

SCIENTIFIC PROGRAM

September 12, Wednesday

Satellite Symposium

12.00 – 18.00

A genome-wide strategy to define disease expressivity in DM

Allen D. Roses, MD
*Senior Vice President, Pharmacogenetics
GlaxoSmithKline*

Coffee break

Registration

17.00 – 18.30

Welcome

18.30 – 19.00

Prof. Giovanni Meola

Chairman of IDMC-6

Prof. Enrico Decleva

Chancellor of University of Milan

Prof. Virgilio Ferruccio Ferrario

Dean of Medical School - University of Milan

Prof. Giuseppe Rotelli

President of IRCCS Policlinico San Donato

Prof. Paolo Cabitza

*Head of Department of Medical and Surgical Sciences
University of Milan – IRCCS Policlinico San Donato*

Special lecture

19.00 – 20.00

Phenotype, genotype, and molecular basis of Myotonic Dystrophy: past, present, and future

Prof. Giuseppe Novelli

*Professor and Chair of Genetics
Tor Vergata University
Rome - Italy*

Welcome Cocktail

20.00

DNA INSTABILITY AND MODELS OF MYOTONIC DYSTROPHIES

Session Chairs: Christopher Pearson and Be Wieringa

- 8.00 01 **Bidirectional transcripts at the DM1 locus and other triplet repeat diseases: implications for disease mechanisms**
SJ Tapscott, GN Filippova, S Mahoney, P Ladd, DH Cho
-
- 8.15 02 **Saturation effects of binding of miR-1 and miR-206 to the 3' UTR target site in DMPK mRNA: Yet another explanation for pathophysiological findings in cell and animal models for Myotonic Dystrophy type 1 (DM1)?**
Walther JA, A.van den Broek, Derick G Wansink and Bé Wieringa
-
- 8.30 03 **Modelling CUG expansion toxicity in Drosophila: what role for expansion context?**
G Le Mée, N Ezzeddine and O Aït-Ahmed
-
- 8.45 04 **Modelling Congenital Myotonic Dystrophy**
V Srinivasan, Q Yu and Mi Mahadevan
-
- 9.00 05 **CTG repeat “big jumps” in transgenic mice**
L Foiry, M Gomes-Pereira, A Nicole, S Tomé, C Junien, A Munnich and G Gourdon
-
- 9.15 06 **CUG length-dependent, brain region- and muscle type-specific splicing abnormalities in DM1 transgenic mice**
M Gomes-Pereira, A Huguet, A Nicole, J Acquire, A Munnich, G Gourdon
-
- 9.30 07 **Insights from an Inducible/Reversible Mouse Model of RNA Toxicity in DM1**
MS Mahadevan, R S Yadava, V Srinivasan, Q Yu, C Frenzel-McCardell, J Puymirat, CA Thornton, AL Tucker, OW Prall, R.P Harvey
-
- 9.45 09 **Severe skeletal muscle wasting in a tissue-specific, inducible mouse model for Myotonic Dystrophy**
JP Orengo and TA Cooper
-
- 10.00 010 **Somatic mosaicism and genotype-phenotype correlations in DM1.**
F Morales, G Hogg, P Cuenca, G del Valle, R Brian, M Sittenfeld, T Ashizawa, A Wilcox, DE Wilcox and DG Monckton

- 10.30 011 **CTCF binding in cis regulates CAG/CTG repeat instability**
KA Hagerman, RT Libby, VV Pineda, R Lau, JD Cleary, BL Sopher, DH Cho, S Baccam, SJ Tapscott, GN Filippova, CE Pearson, AR. La Spada
-
- 11.45 012 **CNS RNA gain-of-function effects induced by CUG and CCUG transcripts**
R Daughters, Y Kang, D Tuttle, J Margolis, T Zu, M Moseley, J Day, M Swanson, L Ranum
-
- 11.00 013 **Pre-mutation allele pool in DM2**
LL Bachinski, T Czernuszewicz, LS Ramagli, T Suominen, O Raheem, MD Shriver, CA Thornton, B Udd, MJ Siciliano & R. Krahe

11.15 O14 **A Transgenic Mouse Model for Myotonic Dystrophy Type 2 (DM2)**
R Krahe, M Sirito, M Wojciechowska, S Hajibashi, SE Olufemi, DR Mosier, O Raheem, R Randen, B Udd, & LL Bachinski

11.30 O15 **REVERSIBLE MULTISYSTEMIC MOUSE MODELS OF DM1 and DM2**
Y Kang, J Margolis, J Day, L Ranum

Session 1 Poster session

11.45 – 12.15

DNA INSTABILITY AND MODELS OF MYOTONIC DYSTROPHIES

Session Chair: Genevieve Gourdon

P1 **Sustained expression of CUG repeat RNA in Drosophila muscles is degenerative**

A Garcia-Lopez, L Monferrer and R Artero

P2 **Cis-elements, DNA replication and repeat instability at the human myotonic dystrophy type 1 locus**

JD Cleary, L Foiry, G Gourdon & CE Pearson

P2B **CTG repeat instability in transgenic mice: investigating the role of Msh2 and Ligase I**

S Tomé, L Foiry, A Huguet, DW Melton, A Munnich and G Gourdon

P3 **Progressive atrophy of the skeletal muscles in DM1 mice**

A Vignaud, A Ferry, G Gourdon, A Huguet, GS Butler-Browne and D Furling

P4 **Variables acting upon the ctg expansion over time in DM1 patients**

A Lopez de Munain, Cobo AM, Poza JJ

P5 **Identification of abnormal gene expression in myotonic dystrophy type 1 using a human PGD-derived embryonic stem cell line exhibiting intranuclear foci**

G Pietu, C Rochon, K Giraud-Triboult, L Kassar-Duchossoy, D Furling, J Denis, B Champon, C Martinat, K Sermon, M Peschanski

P6 **CUG repeats in DM1 are located within a retained intron of the DMPK 3'UTR**

ET Wang, D Housman, C Burge

Lunch

12.15 – 13.15

Session 2 Oral session

13.15 – 16.15

CELLULAR AND MOLECULAR ASPECTS OF MYOTONIC DYSTROPHIES

Session Chairs: Tee Ashizawa and Jack Puymirat

13.15 O16 **Form and function of short (CUG)_n-RNA in cells transcribing extended CTG repeats characteristic of myotonic dystrophy type 1**

G Pall and A Hamilton

13.30 O17 **Defective mRNA in myotonic dystrophy accumulates at the periphery of nuclear splicing speckles.**

I Holt, S Mittal, D Furling, GS Butler-Browne, JD Brook, GE Morris

- 13.45 **O18 A bioinformatics approach to identify novel genes mis-spliced in myotonic dystrophy**
R Voelker & JA Berglund
-
- 14.00 **O19 Cytoplasmic CUG RNA foci are insufficient to result in aberrant RNA splicing**
P Sarkar, C Wolf, W Dansithong, S Paul, A Chiang, D Branco, MC Sherwood, I Holt, GE Morris, L Comai, CI Berul and S Reddy
-
- 14.15 **O20 Aberrantly spliced alpha-dystrobrevin alters alpha-syntrophin binding in myotonic dystrophy type 1**
M Nakamori, T Kimura, T Matsumura, H Fujimura, MP Takahashi and S Sakoda
-
- 14.30 **O21 Amphiphysin 2 splicing alteration as a possible cause of muscle atrophy in myotonic dystrophic patients**
C Hammer, L Guigou, C Sellier, C Fugier, MC Hummel, C Thibault, N Sergeant, J Laporte, D Furling, B Udd and N Charlet-Berguerand
-
- 14.45 **O22 The Response to Serum Starvation in DM1 Lens Cells Involves FGF Receptor Signalling and Ca²⁺ Channel Activation.**
JD Rhodes, S L Russell and A R Prescott
-
- 15.00 **O23 Inhibition of prostaglandin E2 (PGE2) production restores the differentiation of congenital human myotonic dystrophy type 1 myoblasts**
J Puymirat, D Beaulieu, P Chapdelaine
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- 15.15 **O24 Significant alteration of gene expression in DM1 myoblasts is elicited by molecular events unlinked to RNA splice defects**
W Dansithong, S Paul, K Promnares, MP Takahashi, L Comai and S Reddy
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- 15.30 **O25 Mutant DMPK transcripts activates Notch signaling, impairing myogenesis in myotonic dystrophy type 1**
Xu W, Gao R, Takeuchi T, Furihata M, Puymirat J, Furling D, Ashizawa T and PS Sarkar
-
- 15.45 **O26 Differential expression of elongation factor 1 alpha (EF1A) in DM muscle cells.**
R Pelletier, F Hamel and J Puymirat
-
- 16.00 **O27 Spinocerebellar ataxia type 10 – parallel with and disparity from myotonic dystrophies in the RNA-mediated pathogenic mechanism**
MC White, R Gao, W Xu, SF Edwards, S Raskin, HAG Teive, G Schuster, HY Zoghbi, PS Sarkar, T Ashizawa

Coffee break **16.15 – 16.30**

Special lecture: Leonardo and Last Supper: restoration works **16.30 – 17.00**

dott. E De Palmieri

Direttore Soprintendenza per il Patrimonio Storico, Artistico e Etnoantropologico di Milano

Visit to “Last Supper” **17.30**

“Pizzata” **20.00**

CELLULAR AND MOLECULAR ASPECTS OF MYOTONIC DYSTROPHIES

Session Chair: Stephen Tapscott

- P7 The p16 pathway mediates premature senescence of DM1 myoblasts**
Bigot A, Francois V, Butler-Browne GS, Mouly V and Furling D
-
- P8 Differential expression of splicing regulators and effects of CUG repeats in DM1 cerebral cell models**
D Ghanem, O Leroy, H Tran, CM Dhaenens, S Schraen-Maschke, B Sablonnière, L Buée, A Andreadis, N Sergeant, ML Caillet-Boudin
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- P9 Visualization of the alternative splicing in living cells**
D Furutama, T Tanaka, R Sakai, T Maeda, T Hanfusa, N Ohsawa
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- P10 In vitro study of DM1 primary myotubes**
E Loro, A Botta, C Catalli, V Romeo, F Rinaldi, C Angelini, L Vergani
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- P11 Woodchuck post-transcriptional regulatory element induces nuclear export of myotonic dystrophy transcripts and repairs muscle cell differentiation**
NP Mastroiannopoulos, E Chrysanthou, J Uney, MS Mahadevan & LA Phylactou
-
- P12 Analysis of MTMR1 pre-mRNA splicing in DM1 and DM2 muscle biopsies**
M Santoro, A Modoni, M Masciullo, E Ricci, PA Tonali, G Silvestri
-
- P13 Oxidative stress in DM1: role of NFkB and related proteins**
Valenti F, Aguenouz M, Musumeci O, Rodolico C, Lanzano N, Ciranni A, Crupi R, Toscano A, Vita G
-
- P14 Comparative studies of DM1 and DM2 in muscular biopsies**
S Salvatori, M Fanin, S Furlan, A Picard, E Pastorello, V Romeo, CP Trevisan and C Angelini
-
- P15 Abnormal expression of DMPK substrate phospholamban in DM2**
O Raheem, J Holmlund-Hapf, T Suominen, A Vihola, H Haapasalo, R Krahe and B Udd
-
- P16 Differences in aberrant expression and splicing of genes involved in Ca²⁺ metabolism between DM2 and DM1**
A Vihola, M Siritto, LL Bachinski, S-E Olufemi, O Raheem, T Suominen, B Udd and R Krahe
-
- P17 A variably spliced region of Ryanodine receptor 1 may be involved in excitation-contraction coupling.**
T Kimura, M Nakamori, MP Takahashi, H Yoshikawa, S Sakoda and AF Dulhunty
-
- P18 The CTG repeat expansion size correlates with the splicing defects observed in muscles from myotonic dystrophy type 1 patients**
Botta A, Rinaldi F, Catalli C, Bonifazi E, Loro E, Vergani L, Romeo V, Angelini C, Novelli G
-
- P19 Gene expression analysis in Myotonic Dystrophy: Indications for a common molecular pathogenic pathway in DM1 and DM2**
Rinaldi F, Botta A, Vallo L, Bonifazi E, Gambardella S, Mancinelli E, Angelini C, Meola G, Novelli G
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RNA-BINDINGS PROTEINS

Session Chairs: Charles Thornton and Tom Cooper

- 8.45 O28 **Role of PKC pathway in DM1 pathogenesis by regulating CUGBP1 phosphorylation and steady state levels**
NM Kuyumcu-Martinez and TA Cooper
- 9.00 O29 **Signal transduction pathways regulating CUGBP1 RNA-activity in DM1 patients**
E Salisbury, K Sakai, B Schoser, H Nguen, M Gu, CH Huichalaf, L Wang, NA Timchenko, L Timchenko
- 9.15 O30 **MBNL binds similar RNA structures in the CUG repeats of myotonic dystrophy and its pre-mRNA substrate cardiac troponin T**
MB Warf and JA Berglund
- 9.30 O31 **The RNA binding specificity of Drosophila muscleblind**
E Goers and JA Berglund
- 9.45 O32 **The overexpression of MBNL1 fetal isoform observed in myotonic dystrophy brain does not modified tau splicing**
C Dhaenens, S Schraen-Maschke, V Vingtdeux, H Tran, D Ghanem, O Leroy, J Delplanque, E Vanbrussel, A Delacourte, P Vermersch, CA Maurage, H Gruffat, A Sergeant, M Mahadevan, S Ishiura, L Buée, TA Cooper, M-L Caillet-Boudin, N Charlet-Berguerand, B Sablonnière, N Sergeant
- 10.00 O33 **Biochemical analyses of MBNL1 complexes in myotonic dystrophy**
S Paul, W Dansithong, I Holt, JD Brook, MP Takahashi, GE Morris, L Comai and S Reddy
- 10.15 O34 **Multiprotein complexes as targets for RNA CCUG repeats expanded in patients with DM2**
E Salisbury, B Schoser, C Schneider-Gold, G-L Wang, NA Timchenko, L Timchenko

Coffee break

10.30 – 10.45

- 10.45 O35 **MBNL3 Down- Regulates MEF2 dependent Genes by Alternative Splicing of MEF2 Transcription Factor**
KS Lee, Y Cao, SJ Tapscott and EH Wang
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- 11.00 O36 **Structural determinants for the molecular properties of MBNL proteins**
Y Kino, Y Oma, H Onishi, N Sasagawa, N Nukina, S Ishiura
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- 11.15 O37 **Proteins that interact with MBNL1**
H Onishi, Y Kino, N Sasagawa, S Ishiura
-
- 11.30 O38 **Interactions of Muscleblind Proteins with Splicing Target and Pathogenic RNAs**
Y Yuan, S Compton, K Sobczak, M Stenberg, C Thornton, J Griffith, M Swanson
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RNA-BINDINGS PROTEINS

Session Chair: Maurice Swanson

- P20 **Zebrafish knock-down model for muscleblind-like 2**
LE Machuca-Tzil and JD Brook
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- P21 Functional studies of Muscleblind-like protein 1 (MBNL1)**
S Mittal and JD Brook
-
- P22 Characterization of proteins that bind to the CUG repeats**
J Marie and F-X Laurent
-
- P23 Colocalization of ribonuclear inclusions and MBNL1 foci with no impairment of in vitro adult DM1 and DM2 myoblasts differentiation**
R Cardani, E Mancinelli, E Bonifazi, A Botta, G Novelli, G Meola
-
- P24 A putative role of ribonuclear inclusions and MBNL1 in the impairment of gallbladder smooth muscle contractility with colelithiasis in myotonic dystrophy type 1.**
R Cardani, E Mancinelli, G Saino, L Bonavina, G Meola
-
- P25 Study on differential binding properties of MBNL1 isoforms and of CELF proteins on RNAs containing CUG repeats or potential splicing target sequences.**
N Marmier-Gourrier, A Vautrin, N Charlet, I Behm-Ansmant and C Branlant
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- P26 Studies on the RNA recognition properties of MBNL1 and CUG-BP1**
A Vautrin, N Marmier-Gourrier, I Behm-Ansmant and C Branlant
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- P27 RNAi-mediated silencing of Drosophila muscleblind**
JM Fernandez-Costa and RD Artero
-
- P28 The function and expression of K02H8.1 (CeMBNL), the ortholog of mammalian MBNLs.**
N Sasagawa, E Ohno, Y Kino and S Ishiura
-
- P29 Imaging mRNAs involved in Myotonic Dystrophy type 1 (DM1) using Atomic force Microscopy (AFM)**
F Meullenet, S Allen, S Tandler and D Brook
-
- P30 Loss of Tau exon 2/3 inclusion in DM1 implies the carboxy-terminal tail of MBNL1**
H Tran, CM Dhaenens, D Ghanem, N Charlet, TA Cooper, A Andreadis, E Van Brussels, B Sablonnière, L Buée, ML Caillet-Boudin, S Shraen-Maschke and Nicolas Sergeant

Lunch **12.30 – 13.30**

Session 4 Oral session **13.30 – 15.45**

CHARACTERIZATION AND MANAGEMENT OF SYSTEMIC ASPECTS OF MYOTONIC DYSTROPHIES

Session Chairs: John Day and Bruno Eymard

- 13.30 **O39 Neuropsychological and Adaptive Skills in Myotonic Dystrophy type 1- A Study on 57 Individuals with Congenital and Childhood Forms**
L Hakenäs-Plate, A-B Ekström, M Tulinius, E Wentz
-
- 13.45 **O40 Autism Spectrum Disorders in Myotonic Dystrophy type 1-A Study on 57 Individuals with Congenital and Childhood Forms**
A-B Ekström, L Hakenäs-Plate, M Tulinius, E Wentz
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- 14.00 **O41 Cognitive deficits bridging ages: a behavioural phenotype in Myotonic dystrophy type 1?**
S Winblad, A-B Ekström, L Hakenäs-Plate, M Tulinius, E Wentz, C Lindberg

- 14.15 042 **Cognitive impairment in myotonic dystrophy type 1 (DM1): a longitudinal follow-up study**
A Modoni, C Marra, MG Vita, M Masciullo, PA Tonali, E Ricci, G Silvestri
-
- 14.30 043 **Cross-Sectional Analysis of CNS Imaging and Function in DM1 and DM2.**
JW Day, J Dalton, JR Wozniak, D Franc, L Hemmy, KO Lim, LPW Ranum
-
- 14.45 044 **DM2 is predicted by combined type 2 fibre centronucleation and atrophy**
G Bassez, E Chapoy, S Bastuji-Garin, H Radvanyi-Hoffman, F-J Authier, J-F Pellissier, B Eymard, RK Gherardi
-
- 15.00 045 **Myotonic dystrophy type 2 (DM2); a diagnostic alternative to fibromyalgia**
S Auvinen, Suominen T, Hannonen P, Bachinski L, Krahe R, Udd B
-
- 15.15 046 **Disproportionately high prevalence of co-segregating CLCN1 mutations among myotonic dystrophy type 2 patients from Finland and Germany**
T Suominen, B Schoser, O Raheem, S Auvinen, M Walter, R Krahe, H Lochmüller, W Kress and B Udd
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- 15.30 047 **Mapping of muscular involvement of lower legs in myotonic dystrophy : a magnetic resonance study and correlation to clinical data**
C Côté, H Bassez, JM Janier, L Herbert and J Puymirat
-

Coffee break 15.45 – 16.00

Session 5 Oral session 16.00 – 17.45

CHARACTERIZATION AND MANAGEMENT OF SYSTEMIC ASPECTS OF MYOTONIC DYSTROPHIES

Session Chairs: Nakaaki Ohsawa and Bjarne Udd

- 16.00 048 **Pacemakers do not prevent sudden death in myotonic dystrophy type 1**
WJ Groh, MR Groh, RM Pascuzzi
-
- 16.15 049 **Cardiac Arrhythmias in Type I Myotonic Dystrophy patients with Sleep Apnoea An Implantable Monitoring Device Prospective Study**
K Wahbi, A Lazarus, B Eymard, C Meune, J Varin, P Laforêt, D Duboc
-
- 16.30 050 **Progression of muscle weakness and cardiac involvement in myotonic dystrophy type 1 (DM1) vs type 2 (DM2): a longitudinal study.**
V Sansone, S Briganti, M Panzeri, R Mauro, A Degrade, V Montericcio, L De Ambroggi and G Meola
-
- 16.45 051 **Cardiac Involvement in DM2 : A 30 Patients Follow-up Study**
K Wahbi, G Bassez, C Meune, P Laforêt, A Lazarus, H Radvanyi, B Eymard, D Duboc
-
- 17.00 053 **Gastrointestinal (GI) symptoms in myotonic dystrophy type 1 (DM1) patients enrolled in the NIH Registry**
J Hilbert, W Martens, A Parkhill, A Smirnow, R Moxley III & the Registry Scientific Committee
-
- 17.15 054 **Erectile dysfunction in myotonic dystrophy type 1 (DM1)**
A Clemenzi, AF Radicioni, E Bucci, P Latino, S Morino, M Garibaldi, A Di Pasquale, A Anzuini, A Lenzi, G Novelli, G Antonini
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Gala dinner 20.00

CHARACTERIZATION AND MANAGEMENT OF SYSTEMIC ASPECTS OF MYOTONIC DYSTROPHIES

Session Chair: Bjarne Udd

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- P31 **Investigation of dementia in a patient with Myotonic Dystrophy type 1: is it a DM1 associated phenomenon or is it Alzheimer's disease ?**
C Lindberg and S Winblad
-
- 32 **Cerebrospinal fluid tau and amyloid β 42 protein in patients with myotonic dystrophy type 1 (DM1)**
S Winblad, JE Månsson, K Blennow, C Jensen, L Samuelsson, C Lindberg
-
- P33 **Cognitive and personality profile in Myotonic Dystrophy type1 (DM1)**
A Sistiaga, I Urreta, M Jodar, AM Cobo, J Emparanza, J Poza, A Imaz, JF Martí-Masso y A López de Munain
-
- P34 **Intellectual functioning in a large sample of adult and late-onset DM1 patients**
S Jean, L Richer, J Mathieu
-
- P 35 **Comprehensive evaluation of sleep-wake cycle and daytime somnolence in myotonic dystrophy type 1 (DM1)**
MB Panico, V Pisani, F Placidi, A Romigi, F Izzi, F Corte, MG Marciani, R Massa
-
- P36 **Cognitive impairment and psychiatric disorders in the juvenile form of myotonic dystrophy**
M Douniol, O Lanthier-Gazzano, A Jacqueline, A Afenjar, N Angeard, D Héron, M Plaza, D Cohen
-
- P37 **Psychopathological and cognitive characteristics in DM1 and DM2**
A Palmieri, V Romeo, F Squarzanti, E Albertini, C Borsato, E Pegoraro, C Angelini
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- P38 **Theory of mind and cognitive disorders in myotonic dystrophy type 1 (DM-1): a preliminary study to understand non-compliance with home ventilation treatment in Steinert's population**
V Havet, P Allain, I Péniisson-Besnier, I Richard, N Meslier
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- P39 **The block of Ca-dependent K⁺channels reduces myotonia in Steinert disease: an in vivo pharmacological study**
C Chisari, R Licitra, B Rossi
-
- P40 **Unbalance in Myotonic Dystrophy-1 may follow cervical ataxia and respond to exercise**
L Tesio, C Chessa, M Atanni
-
- P41 **Quantitative evaluation of muscle degeneration in DM1 patients using MRI**
B Hiba, LJ Hébert, C Vial, J Saulnier, M Nejjari, JF Remec, C Coté, F Bouhour, J Puymirat and M Janier
-
- P42 **Different muscle MRI features in myotonic dystrophy type 1 and 2**
C Borsato, V Romeo, E Albertini, C D'Ascenzo, F Squarzanti, A Palmieri, V Beltrame, R Dal Borgo, R Stramare, M Fanin, E Pegoraro, C Angelini
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P43 Myotonic Dystrophies type 1 and type 2: MRI and SPECT comparative study
V Romeo, P Zucchetta, C Ferrati, L Antunovic, R Manara, E Pegoraro, C Angelini

P44 Myotonic dystrophy type 2: clinical, neurophysiological and muscular features of a family with short CCTG expansion
S Lucchiari, S Corti, S Pagliarani, M Servida, E Fruguglietti, M Moggio, N Bresolin, GP Comi

P45 Weakness and fatigue more than myotonia affect physical and mental perception of quality of life in patients with myotonic dystrophies
M Panzeri, V Sansone, S Gandossini, INQoL Group, MR Rose and G Meola

P46 Muscle pathology in myotonic dystrophies – an ultrastructural study
A Nadaj-Pakleza, A Lusakowska, E Szmidt-Salkowska, A Sulek, W Krysa, M Rajkiewicz, A Kaminska

Session 5 Poster session

9.00 – 9.45

CHARACTERIZATION AND MANAGEMENT OF SYSTEMIC ASPECTS OF MYOTONIC DYSTROPHIES

Session Chair: Denis Duboc

P 47 ECG-Holter monitoring is a valuable tool to screen Myotonic Dystrophy type 1 (DM1) patients for advanced conduction defects
E Bucci, C Balla, F Marrara, D Santini, A Clemenzi, P Latino, S Morino, M Testa, G Antonini

P 48 Different phenotypic expression and CTG repeat expansion size in myotonic dystrophy type 1 patients
S Contardi, F Pizza, P Avoni, R Liguori

P 49 RAMYD (Risk of Arrhythmias in MYotonic Dystrophy) study: the design
M Pace, A Dello Russo, M Casella, F Mangiola, A Modoni, G Silvestri, E Ricci, G Nigro, L Politano, MG Bongiorni, P Melacini, P Della Bella, F Belloci

P 50 RAMYD study: baseline cardiological characteristics.
M Pace, A Dello Russo, M Casella, F Mangiola, M Vaccarella, A Modoni, G Silvestri, E Ricci, R Vannicelli, G Nigro, L Politano, MG Bongiorni, P Melacini, P Della Bella, F Belloci

P 51 RAMYD study preliminary results: electrophysiological study and devices implant
M Pace, A Dello Russo, M Casella, G Nigro, MG Bongiorni, P Melacini, P Della Bella, F Belloci

P 52 Comparison between electroanatomic mapping vs cardiac magnetic resonance imaging in myocardial substrate study in Myotonic Dystrophy type 1
C Bisceglia, M Pace, M Casella, A Dello Russo, R Biddau, M Vaccarella, G Pelargonio, F Mangiola, F Belloci

P 53 Assessment of noninvasive ventilation in myotonic dystrophy type 1
MA Hamon, N Meslier, F Dubas, JL Racineux, I Pénisson-Besnier

P 54 Ventilatory function in patients with myotonic dystrophy type 1.
MG Di Gregorio, M Scutifero, F Spina, R Russo, A Palladino, G Fiorentino and L Politano

P54B Artificial ventilatory management in myotonic dystrophy at a Japanese hospital for chronic neuromuscular disorders
S Kon, Y Oyama, H Takada

- P 55 Is heart rate variability a prognostic indicator in patients with dystrophia myotonica type 1**
A Palladino, M Scutifero, VM Ventriglia, P Sannino, MG Di Gregorio, R Russo, G Nigro, L Passamano, VR Petretta, V Cozza, E Bonifazi, G Novelli, G Nigro and L Politano
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- P 56 A cross-sectional study for glucose intolerance of myotonic dystrophy**
T Matsumura, H Iwahashi, MP Takahashi, T Saito, H Fujimura, S Shinno
-
- P 57 Intracellular insulin mediated signalling in Myotonic Dystrophy Type 1 (DM1)**
P Latino, P Castri, E Bucci, A Clemenzi, S Morino, A Di Pasquale, A Fornasiero, M Garibaldi, L Iacovelli, F Orzi, G Antonini
-
- P 58 Multidisciplinary study in patients with myotonic dystrophy type 1**
N Olivero, T Mongini, L Vercelli, G Gai, A Mattei, F Conrotto, A Cicolin, A Farri, L Palmucci
-
- P 59 Abnormal β -cell function in myotonic dystrophy type-1**
F Bouhour, A Brac de la Perrière, H Bassem, C Vial, M Janier, J Puymirat
-
- P 60 Insulin resistance in patients with myotonic dystrophy type 1**
V Rakocovic-Stojanovic, S Popovic, S Peric, A Nikolic, I Basta, Z Stevic, Z Tasic, D Lavrnica
-
- P 61 Advanced oxidation protein products in serum of patients with myotonic disease type 1: correlation with extra-muscular phenotype**
M Falorni, L Volpi, M Mancuso, A Rocchi, G Malvaldi, A Pompella, A Paolicchi, G Siciliano
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- P 62 Intraocular pressure and central corneal thickness study in patients with Steinert myotonic dystrophy**
N Rosa, M Lanza, C Irregolare, A Palladino, L Passamano, F Spina, MR Cecio, L Politano
-
- P 63 A case of patient with coexistent Thomsen's disease and benign hyperbilirubinemia (e.g. Gilbert syndrome (GS)).**
HS Muradyan and SG Khachatryan
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Session 6 Oral session

9.45 – 12.00

SPECIAL ASPECTS OF MANAGEMENT

Session Chairs: Peter Harper and Laura Ranum

- 9.45 **O55 Protocol development for Preimplantation Genetic Diagnosis (PGD) of Myotonic Dystrophy Type 1 (DM1) in the UK: experience from 25 cycles**
Kakourou G, Dhanjal S, Mamas T, Doshi A, Gotts S, Serhal P, Ranieri DM, Delhanty JDA, Harper JC, SenGupta SB
-
- 10.00 **O56 Congenital myotonic dystrophy: Canadian incidence and cohort study.**
C Campbell, SL Venance, P Jacob, V Siu
-
- 10.15 **O57 Presymptomatic testing in Myotonic Dystrophy type I - 6-year experience for 131 candidates**
A Jacquette, C Colas, M Gargiulo, P Laforêt, B Eymard, J Feingold, H Radvanyi, D Héron
-

Coffee break

10.30 – 10.45

- 10.45 **O58 It's genetic, but what does that mean?**
C Downing
-

- 11.00 **059 Review of children diagnosed with myotonic dystrophy over 30 years**
J Fenton-May, C Sampson, MT Rogers
-
- 11.15 **060 Neuropsychological Profile in the childhood form of DM1**
N Angeard, M Gargiulo, A Jacquette, H Radvanyi, B Eymard and D Héron
-
- 11.30 **061 The Saguenay Myotonic Dystrophy Integrated Care Pathway: Development and preliminary validation**
C Gagnon, M-C Chouinard, S Jean, J Mathieu
-
- 11.45 **062 Myotonic dystrophy type 2 in Japan: distinct ancestral origin from Caucasian families**
Y Amakusa, T Matsuura, T Saito, T Kimura, O Yahara, H Aizawa, Y Ikeda, JW Day, LPW Ranum, K Ohno
-

Session 6 Poster session

12.00 - 12.45

SPECIAL ASPECTS OF MANAGEMENT

Session Chairs: Giuseppe Novelli and Ralf Krahe

- P 64 A care-card for myotonic dystrophies: improving management and follow-up**
V Sansone, R Mauro, M Panzeri and G Meola
-
- P 65 From initial symptoms to genetic confirmation: what is the time-lag for myotonic dystrophies in Italy?**
V Sansone, R Mauro, M Panzeri and G Meola
-
- P 66 Determinants of genetic knowledge in patients with myotonic dystrophy type 1 (DM1)**
L Laberge, M Perron, J Mathieu, J Auclair, M Gaudreault, S Veillette
-
- P 67 Myotonic dystrophy unlinked to DM1 and DM2 mutations in three siblings**
V Pisani, A Botta, E Bonifazi, MB Panico, C Rocchi, GA Marfia, F Sangiuolo, G Bernardi, G Novelli, R Massa
-
- P 68 Self-reported health problems and health habits in myotonic dystrophy type 1: a patient-oriented perspective**
MC Chouinard, C Gagnon, L Laberge, S Jean, J Mathieu
-
- P 69 What do patients with Myotonic Dystrophy Type 1 know about their disorder: the first study in Bashkortostan (Russia)**
L Akhmadeeva, H Derevyanko, R Magzhanov
-
- P 70 Development of Orofacial Dysfunction in Young Individuals with Myotonic Dystrophy type 1**
L Sjögren, M Engvall, A-B Ekström, S Kiliaridis, M Tulinius, A Lohmander
-
- P 71 Molecular analysis of a family co-segregating myotonic dystrophy type 1 and Charcot-Marie-Tooth disease**
C Braida, F Spaans, CG Faber, HJM Smeets, P Hofman, CEM de Die-Smulders and DG Monckton
-
- P 72 An improved method for Southern DNA and Northern RNA blotting using a Mupid®-2 Mini-Gel electrophoresis unit for diagnosis of DM1 and 2**
H Furuya, T Yamada, K Ikezoe, T Arahata, Y Fukumaki, N Fujii
-

P 73 Myasthenic phenotype as possible manifestation of myotonic dystrophy
V Milic Rasic, J Mladenovic, R Dimitirijevic, V Dobricic, S Romac

P 74 Clinical and Neuroimaging features of Myotonic Dystrophy in childhood
K Gorni, S Orcesi, C Uggetti, E Fazzi, A Berardinelli

Lunch 12.45 – 13.45

Session 7 Oral session 13.45 - 16.00

THERAPEUTIC TRIALS AND FUTURE ADVANCES

Session Chairs: Dick Moxley and Giovanni Meola

- 13.45 **O63 Correlation between Measures of Muscle Mass, Strength, Function and Quality of Life (QOL) in Patients with Myotonic Dystrophy Type 1 (DM-1): Implications for Clinical Trials.**
S Panda, N Dilek, B Martens, C Quinn, M McDermott, C Heatwole, C Thornton, R Moxley III
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- 14.00 **O64 Cardiac Safety and Efficacy of Mexiletine in Myotonic Dystrophy Type 1 (DM1)**
R Moxley III, E Logigian, M McDermott, W Martens, S Pandya, A Wiegner, C Thornton, R Tawil, R Moxley IV, C Barbieri, C Annis, N Dilek
-
- 14.15 **O65 Effects of Mexiletine on myotonia, muscle strength, and cardiac parameters in myotonic dystrophies over time**
V Sansone, S Briganti, M Panzeri, R Mauro, A Degrade, L De Ambroggi and G Meola
-
- 14.30 **O66 Safety and Tolerability of Recombinant Human Insulin-Like Growth Factor 1 Complexed with IGF Binding Protein 3 (rhIGF1/IGFBP3) in Myotonic Dystrophy Type 1 (DM1)**
C Heatwole, W Martens, C Quinn, J Hilbert, S Pandya, C Jackson, C Thornton, M McDermott, R Moxley
-
- 14.45 **O67 Development of RNA-based therapeutic model system for myotonic dystrophy (DM)**
RS Yadava, EG Ames, V Srinivasan and MS Mahadevan
-
- 15.00 **O68 Oligonucleotide-mediated silencing of expanded DMPK transcripts in a DM1 myoblast-myotube cell model**
SAM Mulders, WJAA van den Broek, G Gourdon, Bé Wieringa and DG Wansink
-
- 15.15 **O69 Correction of CIC-1 splicing eliminates the myotonia in mouse models of myotonic dystrophy**
TM Wheeler, JD Lueck, MS Swanson, RT Dirksen and CA Thornton
-
- 15.30 **O70 Biochemical screening methods to find inhibitors of RNA/protein interaction: CUG^{exp}/MBNL1 example**
K Sobczak and CA Thornton
-
- 16.00 **O71 Antisense RNA-based gene therapy reverses muscle atrophy in a mouse model of myotonic dystrophy type 1**
J Puymirat, G Doucet, A Vignaud, A Huguet, D Furling, G Gourdon, A Ferry
-

Session 7 Poster session 16.00 – 16.15

THERAPEUTIC TRIALS AND FUTURE ADVANCES

Session Chair: Benedikt Schoser

- P75 Reduced oxidative stress markers after cysteine donor enriched dietary intake in patients with myotonic dystrophy type 1**
L Volpi, M Falorni, C Carlesi, G Ricci, M Mancuso, L Petrozzi, M Franzini, A Paolicchi, G Siciliano
- P76 Dehydroepiandrosterone in myotonic dystrophy type 1**
I Pénisson-Besnier, M Devillers, R Porcher, D Orlikowski, V Doppler, C Desnuelle, X Ferrer, MC Arne Bes, F Bouhour, C Tranchant, E Lagrange, A Vershueren, D Uzenot, C Vial, A Labarre Vila, J Pouget, B Eymard, D Annane
- P77 Abnormal glucose metabolic disorder in myotonic dystrophy type 1 (DM1): hyperinsulinemia in DM1 were inhibited using voglibose**
M Kinoshita, M Shigeta, K Hirose
- P78 Characterization of MBNL1 RNA ligands and search for molecules disrupting the (CUG)n-MBNL1 interaction**
L Guigou, P Villa and N Charlet
- P79 High-throughput screen of chemical compounds to identify candidates that relieve the nuclear retention of CUG-rich mRNA**
E Querido and P Chartrand
- P80 Non Invasive Assessment of Mouse Muscle Volume Using 3D μ -Echography**
M Nejjari, M Janier, G Gourdon, J Puymirat and B Hiba

Coffee break

16.15 – 16.30

Session 8 INTERACTIVE SESSION AND HIGHLIGHT

Interactive session: Special meeting with advocacy groups, patients and families 16.30 – 18.30

- **Representative advocacy groups**
 - Shannon M. Lord
Hunter Research Fund (USA)
How the Myotonic Dystrophy Foundation started
 - Margaret Bowler
Myotonic Dystrophy Support Group (UK)
Encouragement to look after our own health needs day by day (patients and carers)
 - Claude Bourlier
French Myotonic Dystrophy Support Group (France)
Living with a DM1, patient point of view
 - Toba Balaban
DM2 Support/Advocacy Group (International)
Myotonic Dystrophy Toronto Regional Support Group (Canada)
The DM2 Experience: Web-based patient support
 - Caterina Campanelli
Associazione Italiana contro le Miopatie Rare – AIM (Italy)
Migliorare la conoscenza dei bisogni fondamentali delle famiglie colpite da miopatie per fornire loro risposte più efficaci
- **Questions from patients group to investigators and physicians**
- **Myotonic Dystrophy Foundation Excellence in Research Award**

- John Brekka
- **Young investigator's Poster Award**
Representative AIM (Associazione Italiana di Miologia)
- **Guidelines and standards of care for DM**
Dick Moxley and Chairmen

Highlight: Chris Pearson, Ralf Krahe, Bruno Eymard, Dick Moxley

18.30 – 19.30

Concluding Remarks

19.30

Milan, 2007

I ***t would be a wonderful***

D ***elight to welcome in***

M ***ilan as the perfect host for the***

C ***onsortium meeting***