Welcome to the 7th 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 100th 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 biannual 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                             Tiemo Grimm                             Benedikt Schoser
Houston, USA                          Würzburg, Germany               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.

Welcoming Address

100 Years of Myotonic Dystrophy and Steinert Disease

7th International Myotonic Dystrophy Consortium Meeting in Würzburg, September 9-12, 2009

I cordially welcome all participants of the 7th 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.

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

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Brigitte Wolf
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Congress Organisation
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www.carlo-praetorius-de

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 vauling 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.

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
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

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, Hélène (Lille, France)

S2-04 A (CCUG)n-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
<table>
<thead>
<tr>
<th>Session</th>
<th>Title</th>
<th>Presenter</th>
<th>Time</th>
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<tr>
<td>S2-07</td>
<td>Identification of novel candidates that intervene in Myotonic Dystrophy Type 1 (DM1)</td>
<td>Francois-Xavier Laurent (Gif sur Yvette, France)</td>
<td>09:45 – 10:00</td>
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<td>S2-08</td>
<td>The DMSXL transgenic mice carrying very large expansions exhibit molecular and physiological defects: an animal model for gene therapy experiments?</td>
<td>Gourdon, Geneviève (Paris, France)</td>
<td>10:30 – 10:45</td>
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<td>S2-09</td>
<td>A second example of RNA-gain-of-function disease: Sam68 and MBNL1 sequestration by CGG repeats in Fragile X Tremor Ataxia Syndrome</td>
<td>Sellier, Chantal (Illkirch, France)</td>
<td>10:45 – 11:00</td>
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<td>S2-10</td>
<td>Development of a muscleblind-like 1 (Mbnl1) overexpression model to assess the use of MBNL1 as a potential therapeutic for myotonic dystrophy</td>
<td>Chamberlain, Christopher (Minneapolis, USA)</td>
<td>11:00 – 11:15</td>
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<td>S2-11</td>
<td>Misregulation of diacylglycerol kinase eta (DGKη) splicing as a potential cause of neuropsychiatric symptoms in myotonic dystrophy type 1</td>
<td>Matsuura, Tohru (Okayama, Japan)</td>
<td>11:15 – 11:30</td>
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<td>S2-12</td>
<td>Study on the mechanisms of regulation and deregulation of hcTNT exon 5 inclusion</td>
<td>Behm-Ansmant, Isabelle (Vandoeuvre-les-Nancy, France)</td>
<td>11:30 – 11:45</td>
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<td>S2-13</td>
<td>MBNL1 associates with YB-1 in cytoplasmic stress granules</td>
<td>Ishiura, Shoichi (Tokyo, Japan)</td>
<td>11:45 – 12:00</td>
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<td>S2-14</td>
<td>Abnormal secretion of prostaglandin E2 inhibits differentiation of congenital myotonic dystrophy muscle cells</td>
<td>Furling, Denis (Paris, France)</td>
<td>12:00 – 12:15</td>
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<td>P1</td>
<td>Poster Session 1: Basic Research—Part 1</td>
<td>Müller-Reible, Clemens R. (Würzburg, Germany)</td>
<td>13:00 – 14:00</td>
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<td>Pearson, Christopher (Toronto, Canada)</td>
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<td>P2</td>
<td>Poster Session 2: Basic Research—Part 2</td>
<td>Timchenko, Lubov (Houston, USA)</td>
<td>13:00 – 14:00</td>
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<td>Thornton, Charles (Rochester, USA)</td>
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<td>P3</td>
<td>Poster Session 3: Clinical Issues—Part 1</td>
<td>Van Engelen, Baziel (Nijmegen, The Netherlands)</td>
<td>13:00 – 14:00</td>
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<td>Eymard, Bruno (Paris, France)</td>
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<td>P4</td>
<td>Poster Session 3: Clinical Issues—Part 2</td>
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<td>13:00 – 14:00</td>
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Poster Session 4: Translation and Therapy

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

Pathomechanisms in DM—Part 2

Chairs: Gourdon, Genevieve (Paris, France)
        Wieringa, Be (Nijmegen, The Netherlands)

S3-01 Investigating cellular and molecular abnormalities in the central nervous system of mice carrying large CTG repeat expansions
Gomes-Pereira, Mario (Paris, France)

S3-02 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
Braida, Claudia (Glasgow, UK)

S3-03 De novo base substitution mutations at the myotonic dystrophy type 1 locus
Couto, Jillian (Glasgow, UK)

S3-04 Molecular pathophysiology and CNS effects in mouse models of myotonic dystrophy types 1 & 2
Kang, Yuan-Lin (Minneapolis, USA)

S3-05 A novel role for the protein RFDM in restoring fusion of fetal DM1 muscle satellite cells
Pelletier, Richard (Quebec, Canada)

S3-06 The role of microRNAs in the regulation of gene expression in Myotonic Dystrophy
Timchenko, Lubov (Houston, USA)

S3-07 Study on human MBNL1 RNA binding properties
Vautrin, Audrey (Vandoeuvre-Les-Nancy, France)

S3-08 Testing the role of CUGBP1 in skeletal muscle wasting in DM1
Ward, Amanda (Houston, USA)

S3-09 Accumulation of Mutant DM2 (CCUG)n Transcripts Triggers Activation of PKR and ER Stress Response
Wojciechowska, Marzena (Houston, USA)

Coffee break / Poster Viewing / Exhibition

Models for DM

Chairs: Maurice Swanson, Maurice (Gainesville, USA)
        Mahadevan, Mani (Charlottesville, USA)

S4-01 Animal models for DM
Cooper, Tom (Houston, USA)

S4-02 Muscleblind Loss-of-Function Models for Myotonic Dystrophy
Swanson, Maurice (Gainesville, USA)

S4-03 Insights from mouse models of RNA toxicity
S4-04  
**Mouse Models for Myotonic Dystrophy Type 2 (DM2) Lacking Aberrant Splicing Implicate Novel Cytoplasmic Pathomechanisms**

Mahadevan, Mani (Charlottesville, USA)

17:45 – 18:00

S4-05  
**CCUG RNA gain-of-function effects in a conditional mouse model of myotonic dystrophy type 2 (DM2)**

Krahe, Ralf (Houston, USA)

18:00 – 18:15

S4-06  
**Mouse model of congenital myotonic dystrophy type 1**

Margolis, Jamie M. (Minneapolis, USA)

18:15 – 18:30

S4-07  
**A Drosophila model for Myotonic Dystrophy Type 2 (DM2)**

Srinivasan, Varadamurthy (Charlottesville, USA)

18:30 – 18:45

S4-08  
**Towards a Zebrafish Model of Myotonic Dystrophy**

Bergmann, Andreas (Houston, USA)

18:45 – 19:00

S4-09  
**Toxicity of noncoding CUG and CCUG repeats**

Timchenko, Lubov (Houston, USA)

19:00 – 19:15

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**Friday, September 11, 2009**

Lecture Hall 1 (HS216, Audimax)

S5  
**Clinical Issues in DM**

Chairs:  
Eymard, Bruno (Paris, France)
Meola, Giovanni (Milan, Italy)

08:00 – 12:00

S5-01  
**Unusual presentations and a large proportion of mild phenotypes expand the spectrum and epidemiology of DM2**

Udd, Bjarne, (Tampere, Finland)

08:00 – 08:20

S5-02  
**Brain imaging in DM**

Day, John (Minneapolis, USA)

08:20 – 08:40

S5-03  
**Congenital myotonic dystrophy: Canadian surveillance and cohort study**

Campbell, Craig (London, Canada)

08:40 – 08:55

S5-04  
**Spectrum of disease manifestations of juvenile myotonic dystrophy type 1 (JDM) patients**

Luebbe, Elizabeth (Rochester, USA)

08:55 – 09:10

S5-05  
**Visual function in Congenital and Childhood Myotonic Dystrophy Type 1**

Ekström, Anne-Berit (Gothenburg, Sweden)

09:10 – 09:25

S5-06  
**Sleep-Disordered Breathing in a cohort of 40 Italian Steinert's Dystrophy patients. A clinical and polysomnographic study**

Morandi, Lucia (Milan, Italy)

09:25 – 09:40

S5-07  
**Cerebral white matter affection in myotonic dystrophy type 1 and 2 - A Diffusion-Tensor-Imaging Study at 3T**

Minnerop, Martina (Juelich, Germany)

09:40 – 09:55

*Coffee break / Poster Viewing / Exhibition*  
09:55 – 10:30

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S5-08  White Matter Microstructural Abnormalities in DM Observed with Diffusion Tensor Imaging  
     Franc, Daniel (Minneapolis, USA)  
     10:30 – 10:45

S5-09  Depression in Myotonic dystrophy type 1: clinical and neuronal correlates  
     Lindberg, Christopher (Göteborg, Sweden)  
     10:45 – 11:00

S5-10  French registry for myotonic dystrophies (DM1-DM2): toward the characterization of a large DM population  
     Guiraud-Dogan, Céline (Créteil, France)  
     11:00 – 11:15

S5-11  Efficacy And Limitation Of Group Exercise For Swallowing Training In Myotonic Dystrophy  
     Saito, Akiko (Aomori, Japan)  
     11:15 – 11:30

S5-12  General anesthesia in myotonic dystrophy type 2  
     Kirzinger, Lukas (Munich, Germany)  
     11:30 – 11:45

S5-13  Double genetic trouble (DM2/FSHD) in a Sardinian DM2/PROMM family  
     Meola, Giovanni (Milan, Italy)  
     11:45 – 12:00

Lunch / Poster Viewing with Presenters (even numbers)/ Exhibition  
     12:00 – 14:00

P1  Poster Session 1: Basic Research—Part 1  
     Chairs:  Müller-Reible, Clemens R. (Würzburg, Germany)  
              Pearson, Christopher (Toronto, Canada)  
     13:00 – 14:00

P2  Poster Session 2: Basic Research—Part 2  
     Chairs:  Timchenko, Lubov (Houston, USA)  
              Thornton, Charles (Rochester, USA)  
     13:00 – 14:00

P3  Poster Session 3: Clinical Issues—Part 1  
     Chairs:  Van Engelen, Baziel (Nijmegen)  
              Eymard, Bruno (Paris, France)  
     13:00 – 14:00

P4  Poster Session 3: Clinical issues—Part 2  
     Chairs:  Udd, Bjarne (Helsinki; Finland)  
              Reiners, Karlheinz (Würzburg, Germany)  
     13:00 – 14:00

P5  Poster Session 4: Translation and Therapy  
     Chairs:  Ishiura, Shoichi (Tokyo, Japan)  
              Schneider-Gold, Christiane (Bochum, Germany)  
     13:00 – 14:00

S6  TREAT-NMD / Marigold Foundation Working group Summaries  
     14:00 – 15:00

S7  Keynote Lecture  
     Chairs:  Ashizawa, Tetsuo (Gainesville, USA)  
              Krahe, Ralf (Houston, USA)  
     15:00 – 16:00

S7-1  Developing treatment for hereditary neuromuscular disease  
     Fischbeck, Kenneth H. (Bethesda, USA)  
     Coffee break / Poster Viewing / Exhibition  
     16:00 – 16:30

Walk to the Residence  
     16:30 – 16:50
Visiting the Residence
Lift available; admission by ticket only.
Pre-booking at registration requested

Winetasting Reception at the Cellar of the Residence
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)n 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
S9-03  Cardiac abnormalities in Congenital Childhood Myotonic muscular dystrophy (DM1)  
Mishra, Shri (Los Angeles, USA)  
11:30 – 11:45

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  
Wahbi, Karim (Paris, France)  
11:45 – 12:00

S9-05  Protein Kinase C inhibition ameliorates the cardiac phenotype of a mouse model for myotonic dystrophy type 1  
Kuyumcu-Martinez, Muge N. (Houston, USA)  
12:00 – 12:15

S9-06  The progression of Muscular Impairment Rating Scale (MIRS) and the development of cardiac conduction abnormalities in DM1  
Antonini, Giovanni (Rome, Italy)  
12:15 – 12:30

Lecture Hall 2 (HS224)

M1  Patient Session in German / Vorträge für Patienten und Angehörige auf Deutsch  
08:30 – 12:30

M1-1  Einführung  
Schoser, Benedikt (München)  
08:30 – 09:00

M1-2  Klinische Symptome der DM1 und DM2  
(klinischer Verlauf, CDM, Herz, Auge, Hirn, Muskel, endokrine Organe, Immunsystem)  
Schneider-Gold, Christiane (Bochum)  
09:00 – 09:45

M1-3  Grundlagen und Diagnose der DM1 und DM2 (Ursache, Genetik, Beratung, Pränataldiagnostik)  
Kress, Wolfram (Würzburg), Grimm, Tiemo (Würzburg)  
09:45 – 10:30

Kaffeepause  
10:30 – 11:00

M1-4  Komplikationen und Behandlungsmöglichkeiten der DM1 und DM2 (Herz, Müdigkeit, Schmerzen, Operationen, Darm)  
Schoser, Benedikt (München)  
11:00 – 11:45

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  
Chairs: Grimm, Tiemo (Würzburg, Germany)  
Schoser, Benedikt (Munich, Germany)  
13:30 – 14:50
S10-1 Genetic counseling in DM1
Rogers, Mark (Cardiff, UK)  
13:30 – 14:10

S10-2 Genetic counseling in DM2
Kress, Wolfram (Würzburg, Germany)  
14:10 – 14:45

Coffee break / Poster Viewing / Exhibition  
14:45 – 15:30

S11 Messages from the patient organizations
Chairs: Moxley III, Richard (Rochester, USA)  
Monckton, Darren G (Glasgow, UK)  
15:30 – 16:45

S11-1 Awards (award committee)  
15:30 – 16:00

S11-2 Presentation of the patients organizations  
16:00 – 16:45

S12 Conference Highlights  
Chairs: Moxley III, Richard (Rochester, USA)  
Monckton, Darren G (Glasgow, UK)  
16:45 – 18:00

S12-1 Late-breaking Session  
16:45 – 17:15

S12-2 Conference Highlights and Concluding Remarks  
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)  
Schoser, Benedikt (München)  
17:15 – 18:00

Farewell-Dinner Riverboat Cruise on the Main River  
19:00 – 23:00

Meeting point: Alter Kranen - The Old Crane” at the river Main; admission by ticket only. Pre-booking requested

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 Mastroyiannopoulos (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., Sulek-Piatkowska A., Froster U. G. (Leipzig, Germany)

P3  Poster Session 3: Clinical Issues—Part 1
Chairs: Van Engelen, Baziel (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)

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
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 Roebling (Ulm)

P4-04 The lived experience of patients with Myotonic Dystrophy Type 1
Maud-Christine Chouinard (Jonquière)

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 (Jonquières)

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 (Jonquière)

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)
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(List in alphabetical order):

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