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Welcome to Switzerland, home of Swiss watches, Swiss cheeses, Swiss chocolates, Swiss bank accounts; the playground of Europe, for those who can afford it, such as bankers and politicians with expense accounts, and of course participants in our cut-price WMS congresses.

We are once again full house and everything seems set for another superlative Triple E congress. We have had a few hiccoughs on the way, due to servers developing a firewall ("antibodies") to each other, so that some participants, including myself and two other members of our program committee, who thought their registration had gone through on line, were only virtual registrants, and not officially clocked in by Symporg. Hopefully this has now all been resolved and there are no more virtual participants arriving unexpectedly in Geneva. They will of course still be very welcome.

As I have mentioned in previous years, the growing popularity of the WMS Congress year on year has posed major organisational problems for the Programme Committee, not only to accommodate the increasing numbers of poster presentations but also to ensure adequate conditions for their presentation and discussion. Our earlier ceiling of 400 participants is now past history, and for the past 3 years we have passed the 500 mark 3 months ahead of the congress.

At Newcastle last year we implemented one of the suggestions of our advisory subcommittee (Flanigan, Bonnemann and Hilton-Jones) to have 2 open sessions of poster presentations, without formal discussion, on days 1 and 2 of the congress. This proved very successful and took a lot of pressure off the formal poster sessions and is being repeated again this year.

Their second proposal was to extend the meeting into a fourth day, which would allow two more formal poster sessions on day 3. We were unable to extend this year’s meeting beyond the traditional 3-day format, but shall introduce the 4-day meeting in Japan next year. If successful it may become the norm for future years.

Each annual WMS takes a lot of hard work to get the show on the road, and I would firstly like to offer a special word of appreciation to Urs Ruegg and his energetic Geneva team for getting all the cogs in gear and putting together an exciting overall program. I feel assured we are heading for a superb Triple E congress.

Once again, I would like to commend our programme committee for their intense efforts of getting everything into shape over a hectic weekend in May in Geneva and also, in particular, to Anna Lancehielm and Camilla Wahlin of Congrex, who once again worked wonders in updating the programme pari passu with the activities of the program committee, and finally getting it ready for publication.

We have again made provision on the final afternoon for 6 oral late breaking presentations, with a late submission date about a month ahead of the congress, and no prior publication of the abstracts ahead of the meeting, but inclusion in the local program publication. We hope this special opportunity of presenting exciting new data will once again generate a lively and interesting session.

The fellowships for young scientists (based on the quality of their abstracts) which we introduced some years back, are now a regular annual commitment and this year we have been able to award 40 fellowships of 500 euros each. I hope we shall be able to increase the number of fellowships in future years if we can augment the WMS Education Fund. We would once again like to express our gratitude to Elsevier, for their magnanimity, and foresight, in agreeing to pro-
provide a royalty to our society each year from the income of the journal, Neuromuscular Disorders. This formed the basis for our special Education Fund and the first call each year on this fund has been the fellowships. The Fund also provides a fixed honorarium of 500 euros for all invited keynote speakers towards their travel and accommodation costs.

We are also grateful to Elsevier for the four annual Elsevier prizes of 500 euros each for the best oral or poster presentations by a young researcher, to be decided by an international multidisciplinary panel of judges (we define “young” as “not yet definitive”, i.e. still in training). It is at the discretion of the panel to select either poster or oral presentations. In addition Elsevier have awarded 14 one-year subscriptions to WMS (one for each year of the Society’s existence), for 14 worthy runners-up for the Elsevier prizes.

At the 10th Congress in Brazil, I decided to donate all proceeds from my memoirs, Ramblings of a Peripatetic Paediatrician, to WMS to provide awards and prizes for young researchers, and established on an annual basis a President’s prize of 500 euros for the most innovative contribution by a young scientist to the congress. This was initiated in 2006 in Bruges. In 2007 I established an additional President’s Prize for the best presentation, poster or oral, by a first time young presenter. These two prizes will continue to be awarded annually and are underwritten for the next 10 years (unless our WMS Bankers go belly-up!).

We also hope to establish as an annual prize the Léa Rose SMA prize, endowed by Natalie van der Mersch after the Bruges meeting, in memory of her infant daughter whom she lost to SMA.

Your program committee strive each year to achieve the three goals of our Triple E society, Education, Enjoyment and Excitement, and each year welcomes all feedback and constructive criticisms and suggestions. Please do complete the appraisal forms in your congress bags and return them by lunchtime on day 3. This is now obligatory if you wish to have a CME certificate!

Next year we will be meeting in the delightful university town of Kumamoto, on Kyushu Island, the most southwest part of Japan. It has Japan’s most active volcano Mt Aso nearby, which we plan to visit. (There are reassuring concrete bunkers alongside should it become angry.) The four-day congress will run from Wednesday 13th to Saturday 16th October, with the opening reception on Tuesday 12th October. The pre-congress course will be on Monday 11th and Tuesday morning 12th October. The deadline for abstracts will, as usual, be 31st March.

You should have a flyer in our congress bag. Watch the dedicated website www.wms2010.com for further information or the WMS website www.worldmusclesociety.org. As we anticipate another full house please register early. And don’t forget the Dubowitz two times rule and allow yourself an extra week to explore Japan. And do look into the very generous Japan Rail Pass. A week’s unlimited travel between Kyushu Island in the south and Hokkaido in the north may not cost you much more than a single fare from Tokyo to Kyushu.

Meanwhile have a great time in Geneva

Victor Dubowitz, President of the World Muscle Society
When a patient presents with proximal muscle weakness, do you suspect a potentially fatal disease?

Include Pompe Disease in the differential diagnosis.
also known as Acid Maltase Deficiency or Glycogen Storage Disease (type II)


Pompe disease is a progressive, debilitating, often fatal neuromuscular disease. Early diagnosis and intervention are critical.

For more information visit the Genzyme booth.

TREAT-NMD
Neuromuscular Network

TREAT-NMD is a global network uniting the key opinion leaders in the neuromuscular field to provide the focus, tools and expertise to accelerate new therapies and deliver optimal care.

From patient registries to standardised operating procedures to care standards, the network is creating the tools that the neuromuscular field needs to bring promising therapies more quickly from the lab to the clinic.

Join TREAT-NMD! Visit www.treat-nmd.eu/membership to find out how you can get involved.
Dear Colleagues,

WELCOME TO THE WORLD MUSCLE SOCIETY CONGRESS IN GENEVA!

On behalf of the Organizing Committee, I gladly welcome you to Geneva.

As many of you might know, this city is the birthplace of the Red Cross, the home of the headquarters of the United Nations World Health Organization and is considered to be one of the top cities to live in worldwide. Our University, which will house us in the newly constructed UniMail building, celebrates its 450th anniversary this year.

If you have any spare time, we suggest you stroll around Geneva and visit some of its museums and, perhaps, take a tour around Switzerland looking at other attractive locations such as Zermatt, Berne or Basel.

You will hear more about the history of Switzerland and its sights at the opening ceremony. The boat cruise on Lac Léman scheduled for Friday evening, will give you a taste of the beautiful Swiss scenery.

We have been able to attract exceptional speakers to WMS 2009 and have selected additional speakers from the over 450 abstracts that were submitted. The poster sessions will provide a basis for in-depth discussions and the posters will be on display throughout the meeting.

At the time of writing this text in early June 2009, we are once again close to a full house with registrations, highlighting the interest of the topic and making the congress a melting pot for scientific exchange of results and ideas.

I would like to acknowledge the help provided by the Program Committee of the WMS as well as the support of our sponsors and Symporg.

WMS congresses are well known for the triple “E”: Education, Excitement and Enjoyment - we are sure you will find them all in Geneva!

Urs T. Ruegg, President of the Local Organising Committee
The Congress offers a unique opportunity for delegates to learn about leading edge innovations from all over the world. The format of the program includes keynote speakers, oral and poster presentations from submitted abstracts. The social program offers the opportunity for networking, meeting the delegates in addition to enjoying leisureful moments.

The main aims of the WMS are to:

- Provide a multidisciplinary scientific forum to advance and disseminate knowledge in the neuromuscular field for the benefit of patients.
- Stimulate, encourage and help develop programs for professionals working in the neuromuscular field.
- Encourage multidisciplinary collaborations.
- Provide opportunities for young investigators in the neuromuscular field.
- Promote the achievement of high standards in clinical practice.

**PROGRAM COMMITTEE**
- Victor Dubowitz London (United Kingdom)
- Urs T. Ruegg Geneva (Switzerland)
- Haluk Topaloglu Ankara (Turkey)
- Peter Van den Bergh Brussels (Belgium)
- Thomas Voit Paris (France)

**LOCAL ORGANISING COMMITTEE**
- Urs T. Ruegg (President) University of Geneva
- Pierre-Yves Jeannet University of Lausanne
- Thierry Kuntzer University of Lausanne
- Thomas Meier Santhera Pharmaceuticals Liestal
- Susan Treves University of Basel, University of Ferrara
- Pierre Maechler University of Geneva
- Markus A. Ruegg University of Basel

**CME CREDITS**
The Swiss Neurological Society have granted 12 CME Credits for the WMS Congress.

**ADMINISTRATIVE SECRETARIAT**
SYMPORG SA is the official conference organiser and will be on duty during the conference hours at the Uni-Mail.

SYMPORG SA
Avenue Krieg 7
1208 Geneva
Switzerland
info@wms2009.com
Tel +41 (0) 22 839 84 84
Fax +41 (0) 22 839 84 85
**UNI-MAIL**

Uni-Mail is situated in the center of the city, next to the Arve River, at the end of the Plainpalais and can be easily reached by car, from the train station and from the airport. Uni-Mail is a recent addition of the Geneva University, offering several auditoriums and numerous conference rooms, all of them with the most up-to-date equipment.

**Address:**

Université de Genève, 40 boulevard du Pont d’Arve
1205 Genève - Switzerland

**ACCESS**

*From the airport:*

Bus 10 - (stop 22 Cantons-Gare Cornavin), then tram 15 (stop Uni-Mail)

*From the Cornavin railway station:*

Tram 15 and 17 (stop Uni-Mail)

*Other possibilities:*

Tram 12 and 13 (stop Pont d’Arve)

Bus 1 (stop Pont d’Arve)

**BADGES AND TICKETS**

The badge is your passport to all conference activities as well as your tickets for all social activities. Be sure to have them with you at all times.

**CLIMATE AND CLOTHING**

The climate is usually mild in September with temperatures of around 17 to 20°C Celsius (63 to 70°F). However, in addition to light clothing, don’t forget warmer clothes and a raincoat for rainy days and good walking shoes as well as adapted clothing if you plan excursions to the mountains.

**CURRENCY**

The local currency is the Swiss Franc (CHF). One Swiss Franc equals approximately 0.60 Euro. Banking hours are from Monday to Friday, 08:30 to 16:30. Mastercard and Visa are accepted almost everywhere and many places take American Express. There are several ATMs in the City, as well as at the congress location.

**DRESS CODE**

Dress informally for all conference activities, as well as for the Gala Dinner, which does not require evening dress.

**INTERNET**

A personal code has been sent to all participants by e-mail to access the wifi of the Uni-Mail.

**LOCAL TRANSPORTATION**

Your hotel in Geneva will give you a pass entitling you to free use of the Geneva public transportation system (TPG) during the week of the congress: buses, trams, train between downtown and airport and yellow boats (Mouettes) crossing the Geneva harbour.

**OFFICIAL LANGUAGE**

English is the official language of the congress. No simultaneous interpretation will be provided.

**SECRETARIAT OPENING HOURS**

<table>
<thead>
<tr>
<th>Day</th>
<th>Time</th>
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<tbody>
<tr>
<td>WEDNESDAY 9TH SEPTEMBER 2009</td>
<td>12:00 – 21:00</td>
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<tr>
<td>THURSDAY 10TH SEPTEMBER 2009</td>
<td>07:00 – 11:00</td>
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<td>12:00 – 21:00</td>
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<tr>
<td>FRIDAY 11TH SEPTEMBER 2009</td>
<td>08:00 – 11:00</td>
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<td>12:00 – 18:00</td>
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<tr>
<td>SATURDAY 12TH SEPTEMBER 2009</td>
<td>08:00 – 11:00</td>
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<td>12:00 – 18:00</td>
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**SHOPPING**

Shops in Geneva are open Monday to Friday from 08:30 to 19:00 and Saturday from 08:30 to 17:00.
WMS Satellite Teaching Course
Location: Lausanne University Hospital, 8-9 September 2009

**MONDAY 7 SEPTEMBER 2009**

19:00  Pre-course dinner in Lausanne at the Brasserie Lausanne Moudon, rue du Tunnel 20, CH – 1005 Lausanne

**TUESDAY 8 SEPTEMBER 2009**

08:00  Coffee and Registration, Auditorium Jequier-Doge, 8th floor

08:45  Learning from the history and examination, part 1: inherited muscle disease
       *Kate Bushby*

09:30  Learning from the history and examination, part 2: acquired muscle disease
       *David Hilton-Jones*

10:15  How do we perform a clinical examination on children?
       *Carsten Bonnemann*

11:00  Coffee break

11:30  What can we learn from the muscle biopsy, part 1: muscular dystrophies, congenital myopathies, myofibrillar myopathies
       *Caroline Sewry*

12:00  What can we learn from the muscle biopsy, part 2: metabolic, mitochondrial, inflammatory and other acquired myopathies
       *Anders Oldfors*

12:30  Molecular diagnostic testing and what does it mean?
       *Volker Straub*

13:00  Lunch

13:45  Delegates are split into two (group A and group B)

14:00  Group A - guide to muscle examination in adults and children and opportunity to meet patients with different disorders and diagnosis
       Group B - muscle pathology session in Histopathology suite

16:00  Coffee

16:15  Group A - muscle pathology session in Histopathology suite
       Group B - guide to muscle examination in adults and children and opportunity to meet patients with different diagnoses

18:15  Close of session

19:30  Evening cocktail party at the Domaine Wannaz, La Tour de Chenaux, 1091 Chenaux
WMS Satellite Teaching Course
Location: Lausanne University Hospital, 8-9 September 2009

**WEDNESDAY 9 SEPTEMBER 2009**

<table>
<thead>
<tr>
<th>Time</th>
<th>Topic</th>
<th>Presenter</th>
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<tbody>
<tr>
<td>8:00</td>
<td>Systematic overview on the floppy infant presentation and diagnostic approach</td>
<td>Thomas Voit</td>
</tr>
<tr>
<td>9:00</td>
<td>Systematic overview on patients presenting with limb-girdle and distal muscle weakness and diagnostic approach</td>
<td>Marianne de Visser</td>
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<tr>
<td>10:00</td>
<td>Coffee</td>
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<tr>
<td>10:15</td>
<td>Presentation of difficult, unusual or unsolved cases by participants</td>
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<tr>
<td>12:15</td>
<td>End of the meeting</td>
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Fondation Suisse de Recherche sur les Maladies Musculaires

24 years ago, the parents of 2 myopathic children decided to establish the Swiss Foundation on Muscular Disease Research or Fondation Suisse de Recherche sur les Maladies Musculaires (FSRMM). Their decision was motivated at the time by the lack of interest of research in this field in Switzerland.

Since the mid eighties, the foundation (FSRMM) has funded approximately one hundred fellowships for a global amount of CHF 18 million, funding researchers active in the 5 Medical Universities of Switzerland. These researchers, having spent endless efforts in finding the fundamental mechanisms of neuromuscular diseases, have just begun to collect the fruit of their dedication. The FRSMM is also at the origin of the creation of the pharmaceutical company Santhera, based in Liestal, just 5 years ago. The company's aim is to develop drugs in the field of rare diseases.

The Foundation supports symposia gathering researchers working in the field of neuromuscular diseases in Switzerland. Further, the foundation organises every 2 years, a symposium in Macolin where fellowships grantees meet each other during 3 days.

The FSRMM works almost exclusivity on a voluntarism basis. Administrative costs are therefore maintained under 5%.

www.fsrmm.ch
**Program at a Glance**

**WEDNESDAY 9TH SEPTEMBER 2009**

<table>
<thead>
<tr>
<th>Schedule</th>
<th>Main Auditorium</th>
<th>Poster rooms</th>
<th>Main Hall</th>
<th>Meeting room</th>
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<tbody>
<tr>
<td>12:30 - 15:00</td>
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<td></td>
<td>WMS Executive Board Meeting</td>
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<tr>
<td>15:00 - 16:00</td>
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<td>Setting up of posters</td>
<td>Registration</td>
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<td>16:00 - 18:30</td>
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<td></td>
<td>Welcome Ceremony and Reception</td>
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<td>18:30 - 21:00</td>
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**THURSDAY 10TH SEPTEMBER 2009**

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<th>Schedule</th>
<th>Main Auditorium</th>
<th>Poster rooms</th>
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<tbody>
<tr>
<td>07:00 - 08:15</td>
<td></td>
<td>Setting up of posters</td>
<td>Registration</td>
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<tr>
<td>08:30 - 09:00</td>
<td>Opening of Congress</td>
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<tr>
<td>09:00 - 10:30</td>
<td>New methods for assessing disease progression in neuromuscular disorders</td>
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<td>Coffee break</td>
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<td>10:30 - 11:00</td>
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<td></td>
<td>Commercial exhibition</td>
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<tr>
<td>11:00 - 12:00</td>
<td>New insights into neuromuscular diseases</td>
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<td>12:00 - 13:00</td>
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<td>Lunch break</td>
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<td>13:00 - 14:30</td>
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<td>Editorial Board Meeting</td>
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<tr>
<td>14:30 - 15:30</td>
<td>Chaired poster discussion (Session 1)</td>
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<tr>
<td>15:30 - 16:00</td>
<td>Poster viewing: Presenters of posters, Session 1</td>
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<td>16:00 - 16:30</td>
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<td>Coffee break</td>
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<tr>
<td>16:30 - 17:00</td>
<td>Poster viewing: Presenters of posters, Session 2</td>
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<tr>
<td>17:00 - 18:00</td>
<td>Chaired poster discussion (Session 2)</td>
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<tr>
<td>18:00 - 20:30</td>
<td>Special Industry Forum: Emerging Therapies for Neuromuscular Diseases</td>
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<tr>
<td>20:30 - 21:00</td>
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<td>Special Industry Forum: Snacks and drinks</td>
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### FRIDAY 11TH SEPTEMBER 2009

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<th>Schedule</th>
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<th>Meeting room</th>
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<tbody>
<tr>
<td>09:00 - 10:30</td>
<td>The extracellular matrix in normal and diseased muscle</td>
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<td>Coffee break Commercial exhibition</td>
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<td>10:30 - 11:00</td>
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<td>11:30 - 12:30</td>
<td>New insights into neuromuscular diseases</td>
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<td>12:30 - 14:00</td>
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<td>Lunch break Commercial exhibition</td>
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<td>14:00 - 15:00</td>
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<td>Chaired poster discussion (Session 3)</td>
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<td>15:00 - 15:30</td>
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<td>Poster viewing: Presenters of posters, Session 3</td>
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<td>15:30 - 16:00</td>
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<td>Coffee break Commercial exhibition</td>
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<td>16:00 - 17:00</td>
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<td>Chaired poster discussion (Session 4)</td>
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<tr>
<td>19:30 - 22:30</td>
<td>Gala Dinner on the Lake of Geneva (Lac Léman) aboard the boat “Lausanne”</td>
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### SATURDAY 12TH SEPTEMBER 2009

<table>
<thead>
<tr>
<th>Schedule</th>
<th>Main Auditorium</th>
<th>Poster rooms</th>
<th>Main Hall</th>
<th>Meeting room</th>
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</thead>
<tbody>
<tr>
<td>09:00 - 10:30</td>
<td>Advances in treatment of neuromuscular disorders</td>
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<td>Coffee break Commercial exhibition</td>
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<td>10:30 - 11:00</td>
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<td>11:00 - 12:00</td>
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<tr>
<td>12:00 - 13:00</td>
<td>WMS General Assembly</td>
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<td>13:00 - 14:30</td>
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<td></td>
<td>Lunch break Commercial exhibition</td>
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<tr>
<td>14:30 - 15:00</td>
<td>Advances in treatment of neuromuscular disorders</td>
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<tr>
<td>15:00 - 16:30</td>
<td>Late breaking news</td>
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<tr>
<td>16:30 - 17:00</td>
<td></td>
<td></td>
<td>Coffee break Commercial exhibition</td>
<td></td>
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<tr>
<td>17:00 - 17:30</td>
<td>Prize giving and welcome to WMS 15, Kumamoto - Japan</td>
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<tr>
<td>17:30</td>
<td>Handover of the WMS flag and closing of the Congress</td>
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Social Program

WELCOME RECEPTION  9th September 2009
The official welcome reception will take place on the 9th of September at Uni Mail, in the exhibition area, in Geneva. The evening will begin with a few welcoming speeches. Complimentary food and wine will be served throughout the evening. Dresscode: informal.

GALA DINNER  11th September 2009
The world famous Geneva "Jet d’eau" will form the backdrop for the Gala Dinner. Participants will leave from Downtown Geneva on a boat for a 3-hour relaxing cruise on Lake Geneva. A welcome drink will be served onboard to help guests unwind in front of the last lights of the day reflecting on the lake. After sunset, main dishes will be served in a musical atmosphere. The boat trip will end at around 22:30 to allow everyone to stroll the streets of Geneva at their own leisure. Meeting point: boarding quay "des Vieux Pâquis" (opposite the Hôtel Beau-Rivage). Dresscode: informal.

Hopefully she'll never hear the name Santhera as she grows up...

...but if she needs us, we'll be with her every step of the way.

As an emerging specialty pharmaceutical company, Santhera believes that the development of small molecules has a big future. Unmet medical need is what drives our work. Particularly rare diseases and the development of orphan drugs where there are no current alternatives.

That's why we are busy developing novel solutions to improve the lives of patients with severe neuromuscular diseases.

At Santhera we care about people – every step of the way.

when it comes to science we know how to move people
### WEDNESDAY 9 SEPTEMBER 2009

<table>
<thead>
<tr>
<th>Time</th>
<th>Location</th>
<th>Event</th>
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<tbody>
<tr>
<td>12:30–16:00</td>
<td>Room R030</td>
<td>WMS Executive Board Meeting – UniMail</td>
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<tr>
<td>15:00–18:30</td>
<td>Poster rooms (1 to 9)</td>
<td>Registration and setting up of posters</td>
</tr>
<tr>
<td>18:30</td>
<td>Main Hall</td>
<td>Welcome Ceremony and Reception</td>
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### THURSDAY 10 SEPTEMBER 2009

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<tr>
<th>Time</th>
<th>Location</th>
<th>Event</th>
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<tbody>
<tr>
<td>07:00–08:15</td>
<td>Poster rooms (1 to 9)</td>
<td>Registration and setting up of posters</td>
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<tr>
<td>08:30–09:00</td>
<td>Auditorium (R380)</td>
<td>Opening of Congress</td>
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<td>Message from the WMS President Victor Dubowitz</td>
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<tr>
<td>09:00–10:30</td>
<td>Auditorium (R380)</td>
<td>New methods for assessing disease progression in neuromuscular disorders; Invited lectures (M.I. 1-3)</td>
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<tr>
<td>10:30–11:00</td>
<td>Main Hall</td>
<td>Morning Tea and Coffee</td>
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<tr>
<td>11:00–12:00</td>
<td>Auditorium (R380)</td>
<td>New methods for assessing disease progression in neuromuscular disorders; Invited lectures (M.I. 4-5)</td>
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<tr>
<td>12:00–13:00</td>
<td>Auditorium (R380)</td>
<td>New insights into neuromuscular diseases; Oral presentations (G.O. 1-4)</td>
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<tr>
<td>13:00–14:30</td>
<td>Main Hall</td>
<td>Lunch and Exhibition</td>
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<tr>
<td>13:00–14:30</td>
<td>Room R030</td>
<td>Meeting of Editorial Board of Neuromuscular Disorders</td>
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<td>14:30–15:30</td>
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<td>Chaired poster discussion session 1: Parallel sessions (1–8)</td>
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<td></td>
<td>Poster room 4</td>
<td>Poster 1 - M.P.1.01–09 Clinical trials, databases and registries</td>
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<td>Poster room 5</td>
<td>Poster 2 - EM.P.1.01- 9 Pharmacological approaches involving TGF-beta signalling</td>
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<td>Poster room 1</td>
<td>Poster 3 – EM.P.2.01 - 13 Alpha-dystroglycanopathies</td>
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<td>Poster room 3</td>
<td>Poster 4 – G.P.1.01 - 10 Congenital myopathies I: RYR-1 and related disorders</td>
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<td>Poster room 7</td>
<td>Poster 5 – G.P.2.01 - 6 Acute quadriplegic myopathy</td>
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<td>Poster room 2</td>
<td>Poster 6 – G.P.3.01 – 11 Mitochondrial myopathies</td>
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<td>Poster room 8</td>
<td>Poster 7 – G.P.4.01 - 9 Charcot-Marie-Tooth disease, Peripheral neuropathy and ALS</td>
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<td>Time</td>
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<td>15:30-16:00</td>
<td>Poster rooms (1 to 9) Poster Viewing session 1: presenters of all posters discussed on Thursday 10 September to be available for poster viewing</td>
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<tr>
<td>16:00-16:30</td>
<td>Main Hall Afternoon Tea and Coffee and Exhibition</td>
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<tr>
<td>16:30-17:00</td>
<td>Poster rooms (1 to 9) Poster Viewing session 2: presenters of all posters discussed on Friday 11 September to be available for poster viewing</td>
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<tr>
<td>17:00-18:00</td>
<td>Chaired poster discussion session 2: Parallel sessions (9–16)</td>
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<td></td>
<td>Poster room 4 Poster 9 – M.P.2.01 - 9</td>
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<td></td>
<td>Motor function and respiratory function assessment</td>
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<td>Poster room 5 Poster 10 – EM.P.3.01 - 8</td>
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<td></td>
<td>Investigations of the extracellular matrix</td>
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<td>Poster room 3 Poster 11 – T.P.1.01 - 11</td>
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<td></td>
<td>Therapeutic approaches using RNA surgery and related technologies</td>
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<td>Poster room 1 Poster 12 – T.P.2.01 - 13</td>
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<td>Pharmacological therapy approaches using mouse models</td>
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<td>Poster room 7 Poster 13 – G.P.6.01 - 9</td>
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<td>Limb girdle muscular dystrophy I: dysferlinopathy</td>
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<td>Poster room 2 Poster 14 – G.P.7.01 - 12</td>
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<td>Congenital myopathies II: nemaline myopathies and related myopathies</td>
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<td>Poster room 8 Poster 15 – G.P.8.01 - 11</td>
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<td>Metabolic myopathy I: Pompe disease</td>
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<td>Poster room 9 Poster 16 – G.P.9.01 - 10</td>
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<td>Spinal muscular atrophies</td>
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<tr>
<td>18:15-21:00</td>
<td>Auditorium (R380) Special Industry Forum: Emerging Therapies for Neuromuscular Diseases Presenting companies and organisations:</td>
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<td>Time</td>
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<td>09:00-10:30</td>
<td>Auditorium (R380)</td>
<td>Invited lectures (EM.I. 1–3)</td>
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<td>The extracellular matrix in normal and diseased muscle</td>
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<td>10:30-11:00</td>
<td>Main Hall</td>
<td>Morning Tea and Coffee</td>
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<td>11:00-11:30</td>
<td>Auditorium (R380)</td>
<td>Oral Presentations (EM.O.1-2)</td>
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<td>The extracellular matrix in normal and diseased muscle</td>
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<td>11:30-12:30</td>
<td>Auditorium (R380)</td>
<td>Oral presentations (G.O. 5-8)</td>
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<td>New insights into neuromuscular diseases</td>
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<td>12:30-14:00</td>
<td>Main Hall</td>
<td>Lunch and Exhibition</td>
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<td>14:00-15:00</td>
<td>Poster rooms (1 to 9)</td>
<td>Chaired poster discussion session 3: Parallel sessions (17–24)</td>
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<td>Poster room 4</td>
<td>Poster 17 – M.P.3.01 - 10</td>
<td>Functional muscle testing and myometry</td>
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<td>Poster room 5</td>
<td>Poster 18 – M.P.4.01 - 7</td>
<td>Methodology: general approaches</td>
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<td>Poster room 2</td>
<td>Poster 19 – EM.P.4.01 - 10</td>
<td>Collagen VI-related muscular dystrophy I: clinical and genetic approaches</td>
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<tr>
<td>Poster room 3</td>
<td>Poster 20 – T.P.3.01 - 8</td>
<td>Clinical trial approaches for Duchenne muscular dystrophy</td>
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<td>Poster room 7</td>
<td>Poster 21 – T.P.4.01 - 10</td>
<td>Further therapeutic approaches to muscular dystrophy</td>
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<td>Poster room 1</td>
<td>Poster 22 – T.P.5.01 - 13</td>
<td>Pharmacological approaches to therapy using mouse models</td>
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<td>Poster room 8</td>
<td>Poster 23 – G.P.10.01 - 9</td>
<td>Limb girdle muscular dystrophy II</td>
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<tr>
<td>Poster room 9</td>
<td>Poster 24 – G.P.11.01 - 11</td>
<td>Metabolic myopathy II and neuromuscular junction disorders</td>
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<tr>
<td>15:00-15:30</td>
<td>Poster rooms (1 to 9)</td>
<td>Poster Viewing session 3: presenters of all posters discussed on Friday 11 September to be available for poster viewing</td>
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<tr>
<td>15:30-16:00</td>
<td>Main Hall</td>
<td>Afternoon Tea and Coffee and Exhibition</td>
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<td>16:00-17:00</td>
<td>Poster rooms (1 to 9)</td>
<td>Chaired poster discussion session 4: Parallel sessions (25–33)</td>
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<td>Poster room 3</td>
<td>Poster 25 – M.P.5.01 - 9</td>
<td>New methods of imaging</td>
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<td>Poster room 4</td>
<td>Poster 26 – EM.P.5.01 - 8</td>
<td>Collagen VI-related muscular dystrophy II: experimental approaches</td>
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<tr>
<td>Poster room 9</td>
<td>Poster 27 – T.P.6.01 - 9</td>
<td>Therapy approaches using stem cells</td>
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Program Summary

Poster room 7
Poster 28 – G.P.12.01 – 11
Congenital myopathies III: centronuclear myopathies and actin myopathies

Poster room 5
Poster 29 – G.P.13.01 – 12
Duchenne muscular dystrophy

Poster room 2
Poster 30 – G.P.14.01 – 12
Myotonic disorders

Poster room 8
Poster 31 – G.P.15.01 – 13
Emery-Dreifuss muscular dystrophy and facioscapulohumeral muscular dystrophy

Poster room 1
Poster 32 – G.P.16.01 – 10
Inflammatory myopathies

Poster room 6
Poster 33 – G.P.17.01 – 11
Diagnostic and treatment approaches and rare disorders

19:30-22:30 Gala Dinner on the Lake of Geneva (Lac Léman)
aboard the boat "Lausanne"
Meeting point: see map on page 65

SATURDAY 12 SEPTEMBER 2009

09:00-10:30 Auditorium (R380) Invited lectures (EM.I. 1-3)
The extracellular matrix in normal and diseased muscle

09:00-10:30 Auditorium (R380) Invited lectures (TI. 1-3)
Advances in treatment of neuromuscular disorders

10:30-11:00 Main Hall Morning Tea and Coffee

11:00-11:30 Auditorium (R380) Invited lectures (TI.4)
Advances in treatment of neuromuscular disorders

11:30-12:00 Auditorium (R380) Oral Presentations (TO.1-2)
Advances in treatment of neuromuscular disorders

12:00-13:00 Auditorium (R380) WMS General Assembly

13:00-14:30 Main Hall Lunch

14:30-15:00 Auditorium (R380) Oral Presentations (TO.3-4)
Advances in treatment of neuromuscular disorders

15:00-16:30 Auditorium (R380) Late Breaking News

16:30-17:00 Main Hall Afternoon Tea and Coffee

17:00-17:30 Auditorium (R380) Prize giving and welcome to WMS 15, Kumamoto, Japan

17:30 Auditorium (R380) Handover of the WMS flag and closing of the Congress
### WEDNESDAY 9 SEPTEMBER 2009

<table>
<thead>
<tr>
<th>Time</th>
<th>Location</th>
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<tbody>
<tr>
<td>12:30-16:00</td>
<td>Room R030</td>
<td>WMS Executive Board Meeting</td>
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<tr>
<td>15:00-18:30</td>
<td>Poster rooms (1 to 9)</td>
<td>Registration and setting up of posters</td>
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<tr>
<td>18:30</td>
<td>Main Hall</td>
<td>Welcome Ceremony and Reception</td>
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### THURSDAY 10 SEPTEMBER 2009

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<th>Time</th>
<th>Location</th>
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<tr>
<td>07:00-08:15</td>
<td>Poster rooms (1 to 9)</td>
<td>Registration and setting up of posters</td>
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<tr>
<td>08:30-09:00</td>
<td>Auditorium (R380)</td>
<td>Opening of Congress – Message from the president Victor Dubowitz</td>
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<tr>
<td>09:00-10:30</td>
<td>Auditorium (R380)</td>
<td>New methods for assessing disease progression in neuromuscular disorders; Invited lectures (M.I. 1-3) Chairpersons: V. Dubowitz, U. Ruegg</td>
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<tr>
<td></td>
<td>M.I.1</td>
<td>New methods for assessing disease progression in neuromuscular disorders: assessment of strength and function E. Mercuri</td>
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<td>M.I.2</td>
<td>Metabolic assessments of muscle disease J. Vissing</td>
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<td>M.I.3</td>
<td>Quantitative assessment of skeletal muscle by NMR: current and future approaches P.G. Carlier</td>
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<tr>
<td>10:30-11:00</td>
<td>Main Hall</td>
<td>Morning Tea and Coffee</td>
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<tr>
<td>11:00-12:00</td>
<td>Auditorium (R380)</td>
<td>New methods for assessing disease progression in neuromuscular disorders; Invited lectures (M.I. 4-5) Chairpersons: I. Nonaka, A. Engel</td>
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<td>M.I.4</td>
<td>Discovering biomarkers for monitoring neuromuscular diseases: a turning point A. Ferlini</td>
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<td>M.I.5</td>
<td>The dystrophic dogs as an excellent animal model of Duchenne muscular dystrophy (DMD) S. Takeda</td>
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<tr>
<td>12:00-13:00</td>
<td>Auditorium (R380)</td>
<td>New insights into neuromuscular diseases; Oral presentations (G.O. 1-4) Chairpersons: N. Levy, N. Laing</td>
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<tr>
<td></td>
<td>G.O.1</td>
<td>Diagnosis of muscular dystrophies at the nanometer scale S. Puttini; M. Lekka; T. Kuntzer; N. Mermod; A. Kulik</td>
</tr>
</tbody>
</table>
Full Program

THURSDAY 10 SEPTEMBER 2009

G.O.2
A primary sequence motif underlying calpain 3 substrate cleavage
A. de Morrée; D. Lutje Hulsik; A. Impagliazzo; H.H.B. van Haagen;
P. de Galan; A. van Remoortere; P.A.C. 't Hoen; R.R. Frants;
S.M. van der Maarel

G.O.3
Contraction-dependent (FSHD1) and independent (FSHD2) epigenetic changes of D4Z4 unify FSHD
J.C. de Greef; R.J.L. Lemmers; B.G.M. van Engelen; S. Sacconi;
S.L. Venance; R.R. Frants; R. Tawil; S.M. van der Maarel

G.O.4
α-Actinin-3 regulates muscle glycogen phosphorylase: a potential mechanism for the metabolic consequences of the common human null allele of ACTN3
K.G.R. Quinlan; J.T. Seto; N. Turner; M. Floetenmeyer; D.G. Macarthur;
J.M. Raftery; N. Yang; R.G. Parton; G.J. Cooney; K.N. North

13:00-14:30
Main Hall
Lunch and Exhibition

13:00-14:30
MS 130
Meeting of Editorial Board of Neuromuscular Disorders

14:30-15:30
Poster session 1: parallel sessions (1-8)

Poster room 4
Posters 1 – Clinical trials, databases and registries
Chairpersons: T. Kuntzer, T. Meier

M.P.1.01
Pilot trial with cyclosporin A in patients with collagen VI myopathies
L. Merini; A. Angelin; T. Tiepolo; P. Braghetta; P. Sabatelli;
A. Zamparelli; A. Armaroli; M.E. Michelini; A. Ferlini; N.M. Maraldi;
P. Bonaldo; P. Bernardi

M.P.1.02
SNT-MC17/idebenone in the treatment of Friedreich’s ataxia:
preliminary safety data from a 12-month European randomized,
placebo-controlled study
J.B. Schulz; T. Meier; G.L. Holder

M.P.1.03
Preliminary results from single subcutaneous administration
of ACE-031, a form of the soluble activin type IIB receptor, in healthy
postmenopausal volunteers
N.G. Borgstein; C.H. Condon; Y. Yang; D.M. Wilson; E. Haltom;
J.L. Lachey; J. Seehra; M.L. Sherman

M.P.1.04
Comparison of disease-causing mutations in Duchenne muscular
dystrophy from the Cooperative International Neuromuscular Research
Group with two large DMD mutation databases
L.P. Hache; E. Feingold; D. Escolar; C. McDonald; P.R. Clemens

M.P.1.05
Frequency of dystrophin gene mutations in the DuchenneConnect
registry
V. Rangel Miller; G. Spinella; P. Furlong

M.P.1.06
An international registry for FKRP (Fukutin-Related Protein) patients-
the first international registry
T.A. Willis; M.C. Walter; K.M.D. Bushby; H. Lochmueller; V. Straub
M.P.1.07  The Smartnet Clinical Network – Creation of a national standardised assessment tool and natural history database for spinal muscular atrophy
A.G. Mayhew; E. Scott; F. Muntoni; A. Manzur

M.P.1.08  Diagnosis of Pompe disease: timing and methods used as reported to the Pompe registry
P.S. Kishnani; T. Miller; S. Prosad; Pompe Boards of Advisors

M.P.1.09  TREAT-NMD Clinical Trials Coordination Centre: efficiency of networking
K. Gramsch; A. Pohl; J. Kirschner; R. Korinthenberg; S. Geismann; A. Tassoni

Poster room 5  Posters 2 – Pharmacological approaches involving TGF–beta signalling
Chairpersons: A. De Luca, L. Dux

EM.P.1.01  Prevention of muscle fibrosis and myonecrosis in mdx mice by suramin, a TGF–β 1 blocker
M.J. Marques; A.P.T. Taniguti; A. Pertille; H. Santo Neto

EM.P.1.02  Long-term activin receptor type IIB inhibition improves strength and function of dystrophic muscle
J.L. Lachey; S. Bogdanovich; E.E. Pistilli; A.E. Pullen; J. Seehra

EM.P.1.03  Potential involvement of angiotensin II in pathological signs of dystrophic muscle: in vivo and ex vivo outcome of a chronic treatment with enalapril, an angiotensin–converting enzyme inhibitor, in the exercise-aggravated mdx mouse model
A. Cozzoli; V. Sblendorio; R.F. Capogrosso; B. Nico; D. Conte Camerino; A. De Luca

EM.P.1.04  Treatment approaches in laminin-α2-deficient congenital muscular dystrophy (MDC1A)
S. Meinen; S. Lin; M.A. Rüegg

EM.P.1.05  Heparan sulfate-dependent interaction of myostatin and syndecan-4
A. Keller-Pinter; L. Mendler; L. Dux

EM.P.1.06  Extracellular matrix metalloproteinase protein inducer (EMMPRIN/CD147) regulates myoblast differentiation through an MMP-mediated control of TGFβ activity
M. Attia; E. Huet; S. Gawrzak; S. Menashi; I. Martelly

EM.P.1.07  Effects of muscle hypertrophy on individual myonuclear domain sizes in single muscle fibers from myostatin deficient or IGF–1 over-expressing mice
R. Qaiasr; K. Morine; E.R. Barton; H.L. Sweeney; L. Larsson

EM.P.1.08  Low temperature-induced inhibition of myogenic differentiation is cancelled by IGF–1 and vitamin C
A. Shima; R. Matsuda
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<tr>
<th>Poster room 1</th>
<th>Posters 3 – Alpha-dystroglycanopathies</th>
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<tr>
<td><strong>EM.P.2.01</strong></td>
<td>Expression analysis of α-dystroglycan glycosyltransferases during myoblast–myotube differentiation</td>
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<td>M. Brockington; S. Torelli; C. Godfrey; F. Muntoni</td>
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<td><strong>EM.P.2.02</strong></td>
<td>Secretion of the N-terminal domain of α-dystroglycan in the human cerebrospinal fluid in physiological and pathological conditions</td>
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<td>K. Matsumura; F. Saito; Y. Saito-Arai; M. Ikeda; A. Nakakmura; T. Shimizu</td>
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<td><strong>EM.P.2.03</strong></td>
<td>An investigation of candidate genes in dystroglycanopathy patients</td>
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<td>C. Godfrey; E. Clement; S. Torelli; M. Brockington; S. Abbs; F. Muntoni</td>
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<td><strong>EM.P.2.04</strong></td>
<td>Unraveling the genetic complexity of alpha-dystroglycanopathies: ethnically diverse pathogenic mutations</td>
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<td>M.C. Manzini; D. Gleason; B.S. Chang; R.S. Hill; J.N. Partlow; B.J. Barry; A. Poduri; L. Basel-Vanagaite; M.Z. Seidhamed; M.A.M. Salih; W.B. Dobyns; C.A. Walsh</td>
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<td><strong>EM.P.2.05</strong></td>
<td>Congenital muscular dystrophy with defective α-dystroglycan glycosylation, cerebellar hypoplasia and severe epilepsy</td>
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<td>S. Messina; G. Tortorella; M. Spanò; D. Concolino; A. d’Amico; C. Bruno; F.M. Santorelli; E. Mercuri; E. Bertini</td>
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<td><strong>EM.P.2.06</strong></td>
<td>Two siblings with alpha-dystroglycan dysglycosylation myopathy, epilepsy and personality disorder</td>
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<td>J. Reimann; S. Moskau-Hartmann; D.J. Morris-Rosendahl; R. von Wrede; C. Kornblum</td>
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<td><strong>EM.P.2.07</strong></td>
<td>Frequency of Fukutinopathy in Japanese cardiomyopathy patients with myopathy</td>
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<td>H. Matsumoto; Y.K. Hayashi; T. Murakami; I. Nonaka; I. Nishino</td>
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<td><strong>EM.P.2.08</strong></td>
<td>Fukutin gene mutations in an Italian patient with early onset muscular dystrophy but no central nervous system involvement</td>
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<td>S. Saredi; E. Mottarelli; A. Ruggieri; A. Ardissone; S. Zanotti; L. Farina; L. Morandi; I. Moroni; M. Mora</td>
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<td><strong>EM.P.2.09</strong></td>
<td>Spectrum of mutations and phenotypic variability in FKRP-related disease in Norway</td>
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<td>E. Stensland; S. Lindal; C. Jonsrud; T. Torbergsen; M. Rasmussen; L. Bindoff; F.M. Thyssen; Ø. Nilsen</td>
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<td><strong>EM.P.2.10</strong></td>
<td>Deficiency of multiple alpha dystroglycan ligand interactions underlie the phenotype of a FKRP-deficient mouse model for muscle eye brain disease</td>
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<td>M.R. Ackroyd; S. Prior; C. Whitmore; M. Kaluarachchi; F. Muntoni; S.C. Brown</td>
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</table>
EM.P.2.11 Fukutin-related protein expression in murine dystrophic models carrying single and double mutations for dystrophin and LARGE.

P.C.M. Martins; D. Ayub-Guerrieri; V.L. Ferreira; P.C. Onofre-Oliveira; G. Monteiro; D. Zilbersztajn; L.U. Yamamoto; C.M.C. Mori; L.E.S. Netto; M. Vainzof


A. d’Amico; C. Bruno; E. Silvestri; P. Alfieri; G. Vasco; A. Tessa; F.M. Santorelli; E. Bertini; E. Mercuri

EM.P.2.13 Overexpression of LARGE strongly increases laminin binding of α-dystroglycan but does not exhibit toxic effects in mice.

F. Saito; Z. Xin; M. Ikeda; H. Hagiwara; T. Shimizu; K. Matsumura

Poster room 3

Posters 4 – Congenital myopathies I: RYR1 and related disorders

Chairpersons: L. Politano, N. Romero

G.P.1.01 Phenotypic variations of central core disease

O. Paciello; A. Tammaro; L. Passamano; M. Scutifero; E. Picillo; A. Di Martino; S. Papparella; L. Politano

G.P.1.02 Phenotypic spectrum of core-rod myopathy caused by dominant or recessive RYR1 mutations

K.G. Claeys; N. Monnier; P. Laforet; G. Brochier; A. Ferreiro; A. Barois; B. Eymard; J. Lunardi; M. Fardeau; N.B. Romero

G.P.1.03 King-Denborough syndrome associated with mutations in the skeletal muscle ryanodine receptor (RYR1) gene

J. Dowling; S. Lillis; K. Amburgey; S. Leber; H. Zhou; S. Al-Sarraj; E. Wraige; S. Abb; C. Sewry; F. Muntoni; H. Jungbluth

G.P.1.04 An autosomal-recessive form of centronuclear myopathy is caused by mutations in the skeletal muscle ryanodine receptor (RYR1) gene

J. Wilmshurst; S. Lillis; H. Zhou; W. Kress; R. Solomon; A. Ndondo; J. Greenberg; C.C. Sinclair-Smith; E. Bertini; C. Boennemann; V. Straub; R. Quinlivan; C.A. Sewry; E. Wraige; S. Abb; F. Muntoni; H. Jungbluth

G.P.1.05 RYR1 is a common cause of congenital fibre type disproportion with ptosis, ophthalmoplegia, scoliosis and pronounced axial muscle weakness

N.F. Clarke; N. Monnier; R.L.L. Smith; A.J. Kornberg; M.A. Farrell; L. Waddell; S. Cooper; J. Lunardi; K.N. North

G.P.1.06 An atypical presentation of lethal neonatal hypotonia associated with a genomic rearrangement of the RYR1 gene

N. Monnier; A. Laquerière; S. Marret; A. Goldenberg; S. Drouhin; J. Lunardi
## Full Program

### THURSDAY 10 SEPTEMBER 2009

| G.P.1.07 | Progressive muscle biopsies morphological changes in long —term follow —up of multiminicore disease related to RYR1 gene  
A.L. Taratuto; M. Saccoliti; F. Lubieniecki; N. Monnier; N. Romero; P. Richard; J. Lunardi |
| G.P.1.08 | Calcium influx analysis by TIRF microscopy on cultured primary myotubes from patients with RYR1 mutations  
S. Treves; P.Y. Jeannet; M. Vukcevic; T. Girard; A. Urwyler; F. Zorzato |
| G.P.1.09 | Clinical, pharmacological and molecular investigation of patients presenting with exertional heat stroke  
N. Monnier; D. Bendahan; G. Kozak-Ribbens; C. Foutrier-Morello; C. Brasset; E. Sagui; M. Aubert; M. Lamaria; N. Tessier; P.J. Cozzone; J. Lunardi |
| G.P.1.10 | DHPR α1 S subunit controls skeletal muscle mass and morphogenesis  
F. Piétri-Rouxel; C. Gentil; D. Baas; E. Mouiel; C. Jourdé; A. Ferry; A. Vignaud; L. Schaeffer; T. Voit; L. Garcia |

### Poster room 7

**Posters 5 - Acute quadriplegic myopathy**  
*Chairpersons: F. Mastaglia, L. Larsson*

| G.P.2.01 | Acute quadriplegic myopathy: underlying mechanisms, improved diagnostic methods and specific intervention strategies  
L. Larsson; A.M. Gustafson; J. Ochala; V. Banduseela; M. Li; S. Aare; Y. Hedström; R. Qaisar; M. Llano Diaz; X. Tong; B.R. Dworkin |
| G.P.2.02 | Aspiration muscle biopsy for monitoring and diagnosis of acute quadriplegic myopathy  
Y. Hedström; V. Banduseela; L. Larsson |
| G.P.2.03 | Partial reversal of ventilator-induced diaphragm muscle cell dysfunction  
J. Ochala; L. Larsson |
| G.P.2.04 | Gene expression and muscle fiber function in a porcine AQM-ICU model  
V. Banduseela; J. Ochala; Y. Chen; H. Goransson; H. Norman; P. Radell; L.I. Eriksson; E. Hoffman; L. Larsson |
| G.P.2.05 | Expression profile and functional analysis of masticatory muscle in porcine AQM-ICU model  
S. Aare; V. Banduseela; J. Ochala; H. Goransson; H. Norman; P. Radell; L.I. Eriksson; L. Larsson |
| G.P.2.06 | Diaphragm muscle weakness in an experimental intensive care unit model  
J. Ochala; M. Llano-Diez; L. Larsson |

### Poster room 2

**Posters 6 - Mitochondrial myopathies**  
*Chairpersons: R. Taylor, P. Maechler*

| G.P.3.01 | An adult case of myopathy due to mutations of the TK2 gene  
A. Béhin; C. Jardel; K. Cloës; T. Beillévaire; A. Lombès; B. Eymard |
### G.P.3.02
Late-onset MNGIE without peripheral neuropathy due to incomplete loss of thymidine phosphorylase activity

*Authors*: R. Massa; A. Tessa; M.A. Margollicci; A. Romigi; C. Terracciano; R. Corrozzo; G. Bernardi; F.M. Santorelli

### G.P.3.03
Recurrent mutations in the NDUFS2 gene causing isolated complex I deficiency in skeletal muscle

*Authors*: R. McFarland; H. Tuppen; L. He; E.L. Blakely; A.A.M. Morris; M. Clarke; S. Jones; A.M. Devlin; R.W. Taylor

### G.P.3.04
Autosomal dominant Progressive External Ophthalmoplegia (adPEO) due to mutations in the PED1 gene: a clinical, histochemical and molecular survey of 33 patients

*Authors*: C. Fratter; G. Gorman; J.D. Stewart; M. Buddles; C. Smith; J. Evans; A. Sellar; J. Poulton; M. Roberts; M.G. Hanna; S. Rahman; S.E. Omer; T. Klopstock; B. Schoser; C. Kornblum; B. Lecky; P.F. Chinnery; D.M. Turnbull; R. Horvath; R.W. Taylor

### G.P.3.05
A novel heteroplasmic mitochondrial MT-ND5 frameshift mutation causing isolated paediatric complex I deficiency

*Authors*: C. Alston; C. Reid; R. Horvath; H. Mundy; R.W. Taylor

### G.P.3.06
Neuromuscular disease presentation with three genetic defects involving two genomes: the characterisation of a novel mitochondrial tRNA mutation exhibiting skewed segregation

*Authors*: M. Al-Dosary; R. Whittaker; J. Hood; R. McFarland; J.A. Goodship; D.M. Turnbull; R.W. Taylor

### G.P.3.07
Novel mutation in SUCLA2 gene: encephalomyopathy, dystonia and deafness associated with mild methylmalonic aciduria and mt DNA depletions

*Authors*: A.E.N. Nascimento Osorio; A.N.S. Navarro-Sastre; J.C. Colomer; C.P. Paredes; A.G. Gutierrez; C.O. Ortez; M.V. Vilaseca; J.M. Montoya; T.R. Ribes

### G.P.3.08
Myoclonic epilepsy, cortical dysplasia and mitochondrial complex I deficiency: a case report

*Authors*: C. Nesti; M. Mancuso; L. Petrozzi; F.M. Santorelli; A. Rocchi; G. Ali; A. Tessa; A. Lo Gerfo; A. Iudice; G. Siciliano

### G.P.3.09
Cerebellar ataxia, myopathy and sensorimotor neuropathy associated with a mitochondrial m.8363G>A tRNA(Lys) gene mutation: a clinical and biochemical study

*Authors*: E. Brusse; A. Korsten; G. Schoonderwoerd; D. Hellebrekers; J.A. Maat-Kievit; H.J.M. Smeets; I.F.M. De Coo

### G.P.3.10
Oxidative stress biomarkers are related to disease severity in mitochondrial myopathies, and may be modified by a cysteine donor

*Authors*: M. Mancuso; D. Orsucci; A. Lo Gerfo; G. Santoro; C. Nesti; A. Rocchi; L. Petrozzi; F. Galetta; G. Siciliano
Full Program

THURSDAY 10 SEPTEMBER 2009

G.P.3.11 Increased protein nitration in mitochondrial diseases: evidence for vessel wall involvement
G. Vattemi; Y. Mechref; M. Marini; P. Tonin; P. Minuz; L. Grigoli; Klouckova; C. Chiamulera; A. Meneguzzi; M. Di Chio; V. Tedesco; L. Lovato; M. Degan; M. Filasto; G. Arcaro; M. Riccadonna; V. Guglielmi; G. Fumagalli; N. Rizzuto; A. Lechi; M. Novotny; G. Tomelleri

Poster room 8 Posters 7 – Charcot-Marie-Tooth disease, peripheral neuropathy and ALS
Chairpersons: P. Van den Bergh, P. Serdaroglu

G.P.4.01 Charcot-Marie-Tooth type 1E in monozygotic twins linked to a rare PMP22 mutation
A. Matos; L. Negrão; A. Geraldo; S. Batista; A. Palmeiro; M.P. Tavares

G.P.4.02 The impact of silicone ankle foot orthosis (SAFO's) in patients with Charcot Marie Tooth (CMT) disease on cadence, balance and falls
N. Cartwright; S.L. Glover

G.P.4.03 Clinical, eletrophysiological and genetic finding in three patients with CMT4A
C. Ortez; A. Nascimento; S. Kapetanovic; A. Gutierrez; F. Palau; J. Colomer

G.P.4.04 A new MTMR2 mutation is responsible for a congenital form of Charcot-Marie-Tooth disease (CMT4B1) and vocal cord paresis
M. Tazir; S. Nouioua; T. Hamadouche; R. Bernard; D. Grid; N. Levy; J.M. Vallat

G.P.4.05 Clinical and electrophysiological findings in the course of childhood Guillain-Barré Syndrome: 10 years experience in a pediatric neuromuscular unit
A.P. Gutiérrez Mata; A.E. Nascimento Osorio; C.I. Ortez González; J. Colomer Oferil

G.P.4.06 Dislocation of neuronal nitric oxide synthase contributes to muscle atrophy in amyotrophic lateral sclerosis
N. Suzuki; M. Aoki; H. Warita; S. Takeda; Y. Itoyama

G.P.4.07 Skeletal muscle regeneration in amyotrophic lateral sclerosis
A. Scaramozza; A. Corbu; A. Aiti; V. Papa; L. Tarantino; L. Badioli De Giorgi; E. Pogoraro; G. Soraru; C. Angelini; G.N. Martinelli; G. Cenacchi

G.P.4.08 Flexor-dominant myopathic phenotype in patients with His46Arg substitution in the Cu/Zn superoxide dismutase gene
S. Yamashita; E. Kimura; F. Yamamoto; A. Migita; S. Mita; H. Teramoto; M. Uchino
G.P.4.09
Multiple analysis of hereditary spastic paraplegia
N.H. Akcakaya ; Ç. Atay ; N. Isik ; F.I. Uludağ ; F. Deymeer ; P. Serdaroglu ; E. Battaloğlu ; Y. Parman

Poster room 9
Poster 8 – Distal myopathies and myofibrillar myopathies
Chairpersons: Z. Argov, S. Noguchi

G.P.5.01
Retrospective study of 29 cases of distal myopathies
J. Franques ; E. Campana-Salort ; S. Attarian ; A. Verschueren ; C. Fernandez ; A. Maues de Paula ; D. Figarella-Branger ; J. Pouget

G.P.5.02
Distal myopathy with rimmed vacuoles (DMRV): Experience of a rare disorder
A. Nalini ; N. Gayathri

G.P.5.03
Genotype–phenotype correlation of DMRV/hIBM patient in Japan
K. Momma ; S. Noguchi ; Y.K. Hayashi ; K. Motoyoshi ; K. Kamakura ; I. Nonaka ; I. Nishino

G.P.5.04
Ultrastructural evidence of amyloidogenesis: an upstream event to myofiber degeneration in a mouse model of DMRV/hIBM
T. Tokutomi ; M.C. Malicdan ; S. Noguchi ; I. Nonaka ; Y.K. Hayashi ; I. Nishino

G.P.5.05
The evaluation of N-acetylmannosamine in model mouse towards the development of therapeutic strategy in DMRV/hIBM
S. Noguchi ; M.C.V. Malicdan ; Y.K. Hayashi ; I. Nonaka ; I. Nishino

G.P.5.06
In vitro assessment of AAV8 based gene delivery for hereditary inclusion body myopathy
M. Telem ; Z. Shlomai ; S. Mitrani-Rosenbaum

G.P.5.07
New monoclonal antibodies against the C-terminal M10 domain of titin
J. Sarparanta ; O. Raheem ; P. Hackman ; B. Udd

G.P.5.08
Myofibrillar myopathies: The Lausanne experience with DES mutations in 11 patients
M. Dunand ; J.A. Lobrinius ; F. Thonney ; X. Jeanrenaud ; D. Selcen ; S. Rudnik-Schöneborn ; J.M. Burgunder ; T. Kuntzer

G.P.5.09
Screening the FHL1 mutation in Japanese patients with non-4q35 FSHD
X.J. Xiao ; K. Goto ; S. Noguchi ; Y.K. Hayashi ; I. Nishino

G.P.5.10
Novel FHL1 mutation in familial mixed reducing body myopathy with rigid spine
J. Schessl ; A. Columbus ; Y. Hu ; Y. Zou ; T. Voit ; H.H. Goebel ; C.G. Bönnemann

15:30-16:00 Poster rooms (1 - 9)
Poster Viewing session 1: presenters of all posters discussed on Thursday 10 September to be available for poster viewing

16:00-16:30 Main Hall
Afternoon Tea and Coffee and Exhibition
THURSDAY 10 SEPTEMBER 2009

16:30-17:00 Poster rooms (1–9)
Poster Viewing session 2: presenters of all posters discussed on Friday 11 September to be available for poster viewing

17:00-18:00 Poster room 4
Posters 9 – Motor function and respiratory function assessment
Chairpersons: S. Iannaccone, J. Florence

M.P.2.01 Reliability of the North Star Ambulatory Assessment in a multicentric setting
E.S. Mazzone; S. Messina; G. Vasco; M. Main; M. Eagle; A. D’Amico; C. Bruno; L. Politano; A. Berardinelli; T. Mongini; A. Pini; R. Battini; G. Comi; E. Pegoraro; L. Morandi; M. Villanova; G. Vita; E. Bertini; E. Mercuri

M.P.2.02 Motor Function Measure: construction of a short form (MFM-20) for children with neuromuscular disease aged between 2 and 6
C. de Lattre; C. Payan; C. Payet; C. Fafin; F. Girardot; A. Jouve

M.P.2.03 The Motor Function Measure (MFM): sensitivity to change
C.A.M. Payan; C. Vuillerot; J. Fermanian; C. Bérard; and the MFM study group

M.P.2.04 Bioengineering and muscular dystrophy
M.G. d’Angelo; M. Romei; S. Gandossini; S. Bonato; D. Colombo; E. Marchi; A. Lo Mauro; A.C. Turconi; G.P. Comi; A. Aliverti; N. Bresolin

M.P.2.05 Optoelectronic plethysmography in respiratory assessment in Duchenne muscular dystrophy
M. Romei; M.G. d’Angelo; S. Gandossini; S. Bonato; G.P. Comi; D. Colombo; E. Marchi; A. Lo Mauro; A. Aliverti; N. Bresolin

M.P.2.06 External control of exhalation for cough assistance: a method for patients with glottis dysfunction
S.C. Lee; S.W. Kang; S.H. Im

M.P.2.07 The benefits of lung inflation training using positive end-expiratory pressure valve for patients with neuromuscular disorders
T. Matsumura; T. Saito; H. Fujimura; S. Shinno

M.P.2.08 Use of the Motor Function Measure to assess motor function in patients with spinal muscular atrophy
C. Vuillerot; C. Payan; C. Bérard

M.P.2.09 Mdx diaphragm muscle as a target of dystrophin gene therapy
F. Kimura; T. Suga; M. Ishizaki; K. Uchino; T. Koide; Y. Uchida; Y. Maeda; J.S. Chamberlain; M. Uchino

Poster room 5
Posters 10 – Investigations of the extracellular matrix
Chairpersons: S. Brown, M. Grounds
| EM.P.3.01 | The adult phenotype of congenital muscular dystrophy (MDC1A) due to mutation of LAMA2  
N. Canki-Klain; C. Béroud; N.F. Clarke; I. Kovac; S. Chambert; P. Guicheney |
| EM.P.3.02 | Intracellular signaling pathway alterations in laminin α2 chain deficient skeletal muscle and brain  
V. Carmignac; M. Durbeej-Hjalt |
| EM.P.3.03 | Differential expression of genes involved in muscular degeneration in four dystrophic mouse models  
P.C.G. Onofre-Oliveira; P.C.M. Martins; V.L. Ferreira; D. Ayub-Guerrieri; M. Vainzof |
| EM.P.3.04 | Muscle-derived Duchenne muscular dystrophy fibroblasts show altered production of extra-cellular matrix components  
S. Zanotti; S. Gibertini; S. Saredi; F. Blasevich; R. Mantegazza; L. Morandi; M. Mora |
| EM.P.3.05 | Expression of fibulins 1-5 during myogenesis in vitro and in skeletal muscle regenerating in vivo, and in dystrophic mdx muscles  
M.D. Grounds; T. Shavlakadze; J. Vukovic |
| EM.P.3.06 | Transient upregulation of matrilin-2 gene expression suggests a role in early steps of skeletal muscle regeneration  
É. Korpos; L. Mátés; L. Mendler; M. Kiricsi; Á. Zvara; F. Deák; Z. Rottenberger; A. Keller-Pintér; L. Puskás; L. Dux; L. Kiss |
| EM.P.3.07 | The fate of glycosaminoglycans (GAGs) during skeletal muscle regeneration and myoblast differentiation  
M. Oudghir; A. Duchesnay; T. van Kuppevelt; I. Martelly |
| EM.P.3.08 | Matrix matters in man and mouse  
N.C. Voermans; B.G. van Engelen |

Poster room 3 Posters 11 – Therapeutic approaches using RNA surgery and related technologies  
Chairpersons: L. Garcia and W.C. Yee

| T.P.1.01 | Pre-trial antisense screening of myogenic cells from boys with Duchenne muscular dystrophy and genomic and transcriptomic biomarkers discovery for treatment monitoring  
M. Neri; M.S. Falzareno; M. Fabris; M. Bovolenta; E. Bassi; D. Perrone; A. Medici; P. Sabatelli; L. Merlini; F. Gualandi; P. Rimessi; A. Ferlini |
| T.P.1.02 | Importance of checking exon skipping events prior to clinical trials using systemically delivered antisense morpholino in Duchenne muscular dystrophy patients  
S. Nakano; S. Ozasa; H. Kosuge; K. Nomura; I. Fujii; K. Ito; M. Matsukura; S. Kimura |
### Full Program

#### THURSDAY 10 SEPTEMBER 2009

| T.P.1.03 | In vitro splicing analysis reveals that availability of a cryptic splice site is not a determinant for alternative splicing patterns caused by +1G>A mutations in introns of the dystrophin gene  
* M. Matsuo; Y. Habara; Y. Takeshima; H. Awano; Y. Okizuka; Z. Zhang; M. Yagi |
| T.P.1.04 | Dual exon skipping in myostatin and dystrophin as a potential therapy for Duchenne muscular dystrophy  
* D.U. Kemaladewi; W.M.H. Hoogaars; S.H. van Heiningen; J.C.T. van Deutekom; J.T. den Dunnen; G.J.B. van Ommen; A.R. Aartsma-Rus; P. ten Dijke; P.A.C. ’t Hoen |
| T.P.1.05 | Trans-splicing approaches to repair Duchenne dystrophin transcripts  
* S. Lorin; C. Peccate; G. Griffith; T. Voit; L. Garcia |
| T.P.1.06 | Induced non-productive splicing to study muscle gene expression  
* S.D. Wilton; S. Fletcher; A.M. Adams; R. Johnsen; K. Greer |
| T.P.1.07 | Correcting SMN2 splicing with tailed antisense oligoribonucleotides: a promising therapeutic strategy for spinal muscular atrophy  
* H. Zhou; N. Owen; I.C. Eperon; F. Muntoni |
| T.P.1.08 | The use of antisense oligomer for splice switching in spinal muscular atrophy fibroblasts  
* C. Mitrpant; C. Fragall; R. Johnsen; A.H. Burghes; S. Fletcher; S.D. Wilton |
| T.P.1.09 | Oculopharyngeal muscular dystrophy (OPMD): physiopathological mechanisms and gene therapy approaches  
* C. Trollet; O. Bailes; Y. Anvar; K. Foster; K. Mamchou; P.A. ’t Hoen; V. Raz; S. van der Maarel; M. Antoniou; V. Moul; G. Butler-Browne; G. Dickson |
| T.P.1.10 | Exon-skipping of dysferlin in CD133+ stem cells isolated from normal and patient affected with dysferlinopathies  
* C. Navarro; A. Farini; P. Razini; M. Krahn; L. Garcia; N. Lévy; Y. Torrente |
| T.P.1.11 | Correction of endogenous DMPK transcripts by chimeric U2 sma nuclear RNA – artificial trans-splicing molecules as therapeutic strategy in dystrophia myotonica type 1  
* H.Y. Chen; P. Kathirvel; Q.B. Xiong; P.S. Lai; W.C. Yee |

**Poster room 1**  
**Posters 12 – Pharmacological therapy approaches using mouse models**  
Chairpersons: I. Richard, D. Conte Camerino

| T.P.2.01 | Assessment of treatment efficiency using the mdx model: how to interpret the data  
* M. Carre-Pierrat; L. Ségalat; L. Ségalat |
Full Program

TP.2.02 Effect of 20 approved drugs on a wide range of mdx parameters
M. Carre-P ierrat; A. Lafoux; F. Fougerousse; C. Huchet-Cadiou; L. Ségalat

TP.2.03 Erythropoietin application in mdx mice: a pilot trial
A.A.S. Carvalho; M.R. Ugollini; A.C. Santomauro JR; L.Z.P. Oliveira; V.P.P. Lioi; R.A. dos Santos; M.H.C. Carvalho; L.G. Ferreira; M.T. Nunes; S.B. Zyngier; D. Feder

TP.2.04 Oral TNF alpha inhibitor in mdx dystrophic mice: a pilot trial
D. Feder; L.H. Brito; L.S. Soad; V. Oliveira; E.A.C. Barros; R.A. dos Santos; L.G. Ferreira; M.H.C. Carvalho; M.T. Nunes; S.B. Zyngier; A.A.S. Carvalho

TP.2.05 Tamoxifen is more potent than raloxifene to ameliorate skeletal muscle function of adult mdx5Cv mice
O.M. Dorchies; J. Reutenauer; O. Patthey-Vuadens; U.T. Ruegg

TP.2.06 Exploring the role of phospholipase A2 in Duchenne muscular dystrophy
A.K. Johansson; O. Petermann; O.M. Dorchies; E. Roulet; U.T. Ruegg

TP.2.07 Attenuation of adverse effects of prednisolone on α-sarcoglycan-deficient cardiomyopathy by mineralocorticoid-receptor-antagonism
A.M. Blain; R. Bauer; G. MacGowan; E. Greally; S. Laval; K. Bushby; V. Straub

TP.2.08 The stretch-activated calcium channel candidate proteins TRPC1 and 3 present a distinctly different expression pattern in mdx mice
L.H. Jorgensen; S.H. Laval; H. Brinkmeier; V. Straub; K. Bushby; H. Lochmueller

TP.2.09 Role of the cationic channel TRPC1 in Duchenne muscular dystrophy: analysis of double knock-out TRPC1-/- dystrophin minus mice
C. Gallo; G. Shapovalov; O.M. Dorchies; O. Petermann; N. Zanou; P. Gailly; U.T. Ruegg; E. Roulet

TP.2.10 Identification of in-vivo TRPC1 activity in skeletal muscle at single-channel level; role in Ca2+ entry
G. Shapovalov; N. Zanou; C. Gallo; I. Anguish; E. Roulet; P. Gailly; U.T. Ruegg

TP.2.11 Comparison of subcutaneous injection and oral routes of administration of EGCG for ameliorating dystrophic lesions in a mouse model of Duchenne muscular dystrophy
Y. Nakae; O.M. Dorchies; P.J. Stoward; U.T. Ruegg

TP.2.12 Nuclear factor-kappa B, mitogen-activated protein kinases and cyclooxygenase/5-lipoxygenase pathway inhibition by flavocoxid improves muscle function and morphology in mdx mice: a comparison study with methylprednisolone
S. Messina; M. Aguennouz; A. Mazzeo; A. Migliorato; A. Bitta; G.L. Vita; M. Russo; E. Rizzuto; A. Musarò; F. Squadrito; G. Vita
### Evaluation of time-dependent effects of statins and fibrate ‘in vivo’ treatment on ClC-1 chloride channel expression and function and on proteomic profile of rat skeletal muscle

- **G.M. Camerino**
- **S. Pierno**
- **C. Digennaro**
- **M.M. Dinardo**
- **M.A. Pellegrino**
- **A.L. George**
- **R. Bottinelli**
- **D. Conte Camerino**

**Poster room 7**

**Posters 13 – Limb girdle muscular dystrophy I: dysferlinopathy**

**Chairpersons:** G. Vita, L. Negrão

**G.P.6.01**

Measurements of progression in dysferlin myopathies: a preliminary prospective quantitative study

- **C. Paradas**
- **C. Serrano-Munuera**
- **J. Llauger**
- **M. Martínez-Lage**
- **C. Márquez**
- **P. Gallano**
- **N. De Luna**
- **R. Rojas-Garcia**
- **E. Gallardo**
- **I. Illa**

**G.P.6.02**

Dysferlinopathy: unilateral foot drop of late onset

- **L. Negrão**
- **A. Matos**
- **A. Geraldo**
- **O. Rebela**
- **E. Vieira**
- **C. Marques**
- **R. Santos**

**G.P.6.03**

Dysferlinopathy in Egypt: clinical and pathological characteristics

- **N. Fahmy**
- **A. Abd-elhady**
- **A. Abd El-Naser**
- **S. Ashour**
- **I. Nonaka**
- **A. Etribi**

**G.P.6.04**

Nitric oxide signalling in selective muscle wasting of Miyoshi myopathy

- **R. Dhanarajan**
- **M. Alexander**
- **A. Oommen**

**G.P.6.05**

Attenuated muscle regeneration is a key factor in dysferlinopathy

- **Y.H. Chiu**
- **S.H. Laval**
- **L.H. Jorgensen**
- **M.A. Hornsey**
- **L. Klinge**
- **R. Charlton**
- **R. Barresi**
- **V. Straub**
- **H. Lochmueller**
- **K. Bushby**

**G.P.6.06**

Systematic screening for genomic deletions and duplications in the dysferlin gene using multiplex ligation-dependant probe amplification and CGH microarrays

- **M. Krahn**
- **N. Wein**
- **A. Borges**
- **P. Bourgeois**
- **V. Labelle**
- **P. Negre**
- **C. Pecheux**
- **M. Bartoli**
- **N. Lévy**

**G.P.6.07**

The association of dysferlin and affixin is regulated by calcium concentration

- **C. Matsuda**
- **K. Kameyama**
- **I. Nishino**
- **Y.K. Hayashi**

**G.P.6.08**

The evaluation of novel therapeutic strategies for the treatment of dysferlinopathy

- **M. Hornsey**
- **Y. Chiu**
- **L. Jorgensen**
- **L. Klinge**
- **S. Laval**
- **R. Barresi**
- **V. Straub**
- **H. Lochmueller**
- **K. Bushby**

**G.P.6.09**

Immunolabelling and FACS as new tools to explore dysferlinopathies

- **W.N. Wein**
- **K.M. Krahn**
- **C.S. Courrier**
- **S.C.E. Salort-Campana**
- **N.K. Nguyen**
- **F.C. Fernandez**
- **P.J. Pouget**
- **F.C. Fossat**
- **D.D. Depetris**
- **L.F. Leturcq**
- **L.N. Lévy**

**Poster room 2**

**Poster 14 – Congenital myopathies II: nemaline myopathies and related myopathies**

**Chairpersons:** C. Sewry
<table>
<thead>
<tr>
<th>Poster Number</th>
<th>Title</th>
<th>Authors</th>
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</thead>
<tbody>
<tr>
<td>G.P.7.01</td>
<td>Analysis of the nebulin gene by multiplex-ligation dependent probe</td>
<td>M. Lunkka-Hytönen; V.L. Lehtokari; C. Wallgren-Pettersson; K. Pelin</td>
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<tr>
<td></td>
<td>amplification (MLPA) and methylation-specific MLPA</td>
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<tr>
<td>G.P.7.02</td>
<td>Studying the functional pathogenesis of nebulin: A giant myofibrillar</td>
<td>M. Hanif; M. Marttila; K. Pelin; M. Grönholm; C. Wallgren-Pettersson</td>
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<td>protein</td>
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<tr>
<td>G.P.7.03</td>
<td>Mutations in TPM2 and TPM3 causing nemaline myopathy or arthrogryposis</td>
<td>V.L. Lehtokari; H. Kalimo; K. Pelin; C. Wallgren-Pettersson</td>
</tr>
<tr>
<td>G.P.7.04</td>
<td>Mosaic of an ACTA1 mutation in a child with nemaline myopathy and</td>
<td>H. Amthor; S. Quijano-Roy; S. Drouhin; K.G. Claey; J. Bataille; B.</td>
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<td></td>
<td>cardiomyopathy</td>
<td>Estournet; N.B. Romero; N. Monnier; J. Lunardi</td>
</tr>
<tr>
<td>G.P.7.05</td>
<td>Prognosis of neonatal onset nemaline myopathy without molecular</td>
<td>L. Servois; S. Quijano-Roy; B. Estournet; N. Laing; T. Voit; N.B.</td>
</tr>
<tr>
<td></td>
<td>diagnosis</td>
<td>Romero</td>
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<tr>
<td>G.P.7.06</td>
<td>Infantile onset muscle stiffness with marked myofibrillar disruption</td>
<td>K. Forrest; I. Bodi; S. Al-Sarraj; S.V. Tan; M. Pitt; R. Kayani; A.</td>
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<td></td>
<td>on muscle biopsy</td>
<td>Durward; M. McDougall; M. Irving; H. Jungbluth; E. Wraige</td>
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<tr>
<td>G.P.7.07</td>
<td>A case of nemaline myopathy with dilated cardiomyopathy, who was</td>
<td>K. Nomura; S. Kimura; S. Ozasa</td>
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<td></td>
<td>treated with home mechanical ventilation</td>
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<tr>
<td>G.P.7.08</td>
<td>A new form of myopathy in four siblings with a distinctive muscle MRI</td>
<td>J.M. Cuisset; N.F. Clarke; C.A. Maurage; G. Vaksmann; S. Maugenre; S.</td>
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<td></td>
<td>pattern</td>
<td>Quijano-Roy; N.B. Romero; P. Guicheney</td>
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<tr>
<td>G.P.7.09</td>
<td>Functional studies of aberrant beta-tropomyosin causing nemaline</td>
<td>M. Marttila; E. Nuutinen; M. Hanif; T. Nyman; K. Donner; K. Pelin; M.</td>
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<td>myopathy and cap myopathy</td>
<td>Grönholm; C. Wallgren-Pettersson</td>
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<tr>
<td>G.P.7.10</td>
<td>Investigation of the patho-biology of MYH7 myopathy mutations</td>
<td>W. Wallefeld; K.J. Nowak; E. Ingle; W. Stanley; C. Bond; N. Laing</td>
</tr>
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<td>G.P.7.11</td>
<td>Central core and cap disease in a case of congenital myopathy</td>
<td>H. Karasoy; O.E. Ozbay; A.N. Yuceyar</td>
</tr>
<tr>
<td>G.P.7.12</td>
<td>Multiminicore myopathy caused by a mutation in MYH7</td>
<td>P.J. Lamont; W. Wallefeld; R. Junckerstorff; N.G. Laing</td>
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<td>Poster room 8</td>
<td>Poster 15 – Metabolic myopathy I: Pompe disease</td>
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<td>Chairpersons: A. van der Ploeg, P. Laforet</td>
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### THURSDAY 10 SEPTEMBER 2009

| G.P.8.01 | Diagnosis of Pompe disease using a rapid dried blood spot assay in patients with muscle weakness  
J.L. Goldstein ; S.P. Young ; M. Changela ; G.H. Dickerson ; H. Zhang ; J. Dai ; D. Peterson ; D.S. Millington ; S. Prasad ; P.S. Kishnani ; D. Bali |
| G.P.8.02 | High density on skeletal muscle CT imaging indicates accumulation of calcium in autophagic vacuoles in childhood-onset Pompe disease  
K. Ishigaki ; I. Nonaka ; R. Kuwatsuru ; T. Murakami ; K. Shishikukra ; H. Suzuki ; Y. Hirayama ; M. Osawa |
| G.P.8.03 | Misleading muscle biopsies in late onset Pompe's disease  
A.L. Taratuto ; A. Dubovsky ; J. Corderi |
| G.P.8.04 | Vascular glycogen storage in Pompe disease demonstrated by epon-embedded muscle section  
S. Mitsuhashi ; I. Nonaka ; S. Noguchi ; Y.K. Hayashi ; H. Sugie ; T. Fukuda ; I. Nishino |
| G.P.8.05 | The pharmacological chaperone AT2220 increases mutant acid alpha-glucosidase levels and reduces tissue glycogen in a mouse model of Pompe disease  
R. Khanna ; R. Soksa ; J. Feng ; Y. Lun ; A.C. Powe ; J. Flanagan ; H. Do ; D.J. Lockhart ; K.J. Valenzano |
| G.P.8.06 | The Rotterdam Motor function Test: a valid and reliable motor function scale for Pompe disease  
C.I. van Capelle ; J.M. de Vries ; N.A.M. van der Beek ; R.P.M. Gadiot ; P.A. van Doorn ; A.T. van der Ploeg |
| G.P.8.07 | Rate of disease progression and response to enzyme replacement therapy in children and adults with Pompe disease  
N.A.M. Van der Beek ; J.M. De Vries ; C.I. Van Capelle ; A.J.J. Reuser ; P.A. van Doorn ; A.T. van der Ploeg |
| G.P.8.08 | Early results in a cohort of late-onset Pompe patients treated with enzyme replacement  
D. de Castro ; K. Lalou ; V. Doppler ; C.A.M. Payan ; A. Thomé ; P. Laforêt ; and the Pompe Registry Study Group |
| G.P.8.09 | Late onset Pompe disease: Clinical and genetic characterization in five Korean patients  
Y.E. Park ; J.H. Choi ; H.S. Kim ; D.S. Kim ; C.M. Kim ; C.H. Lee |
| G.P.8.10 | Severe sleep apnea associated with adult onset Pompe disease: Improvement with alglucosidase alfa  
E. Kiyani ; R. Engin-Unver ; F. Deymeier ; Y. Parmar ; P. Serdaroglu-Oflazer |
| G.P.8.11 | Rigid spine syndrome revealing late-onset Pompe disease  
P. Laforêt ; V. Doppler ; C. Cailloud ; K. Claey ; P. Richard ; K. Lalou ; A. Ferreiro ; B. Eymard |

**Poster room 9**

**Poster 16 – Spinal muscular atrophies**  
Chairpersons: R. Finkel, E. Bertini
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| G.P.9.01 | Albuterol promotes sustained improvement in motor function in spinal muscular atrophy type III  
Setayesh, M.; Sheth, S.; Labus, J.; Arango, C. |
| G.P.9.02 | In vitro evaluation of HDAC inhibitors for the treatment of spinal muscular atrophy  
M. Muchir; H. Deppe; B. Hoffmann; M. Soeberdt; N. Güven |
| G.P.9.03 | Quality of life and motor ability in SMA  
A.P.Q. Araujo; C.M. Oliveira |
| G.P.9.04 | Dysphagia in spinal muscular atrophy type II, more than a bulbar problem?  
L. van den Engel-Hoek; C.E. Erasmus; H.W. van Bruggen; B.J.M. de Swart; L.T. Sie; M.H. Steenks; I.J.M. de Groot |
| G.P.9.05 | Prenatal delay of muscle maturation in spinal muscular atrophy  
R. Martínez-Hernández; S. Bernal; L. Alias; E. Also; C. Soler-Batija; E.F. Tizzano |
| G.P.9.06 | Infantile inflammatory myopathy presenting as SMARD 1  
V. Crugnola; I. Colombo; G. Rossetti; P. Ciscato; A. Prelle; G. Comi; N. Bresolin; M. Moggio; C. Lamperti |
| G.P.9.07 | DNAJB2 gene expression in normal and diseased human and mouse skeletal muscle  
K.G. Claeys; M. Sozanska; J.J. Martin; E. Lacene; L. Vignaud; A. Kichler; D. Scherman; T. Voit; D. Israeli |
| G.P.9.08 | Insights into the pathological basis of autosomal dominant distal spinal muscular atrophy from a large Australian family  
E.C. Oates; N.F. Clarke; S.W. Reddel; K.N. North |
| G.P.9.09 | A new type of autosomal dominant adult onset spinal motor neuronopathy  
M. Jokela; B. Udd; S. Penttilä; A.M. Saukkonen; J. Toivanen; T. Suominen |
| G.P.9.10 | Standardized platform for testing therapeutics in a neonatal mouse model of spinal muscular atrophy  
B. El-Khodor; S. Ramboz; A. Chen; H. Bowling; K. Chen; M. Winberg |

18:00-20:30  
Auditorium (R380)  
**Special Industry Forum:**  
Emerging Therapies for Neuromuscular Diseases  
Presenting companies and organisations:

- Genzyme  
- Santhera Pharmaceuticals  
- Treat-NMD  
- ProSensa

Food and drinks offered
### Full Program

**FRIDAY 11 SEPTEMBER 2009**

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| 09:00-10:30| Auditorium (R380) | The extracellular matrix in normal and diseased muscle; Invited lectures (EM.I. 1–3)  
*Chairpersons: K. Flanigan, M. Rüegg* |
|            |                | EM.I.1 Laminin functions along the neuromuscular axis  
*P.D. Yurchenco; K.M. McKee; S. Capizzi; D.H. Yang* |
|            |                | EM.I.2 Toward a mitochondrial therapy of collagen VI muscular dystrophies  
*P. Bernardi; P. Bonaldo; P. Sabatelli; N.M. Maraldi; L. Merlini* |
|            |                | EM.I.3 Muscle and its extracellular matrix: Genetics disorders of the “Myomatrix”  
*C.G. Bonnemann* |
| 10:30-11:00| Main Hall      | Morning Tea and Coffee                                                   |
| 11:00-11:30| Auditorium (R380) | The extracellular matrix in normal and diseased muscle; Oral presentations (EM.O.1–2)  
*Chairpersons: K. Bushby, T. Toda* |
|            |                | E.M.O.1 E3 domain of laminin α1 chain that binds to dystroglycan is not essential for survival and muscle regeneration in laminin α1 chain mediated correction of laminin α2 chain deficiency  
*K.I. Gawlik; V. Carmignac; M. Åkerlund; H. Elamaa; M. Durbeej* |
|            |                | E.M.O.2 Residual laminin-binding activity and enhanced dystroglycan glycosylation by LARGE in novel model mice to dystroglycanopathy  
*M. Taniguchi; M. Kanagawa; S. Takeda; S. Yuko Miyagoe-Suzuki; S. Takeda; T. Endo; K. Kobayashi; K.P. Campbell; T. Toda* |
| 11:30-12:30| Auditorium (R380) | New insights into neuromuscular diseases; Oral presentations (G.O. 5–8)  
*Chairpersons: K. North, D. Hilton-Jones* |
|            |                | G.O.5 A new autosomal dominant distal vacuolar myopathy associated with mutation of the nuclear matrix protein, matrin 3  
*J. Senderek; S.M. Garvey; M. Krieger; I. Tournev; M. Elbracht; A. Roos; C. Stendel; A. Urtizberea; V. Guergueltcheva; V. Mihailova; H. Feit; J. Tramonte; P. Hedera; C. Bergmann; S. Rudnik-Schöneborn; K. Zerres; H. Lochmüller; E. Seboun; J.S. Beckmann; M.A. Hauser; C.E. Jackson; J. Weis* |
|            |                | G.O.6 New variant of myofibrillar myopathy with CNS involvement  
*S. Shalaby; Y.K. Hayashi; H. Mitsuhashi; K. Goto; I. Nonaka; S. Noguchi; I. Nishino* |
|            |                | G.O.7 A homozygous desmin deletion causes an Emery-Dreifuss like recessive myopathy with desmin depletion  
*V. Carmignac; S. Sharma; S. Arbogast; D. Fischer; C. Serreri; M. Serria; G. Stoltenburg; C.A. Maurage; H. Herrmann; J.M. Cuisset; H. Bär; A. Ferreiro* |
**Full Program**

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<th>Event Description</th>
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<tr>
<td>12:30-14:00</td>
<td>Main Hall</td>
<td>Lunch and Exhibition</td>
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<tr>
<td>14:00-15:00</td>
<td>Poster room 4</td>
<td>Poster session 3: parallel sessions (17–24)</td>
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**Poster room 4**

**Poster 17 – Functional muscle testing and myometry**
*Chairpersons: T. Sejersen, P.Y. Jeannet*

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<th>Authors</th>
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<tbody>
<tr>
<td>G.O.8 Impaired neuromuscular transmission and response to acetylcholinesterase inhibitors in centronuclear myopathy</td>
<td>S.A. Robb; C.A. Sewry; A.Y. Manzur; R.K. Knight; K.R. Mills; M. Pike; C. Muller; M.M. Lees; T. Cullup; H. Jungbluth; M.C. Pitt; F. Muntoni</td>
</tr>
<tr>
<td>M.P.3.01 Gait steadiness and upper-body kinematics in DMD children</td>
<td>R. Ganea; N. Goemans; M. van den Hauwe; K. Aminian; A. Paraschiv-Ionescu P.Y. Jeannet</td>
</tr>
<tr>
<td>M.P.3.02 Detailed analysis of daily-life physical activity patterns in DMD children</td>
<td>P.Y. Jeannet; R. Ganea; C. Piot; N. Goemans; M. van den Hauwe; K. Aminian; A. Paraschiv-Ionescu</td>
</tr>
<tr>
<td>M.P.3.03 6-minute walk test in Duchenne muscular dystrophy: longitudinal observations</td>
<td>C. McDonald; E. Henricson; R.T. Abresch; A. Nicorici; L. Atkinson; A. Reha; G.L. Elfring; L.L. Miller</td>
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<tr>
<td>M.P.3.04 Assessment of StepWatch™ activity monitoring in phase 2b study of ataluren (PTC124™) in nmDMD/BMD</td>
<td>C. McDonald; K. Coleman; E. Henricson; R.T. Abresch; M. Eagle; J. Florence; E. Gappmoier; A.M. Glanzman; A. Reha; G.L. Elfring; L.L. Miller</td>
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<tr>
<td>M.P.3.05 Use of a novel system for grading timed function test performance in phase 2b study of ataluren (PTC124™) in nonsense mutation Duchenne and Becker muscular dystrophy (nmDMD/BMD)</td>
<td>M. Elfring; R.T. Abresch; J. Florence; E. Gappmoier; A.M. Glanzman; E. Henricson; A. Reha; G.L. Elfring; L.L. Miller; L. Atkinson</td>
</tr>
<tr>
<td>M.P.3.06 Quantification of upper limb activity in non ambulant boys with Duchenne muscular dystrophy using accelerometry: a proof-of-concept study</td>
<td>L. Servais; A. Canal; J.Y. Hogrel; T. Voit</td>
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<td>M.P.3.07 Isometric dynamometry with a Biodex system for the follow-up of patients suffering from LGMD2A (calpainopathy)</td>
<td>J.Y. Hogrel; G. Olivier; A. Canal; Calpain Study Group</td>
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<td>M.P.3.08 How can measurement of muscle torque in children by hand-held dynamometry enhance clinical decision making</td>
<td>J. Saulnier; L.J. Hébert; C. Lepage; D.B. Maltais</td>
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<td>M.P.3.09 Relation between total power estimated from 3D accelerometric data and distance covered during the six-minute walk test</td>
<td>I. Ledoux; V. Decostre; A. Canal; J.Y. Hogrel</td>
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Poster room 5

Poster 18 – Methodology: general approaches
Chairpersons: B.T. Darras, V. Nigro

M.P.4.01 The value of immunolabelling of myosin heavy chains in the assessment of muscle biopsies
L.H. Feng; N. Bhardwaj; F. Muntoni; C.A. Sewry

M.P.4.02 Perioral skin biopsy to study skeletal muscle protein expression
L. Santoro; C. Fiorillo; M. Nolano; V. Provitera; S. Faraso; C. Vitiello; S. Aurino; V. Nigro

M.P.4.03 Analysis of dynamic muscle properties of FES-activated denervated m. quadriceps
M. Krenn; D. Rafolt; E. Gallasch; H. Kern; W. Mayr

M.P.4.04 Force-generating capacity of human myosin isoforms: results from a modified single muscle fiber in vitro motility assay
M. Li; L. Larsson

M.P.4.05 Electrical impedance myography for non-invasive quantification of spinal muscular atrophy
B.T. Darras; C. Lin; P. Fogerson; H. Butler; J. Caracciolo; S.B. Rutkove

M.P.4.06 Effects of ageing and gender on the spatial organization of nuclei in single human skeletal muscle cells
A. Cristea; P. Karlsson Edlund; J. Lindblad; R. Qaisar; E. Bengtsson; L. Larsson

M.P.4.07 Reverse protein arrays for efficient protein diagnosis of muscular dystrophies in less than 10 mg muscle tissue
C.A. Escher; H. Lochmüller; M.C. Walter; M. Ehrat; J. Reimann; M.A. Ruegg; D. Gygax

Poster room 2

Poster 19 – Collagen VI – related muscular dystrophy I: clinical and genetic approaches
Chairpersons: B. Talim, R. Barresi

EM.P.4.01 Collagen VI related myopathies: exploring the phenotypic spectrum
A.R. Foley; A. Columbus; J. Schlessl; Y. Hu; Y. Zou; S. Iannaccone; K. Mathews; A. Connolly; B. Wong; P. Kaufmann; M. Scavina; K. Flanagan; R. Weiss; R. Finkel; C.G. Bönnemann

EM.P.4.02 Comprehensive clinical, cellular and molecular assessment of 64 French families with COL6-related muscle disorders: clues for genotype/phenotype correlations
P. Richard; L. Briñas; S. Quijano-Ray; C. Ledeuil; A. Ferreiro; C. Gartioux; I. Pénisson-Besnier; A. Béhin; P. Lafolet; M. Mayer; L. Violette; P. Guicheney; B. Eymard; B. Estournet; T. Stojkovic; V. Allamand
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<th>Session</th>
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<tr>
<td>EM.P.4.03</td>
<td>Extensive sequencing of COL6A genes in a cohort of 65 patients with collagen type VI related myopathies. Focus on splicing mutations causing Ullrich congenital muscular dystrophy</td>
<td>E. Martoni; A. Urciuolo; C. Trabandelli; M. Fabris; P. Sabatelli; E. Bertini; E. Mercuri; N.M. Maraldi; P. Bernardi; P. Bonaldo; L. Merlini; A. Ferlini; F. Gualandi</td>
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<tr>
<td>EM.P.4.04</td>
<td>Unusual Col6A mutations in two Ullrich CMD patients</td>
<td>R.J. Butterfield; G. Ascadi; K.J. Swoboda; K.M. Flanigan; R.B. Weiss</td>
</tr>
<tr>
<td>EM.P.4.05</td>
<td>Bethlem myopathy; new insights on prevalence, phenotypic variability and genetic heterogeneity</td>
<td>J. Collins; D. Hicks; A. Sarcozy; A. Lampe; F. Norwood; V. Straub; H. Lochmüller; K. Bushby</td>
</tr>
<tr>
<td>EM.P.4.06</td>
<td>Autosomal recessive inheritance of classic Bethlem Myopathy</td>
<td>A.R. Foley; J. Shoffner; R. Weiss; C.G. Bönnemann</td>
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<td>EM.P.4.07</td>
<td>Autosomal recessive Bethlem myopathy</td>
<td>F. Gualandi; A. Urciuolo; E. Martoni; P. Sabatelli; S. Squarzoni; M. Bovolenta; S. Messina; E. Mercuri; A. Franchella; A. Ferlini; P. Bonaldo; L. Merlini</td>
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<tr>
<td>EM.P.4.08</td>
<td>Chain-specific antibody testing of collagen VI: an additional pre-screening strategy for the diagnosis of Bethlem myopathy</td>
<td>R. Charlton; M. Henderson; J. Richards; D. Hicks; M. Reza; V. Straub; H. Lochmüller; K. Bushby; R. Barresi</td>
</tr>
<tr>
<td>EM.P.4.09</td>
<td>Immunofluorescence and morphological alterations of capillary wall in skeletal muscle of two myosclerosis myopathy patients</td>
<td>P. Sabatelli; P. Grumati; E. Martoni; A. Zamparelli; S. Squarzoni; A. Urciuolo; F. Gualandi; A. Ferlini; L. Merlini; W.B. Stallcup; P. Bonaldo</td>
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<tr>
<td>EM.P.4.10</td>
<td>Severe hand contractures and abnormal collagen VI secretion not due to mutations in the COL6 genes: a novel entity?</td>
<td>O. Dubourg; B. Eymard; C. Gartioux; C.G. Bönnemann; C. Job-Deslandre; V. Allamand; P. Richard; A. Ferreiro</td>
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**Poster room 3**

**Poster 20 – Clinical trial approaches for Duchenne muscular dystrophy**

Chairpersons: G. Buyse, B. Wong

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<th>Authors</th>
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<tr>
<td>T.P.3.01</td>
<td>Glucocorticoids influence therapeutic efficacy of idebenone (Catena®) on peak expiratory flow in patients with Duchenne muscular dystrophy (DMD)</td>
<td>G.M. Buyse; N. Goemans; M. van den Hauwe; I.J.M. de Groot; U. Schara; B. Ceulemans; T. Meier</td>
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<tr>
<td>T.P.3.02</td>
<td>Use of glucocorticoids in Duchenne MD: Consensus Report of the CDC Duchenne Care Considerations Neurology Panel</td>
<td>R. Finkel; D. Biggar; C. Bönnemann; C. Constantin; D. Escolar; E. Massey; T. Miller; J. Pascual; J. Sladky; K. Wagner; B. Wong; K. Bushby</td>
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<tr>
<td>T.P.3.03</td>
<td>Long-term, low-dosage, early steroid therapy in Duchenne muscular dystrophy prolongs ambulation and preserves respiratory and cardiac function. 13 years follow-up</td>
<td>L. Merlini ; E. Malaspina ; A. Cicognani ; E. Franzoni ; A.R. Armaroli ; B. Talim ; F. Gualandi ; M.E. Michelini ; A. Ferlini</td>
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<td>T.P.3.04</td>
<td>Clinical trial using nitric oxide releasing drug and nonsteroidal anti-inflmmatory drugs in muscular dystrophy: design of a study</td>
<td>S. Gandossini ; M.G. d’Angelo ; S. Bonato ; G.P. Comi ; F. Magri ; M. Maggio ; M. Sciaccio ; A.C. Turconi ; C. Sciarati ; N. Bresolin ; E. Clementi</td>
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<td>T.P.3.05</td>
<td>Reproducibility and correlation of pretreatment outcome measures in phase 2b study of ataluren (PTC124™) in nonsense mutation Duchenne and Becker muscular dystrophy (nMMD/BMD)</td>
<td>J. Florence ; R.T. Abresch ; M. Eagle ; E. Gappmaier ; A.M. Glanzman ; E. Henricson ; A. Reha ; G.L. Elfring ; L.L. Miller ; L. Atkinson</td>
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<td>T.P.3.06</td>
<td>Metformin improves weight gain and body mass index in Duchenne muscular dystrophy</td>
<td>M.M. Rutter ; B.L. Wong ; S.R. Rose ; D.J. Klein</td>
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<td>T.P.3.07</td>
<td>Design of an international phase III study with idebenone (Catena®) in patients with Duchenne muscular dystrophy (DMD) - the DELOS study</td>
<td>G.M. Buyse ; T. Meier ; R.S. Finkel</td>
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<td>T.P.3.08</td>
<td>Clinical trial of epigallocatechin-3-gallate in Duchenne muscular dystrophy</td>
<td>A. von Moers ; F. Paul ; M. Schülke ; S. Ohlraum ; Y. Nakae ; O.M. Dorchies ; U.T. Ruegg</td>
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<td>T.P.4.01</td>
<td>Effects of idebenone on mitochondrial function in cells from muscular dystrophy patients and healthy individuals</td>
<td>R. Haefeli ; A. Gemperli ; N. Güven</td>
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<td>T.P.4.02</td>
<td>Effects of rituximab treatment in two patients affected by dysferlin-deficient muscular dystrophy</td>
<td>A. Lerario ; F. Cogiamanian ; C. Marchesi ; S. Bonfiglio ; M. Belicchi ; L. Porretti ; N. Bresolin ; Y. Torrente</td>
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<td>T.P.4.03</td>
<td>Treatment with bortezomib (PS-341) of Golden retriever muscular dystrophy (GRMD): analysis of proteasome inhibition and morphology of dystrophic skeletal muscle</td>
<td>K.P.C. Araújo ; D.F. Moreira ; T.P. Gaiad ; M.A. Miglino ; S.L. Gorniak ; D. Feder ; J.E. Belizário ; C.E. Ambrosio</td>
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| T.P.4.04 | Automated drug screening with contractile muscle tissue engineered from dystrophic myoblasts  
*H. Vandenburgh; J. Shansky; F. Benesch-Lee; K. Skelly; J. Spinazzola; Y. Saponjian; B.S. Tseng* |
| T.P.4.05 | Jaw exercise in patients with Duchenne muscular dystrophy (multi center study) - therapeutic intervention to the masseter muscle  
*S. Nozaki; M. Kawai; R. Shimoyama; N. Futamura; T. Matsumura; Y. Kikuchi* |
| T.P.4.06 | Does physiotherapy change the fibrosis amount on canine dystrophic muscle and dynamical features of gait?  
*T.P. Gaiad; T.M. Guimarães; K.P.C. Araujo; F.A. Caromano; J.C. Serrão; M.A. Migliino; C.E. Ambrosio* |
| T.P.4.07 | The opening of pulmonary respiration resulted in muscle degeneration of diaphragm in canine X-linked muscular dystrophy  
*A.N. Nakamura; M.K. Kobayashi; K.Y. Yuasa; N.Y. Yugeta; S.T. Takeda* |
| T.P.4.08 | Oxidative stress as a therapeutic target: efficacy of antioxidants in the ex vivo treatment of SEPN1-related myopathy  
*S. Arbogast; A. Ferreiro* |
| T.P.4.09 | Measuring restored dystrophin in treated muscle: an immunohistological intensity measurement method  
*V. Arechavala-Gomeza; L. Feng; A. Malerba; I.R. Graham; S.C. Brown; C. Sewry; J. Morgan; F. Muntoni* |
| T.P.4.10 | Towards a better understanding of truncated dystrophin instability  
*V. Robin; C. Beley; M. Reboud; T. Voit; L. Garcia* |

**Poster room 1**  
**Poster 22 – Pharmacological approaches to therapy using mouse models**  
Chairpersons: O. Dorchies, Y. Sunada

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*Y. Hibaoui; O.M. Dorchies; J. Reutenauer; O. Patthey-Vuadens; U.T. Ruegg* |
| T.P.5.02 | Melatonin prevents oxidative-stress mediated mitochondrial permeability transition and death in mouse skeletal muscle cells  
*Y. Hibaoui; E. Roulet; U.T. Ruegg* |
| T.P.5.03 | Restoring mitochondrial function in Duchenne muscular dystrophy by idebenone  
*A. Gemperli; M. Hufschmid; I. Courdier-Fruh; R. Haefeli; M. Erb; R. Dallmann; N. Güven* |
| T.P.5.04 | Effect of a mild exercise regime on disease parameters in the mdx mouse model  
*M. van Putten; C.L. de Winter; W.M.C. van Roon-Mom; G.J.B. van Ommen; P.A.C. ‘t Hoen; A.M. Aartsma-Rus* |
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<td>Muscle-specific expression of myotubularin ameliorates the phenotype of targeted muscles in mouse models of X-linked myotubular myopathy</td>
<td>T. Jamet; F. Fougerousse; N. Guerchet; A. Douar; J.L. Mandel; M. Montus; A. Buj-Bello</td>
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<td>Pharmacological characterization of phospholipase A2 activities in primary cultures of mouse dystrophic muscle</td>
<td>O.M. Dorchies; A.K. Johansson; U.T. Ruegg</td>
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<td>Substantial improvement of laminin α2 chain deficiency by laminin α1 chain overexpression persists throughout life</td>
<td>K.I. Gawlik; M. Durbeej</td>
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<td>T.P.5.08</td>
<td>Contribution of reactive oxygen species generated through NADPH oxidase to abnormal Ca2+ signals in dystrophic skeletal muscle cells</td>
<td>Y. Hibaoui; K. Bedard; K.H. Krause; U.T. Ruegg</td>
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<td>T.P.5.09</td>
<td>Reduced mitochondrial copy number in mdx diaphragm – an adaptive response to reduce oxidative stress?</td>
<td>M. Erb; L. Sumanovski; I. Courdier-Fruh; N. Güven</td>
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<td>Y. Ohsawa; M. Fujino; T. Okada; A. Kuga; S. Hayashi; M. Rikimaru; Y. Sunada</td>
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<td>Lamp1 overexpression may rescue cardiomyopathy in Lamp2 deficient mice</td>
<td>S. Honda; S. Noguchi; M.C.V. Malicdan; Y.K. Hayashi; P. Saftig; I. Nishino</td>
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<td>T.P.5.12</td>
<td>Regeneration is up-regulated in glatiramer acetate treated dy2J/dy2J mice with congenital muscular dystrophy</td>
<td>O. Dadush; S. Aga-Mizrachi; K. Ettinger; R. Tabakman; M. Elbaz; J. Fellig; V. Barak; Y. Nevo</td>
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<td>T.P.5.13</td>
<td>Oxidative stress in mdx muscle: source and consequences</td>
<td>I. Courdier-Fruh; R. Dollmann; N. Güven</td>
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**Poster room 8**

**Poster 23 – Limb girdle muscular dystrophy – II**

Chairpersons: P. Hackman, A. van der Kooij

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<td>G.P.10.02</td>
<td>Does 6-sarcoglycan-associated autosomal dominant cardiomyopathy exist?</td>
<td>A. Sarkozy; R. Buer; J. Hudson; H.D. Müller; C. Sommer; G. Dekomien; J. Bourke; D. Routledge; K. Bushby; J. Klepper; V. Straub</td>
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G.P.10.03 Refinement of the LGMD1 locus on 7q36 by genotyping new Finnish families
P. Hackman; S. Sandell; J. Sarparanta; H. Luque; S. Huovinen; O. Raheem; P.H. Jonson; I. Mahjneh; B. Udd

G.P.10.04 PLEC1-related epidermolysis bullosa simplex-muscular dystrophy (EBS-MD) with early onset and associated myasthenic symptoms
K. Forrest; J. Melero; S. Robb; S. Goyal; H. Jungbluth; E. Wraige

G.P.10.05 Pathogenic caveolin-3 mutations: influence on canonical signalling pathways in vitro and effects in a rat muscle in vivo model of caveolinopathy
E. Brauers; P. Martinez; M. De Baets; A. Krüttgen; J. Weis

G.P.10.06 Caveolinopathy presenting as neonatal hypotonia
M. Olivé; L. Gonzalez-Quereda; P. Gallano; M. Roig; F. Munell; A. Sanchez; A. Macaya; I. Ferrer

G.P.10.07 Differential expression of microRNAs in calpainopathies
A.M. Aguennouz; M.O. Musumesci; R.C. Rodolico; L.N. Lanzano; G.A. Garufi; D.P.M. De Pasquale; V.G. Vita; T.A. Toscano

G.P.10.08 Myophosphorylase deficiency and calpainopathy in the same patient
N. Pulur; Y. Parman; F. Deymeer; P. Serdaroglu-Öflazer

G.P.10.09 Description of a family associating a calpainopathy and a Duchenne myopathy
S. Nouioua; A.M. Cobo; T. Benhassine; A. Urtizberea; S. Slimani; N. Terki; S. Assami; M. Tazir

Poster room 9 Poster 24 – Metabolic myopathy II and neuromuscular junction disorders
Chairpersons: J. Colomer, C. Bruno

G.P.11.01 RNA processing differences explain tissue specificity in exercise intolerance myopathy due to ISCU intronic mutation
P.S. Sanaker; M. Toompuu; Z. Chrzanowska-Lightowlers; L. He; V. Hogan; R.W. Taylor; C. Tzoulis; L.A. Bindoff

G.P.11.02 Early-onset neutral lipid storage disease with myopathy due to PNPLA2 mutations
D. Cassandrini; F. Trucco; S. Scapolan; G.M. Magnano; S. Pessano; F. Scuderi; A. Rossi; M. Pedemonte; C. Minetti; C. Bruno

G.P.11.03 Reduced oxygen availability in muscle of exercising glycogenosis type III patients shown by multi-parametric functional NMR
C. Wary; A. Nadaj-Pakleza; P. Lafort; A. Monnet; S. Fleury; R. Carlier; C. Baligand; B. Eymard; P. Labrune; P.G. Carlier

G.P.11.04 Pattern and evolution of skeletal muscle involvement in glycogenosis type III (debrancher deficiency): muscle imaging findings from childhood to adult age
A.A. Nadaj-Pakleza; J. Renoux; P. Carlier; A. Mollet; P. Labrune; A. Behin; B. Eymard; P. Laforet
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<td>A life threatening case of α-enolase deficiency</td>
<td>O. Musumeci; C. Rodolico; A. Ciranni; M. Aguennouz; N. Lanzano; A. Toscano</td>
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<td>Clinical correlations of binding and blocking acetylcholine receptor antibodies in myasthenia gravis</td>
<td>S.Y. Kang; J.H. Kang; J.C. Choi; J.S. Lee</td>
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<td>Elevated serum level of interleukin-32 in the patients with myasthenia gravis</td>
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<td>Long term remissions in MuSK-positive myasthenia gravis after a single course of rituximab</td>
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<td>G.P.11.09</td>
<td>Congenital end-plate acetylcholinesterase deficiency and the effect of ephedrine on clinical findings, lung functions and nocturnal parameters</td>
<td>E. Kiyan; B. Kara; P. Serdaroglu Oflazer; Y. Parman; N. Tosdemir; F. Deymeer; A.G. Engel</td>
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<td>G.P.11.10</td>
<td>Partial genomic deletions of RAPSN account for 15% of congenital myasthenic syndrome after negative DNA Sequencing</td>
<td>P. Richard; K. Gaudon; I. Pénisson-Besnier; B. Chabrol; F. Bouhour; C. Vial; A. Ben Ammar; S. Bauché; B. Eymard; D. Hantai</td>
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<td>G.P.11.11</td>
<td>Myasthenic syndrome due to defects in rapsyn: clinical and molecular findings in 39 patients</td>
<td>M. Milone; X.M. Shen; D. Selcen; K. Ohno; J. Brengman; S.T. Iannaccone; C.M. Harper; A.G. Engel</td>
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**15:00-15:30** Poster rooms (1 - 9)  
Poster Viewing session 3: presenters of all posters discussed on Friday 11 September to be available for poster viewing

**15:30-16:00** Main Hall  
Afternoon Tea and Coffee and Exhibition

**16:00-17:00** Poster session 4: parallel sessions (25–33)

**Poster room 3**  
Poster 25 – New methods of imaging  
Chairpersons: D. Fischer, S. Quijano-Ray

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<td>Magnetic resonance imaging of cardiac function in mouse models for muscular dystrophy associated cardiomyopathy</td>
<td>E. Greally; B.J. Davison; R. Patel; A. Blain; S. Laval; H. Lochmüller; K. Bushby; A. Blamire; G. MacGowan; V. Straub</td>
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<td>M.P.5.02</td>
<td>Evaluating muscle function in mice lacking myostatin by functional nuclear magnetic resonance in vivo: preliminary results</td>
<td>C. Baligand; H. Gilson; J.C. Ménard; O. Schakman; C. Wary; J.P. Thissen; P.G. Carlier</td>
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M.P.5.03 Specificity and sensitivity of patterns of muscle MRI involvement in muscular dystrophies with rigidity of the spine
E. Mercuri; E. Clement; A. Offiah; A. Pichiecchio; J. Alsopp; G. Vasco; F. Bianco; A. Manzur; S. Messina; E. Ricci; M. Rutherford; F. Muntoni

M.P.5.04 Evaluation of in-phase and out-of-phase and FISP MRI to quantify muscle fat content in OPMD
D. Fischer; K. Scheffler; K. Heinimann; M. Tolnay; R. Rodoni; A. Fischmann; M. Gloor

M.P.5.05 Whole-body muscle MRI in collagen type VI-related myopathies (Ullrich CMD and Bethlem myopathy)
S. Quijano-Roy; D. Avila-Smirnow; M. Zayani; S. Chaabane; M. Hamida; B. Estournet; L. Viollet; D. Fischer; P. Cuvelier; A. Ferreiro; N. Dehlinger; N.B. Romero; L. Briñas; C. Gartioux; P. Guicheney; P. Richard; V. Allamand; P.G. Carlier; R. Carlier

M.P.5.06 Steroids and T2 relaxation time mapping of the gluteal muscles in patients with Duchenne muscular dystrophy (DMD): longitudinal and quantitative assessment
H. Kim; T. Laor; B. Dardzinski; M. Chadehumbe; B.L. Wong

M.P.5.07 Serial high-resolution muscle ultrasonography
A. van Baalen; G. Wiegand; U. Stephani

M.P.5.08 In vivo imaging of muscle fibers under normal and pathological conditions
J. Böhm; M. Koch; Y. Lutz; J.L. Vonesch; D. Hentsch; J.L. Mandel; J. Laporte

M.P.5.09 Non-invasive assessment of muscle stiffness with magnetic resonance elastography
L. Debernard; J.Y. Hogrel; S.F. Bensamoun

Poster room 4

Poster 26 – Collagen VI – related muscular dystrophy II: experimental approaches
Chairpersons: H. Topaloglu, V. Allamand

EM.P.5.01 Gene expression and proteome profiles in Col6a1-/- mice, a model of Ullrich congenital muscular dystrophy (UCMD)
M. Bovolenta; S. De Palma; M. Vasso; N.M. Maraldi; F. Gualandi; L. Merlini; P. Sabatelli; T. Tiepolo; P. Bonaldo; P. Bernardi; C. Gelfi; A. Ferlini

EM.P.5.02 Role of mitochondria in the pathogenesis of muscular dystrophies
F. Palma; A. Angelin; T. Tiepolo; P. Sabatelli; N.M. Maraldi; L. Merlini; P. Bonaldo; P. Bernardi
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**EM.P.5.03**
The cyclophilin inhibitor Debio 025 normalizes mitochondrial function, muscle apoptosis and ultrastructural defects in Col6a1-/- myopathic mice

_A. Angelin; T. Tiepolo; E. Palma; P. Sabatelli; L. Merlini; L. Nicolosi; F. Finetti; P. Braghetta; G. Vuagniaux; J.M. Dumont; C.T. Baldari; P. Bonaldo; P. Bernardi_

**EM.P.5.04**
Genetic ablation of cyclophilin D rescues mitochondrial defects and prevents muscle apoptosis in collagen VI myopathic mice

_T. Tiepolo; E. Palma; A. Angelin; P. Sabatelli; N.M. Maraldi; E. Basso; A.M. Forte; P. Bernardi; P. Bonaldo_

**EM.P.5.05**
Zebrafish models of collagen VI myopathies

_M.J. Blankinship; A.S. Busta; E.L. Feldman; J.J. Dowling_

**EM.P.5.06**
Collagen VI alpha5 chain exhibits a restricted localization at junctions in human skeletal muscle and skin

_R. Wagener; S.K. Gara; S. Squarzoni; A. Zamparelli; S. Santi; P. Grumati; A. Urciuolo; O. Donzelli; E. Martoni; F. Gualandi; L. Merlini; P. Bonaldo; P. Sabatelli; M. Paulsson_

**EM.P.5.07**
Abnormal elastin deposits and altered organization of elastic fibers in collagen VI related disorders

_S. Petrini; P. Sabatelli; A. d'Amico; F. Gualandi; P. Braghetta; M. Coccetti; T. Rizza; R. Carrozzo; G. Pepe; P. Bonaldo; A. Ferlini; L. Merlini; E. Bertini_

**EM.P.5.08**
Novel collagen VI alpha chains distribution in murine skeletal muscle: possible implications for neuromuscular disorders

_P. Sabatelli; S.K. Gara; S. Squarzoni; A. Zamparelli; P. Grumati; A. Urciuolo; A. Ferlini; L. Merlini; R. Wagener; M. Paulsson; P. Bonaldo_

**Poster room 9**
**Poster 27 – Therapy approaches using stem cells**

_Chairpersons: J-T. Vilquin, J.P. Tremblay_

**T.P.6.01**
Possible role of RTX in the differentiation of pericyte-like cells in Duchenne muscular dystrophy

_A. Farini; M. Meregalli; M. Belicchi; D. Parolini; G. D’Antona; S. Sangiorgi; G. Cosso; R. Bottinelli; N. Bresolin; Y. Torrente_

**T.P.6.02**
Aldehyde dehydrogenase activity identifies distinct populations of progenitors within human skeletal muscle

_K. Vauchez; J.P. Marolleau; M. Schmid; P. Khattar; A. Chapel; C. Catelain; S. Lecourt; J. Larghero; M. Fiszman; J.T. Vilquin_

**T.P.6.03**
Restoring cell-basal lamina interaction to rescue tissue degeneration in congenital muscular dystrophy

_E. Porrello; T. Domi; A. Capotondi; D. Triolo; M. Sampaolo; S. Brunelli; G. Comi; M.A. Ruegg; G. Cosso; A. Biffi; A. Quattrini; S.C. Previtali_
### T.P.6.04
**Comparison of skeletal muscle potential of mesenchymal stem-cells from different sources injected in SJL mice**
- E. Zucconi
- N.M. Vieira
- C.R. Bueno Junior
- V. Brandalise
- M. Secco
- M.F. Suzuki
- P. Bartolini
- P.C. Brum
- M. Vainzof
- M. Zatz

### T.P.6.05
**Poor maintenance of eGFP- mesenchymal stem cells in the dystrophic muscle**
- D. Ayub-Guerrieri
- P.C.G. Onofre-Oliveira
- V.F. Lopes
- P.C.M. Martins
- M. Vainzof

### T.P.6.06
**Real-time monitoring of cell transplantation in mouse dystrophic muscles by a secreted alkaline phosphatase reporter gene**
- X. Gerard
- L. Vignaud
- S. Charles
- C. Pinset
- D. Scherman
- A. Kichler
- D. Israeli

### T.P.6.07
**Lin(-) bone marrow stem cells transplantation repairs structure of mdx mice neuromuscular junctions**
- A.V. Sokolova
- V.V. Zenin
- V.M. Mikhailov

### T.P.6.08
**Effect of MAPK inhibition on the rhabdomyosarcoma cell line TE671: An in vitro model for the study of human muscle differentiation**
- E. Schirwis
- N. De Luna
- I. Illa
- E. Gallardo

### T.P.6.09
**Will it be possible to do a phase 2 clinical trial of myoblast transplantation for DMD patients?**
- J.P. Tremblay
- D. Skuk

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**Poster room 7**

**Poster 28 – Congenital myopathies III: centronuclear myopathies and actin myopathies**
*Chairpersons: H.H. Goebel, G.D. Shelton*

### G.P.12.01
**Immunodetection of myotubularin in human tissues: a diagnostic tool for X-linked myotubular myopathy**
- L. Al-Qusairi
- M.T. Bui
- C. Kretz
- K. Tomczak
- D. Furling
- A.H. Beggs
- J.L. Mandel
- N. Romero
- J. Laporte
- A. Buj-Bello

### G.P.12.02
**T-tubule disorganisation and defective excitation-contraction coupling in muscle fibres lacking myotubularin lipid phosphatase**
- L. Al-Qusairi
- N. Weiss
- C. Berbey
- N. Messaddeq
- C. Kretz
- D. Sanoudou
- A.H. Beggs
- B. Allard
- J.L. Mandel
- J. Laporte
- V. Jacquemond
- A. Buj-Bello

### G.P.12.03
**Centronuclear myopathy with cataracts due to a novel heterozygous mutation in the dynamin 2 (DNM2) gene**
- H. Jungbluth
- T. Cullup
- S. Lillis
- H. Zhou
- C. Sewry
- S. Abbs
- F. Muntoni

### G.P.12.04
**A missense variant in the MTM1 gene associated with X-linked myotubular myopathy in Labrador retrievers**
- G.D. Shelton
- J. Bohm
- E. Snead
- M. Kozlowski
- K. Minor
- L. Tiret
- M.K. Childers
- S.M. Taylor
- J.R. Mickelson
- L.T. Guo
- A.P. Mizisin
- J. Laporte
- A.H. Beggs
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<td>G.P.12.05</td>
<td>Myotubularins and the pathogenesis of centronuclear myopathies</td>
<td>J.J. Dowling; A. Busta; A.V. Vreede; J.Y. Kuwada; M. Wishart; E.L. Feldman</td>
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<td>G.P.12.06</td>
<td>Progressive muscle biopsies morphological changes in MTM1-related centronuclear congenital myopathy</td>
<td>F. Lubieniecki; V. Biancalana; N. Romero; S. Monges; M. Saccoliti; A.L. Taratuto</td>
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<td>G.P.12.07</td>
<td>A new ACTA1 mutation in two unrelated sporadic cases of neonatal form of CFTD</td>
<td>N. Deconinck; N. Monnier; M.C. Commare; P. Mezin; A. Michotte; J. Lunardi</td>
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<td>G.P.12.08</td>
<td>Zebra body myopathy resolved</td>
<td>C.A. Sewry; J. Holton; D.J. Dick; T. Jacques; F. Muntoni; M. Hanna</td>
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<td>G.P.12.09</td>
<td>Actin myopathy with myofibrillar dysgenesis and abnormal ossification</td>
<td>A. Arai; I. Nonaka; Y. Saito; F. Komaki; H. Sakuma; K. Sugai; M. Sasaki; S.P. Robertson; G. Nishimura; I. Nishino</td>
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<tr>
<td>G.P.12.10</td>
<td>Repeat muscle biopsy in the diagnosis of cap myopathy with ACTA1 gene mutation</td>
<td>R.M. Hung; C.E. Hawkins; G. Yoon; D. Biggar; J. Vajsar</td>
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<td>G.P.12.11</td>
<td>In vivo manipulation of skeletal muscle to characterize the mechanisms underlying centronuclear myopathies</td>
<td>B. Cowling; L. Amoasii; V. Tosch; P. Koebel; J.L. Mandel; J.F. Laporte</td>
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**Poster room 5**

**Poster 29 – Duchenne muscular dystrophy**

Chairpersons: B. Kakulas, H.B. Ginjaar

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<td>G.P.13.01</td>
<td>DYC-1 and ZYX-1, new actors of muscle degeneration in caenorhabditis elegans</td>
<td>C. Lecroisey; M.C. Mariol; Y. Schwab; M. Labouesse; L. Ségolat; K. Gieseler</td>
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<td>G.P.13.02</td>
<td>Non-coding RNAs within the DMD gene</td>
<td>M. Bovolenta; M. Neri; S. Brioschi; M. Fabris; C. Scotton; M.S. Falzarano; P. Rimessi; G. Perini; F. Gualandi; A. Ferlini</td>
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<td>G.P.13.03</td>
<td>Can VLA-4 be applied as a biomarker for prognosis and therapeutic target in Duchenne muscular dystrophy?</td>
<td>F. Pinto-Mariz; L.R. Carvalho; W. de Mello; A. Prufer; M.G. Ribeiro; M.C.S. Alves-Cunha; I. Riederer; E. Negroni; T. Voit; I. Desguerre; G. Butler-Browne; W. Savino; S.D. Silva-Barbosa</td>
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<td>G.P.13.04</td>
<td>In-frame deletion of the entire dystrophin rod domain prevents compensation by utrophin and causes an unusually severe muscular dystrophy</td>
<td>J. Schessl; M.L. Yang; P.T. Heydemann; R.L. Sufit; L. Medne; A. Attia; R.S. Finkel; C.G. Bönemann</td>
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G.P.13.05 The diagnosis and management of Duchenne muscular dystrophy: internationally generated care recommendations
K. Bushby; D. Birnkrant; L. Case; P. Clemens; L. Cripe; R. Finkel; A. Kaul; K. Kinnett; C. McDonald; S. Pandya; J. Poysky; F. Shapiro; J. Tomezsko; C. Constantin

G.P.13.06 Clinical phenotype of Becker muscular dystrophy patients with deletions of exons 45-51 and 50-51
C.S.M. Straathof; A.T.J. Helderman-van den Enen; A. Aartsma-Rus; J.T. den Dunnen; B.M. Verbiest; E. Bakker; J.J.G. Verschuuren; H.B. Ginjaar

G.P.13.07 Paediatric females with dystrophinopathy
L.C. McAdam; N. Seemann; K. Shelby; H. Kolski; C. Campbell; D. Biggar

G.P.13.08 Dystrophin gene analysis for identification of DMD/BMD carrier status in Egyptian symptomatic and asymptomatic females
R.M. El Sherif; I. Nonaka; N. Minami

G.P.13.09 Muscle biopsy mRNA-based analysis of point mutations in DMD gene in Spanish patients
J. Juan-Mateu; M.J. Rodriguez; L. González-Quereda; J. Colomer; A. Nascimento; A. Cabello; E. Rivas; M. Madruga; C. Paradas; M. Olivé; P. Gallano

G.P.13.10 Early cardiomyopathy in DMD
G.L. Vita; R. Kirk; H. Lochmüller; K. Bushby; V. Straub

G.P.13.11 Oral bisphosphonates as prophylaxis of steroid-induced osteoporosis in Duchenne muscular dystrophy
A. Sarkozy; T. Cheetham; S. Pearce; D. Rawlings; M. Eagle; G.L. Vita; M. Guglieri; V. Straub; K. Bushby

G.P.13.12 Parents vision on Duchenne muscular dystrophy (DMD) research
D. Feder; B.S. Seiguan

Poster room 2

Post 30 – Myotonic disorders
Chairpersons: V. Straub, C. Sewry

G.P.14.01 Holter monitoring versus Electrocardiogram (ECG) for the detection of cardiac arrhythmia in myotonic dystrophy type 1 (DM1)
J. Whitesell; J. Sampson; K. Whitehead

G.P.14.02 A high prevalence of Brugada syndrome among patients with Steinert’s disease: A new insight into the pathophysiology of Steinert’s disease’s cardiac complications
K. Wahbi; V. Fressart; H.M. Bécane; C. Meune; P. Richard; A. Lazarus; P. Laforté; N. Benammar; H. Radvanyi-Hoffman; B. Eymard; D. Duboc

G.P.14.03 Corneal thickness and endothelial cell characteristics in patients with myotonic dystrophy
N. Rosa; M. Lanza; M. Borrelli; M.L. Filosa; M. De Bernardo; M.R. Cecio; M.G. Di Gregorio; A. Palladino; L. Politano
### Full Program

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| G.P.14.04 | Congenital myotonic dystrophy in patients diagnosed as congenital fiber type disproporion  
K. Tominaga; Y.K. Hayashi; K. Goto; N. Minami; S. Noguchi; I. Nonaka; I. Nishino |
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| G.P.14.05 | Quantitative isometric muscle strength at the ankle in myotonic dystrophy type 1 Patients: A discriminative biological marker  
L.J. Hébert; J.F. Remec; J. Saulnier; C. Vial; J. Puymirat |
| G.P.14.06 | Sleep disturbances in myotonic dystrophy type 2: a comparison with myotonic dystrophy type 1 and healthy controls  
A.A. Tieleman; H. Knoop; A. van de Logt; G. Bleijenberg; B.G.M. van Engelen; S. Overeem |
| G.P.14.07 | In situ hybridization analysis of mRNA in differentiated tissues of adult patients with myotonic dystrophy 1 and 2  
Z. Lukáš; I. Falková; M. Falk; J. Zařídil; R. Hrabáková |
| G.P.14.08 | Analysis of the CLCN1 gene in Czech patients with myotonia congenita  
J. Sedlackova; S. Vohanka; M. Hermanova; P. Vondracek; L. Fajkusova |
| G.P.14.09 | Successful flecainide therapy in paramyotonia congenita  
D. Diodato; O. Farina; G. Ciccone; M. Mayer; S. Sampaoolo; G. Di Iorio |
| G.P.14.10 | Clinical experience with acetazolamide treatment in children; good response in both sodium and chloride channelopathies  
L.T.L. Sie; B. Stunnenberg; J. Trip; I.B. Ginjaar; G. Drost |
| G.P.14.11 | Newly synthesized mexiletine and tocainide analogues are potent use-dependent blockers of skeletal muscle sodium channels: potential implication for the antimyotonic activity  
M. De Bellis; A. De Luca; G. Lentini; A. Carocci; F. Corbo; C. Franchini; D. Conte Camerino |
| G.P.14.12 | Neuromyotonia in childhood: report of a case  
S. Makri; S. Maghnouche; B. Ghiar; M. Ait Kaci-Ahmed |
| Poster room 8 | **Poster 31 – Emery–Dreifuss muscular dystrophy and facioscapulo humeral muscular dystrophy**  
Chairpersons: G. Bonne, Y.K. Hayashi |
| G.P.15.01 | Inflammatory myopathy in early childhood is frequently associated with LMNA mutations  
H. Komaki; Y.K. Hayashi; M. Kata; H. Sakuma; Y. Saito; E. Nakagawa; K. Sugai; M. Sasaki; I. Nonaka; I. Nishino |
| G.P.15.02 | Autophagic degradation of nuclear component in nuclear envelopathy  
Y.F. Park; Y.K. Hayashi; G. Bonne; T. Arimura; S. Noguchi; I. Nonaka; I. Nishino |
| G.P.15.03 | A new antibody specifically recognizes muscles from laminopathy patients  
H. Mitsuhashi; Y.K. Hayashi; S. Noguchi; I. Nishino |
G.P.15.04 Collagen VI deficiency in skin fibroblasts from progeroid laminopathies
G. Lattanzi; A. Zamparelli; M. Stefanini; D. Orioli; G. Novelli; M. Wehnert; P. Sabatelli; S. Squarzoni

G.P.15.05 Characterization of Emd−/−/LmnaH222P/H222P double mutant mice
W.C. Liang; Y.K. Hayashi; Y.E. Park; H. Mitsuhashi; T. Arimura; G. Bonne; S. Noguchi; I. Nishino

G.P.15.06 Inhibition of c-Jun N-terminal kinases signalling to prevent cardiomyopathy caused by mutation in LMNA gene
A. Muchir; W. Wu; J. Shan; G. Bonne; H.J. Worman

G.P.15.07 Clinical features and genetic analysis of fascioscapulohumeral muscular dystrophy in Korean patients
J.M. Hong; D.S. Shim; B.C. Kim; D.S. Kim; I. Nishino; Y.C. Choi

G.P.15.08 About the autosomal dominant muscular dystrophy in the K. kindred once again. Clinical and molecular genetic study
V.M. Kazakov; D.I. Rudenko

G.P.15.09 High genetic variability in European population: the FSHD complex puzzle
M. Govi; I. Scionti; E. Signaroldi; S. Bennardo; F. Greco; G. Fabbri; L. Palmucci; M. Moggio; L. Santoro; G. Tomelleri; C. Angelini; C. Rodolico; G. Siciliano; M. Alù; G. Ferri; E. Bonifazi; R.G. Tupler

G.P.15.10 Unexpected high percentage of asymptomatic subjects carrying the FSHD molecular defect: which factors contribute to the disease mechanism?
E. Bonifazi; C. Lamperti; C. Fiorillo; L. Vercelli; C. Borsato; R. Frusciante; M. Servida; F. Greco; I. Frambolli; L. Colontoni; G. Ricci; L. Volpi; R. Di Leo; C. Manzoli; P. Cudia; E. Pastorello; L. Ricciardi; M. Govi; I. Scionti; M. Cao; G. Siciliano; G. Galluzzi; L. Morandi; A. Di Muzio; C.P. Trevisan; E. Ricci; C. Rodolico; L. Santoro; G. Tomelleri; C. Angelini; L. Palmucci; M. Moggio; R.G. Tupler

G.P.15.11 Patients with facioscapulohumeral muscular dystrophy-like phenotype not linked to 4q35
L. Leonardis; J. Zidar

G.P.15.12 Subcellular trafficking of DUX4, a pro-apoptotic protein encoded at the fascioscapulohumeral muscular dystrophy locus FSHD1A
E.D. Corona; A.L. Rosa

G.P.15.13 Dux4 over-expression in normal mouse: generation of a mouse model with an FSHD phenotype?
J. Dumonceaux; S. Marie; A. Ferry; F. Coppeè; A. Belayew; T. Voit; G. Butler-Browne

Poster room 1 Poster 32 – Inflammatory myopathies
Chairpersons: A. Oldfors, O. Benveniste
## Full Program

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| G.P.16.01 | Expression of myogenic regulatory factors and myo-regeneration in inflammatory myopathies  
J.V. Wanschitz; O. Dubourg; B. Eymard; R. Hoeftberger; M.B. Fischer; E. Lacène; N.B. Romero; S. Herson; G. Butler-Browne; T. Voit; O. Benveniste |
| G.P.16.02 | Expression of TDP-43 in inflammatory myopathies and other muscle diseases  
A. Ishii; H. Takuma; H. Tsuji; N. Ohkoshi; A. Tamaoka |
| G.P.16.03 | Aggregation of TDP-43 in patients of distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy  
K. Sugie; S. Noguchi; M.C.V. Malicdan; M. Ogawa; I. Nonaka; S. Ueno; I. Nishino |
| G.P.16.04 | Cytoplasmic expression of major histocompatibility complex class I in human inflammatory myopathies  
V. Papa; R. Salaroli; R. Rinaldi; C. Ceccarelli; L. Badiali De Giorgi; L. Tarantino; G.N. Martinelli; G. Cenacchi |
| G.P.16.05 | Clinical and pathological features of myopathy associated with anti-signal recognition particle antibodies  
M. Hashimoto; H. Kowa; A. Iwata; S. Tsuji; J. Shimizu |
| G.P.16.06 | Anti-SRP antibody myopathies: clinical characteristics and follow-up of 24 patients  
C. Bloch-Queyrat; A. Rigolet; O. Dubourg; P. Laforet; P. Grenier; A. Behin; T. Stojkovic; Z. Amoura; B. Eymard; S. Herson; L. Musset; O. Benveniste |
| G.P.16.07 | Inflammatory myopathies with anti-Ku antibodies: characteristics and follow up of 28 patients  
A. Rigolet; H. Naffati; O. Dubourg; B. Eymard; P. Cacoub; S. Herson; Z. Amoura; L. Musset; O. Benveniste |
| G.P.16.08 | Expression of HSP90α and HSP90β in the idiopathic inflammatory myopathies and Duchenne muscular dystrophy  
J.L. De Bleecker; B. De Paepe; K.K. Creus; J.J. Martin; J. Weis |
| G.P.16.09 | Epistatic interactions between DRB1 alleles influence susceptibility and clinical phenotype in sporadic inclusion body myositis (sIBM)  
F.L. Mastaglia; M. Needham; A. Scott; I. James; T. Day; L. Kiers; A. Corbett; C. Witt; M. Garlepp; R. Alcock; N. Laing; F. Christiansen |
| G.P.16.10 | Analysis of mtDNA deletions in patients with different forms of idiopathic myositis  
P.R. Joshi; P. Tacik; S. Nikolin; J. Weis; M. Deschauer; S. Zierz |

### Poster room 6
**Poster 33 – Diagnostic and treatment approaches and rare disorders**  
Chairpersons: M. Tulinius, Y. Shapira
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<td>G.P.17.01</td>
<td>Intravenous inotropic treatment of end stage heart failure in neuro muscular disorders</td>
<td>A. van Baalen; G. Fischer; S. von Spiczak; G. Wiegand; U. Stephani</td>
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<td>G.P.17.02</td>
<td>Efficacy and complications of gastrostomy tube feeding in patients with muscular dystrophies</td>
<td>M. Yoshioka; H. Tanaka; H. Konno; T. Takahashi; H. Onodera; K. Ishida</td>
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<td>G.P.17.03</td>
<td>Muscle histopathology in asymptomatic children with incidentally detected high CK</td>
<td>B. Talim; G. Haliloglu; Z. Akcoren; S. Gucer; D. Orhan; G. Kale; H. Topaloglu</td>
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<td>G.P.17.04</td>
<td>Muscle biopsy as a diagnostic tool in neonates</td>
<td>M.A. Al-Muhaizea; H.N. Al-Hindi; H.A. Al-dhalaan; A.K. Chedrawi</td>
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<td>G.P.17.05</td>
<td>Creatine kinase (CK) in a normal population. Towards new referent values?</td>
<td>H. Lileng; S.H. Johnsen; E. Stensland; K. Abeler; S. Loseth; R. Jorde; Y. Figenschau; S. Lindal; S.I. Bekkelund</td>
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<td>G.P.17.06</td>
<td>Axial localized unilateral muscle hypertrophy combined with paraspinal atrophy: an unusual case</td>
<td>F.X.G. Glocker; J.K. Kirschner; G.M. Meng; M.K. Kottlors</td>
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<td>G.P.17.07</td>
<td>Acquired focal rippling muscle syndrome (RMS)</td>
<td>A. Magot; G. Fayet; A. Terrier; P. Chevallier; Y. Pereon</td>
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<td>G.P.17.08</td>
<td>A case report of reversible hydroxychloroquine cardiomyopathy and myopathy</td>
<td>J.B. Sampson; G. Kunkel; J.B. Sampson; S. Chin; K.M. Flanigan</td>
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<td>G.P.17.09</td>
<td>The effect of hypoxia on the proliferation and differentiation of human primary and rat L6 myoblasts is not counteracted by Epo</td>
<td>T. Launay; L. Hagström; S. Lottin-Divoux; D. Marchant; P. Quidu; F. Favret; A. Duvallet; T. Darribère; J.P. Richelet; M. Beaudry</td>
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<td>G.P.17.10</td>
<td>Effects of anemia on gastrocnemius muscle in a model of erythropoietin deficient mice exposed to hypoxia</td>
<td>L. Bex-Hagström; R. El-Hasnaoui-Saadani; F. Favret; J.P. Richelet; M. Beaudry; T. Launay</td>
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<td>G.P.17.11</td>
<td>The educational and vocational status of muscular dystrophy patients</td>
<td>S.H. Im; J.H. Moon; Y.G. Park; S.C. Lee</td>
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19:30 - 22:30 Gala Dinner on the Lake of Geneva (Lac léman) aboard the boat “Lausanne”
Meeting point: see map on page 65
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| 09:00-10:30      | Auditorium (R380) | Advances in treatment of neuromuscular disorders; Invited lectures (T.I. 1–3)  
Chairpersons: M. Vainzof, T. Voit                                    |
|                  |                   | T.I.1                                                                 |
|                  |                   | Current developments in therapeutics for spinal muscular dystrophy     |
|                  |                   | A. Burghes                                                           |
|                  |                   | T.I.2                                                                 |
|                  |                   | Molecular therapies of Duchenne muscular dystrophy                    |
|                  |                   | F. Muntoni                                                            |
|                  |                   | T.I.3                                                                 |
|                  |                   | Regulating interactions between the immune system and muscle for      |
|                  |                   | the treatment of muscular dystrophies                                  |
|                  |                   | J. G. Tidball                                                        |
| 10:30-11:00      |                   | Morning Tea and Coffee                                               |
| 11:00-11:30      | Auditorium (R380) | Advances in treatment of neuromuscular disorders; Invited lectures (T.I.4)  
Chairpersons: S. Treves, S. Wilton                                    |
| 11:30-12:00      | Auditorium (R380) | Advances in treatment of neuromuscular disorders; Oral Presentations (T.O.1–2)  
Chairpersons: I. Richard, I. Nishino                                  |
|                  |                   | T.O.1                                                                 |
|                  |                   | Sialic acid metabolites preclude the development of myopathic phenotype in the DMRV/hIBM mouse model   |
|                  |                   | M.C. Malicdan ; S. Noguchi ; Y.K. Hayashi ; I. Nonaka ; I. Nishino    |
|                  |                   | T.O.2                                                                 |
|                  |                   | Efficient recovery of dysferlin deficiency by dual adeno associated  |
|                  |                   | vector mediated gene transfer                                        |
|                  |                   | W. Lostal ; M. Bartoli ; N. Bourg ; C. Roudaut ; A. Bentaïb ; N. Guerchet ; F. Fougerousse ; P. McNeil ; I. Richard |
| 12:00-13:00      | Auditorium (R380) | WMS General Assembly                                                 |
| 13:00-14:30      | Main Hall         | Lunch                                                                  |
| 14:30-15:00      | Auditorium (R380) | Advances in treatment of neuromuscular disorders; Oral Presentations (T.O.3–4)  
Chairpersons: C. Wallgren-Pettersson, L. Merlini                      |
<p>|                  |                   | T.O.3                                                                 |
|                  |                   | Restoration of dystrophin expression in Duchenne muscular dystrophy: A single blind, placebo-controlled dose escalation study using morpholino oligomer AVI-4658   |
|                  |                   | M. Kinali ; V. Arechavala-Gomez ; L. Feng ; S. Cirak ; D. Hunt ; C. Adkin ; M. Guglieri ; S. Abbs ; P. Nihoyannopoulos ; M.E. Garralda ; M. Rutherford ; C. McCulley ; L. Popplewell ; I.R. Graham ; G. Dickson ; M.J.A. Wood ; D.J. Wells ; S.D. Wilton ; T. Holt ; R. Kole ; V. Straub ; K. Bushby ; C. Sewry ; J.E. Morgan ; F. Muntoni |</p>
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| 15.00-16.30  | Auditorium (R380) | **Late Breaking News**  
Chairpersons: D. Selcen, V. Straub |
| L.B.N.01     |             | Human PTRF mutations cause secondary deficiency of caveolins resulting in muscular dystrophy with generalized lipodystrophy  
Hayashi Y |
| L.B.N.02     |             | Identification of a mutation in agrin that causes congenital myasthenia and affects synapse function  
| L.B.N.03     |             | Non-dysferlin Miyoshi myopathy linked with defective membrane repair (MMD3) is caused by mutations in the calcium activated chloride channel gene TMEM16E which is also mutated in LGMD2L  
| L.B.N.04     |             | In vitro drug screening assays for muscle strength, fatigue and injury using human DMD bioartificial muscles  
| L.B.N.05     |             | Current progress with the systemic administration trial of AVI-4658, a novel Phosphorodiamidate Morpholino Oligomer (PMO) skipping exon 51 in Duchenne muscular dystrophy (DMD)  
Cirak S, Muntoni F, Guglieri M, Bushby K, Leaw S, Shrewsbury S |
| L.B.N.06     |             | Steroid treatment hinders disease rescue of cardiomyopathic hamsters by gene therapy  
| 16:30-17:00  | Main Hall   | **Afternoon Tea and Coffee** |
| 17:00-17:30  | Auditorium (R380) | **Prize Giving and welcome to WMS 15, Kumamoto, Japan** |
| 17:30        | Auditorium (R380) | **Handover of the WMS flag and closing of the Congress** |
Poster Room Map

Ground Floor (Poster rooms 1 to 3)

First Floor (Poster rooms 4 to 9)
Partners and Exhibitors

We would like to thank all following organisations for their generous support of the 14th International WMS Congress.

Gold Partnership

Genzyme
With many established products and services helping patients in nearly 90 countries, Genzyme is a leader in the effort to develop and apply the most advanced technologies in the life sciences, with the aim to develop new medicines, improve its existing therapies, and ensure that patients have access to these treatments.
www.genzyme.com

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Santhera Pharmaceuticals is a Swiss specialty pharmaceutical company focused on small-molecule therapeutics for orphan neuromuscular diseases. Our first product, Catena® to treat Friedreich’s Ataxia, is marketed in Canada while a pivotal Phase III study is well advanced in this indication.
www.santhera.com

Bronze Partnership

Association Française contre le Myopathies
The Association Française contre les Myopathies (AFM) is a patient organization dedicated to accelerating research for the development of new treatments to prevent and cure neuromuscular diseases. It provides care and support for patients and is a leading advocate for patients with rare diseases.
www.afm-telethon.fr

Prosensa Therapeutics B.V.
Prosensa is a biopharmaceutical company focused on the discovery, development and commercialization of therapeutics correcting gene expression through RNA modulation. Its primary focus is to develop therapeutics for genetic neuromuscular and neurodegenerative disorders including Duchenne Muscular Dystrophy (DMD) and Huntington’s Disease.
www.prosensa.eu
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TREAT-NMD is a global network uniting the key leaders in the neuromuscular field to provide the tools and expertise to accelerate new therapies and deliver optimal care. From patient registries to SOPs to care standards, TREAT-NMD is creating the infrastructure needed to bring new treatments more quickly from the lab to the clinic.

www.treat-nmd.eu

Copper Partnership

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The Swiss Foundation on Muscular Disease Research or Fondation Suisse de Recherche sur les Maladies Musculaires (FSRMM) was established 25 years ago with the aim of developing the research in the field of neuromuscular diseases, neglected for far too long in Switzerland. Since then, the FSRMM has delivered approximately one hundred fellowships - for a global amount of CHF 18 mio - to researchers active in the 5 Medical Universities in Switzerland.

www.fsrmm.ch

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Acceleron is committed to the discovery and development of biotherapeutics that harness the powers of the TGF-beta superfamily of proteins. Acceleron is developing novel therapies that modulate the growth of red blood cells, bone, muscle, fat and the vasculature to treat musculoskeletal, metabolic and cancer-related diseases.

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Charity Organisations

Cure Duchenne Muscular Dystrophy
CureDuchenne is a venture philanthropy non-profit, dedicated to funding research for Duchenne muscular dystrophy and creating awareness among the general public for this disease. Our mission is our name...to save this generation of Duchenne boys.
www.cureduchenne.org

Duchenne Parent Project France
We are a non profit association fighting for a cure for Duchenne Muscular Dystrophy. Our goal is to provide the means to accelerate preclinical research by improving scientific collaborations and raising funds for research labs. Among our actions in partnership with the Association Monégasque contre les Myopathies: organization of the Monaco Round Tables and initiation of an International Collaborative Effort project (ICE) to progress towards clinical trials for DMD using exon skipping combined with gene and cell therapies.
www.duchennefr.com

Jain Foundation
Our mission is to accelerate development of a therapy for LGMD2B/Miyoshi, muscular dystrophies caused by dysferlin deficiency. We fund research laboratories worldwide that work on basic and therapeutic projects relevant to dysferlin. We maintain a global patient registry, and facilitate mutational analysis of our registrants, with the goal of being prepared for a clinical trial. We also organize an annual Dysferlin Conference.
www.jain-foundation.org

Parent Project Muscular Distrophy
Parent Project Muscular Dystrophy (PPMD) is the largest nonprofit organization in the United States focused entirely on Duchenne muscular dystrophy. PPMD’s mission is to improve the treatment, quality of life, and long-term outlook for all individuals affected by Duchenne through research, advocacy, education, and compassion.
www.parentprojectmd.org
The Marigold Foundation

The Marigold Foundation is a private, family foundation located in Calgary, Alberta, Canada. We support new and innovative approaches to solving problems at the root level, whether in public policy or biomedical research. We are not afraid to try new directions. Our current programs can be separated into social programs and health research programs. Our social programs include those aimed at helping people in need by providing support and guidance at difficult times. Our health research programs include those aimed at accelerating a cure for myotonic dystrophy (DM) and other rare diseases.

www.marigoldfoundation.org
## General Conditions

### CONGRESS REGISTRATION FEE

<table>
<thead>
<tr>
<th>Category</th>
<th>Fee</th>
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<tr>
<td><strong>Early bird before April 30th, 2009</strong></td>
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<tr>
<td>Member</td>
<td>CHF 635.-</td>
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<td>CHF 955.-</td>
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<td>Accompanying Person</td>
<td>CHF 300.-</td>
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<td>CHF 300.-</td>
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Participants fee includes:
- Program and abstract book
- 3 lunches
- 6 coffee breaks
- Welcome Reception
- Gala Dinner
- All administration and handling

Accompanying persons fee includes:
- Welcome Reception
- Gala Dinner
- Administration and handling (excl. congress documents, lunches and coffee breaks)

### DISCLAIMER

Neither the WMS Local Organising Committee nor SYMPORG SA accepts liability for damages and/or losses of any kind which may be incurred by congress participants or by any person accompanying them, both during the official activities and the excursions. Participants are advised to take out insurance against loss, accident or damage which could be incurred during the congress.
Map of Geneva

1 Grand Hotel Kempinsky
2 Hotel Montbrillant
3 Hotel Tiffany
4 Hotel Warwick
5 Hotel Astoria
6 Hotel Calvy
7 Hotel Comédie
8 Hotel Cristal
9 Hotel Des Alpes
10 Hotel International & Terminus
11 Hotel Le Grenil
12 Hotel Longemalle
13 Hotel Moderne
14 Hotel Montana
15 Hotel Savoy
16 Hotel Strasbourg-Univers
17 Hotel Suisse
18 Hotel Prince
19 Pension Esmeralda
20 Centre Universitaire protestant
21 Foyer George Williams
22 Unimail
23 Boarding Quay des "Vieux Pâquis"
Exhibitors' introduction

We are pleased to announce that the 15th International Congress of the World Muscle Society (WMS2010), will be held at Kumamoto Civic Auditorium (Kumamoto, Japan) for five days, October 12 through 16, 2010. Topics are: 1) New Therapeutic Targets for Neuromuscular Disorders, 2) Congenital Muscular Dystrophies — Celebrating 50th Anniversary of Fukuyama congenital muscular dystrophy, 3) Distal Myopathies and Protein Aggregation Myopathies, and 4) Advances in All Other Neuromuscular Fields.

Aside from an excellent congress program, participants will also have a great opportunity to experience our rich history and cultural heritage, breathtaking natural vistas, and our world-famous Japan hospitality.

Join us in Kumamoto, JAPAN! For more information please visit www.wms2010.com.

Contact person regarding this form

Secretariat of WMS2010, attn: Junko Miyamoto (Ms.)
c/o ICS Convention Design, Inc.
Tel: +81-3-3219-3541 – Fax: +81-3-3219-3626 – Email: wms2010@ics-inc.co.jp