Riccardo Perbellini (Milan)

**P2-16 Abnormally expressed genes in statin induced myopathy appear dysregulated in myotonic dystrophy type 2.**  
Mark Screen (Helsinki)

**P2-17 Use of human embryonic stem cells as new model to decipher early pathological events involved in Myotonic Dystrophy type 1.**  
Jérôme Denis (Evry)

**P2-18 Altered isoform usage for MADS-domain transcription enhancer factor 2 (MEF2) in myotonic dystrophy and other neuromuscular disorders**  
Linda Bachinski (Houston)

**P2-19 Effects of mexiletine on cardiac parameters, muscles strength and myotonia in myotonic dystrophy type 1**  
Marta Panzeri (Milan)

**P2-20 Non-radioactive detection of repeat expansions in DMPK and ZNF9 genes**

Martina Witsch-Baumgartner (Innsbruck)

**P2-21 Global expression profiling of Myotonic Dystrophy type 1 (DM1) and type 2 (DM2) identifies novel effector genes and cellular pathways**

Mario Sirito (Houston)

**P2-22 New Technique for Rapid and Reliable Analysis of Trinucleotide Repeats in Myotonic Dystrophy Type 1.**

Skrzypczak-Zielinska M.<sup>1</sup>, Sulek-Piatkowska A.<sup>2</sup>, Froster U. G.<sup>1</sup> (Leipzig, Germany)

## **P3 Poster Session 3: Clinical Issues—Part 1**

Chairs: Van Engelen, Baziél (Nijmegen)  
Eymard, Bruno (Paris, France)

**P3-01 Myotonic dystrophy type 2 (DM2) in Italy: spectrum of clinical and laboratory findings**

Marta Panzeri (Milan)

**P3-02 Risk of arrhythmia in type I Myotonic Dystrophy: the role of clinical and genetic variables**

Paola Cudia (Venice)

**P3-03 CLCN1 mutations screening in Italian patients affected by myotonic dystrophy type 2 (DM2)**

Saverio Massimo Lepore (Rome)

**P3-04 The correlation between oral dysfunction and videofluoroscopic swallowing findings in myotonic dystrophy type 1(DM1)**

Hirokazu Furuya (Fukuoka)

**P3-05 Frequency of DM1 and DM2 in Germany**

Tiemo Grimm (Würzburg)

**P3-06 Hearing evaluation in DM2: a prospective study of 10 Patients**

Bruno Eymard (Paris)

**P3-07 White matter pathology and neurocognitive correlates in adolescents with myotonic dystrophy type 1: A Diffusion Tensor Imaging study**

Jeffrey Wozniak (Minneapolis)

**P3-08 Repetitive components of compound motor action potential in DM1 patients.**

Anna Modoni (Rome)

**P3-09 Muscle pathological changes and brain MRI findings in DM1**

Chiara Ferrati (Padova)

**P3-10 Subtle cognitive decline in Myotonic dystrophy type 1: a five-year follow up study**

Stefan Winblad (Göteborg)

**P3-11 Test/retest and machine/machine reliability of Dual Energy X-ray Absorptiometry (DEXA) measurements in patients with DM-1 and DM-2.**

Shree Pandya (Rochester)

**P3-12 Participation in physical activity by people with myotonic dystrophy**

Margaret Phillips (Derby)

**P3-13 Clinical and biomolecular findings in a juvenile onset case of myotonic dystrophy type 2**

Marzia Giagnacovo (Milan)

**P3-14 Health-related quality of life in patients with myotonic dystrophy type 1 and myasthenia gravis: a comparative analysis**

Vidosava Rakocevic Stojanovic (Belgrade)

**P3-15 Development of Scottish Myotonic Dystrophy management guidelines**

Cheryl Longman (Glasgow)

**P3-16 Frequency of DM2 and DM1 mutations in the Finnish population**

Tiina Suominen (Tampere)

**P3-17 Lipid metabolism alteration in myotonic dystrophies**

Anja Schmidt (Munich)

## **P4 Poster Session 4: Clinical Issues—Part 2**

Chairs. Udd, Bjarne (Helsinki; Finland)

Reiners, Karlheinz (Würzburg, Germany)

**P4-01 Clinical, muscle pathology and FISH biomolecular findings correlation in 42 Italian patients with myotonic dystrophy type 2.**

Rosanna Cardani (Milan)

**P4-02 Motor outcome measures in childhood and congenital DM1**

Craig Campbell (London)

**P4-03 Structural and functional brain abnormalities in myotonic dystrophy type 1 and 2**

Robert Roebeling (Ulm)

**P4-04 The lived experience of patients with Myotonic Dystrophy Type 1**

Maud-Christine Chouinard (Jonquiere)

**P4-05 Quality of life and family impact of congenital and childhood DM1**

Craig Campbell (London)

**P4-06 Health supervision in Myotonic Dystrophy Type 1**

Cynthia Gagnon (Jonquière)

**P4-07 Chronic muscle stimulation reverts the abnormal sEMG pattern in Myotonic Dystrophy type 1**

Carmelo Chisari (Pisa)

**P4-08 Diagnostic odyssey of myotonic dystrophy type 2 (DM2) patients**

James Hilbert (Rochester)

**P4-09 Ocular motor function in congenital and childhood Myotonic Dystrophy Type 1**

Eva Aring (Göteborg)

**P4-10 High impact symptoms in Myotonic Dystrophy Type-1 (DM1): A qualitative study**

Chad Heatwole (Rochester)

**P4-11 Scaled down genetic analysis of myotonic dystrophy type 1 and type 2**

Masayuki Nakamori (Rochester)

**P4-12 Decision-making dysfunction in DM1**

Nathalie Angeard (Paris)

**P4-13 Hard to swallow: understanding the lived experience of Caregivers for individuals with Myotonic Dystrophy (DM1) and dysphagia**

Kori LaDonna (London)

**P4-14 Quantitative isometric muscle strength at the ankle in myotonic dystrophy type 1.**

Jack Puymirat (Quebec)

**P4-15 Gait and balance difficulties in individuals with Myotonic Dystrophy type 1.**

Elisabet Hammarén (Göteborg)

**P4-16 No evidence of specific postprandial hyperlipidemia in Myotonic Dystrophy.**

Hiroto Takada (Aomori)

**P4-17 Characteristic features of oral and dental health in myotonic dystrophy**

T. Ishida Aomori, Japan)

## **P5 Poster Session 5: Translation and therapy**

Chairs: Ishiura, Shoichi (Tokyo, Japan)  
Schneider-Gold, Christiane (Bochum, Germany)

**P5-01 Perceptions of professional, lay, and peer facilitators goal-setting and strategies used to promote social support and self-management behavior in face-to-face and online support groups for adults with either Multiple Sclerosis or Myotonic Muscular Dystrophy**

Leslie Krongold (Alameda)

**P5-02 The lived experience of DM1 patients caregivers**

Maud-Christine Chouinard (Jonquière)

**P5-03 Living with Myotonic Dystrophy Type 1 (DM1) Sufferers: How caregivers' experience differs according to gender**

Claudia Bouchard (Jonquière)

**P5-04 Adapting and validating the Stanford Self-Management Program for people with DM1: preliminary results and lessons learned**

Cynthia Gagnon (Jonquière)

**P5-05 Quality of life in myotonic dystrophy type 1**

Eric Gagnon (Jonquiere)

**P5-06 Functioning, disability and health-related quality of life in adults with Myotonic dystrophy type 1 (DM1)**

Marie Kierkegaard (Stockholm)

**P5-07 Myotonic Dystrophy - A Scottish Perspective**

Anne Marie Taylor (Dundee)

**P5-08 Role of oro-pharyngo-oesophageal scintigraphy in the evaluation of swallowing disorders in patients with Myotonic dystrophy type 1 (DM1)**

Venanzio Valenza (Rome)

**P5-09 Myotonic dystrophy: a service improvement survey of quality of life, social integration and community support systems for patients and caregivers**

Margaret Phillips (Derby)

**P5-10 In vivo drug screening of 170 natural compounds in a DM1 fly model**

Irma Garcia-Alcover (Paterna)

**P5-11 Systemic delivery of antisense morpholino corrects CIC-1 splicing and reduces myotonia in a transgenic mouse model of DM1**

Thurman Wheeler (Rochester)

**P5-12 Pluripotent stem cells to explore mechanisms and treatments of monogenic diseases**

Cécile Martinat (Evry)

**P5-13 RNA-based gene therapy to remove toxic expanded CUG-transcripts**

Denis Furling (Paris)

**P5-14 High Throughput Screen Assays for Identifying Inhibitors of Protein-RNA Binding As a Potential Treatment for Myotonic Dystrophy Type-1**

Catherine Chen (Bethesda)

**P5-15 Test/retest reliability of regional lean body mass (LBM) measurements using Dual Energy X-ray Absorptiometry (DEXA) in patients with DM1**

Shree Pandya (Rochester)

# Exhibitors and Sponsors

(Status at printing)

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(List in alphabetical order):

**AFM - Association Francaise contre les Myopathies, Paris, France**

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