Neuromuscular Therapeutic Strategies: Overcoming the Barriers from Microscope to Marketplace
Dear Conference Participants:

I’m delighted to welcome you to MDA’s 2011 National Scientific Conference. This promises to be an extraordinary meeting, highlighting remarkable advances in neuromuscular disease research through work done by you, MDA’s research community. I’m pleased that the Association is able to provide this opportunity for all of you to gain new insights and, importantly, share information with your colleagues. We hope this meeting will generate new ideas and new collaborations that will lead us to effective therapies and eventual cures for muscular dystrophies and related disorders.

My motto has always been “together we can make it happen.” Please know how proud we are to have you and your research teams as part of the MDA family, working tirelessly in our common quest to defeat neuromuscular diseases. Your efforts are making a difference in the lives of individuals served by the Association.

Thanks for all that you do for MDA!

Warm regards…

Gerald C. Weinberg

March 13, 2011

Dear Conference Participants:

Welcome to MDA’s 2011 National Scientific Conference, the first in MDA’s new annual National conference series. MDA is pleased to provide this forum for dialogue and exchange of ideas about the various therapeutic strategies under development for neuromuscular diseases.

Research advances in your laboratories are bringing us ever closer to effective therapies on a number of neuromuscular disease fronts and your participation in this meeting ensures its success. We look forward to three days of outstanding scientific presentations and robust discussion that will reduce barriers to and accelerate therapy development for muscular dystrophies and related disorders.

A special thank you goes out to the program planning committee for their hard work over many months to develop such an outstanding program. We hope that you will return home from this meeting with renewed energy and new ideas that will get us to the goal of defeating neuromuscular diseases.

With warm regards,

Valerie A. Cwik, M.D., FAAP, FACMG

March 13, 2011

Dear Conference Participants:

I’m pleased to welcome you to MDA’s 2011 National Scientific Conference. The breadth and depth of the conference program is an emphatic testament to the progress that’s being made in the development of novel therapies for neuromuscular diseases.

MDA’s long track record of funding basic and translational research in neuromuscular diseases has opened the door to the development of cutting-edge therapies. While it’s heartening to see all the progress that’s been made to date — clearly much remains to be done. MDA’s sponsorship of this conference is designed specifically to identify barriers in therapeutic development and to come up with strategies to overcome them. It’s our belief at MDA that collaborative discussion of the issues at this stage will help spearhead the critical progress that will be necessary to finally see therapies in the clinic.

I want to express my gratitude to the program planning committee and the excellent group of presenters for putting together such a stellar conference. Above all, I wish to thank the many research groups in academia and industry that share MDA’s commitment to finding treatments and cures for neuromuscular diseases.

With best wishes for a successful conference,

R. Rodney Howell, M.D., FAAP, FACMG
March 13, 2011

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With best wishes for a successful conference,

R. Rodney Howell, M.D., FAAP, FACMG

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**Agenda**

**Sunday – March 13**

2:00 p.m. .......................... **REGISTRATION**
6:00 – 8:00 p.m. .......................... **EVENING RECEPTION**

**Monday – March 14**

8:15 a.m. .......................... Welcome

**OLIGONUCLEOTIDE SESSION**

**CHAIRS: CARMEN BERTONI, PH.D. AND FRANK BENNETT, PH.D.**

8:30-9:00 a.m. .......................... Keynote speaker: Steve Wilton, Ph.D.

*Splice switching therapies for Neuromuscular Diseases*

9:00-9:30 a.m. .......................... Thurman Wheeler, M.D.

*Antisense oligonucleotide therapy for myotonic dystrophy*

9:30-9:50 a.m. .......................... Alessandra Belayew, Ph.D.

*Suppression of DUX4 or DUX4c protein expression by antisense strategies in a therapeutic approach for FSHD*

9:50-10:10 a.m. ...................... Carmen Bertoni, Ph.D.

*Oligonucleotide-mediated gene editing of the dystrophin gene using methyl-CpG ssODN in the mdx mouse model for Duchenne muscular dystrophy*

10:10-10:30 a.m. ..................... **COFFEE BREAK**

10:30-11:00 a.m. ..................... Don Cleveland, Ph.D.

*Antisense oligonucleotide therapy for motor neuron diseases*

11:00-11:20 a.m. ..................... Paul Porensky, M.D.

*Antisense morpholino against ISS-N1 corrects SMA mice*

11:20-11:40 a.m. ..................... Adrian Krainer, Ph.D.

*Correction of SMN2 RNA splicing in the CNS with antisense oligonucleotide as a therapeutic strategy for spinal muscular atrophy*

11:40 a.m.-12:00 p.m. ............... Aurelie Goyenvalle, Ph.D.

*AAV-U7snRNA mediated multi exon-skipping for Duchenne Muscular Dystrophy*

12:00-12:30 p.m. ..................... Discussion: What are the hurdles to developing oligonucleotide therapies and how do we overcome those barriers?

12:30-1:30 p.m. ....................... **LUNCH**
Tuesday – March 15

STEM CELL SESSION
CHAIRS: CLIVE SVENSEN, PH.D. AND JOHNNY HUARD, PH.D.

1:30-1:50 p.m. Introduction: Clive Svendsen, Ph.D.
1:50-2:10 p.m.
Nicholas Boulis, M.D.
Practical Considerations in Human Spinal Cord Stem Cell Transplantation
2:10-2:30 p.m.
Letizia Mazzini, M.D.
Long-term follow-up after mesenchymal stem cells transplantation in Amyotrophic Lateral Sclerosis
2:30-2:50 p.m.
Daniel Miller, M.D., Ph.D.
FSHD-patient Derived IPS cells as a Model to Study Developmentally regulated DUX4 Expression and Test the effect of Potential Therapies
2:50-3:10 p.m.
Svitlana Garbuzova-Davis, Ph.D., D.Sc.
Blood-Brain/Spinal Cord Barrier Impairment in ALS: How to Repair?

3:10-3:30 p.m. COFFEE BREAK

3:30-3:50 p.m.
Introduction: Johnny Huard, Ph.D.
3:50-4:10 p.m.
Terence Partridge, Ph.D.
Effects of the dystrophin mutation on growth and regeneration in mdx mouse muscle
4:10-4:30 p.m.
Giulio Cossu, M.D.
A Phase I/II cell therapy trial for Duchenne Muscular Dystrophy
4:30-4:50 p.m.
Alessandra Sacco, Ph.D.
Short Telomeres and Stem Cell Exhaustion Model Duchenne Muscular Dystrophy in mdx/mTR Mice
4:50-5:10 p.m.
Ilona Skerjanc, Ph.D.
Retinoic Acid Enhances Skeletal Myogenesis in Human Embryonic Stem Cells by Expanding the Premyogenic Progenitor Population
5:10-5:40 p.m.
Discussion: Taking stem cells to the clinic for ALS and MD – where are we now?

5:40-7:30 p.m. POSTER SESSION A

7:30 p.m. DINNER

SMALL MOLECULE SESSION
CHAIRS: JASBIR SEEHRA, PH.D. AND JEFFREY ROTHSTEIN, M.D., PH.D.

8:30-9:00 a.m.
James Rusche, Ph.D.
Developing drugs that target gene expression to replace key proteins in single gene neurodegenerative diseases
9:00-9:30 a.m.
John McCall, Ph.D.
Small molecule drug discovery in muscular dystrophy: what’s involved?
9:30-9:50 a.m.
Stanley Froehner, Ph.D.
Sildenafil Improves Skeletal and Cardiac Muscle Function in the mdx Mouse
9:50-10:10 a.m.
Gino Cortopassi, Ph.D.
Alterations in Thioredoxin-related antioxidants in Friedreich's ataxia models, and High-throughput screening
10:10-10:30 a.m. COFFEE BREAK

10:30-11:00 a.m.
Jeffrey Rothstein, M.D., Ph.D.
Small molecules for ALS: Assessing candidate targets and optimizing clinical discovery
11:00-11:20 a.m.
Jiming Kong, M.D., Ph.D.
Rescuing motor neurons by targeting the BNIP3 cell death pathway
11:20-11:40 a.m.
Laxman Gangwani, Ph.D.
JNK is required for neuronal degeneration in spinal muscular atrophy
11:40 a.m.-12:00 p.m.
James Dowling, M.D., Ph.D.
Myotubular Myopathy and the Neuromuscular Junction: a Novel Therapeutic Approach?
12:00-12:30 p.m.
Discussion: Developing Small Molecule Therapies.
12:30-1:30 p.m.   LUNCH

PROTEIN THERAPY SESSION
CHAIRS: ROBERT MATTALIANO, PH.D. AND JUSTIN FALLON, PH.D.
1:30-2:00 p.m.   Keynote Speaker: Robert Mattaliano, Ph.D.
Developing a Muscular Dystrophy Therapy — Overcoming Barriers in Pompe Disease

2:00-2:30 p.m.   Jasbir Seehra, Ph.D.
Inhibition of the myostatin pathway for treatment of neuromuscular diseases

2:30-2:50 p.m.   Dennis Guttridge, Ph.D.
Understanding NF-kB Function and Its Therapeutic Potential in DMD

2:50-3:10 p.m.   Jachinta Rooney, Ph.D.
Laminin-111 protein therapy restores viability, reduces pathology and improves muscle strength in the dyW mouse model of Merosin Deficient Congenital Muscular Dystrophy Type 1A

3:10-3:30 p.m.   COFFEE BREAK

3:30-4:00 p.m.   Justin Fallon, Ph.D.
Biglycan as a therapeutic for DMD

4:00-4:20 p.m.   Thomas Thompson, Ph.D.
How structures of myostatin can help with inhibitor design

4:20-4:40 p.m.   Michael Lawlor, M.D., Ph.D.
Weakness and Responses to Treatment Are Dependent on Fiber Type and Mutation in Murine Models of Myotubularin Deficiency

4:40-5:00 p.m.   Thien Nguyen, M.D., Ph.D.
Axonal protective effects of netrin-1

5:00-5:30 p.m.   Discussion: Why choose a protein therapy — what are the advantages and disadvantages of this modality?

5:30-7:30 p.m.   POSTER SESSION B

Wednesday – March 16

GENE THERAPY SESSION
CHAIRS: LOU KUNKEL, PH.D. AND BRIAN KASPAR, PH.D.
8:15-8:25 a.m.   Presentation of Poster Awards

8:25-9:10 a.m.   Keynote speaker: James Wilson, M.D., Ph.D.
Immunology and delivery – the key challenges for in vivo gene therapy

9:10-9:40 a.m.   Jeffrey Chamberlain, Ph.D.
Gene therapy for the muscular dystrophies: overcoming the remaining challenges

9:40-10:00 a.m.   Joel Chamberlain, Ph.D.
Validity of RNAi-based therapeutics as a treatment for FSHD as demonstrated in a mouse model of muscular dystrophy

10:00-10:20 a.m.   Dawn Delfin, Ph.D.

10:20-10:50 a.m.   COFFEE BREAK

10:50-11:20 a.m.   Brian Kaspar, Ph.D.
Gene Delivery for Motor Neuron Disease: Pathway to the Clinic

11:20-11:40 a.m.   Zejing Wang, M.D., Ph.D.
Long term transgene expression and amelioration of muscle function following large scale rAAV-micro-dystrophin treatment in dogs with Duchenne muscular dystrophy

11:40 –12:00 p.m.   Louise Rodino-Klapac, Ph.D.
rAAV5 Mediated Delivery of Dysferlin as a Therapeutic Strategy for LGMD2B and Miyoshi Myopathy

12:00-12:30 p.m.   Discussion

12:30 p.m.   LUNCH
Speakers, Chairs and Committee Members

**CONFERENCE CO-CHAIRS:**
Robert Mattaliano, Ph.D.
Genzyme Corporation

Charles Thornton, M.D.
Professor of Neurology
Department of Neurology
University of Rochester

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Peggy and Gary Edwards Distinguished Endowed Chair
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Chair, Department of Neurology
Methodist Neurological Institute

Carmen Bertoni, Ph.D.
Assistant Professor
Department of Neurology
David Geffen School of Medicine
University of California, Los Angeles

Kenneth H. Fischbeck, M.D.
NIH Distinguished Investigator
Neurogenetics Branch
National Institutes of Neurological Disorders and Stroke

Adrian R. Krainer, Ph.D.
Professor
Cold Spring Harbor Laboratory

Louis M. Kunkel, Ph.D.
Chairman, MDA Scientific Advisory Committee
Director, Program in Genomics, Children’s Hospital Boston
Professor of Pediatrics and Genetics
Harvard Medical School

Elizabeth McNally, M.D., Ph.D.
Professor of Medicine and Human Genetics
Director, Institute for Cardiovascular Research
Director, Cardiovascular Genetics Clinic
University of Chicago Medical Center

Sally K. Nelson, Ph.D.
Associate Clinical Professor of Medicine
Division of Pulmonary Sciences and Critical Care Medicine
University of Colorado, Denver

John D. Porter, Ph.D.
Program Director, Neuromuscular Disease

**INVITED SPEAKERS AND SESSION CHAIRS:**
Alessandra Belayew, Ph.D.
University of Mons

Frank Bennett, Ph.D.
Isis Pharmaceuticals

Carmen Bertoni, Ph.D.
University of California, Los Angeles

Nicholas Boulis, M.D.
Emory University

Jeffrey Chamberlain, Ph.D.
University of Washington

Joel Chamberlain, Ph.D.
University of Washington

Don Cleveland, Ph.D.
Ludwig Institute for Cancer Research/University of California San Diego

Gino Cortopassi, Ph.D.
University of California, Davis

Giulio Cossu, M.D.
San Raffaele Scientific Institute

Dawn Delfin, Ph.D.
The Ohio State University

James Dowling, M.D., Ph.D.
University of Michigan

Justin Fallon, Ph.D.
Brown University

Stanley Froehner, Ph.D.
University of Washington

Laxman Gangwani, Ph.D.
Texas Tech. University Health Science Center
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<tr>
<th>Name</th>
<th>Affiliation</th>
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<tr>
<td>Svitlana Garbuzova-Davis, Ph.D., D.Sc.</td>
<td>University of South Florida</td>
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<td>Aurelie Goyenvalle, Ph.D.</td>
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<td>Research Institute at Nationwide Children's Hospital</td>
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Our appreciation to the South Point Hotel, Casino & Spa for hosting our conference.