

MONDAY JULY 9

**INTRODUCTION**

*"Welcome to the First Annual Dysferlin Conference"*  
Ajit Jain & Plavi Mittal, Jain Foundation Inc.

9:00 – 9:10 AM

**SESSION I: STEM CELL THERAPY**

Introduction

9:10 – 9:20 AM

Keynote Speaker

*"Towards cell therapy for muscular dystrophies"*  
Giulio Cossu, Stem Cell Research Institute, Milan

9:20 – 9:40 AM

*"Adult Stem Cells derived from perivascular tissues of the skeletal muscle"*  
Morayma Reyes, University of Washington

9:40 – 10:00 AM

*"Myogenic potential of CD133+ cells"*  
Yvan Torrente, University of Milan

10:00 – 10:20 AM

*"Local and systemic factors regulating muscle stem cell function"*  
Amy J. Wagers, Joslin Diabetes Center and Harvard Stem Cell Institute

10:20 – 10:40 AM

**BRAINSTORMING: SESSION I**

Moderator: Terence Partridge

10:40 – 11:30 AM

**LUNCH 11:30 AM – 12:30 PM**

## SESSION II: DYSFERLIN GENE THERAPY

Introduction	12:30 – 12:40 PM
Keynote Speaker <i>"Adeno-associated virus vectors for targeted and repeat delivery"</i> R. Jude Samulski, University of North Carolina	12:40 – 1:00 PM
<i>"A strategy based on splicing between concatemeric AAV vectors for reversal of dysferlin deficiency by gene transfer"</i> Isabelle Richard, Généthron, France	1:00 – 1:20 PM
<i>"The <math>\Phi</math>C31 integrase system for gene therapy of LGMD2B/MM"</i> Michele P. Calos, Stanford University School of Medicine	1:20 – 1:40 PM
<i>"The potential of plasmid electrotransfer"</i> Daniel Scherman, Généthron, France	1:40 – 2:00 PM
<b>BRAINSTORMING: SESSION II</b> Moderator: Robert Brown	2:00 – 2:50 PM

## SESSION III: MUTATION ANALYSIS AND DIAGNOSIS

Introduction	2:50 – 3:00 PM
Keynote Speaker <i>"Clinical and molecular defects in dysferlinopathies: from patients to therapies"</i> Nicolas Lévy, Inserm, CHU Timone, Marseille	3:00 – 3:20 PM
<i>"Dysferlin expression in monocytes and skeletal muscle in different pathological conditions in vivo and in vitro"</i> Eduard Gallardo, Hospital de la Santa Creu i Sant Pau, Barcelona	3:20 – 3:40 PM
<i>"Muscle biopsy evaluation in the diagnosis of dysferlinopathies"</i> Steven A. Moore, University of Iowa	3:40 – 4:00 PM
<b>BRAINSTORMING: SESSION III</b> Moderator: Sandra Cooper	4:00 – 4:30 PM

**DINNER 6:30 – 9:30 PM**

TUESDAY JULY 10

**INTRODUCTION**

*"Background: Other ferlins, other organisms, other tissues"*  
Bradley Williams, Jain Foundation Inc.

8:30 – 8:50 AM

**SESSION IV: DYSFERLIN FUNCTION**

Keynote Speaker

*"Overview and recent functional analysis of dysferlin"*

Robert H. Brown, Jr., Massachusetts General Hospital, Harvard Medical School

9:00 – 9:20 AM

*"LGMD2B (Dysferlin-deficiency) shows compensatory upregulation of vesicle trafficking pathway Rab27A and its effector Synaptotagmin like protein 2"*

Eric P. Hoffman, Children's National Medical Center, Washington DC

9:20 – 9:40 AM

*"Folding, 'shape-shifting' and membrane targeting of dysferlin and dysferlin mutants"*

Sandra Cooper, Children's Hospital at Westmead, Australia

9:40 – 10:00 AM

*"Dysferlin from T-tubules to sarcolemma"*

Rita Barresi, University of Newcastle upon Tyne, UK

10:00 – 10:20 AM

*"Identification of proteins related to dysferlin and those functioning in the dysferlin membrane repair pathway"*

Rumaisa Bashir, University of Durham, UK

10:20 – 10:40 AM

**BRAINSTORMING: SESSION IV**

Moderator: Kevin Campbell

10:40 – 11:30 AM

**LUNCH 11:30 AM – 12:30 PM**

**SESSION V: MEMBRANE FUSION AND REPAIR**

Introduction	12:30 – 12:40 PM
Keynote Speaker <i>"Defective membrane repair in dysferlin-deficient skeletal and cardiac muscle"</i> Kevin P. Campbell, University of Iowa	12:40 – 1:00 PM
<i>"Mechanisms and assays of membrane repair"</i> Paul L. McNeil, Medical College of Georgia	1:00 – 1:20 PM
<i>"Membrane biophysics of phospholipid bilayer pore formation and membrane fusion"</i> Joshua Zimmerberg, Marine Biological Laboratory, Massachusetts	1:20 – 1:40 PM
<i>"Sea Urchins: A model system for studying exocytosis, endocytosis, membrane repair and DYSFERLIN?"</i> Steven S. Vogel, NIAAA, National Institutes of Health	1:40 – 2:00 PM
<i>"FER-1 function in C. elegans"</i> Samuel Ward, University of Arizona	2:00 – 2:20 PM
<i>"Genetic and cell biological analyses of secretory vesicle biogenesis during C. elegans spermatogenesis"</i> Steven L'Hernault, Emory University	2:20 – 2:40 PM
<b>BRAINSTORMING: SESSION V</b> Moderator: Elizabeth McNally	2:40 – 3:30 PM

**POSTER SESSION I**

All conference participants

3:30 – 4:30 PM

**DINNER 6:30 – 9:00 PM**

WEDNESDAY JULY 11

**SESSION VI: OTHER THERAPEUTIC STRATEGIES**

Introduction	8:30 – 8:40 AM
Keynote Speaker <i>"Therapeutic strategies with possible applicability to Dysferlin deficiency"</i> H. Lee Sweeney, University of Pennsylvania	8:40 – 9:00 AM
<i>"Myostatin inhibitors and muscle regeneration"</i> Kathryn R. Wagner, Johns Hopkins School of Medicine	9:00 – 9:20 AM
<i>"Myoferlin in myoblast fusion"</i> Elizabeth McNally, University of Chicago	9:20 – 9:40 AM
<i>"Assessing the prospects of therapeutic exon skipping in dysferlinopathies"</i> Terence A. Partridge, Children's National Medical Center, Washington DC	9:40 – 10:00 AM
<b>BRAINSTORMING: SESSION VI</b> Moderator: Eric Hoffman	10:00 – 11:00 AM

**POSTER SESSION II**

All conference participants

11:00 AM – 12:00 PM

**LUNCH** 12:00 – 1:00 PM

**FREE TIME** 1:00 – 6:00 PM

**DINNER** 6:00 – 7:30 PM

**EVENING BRAINSTORMING SESSION**

<b>Topic 1: Lessons From Other Genetic Diseases</b> Moderator: Jeffrey Sklar, Yale University School of Medicine	8:00 – 8:30 PM
<b>Topic 2: Drug Discovery Process</b> Moderator: Donald Kirsch, Cambria Biosciences, Massachusetts	8:30 – 9:00 PM
<b>Topic 3: Clinical Trials: Patient Recruitment and FDA Guidelines</b> Moderators: Plavi Mittal & Esther Hwang, Jain Foundation Inc.	9:00 – 10:00 PM

THURSDAY JULY 12

**SESSION VII: IMMUNE INVOLVEMENT AND CELL DEATH**

Introduction	8:30 – 8:40 AM
Keynote Speaker <i>"Complement inhibition in dysferlin-deficient muscular dystrophy"</i> Simone Spuler, Charité University Medicine Berlin	8:40 – 9:00 AM
<i>"Muscle inflammation in dysferlin deficiency"</i> Kanneboyina Nagaraju, Children's National Medical Center, Washington, DC	9:00 – 9:20 AM
<i>"Genetic inhibition of mitochondrial-dependent necrosis mitigates muscular dystrophy"</i> Jeffery D. Molkentin, Children's Hospital of Cincinnati, University of Cincinnati	9:20 – 9:40 AM
<i>"Apoptosis in dysferlin deficiency"</i> Christina Jamieson, University of California at Los Angeles School of Medicine	9:40 – 10:00 AM
<b>BRAINSTORMING: SESSION VII</b> Moderator: Kathryn Wagner	10:00 – 10:50 AM

**CONCLUSION**

<i>"Continuing efforts towards a cure"</i> Ajit Jain & Plavi Mittal, Jain Foundation Inc.	10:50 – 11:00 AM
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**LUNCH 11:30 AM – 12:30 PM**