Full Program
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OMMYD-2 Meeting	SCIENTIFIC CONFERENCE	DM PATIENTS AND FAMILIES SESSION

OMMYD-2 Meeting

Wednesday, October 16, 2013

08.00 - 08.30	**Welcome and introduction to OMMYD Meeting** Overview of the meeting mandate and procedures
08.30 - 09.15	Break-out sessions with respective team leaders for each Special Interest Group Presentation of results from OMMYD-1 and resultof the literature review between OMMYD-1 and OMMYD-2
09.15 - 09.30	**Break**
09.30 - 12.00	Group discussion by Special Interest Group
12.00 - 13.00	**Lunch**
13.00 - 15.00	Group discussion by Special Interest Group
15.00 - 16.00	Plenary session with all Special Interest Group and group leader reports

SCIENTIFIC CONFERENCE

15.00 - 17.00	**Registration on site of IDMC-9**
17.00 - 17.30	**Welcome Address** Jon Andoni URTIZBEREA, Paris-Hendaye Adolfo LÓPEZ DE MUNAIN, San Sebastián
17.30 - 19.30	**Session 1 - Opening Keynote Lectures** Chairs: *Eric WANG, Boston and Genevieve GOURDON, París*
17.30 - 18:45	**S1.1 Molecular/cellular mechanisms in repeat expansion disorders** **Berend WIERINGA**, Nijmegen, The Netherlands
18:45 - 19:30	**S1.2 iPS cells in biology and disease** **Angel RAYA,** Barcelona, Spain
19.30 - 21.00	**Opening Ceremony** **Musical entertainment** *Kantakidetza Choir* *Welcome cocktail*

Thursday, October 17, 2013

08.00 - 12.00 Session 2 - Disease Mechanisms I

	Chairs: *Nicolas CHARLET-BERGUERAND, Paris and Christopher PEARSON, Toronto*
08.00 - 08.30	S2.1 Overview Lecture: RNA mediated toxicity in DM1 **Ruben ARTERO,** Valencia, Spain
08.30 - 10.00	S2.2 Platform presentations
10.00 - 10.30	**Coffee Break and poster viewing**
10.30 - 12.00	S2.3 Platform presentations
12.00 - 14.00	**Lunch** **P1 - Poster Viewing with Presenters**
	P1.1 Disease Mechanisms Chairs: *Manuel PÉREZ-ALONSO, Valencia and Lubov TIMCHENKO, Houston*
	P1.2 Tisue specific disease and cell/animal models Chairs: *Mani MAHADEVAN, Charlotesville and Nicolas SERGEANT, Lille*
14.00 - 18.00	**Session 3 – Tissue specific mechanisms** Chairs: *Shoichi ISHIURA, Riken and Derick WANSINK, Nijmegan*
14.00 - 14.30	*S3.1* Overview Lecture **Genevieve GOURDON**, Paris, France
14.30 - 16.00	S3.2. Platform presentations
16.00 - 16.30	**Coffee Break and poster viewing**
16.30 - 18.00	S3.3. Platform presentations
18.00 - 21.00	**Sightseeing Tour in San Sebastián**

Friday, October 18, 2013	
08.00 - 12.30	**Session 4 – Clinical measures and biomarkers** Chairs: *Giovanni MEOLA, Milan and Richard MOXLEY, Rochester*
08.00 - 08.30	S4.1 Overview Lecture **Bruno EYMARD,** Paris, France
08.30 - 10.00	S4.2 Platform presentations
10.00 - 10.30	**Coffee Break**
10.30 - 12.00	S4.3 Platform presentations
12.00 - 15.00	**Lunch** **P2 - Poster Viewing with Presenters**
	P2.1 Clinical measures and biomarkers Chairs: *Gerardo GUTIÉRREZ, Madrid* and C*had HEATWOLE, Rochester*
	P2.2 Therapeutic development in model systems Chairs: *Ralf KRAHE, Houston and Benedikt SCHOSER, Munich*
	P2.3 Therapeutic trials Chairs: *Virginia ARECHAVALA-GOMEZA, London and David BROOK, Nottingham*
	P2.4 Ethical-Legal-Social Issues Chairs: *Cynthia GAGNON, Jonquière*
15.00 - 19.00	**Session 5 – Therapeutic development and model systems** Chairs: *Denis FURLING, Paris and Laura RANUM, Gainesville*
15.00 - 15.30	S5.1 Overview Lecture: Multiple models, diverse approaches and progress on many fronts **Tom COOPER,** Houston, TX, USA
15.30 - 17.00	S5.2 Platform presentations
17.00 - 17.30	**Coffee break**
17.30 - 19.30	S5.3 Platform presentations

Saturday, october 19, 2013

08.00 - 10.00	**Session 6 – Therapeutic Trials** Chairs: *Frank BENNET, Carlsbad and Virginia ARECHAVALA-GOMEZA, London*
08.00 - 08.30	S6.1 Overview Lecture **Charles THORNTON**, Rochester, USA
08.30 - 10.00	S6.2 Platform presentations
10.00 - 10.30	**Coffee break**
10.30 - 12.00	**Session 7 – Ethical-Legal-Social Issues** Chairs: *Tee ASHIZAWA, Gainesville and Chad HEATWOLE, Rochester*
10.30 - 11.00	S7.1 Overview Lecture **Cynthia GAGNON**, Jonquière
11.00 - 12.00	S7.2 Platform presentations
12.00 - 14.00	**Lunch at the Kursaal**

DM PATIENTS AND FAMILIES SESSION

Saturday, October 19, 2013

14.00 - 16.30	**Session 8- Interactive Session with Patients & Family groups** Chairs: *Tee ASHIZAWA, Gainsville, FL, USA and Mª Carmen ÁLVAREZ, Valencia, Spain*
14.00 - 14.30	S8.1 Basic science highlights **Darren MONCKTON**, Glasgow, UK
14.30 - 15.00	S8.2 Clinical science lighlights **Mark ROGERS**, Wales, UK
15.00 - 15.30	S8.3 Therapy: current and future **Bruce WENTWORTH**, Framingham, USA
15.30 - 16.30	S8.4 Questions from DM patients and families (simultaneous rooms) S.8.4.1 Spanish speaking Associates S.8.4.2 English speaking Associates S.8.4.1 Other Associations (tentatively French, Italian and German speaking associates)
16.45 - 18.45	**Session 9- Social and Organizational Issues** Chairs: *Jon Andoni URTIZBEREA, Hendaya, France and Maury SWANSON, Gainesville, Florida, USA*
16.45 - 18.00	**S9.1 Advocacy Group presentations** S9.1.1 MDA S9.1.2 MDF S9.1.3 MDSG S9.1.4 AFM S9.1.5 ASEM
18.00 - 18.45	**S9.2 IDMC-9 and Steinert Awards presentations** S9.2.1 Award for the best platform presentation S9.2.2 Award for the best poster presentation S9.2.3 Award for a Trajectory in Clinical Research in DM1 S9.2.4 Award for a Trajectory in Basic Research in DM1 S9.2.5 Special Award to Shannon Lord (lecture by Larry Lord) S9.2.6 Lifetime Achievement Award (LAA Award) S9.2.7 Annoncement for the next IDMC Meeting location
20.00 - 23.00	***Farewell Gala Dinner at the Miramar Palace*** 20.00-21.00 Cocktail (livened up by "Jazzte Borrazte Band") 21.00-23.00 Dinner

Wednesday, October 16

17:45-19:30 **S1**
Keynote Lectures
Chairpersons: Genevieve GOURDON - Paris, Eric WANG - Boston

17:30-18:45 **S-1.1 Molecular/cellular Mechanisms in Repeat Expansion Disorders**
Be WIERINGA
NCMLS Radboud University Nijmegen Medical Centre, Dept. Cell Biology - Nijmegen

18:45-19:30 **S-1.2 Ips Cells in Biology and Disease**
Angel RAYA
Institute For Bioengineering Of Catalonia (IBEC) - Barcelona

Thursday, October 17

08:00-08:30 **S2.1 Overview Lectures**
Chairpersons: Nicolas CHARLET BERGUERAND - Illkirch, Christopher PEARSON - Toronto

08:00-08:30 **S2.1 RNA Mediated Toxicity in DM1**
Rubén D. ARTERO
Fundación De Investigación Sanitaria INCLIVA. Departamento De Genética, Universidad De Valencia - Burjasot (Valencia)

08:30-10:00 **Disease Mechanisms**
Chairpersons: Nicolas CHARLET BERGUERAND - Illkirch, Christopher PEARSON - Toronto

08:30-08:45 **O-01 Expanded (CUG)n RNA Expression as a Variable in DM1 Patients and Disease Models**
Anke GUDDE, Anchel GONZALEZ, Walther VAN DEN BROEK, Be WIERINGA, Derick G. WANSINK
Department Of Cell Biology, Nijmegen Centre For Molecular Life Sciences, Radboud University Nijmegen Medical Centre - Nijmegen, Prosensa Therapeutics B.V. - Leiden

08:45-09:00 **O-02 Correlation Between Muscle Histopathology Impairment and Spliceopathy in Myotonic Dystrophies**
Rosanna CARDANI, Giulia ROSSI, Laura Valentina RENNA, Enrico BUGIARDINI, Guiseppe NOVELLI, Annalisa BOTTA, Giovanni MEOLA
IRCCS-Policlinico San Donato - San Donato Milanese, Milan, TOR VERGATA UNIVERSITY - Rome, University Of Milan - Milan

09:00-09:15 **O-03 CELF1 regulates SERCA1 splicing via PMA pathway**
Yimeng ZHAO, Shoichi ISHIURA
University Of Tokyo - Tokyo

09:15-09:30 **O-04 Modifications to Toxic CUG RNAs Induce Structural Stability and Rescue mis-splicing in Myotonic Dystrophy**
Elaine DELORIMIER, Jeremy COPPERMAN, Alex TABER, Leslie COONROD, Emily REISTER, Feras ACKALL, Kush SHARMA, Peter TODD, Marina GUENZA, Andy BERGLUND
University Of Oregon, Institute Of Molecular Biology - Eugene, OR 97403, University Of Michigan - Ann Arbor, MI 48109

09:30-09:45 **O-05 Nkx2-5, a Modifier and Marker of RNA Toxicity in Skeletal Muscle in Myotonic Dystrophy**
Ramesh S YADAVA, Jordan GLADMAN, Mahua MANDAL, Qing YU, Mani S. MAHADEVAN
University Of Virginai - Charlottesville, VA, University Of Virginia - Charlottesville, Virginia

09:45-10:00 **O-06 Minigene Tau E1-E13 : a New Tool to Develop DM1 Transgenic Mice with an Associated Tauopathy**
Céline CARPENTIER, Fco. Jose FERNANDEZ-GOMEZ, Fanny JUMEAU, Amélie LABUDECK, Claire-Marie DHAENENS, Susanna SCHRAEN-MASCHKE, Luc BUÉE, Nicolas SERGEANT, Marie-laure CAILLET-BOUDIN
Inserm UMR 837, Alzheimer & Tauopathies, Jean-Pierre Aubert Research Centre, Faculty Of Medicine, IMPRT - Lille

10:30-12:00 **Disease Mechanisms**
Chairpersons: Nicolas CHARLET BERGUERAND - Illkirch, Christopher PEARSON - Toronto

10:30-10:45 **O-07 Polymorphisms in the MSH3 mismatch repair gene modifies levels of CTG somatic instability in myotonic dystrophy type 1**
Fernando MORALES, Melissa VÁSQUEZ, Adriana GUILLÉN, Carolina SANTAMARÍA, Patricia CUENCA, Tetsuo ASHIZAWA, Douglas E. WILCOX, Alison WILCOX, Darren MONCKTON
Universidad De Costa Rica, Instituto De Investigaciones En Salud - San José, University Of Florida, Department Of Neurology - Gainesville, Yorkhill Hospital, Ferguson-Smith Centre For Clinical Genetics - Glasgow, University Of Glasgow, Institute Of Molecular, Cell And Systems Biology, College Of Medical, Veterinary And Life Sciences - Glasgow

10:45-11:00 **O-08 5'-variant Repeats and a Flanking DNA Insertion in an Usual Myotonic Dystrophy Type 1 Family Stabilises the Expansion in Both the Soma and Germline**
Khalidah NASSER, Sarah CUMMING, Daniel TAGOE, Craig NAPIER, Heather SMALL, Josie MCGHIE, Berit ADAM, Cheryl LONGMAN, Alison WILCOX, Alexis M DUNCAN, Anne MCKEOWN, Darren MONCKTON
Institute Of Molecular, Cell And Systems Biology, College Of Medical, Veterinary And Life Sciences, University Of Glasgow - Glasgow, West Of Scotland Regional Genetics Service, Southern General Hospital - Glasgow

11:00-11:15 **O-09 Variant Repeats in Two Atypical Myotonic Dystrophy Type 1 Families Dramatically Stabilise the Expansion and Appear to Abolish Symptoms**
Sarah CUMMING, Jack PUYMIRAT, Darren MONCKTON
Institute Of Molecular, Cell And Systems Biology, College Of Medical, Veterinary And Life Sciences, University Of Glasgow - Glasgow, Department

Of Human Genetics, Centre Hospitalier Universitaire De Quebec - Quebec

11:15-11:30 **O-10 Aberrant Methylation Spreading in DM1-affected Human Embryonic Stem Cells**

Shira YANOVSKY-DAGAN , Michal AVITZOUR , Gheona ALTARESCU , Paul RENBAUM , Talia ELDAR-GEVA , Aharon PERETZ , Stella MITRANI-ROSENBAUM , Ephrat LEVY-LAHAD , Silvina EPSZTEJN-LITMAN , Rachel EIGES

Shaare Zedek Medical Center - Jerusalem, Hadassah Hebrew University Medical Center - Jerusalem

11:30-11:45 **O-11 Ran Proteins From Intronic Cctg Expansions in DM2 Patient Brains**

Tao ZU, John CLEARY, Yuanjing LIU, Tammy REID, Monica BANEZ-CORONEL, Guangbin XIA, Tetsuo ASHIZAWA, Anthony YACHNIS, Laura P. W RANUM

Center For NeuroGenetics, College Of Medicine, University Of Florida - Gainesville FL, Department Of Pathology, College Of Medicine, University Of Florida - Gainesville FL

11:45-12:00 **O-12 Suppression of Somatic Instability in Small Disease Associated Alleles for Myotonic Dystrophy Type 1 and Huntington Disease Linked with Milder Symptoms.**

Catherine HIGHAM, Darren MONCKTON

University Of Glasgow - Glasgow

12:00 **P1.1 Disease Mechanisms**
Chairpersons: Manuel PÉREZ ALONSO - Valencia, Lubov TIMCHENKO - Houston

- **P-001 Alternative Splicing Alterations of Ca2+ Handling Genes are Associated with Ca2+ Signal Dysregulation in DM1 and DM2 Myotubes**

Massimo SANTORO, Roberto PIACENTINI, Marcella MASCIULLO, Maria Laura Ester BIANCHI, Anna MODONI, Maria Vittoria PODDA, Enzo RICCI, Claudio GRASSI, Gabriella SILVESTRI

Fondazione Don Gnocchi-ONLUS - Milan, Institute Of Human Physiology, Università Cattolica - Rome, IRCCS San Raffaele Pisana - Rome, Department Of Geriatrics, Neuroscience And Orthopedics, Center Of Neuromuscular Disorders, Università Cattolica - Rome

- **P-002 Antioxidative Enzymes Activities in Patients with Myotonic Dystrophy Type 1**

Vidosava RAKOCEVIC STOJANOVIC, Stojan PERIC, Aleksandra NIKOLIC KOKIC, Dragan MARINKOVIC, Mihajlo B. SPASIC, Dusko BLAGOJEVIC

Neurology Clinic, Clinical Center Of Serbia, School Of Medicine, University Of Belgrade, Belgrade, Serbia - Belgrade, Department Of Physiology, Institute For Biological Research - Belgrade, Faculty For Special Education And Rehabilitation, University Of Belgrade, Belgrade, Serbia - Belgrade

- **P-004 Aberrant Splicing of Annexin Vii by PTB Impairs Muscle Cell Differentiation in Myotonic Dystrophy Type 1**

Richard PELLETIER, Denis FURLING, mark TARNOPOLSKY, Benedikt SCHOSER, Giovanni MEOLA, Jack PUYMIRAT

CHU Quebec - Quebec, Institut De Myologie - Paris, McMaster University Medical Center - Hamilton, Ludwig Maximilians University Munich - Munich, University Of Milan - Milan

- **P-005 Rbfox1 Cooperates with MBNL1 to Regulate Muscle-specific Splicing Events Altered in Myotonic Dystrophy**

Roscoe KLINCK, Angelique FOURRIER, philippe THIBAULT, Mathieu DURAND, elvy LAPOINTE, julain LAPOINTE, Genevieve GOURDON, Giovanni MEOLA, Denis FURLING, benoit CHABOT, Jack PUYMIRAT

Université De Sherbrooke - Sherbrooke, CHU Quebec - Quebec, INSERM U781, Hôpital Broussais - Paris, University Of Milan - Milan, Institut De Myology - Paris, University Of Sherbrooke - Sherbrooke

- **P-006 Sodium Channel Gene Mutation: a Possible Role in Myotonic Dystrophy Type 2 with Severe Myotonia**

Enrico BUGIARDINI, Alexandra SORIANO CAMINERO, Ilaria RIVOLTA, Rosanna CARDANI, Rea VALAPERTA, Federica CIRILLO, Matthew Paul WICKLUND, Giovanni MEOLA

Department Of Neurology, IRCCS Policlinico San Donato, University Of Milan - San Donato Milanese (MI), Department Of Neurology, Hershey Medical Center - Hershey, PA , Department Of Health Science, University Of Milan Bicocca - Monza (MB), Laboratory Of Muscle Histopathology And Molecular Biology, IRCCS Policlinico San Donato - San Donato Milanese (MI), Research Laboratories, Molecular Biology, IRCCS Policlinico San Donato - San Donato Milanese (MI), Laboratory Of Stem Cells For Tissue Engineering, IRCCS Policlinico San Donato - San Donato Milanese (MI)

- **P-007 DMPK, Hexokinase Ii and Src Form a Multimeric Complex on the Mitochondrial Outer Membrane, That Prevents Ros-induced Apoptosis.**

Sergio SALVATORI, Boris PANTIC, Elena TREVISAN, Anna CITTA, Maria Pia RIGOBELLO, Oriano MARIN, Paolo BERNARDI, Andrea RASOLA

Department Of Biomedical Sciences - Padova

- **P-008 Gangliosides Composition in Patients Affected by Myotonic Dystrophy Type 1 and Type 2**

Rea VALAPERTA, Manuela VALSECCHI, Rosanna CARDANI, Massimo AURELI, Fortunata LOMBARDI, Enrico BUGIARDINI, Elena COSTA, Sandro SONNINO, Giovanni MEOLA

IRCCS Policlinico SanDonato - Sandonato Milanese, Milan, University Of Milan - Milan

- **P-009 Roles of MBLN1 and/or MBLN2 on Tau Exon 2 Splicing in Physiological and Myotonic Dystrophy**

Céline CARPENTIER, Dana GHANEM, Fco. Jose FERNANDEZ-GOMEZ, Fanny JUMEAU, Amélie LABUDECK, Claire-Marie DHAENENS, Ian HOLT, Isabelle BEHM ANSMANT, Christiane BRANLANT, Nathalie GOURRIER, Nicolas CHARLET BERGUERAND, Joelle MARIE, Susanna SCHRAEN-MASCHKE, Luc BUÉE, Nicolas SERGEANT, Marie-laure CAILLET-BOUDIN

Inserm UMR837-1, Alzheimer & Tauopathies - Lille, Centre For Inherited Neuromuscular Disease (CIND) - Oswestry, Shropshire,SY10 7AG, Laboratoire AREMS, UMR 7214 - Vandoeuvre-les-Nancy, IGBMC - Illkrich, Institut De Myologie- Univ. Paris 6 - Paris

- **P-010 Regulation of Staufen1 during skeletal muscle development, differentiation, and regeneration.**

Aymeric RAVEL-CHAPUIS, Tara Ellen CRAWFORD, Marie-Laure BLAIS-CRÉPEAU, Guy BELANGER, Chase RICHER, Bernard Jean JASMIN

University Of Ottawa - Ottawa

- **P-011 Re-expression of PKM2 in Type 1 Myofibers Correlates with Altered Glucose Metabolism in Myotonic Dystrophy**

Zhihua (Tina) GAO, Thomas COOPER
Baylor College Of Medicine - Houston

- **P-012 Early Senescence of DM1 Myoblasts is Mediated by a Cugexp-rna/ros Pathway**
Coralie SAINT-JEAN, Erwan GASNIER, Frédérique RAU, ARNAUD JOLLET, Bodvael FRAYSSE, Arnaud KLEIN, Denis FURLING
UPMC Univ. Paris 06, UM 76, Institut De Myologie And Inserm, U974 And CNRS, UMR7215 - Paris

- **P-013 Splicing of ABLIM1 is Aberrant in DM1 Patients**
Natsumi OHSAWA, Michinori KOEBIS, Shoichi ISHIURA
University Of Tokyo - Tokyo

- **P-016 Biomolecules Modeling Implied in Myotonic Dystrophy Type 1 for Structure Based Drug Design**
Alex LOPEZ, Roger ESTRADA, Jose Ignacio BORRELL, Jordi TEIXIDO
Institut Quimic De Sarria (IQS) - Ramon Llull University (URL) - Barcelona

- **P-014 Staufen1 as a Splicing Modulator: Implications for DM1 Therapy?**
Emma BONDY-CHORNEY, Aymeric RAVEL-CHAPUIS, Tara Ellen CRAWFORD, Bernard Jean JASMIN, Jocelyn COTE
University Of Ottawa, Center For Neuromuscular Disease - Ottawa

- **P-015 Characterization of Complex Variant Expanded DMPK Alleles in Italian Patients with Myotonic Dystrophy Type 1**
Giulia ROSSI, Alessandra MORGANTE, Maria Rosaria D'APICE, Luana FONTANA, Federica SANGIUOLO, Claudio CATALLI, Francesco BRANCATI, Roberto MASSA, Guiseppe NOVELLI, Annalisa BOTTA
Tor Vergata University Of Rome - Rome, Tor Vergata Hospital - Rome

- **P-017 Antagonism Between MBNL and CELF Proteins in the Nucleus and Cytoplasm**
Eric WANG, Amanda WARD, Jimena GIUDICE, Daniel TREACY, Thomas WANG, David HOUSMAN, Thomas COOPER, Christopher BURGE
Massachusetts Institute Of Technology - Cambridge, Baylor College Of Medicine - Houston

- **P-018 CUG RNA Suppresses PGC-1alpha Activity and Perturbs Metabolic Homeostasis Via Activating DNa Damage Response ATM Signaling in DM1**
Xiang FANG, Rui GAO, Yong-ping LIU, Robert G. SMITH, Annalisa BOTTA, Guiseppe NOVELLI, Tetsuo ASHIZAWA, Nicolas CHARLET BERGUERAND, Denis FURLING, Partha SARKAR
University Of Texas Medical Branch - Galveston, Biopathology Department - Rome, University Of Florida - Gainesville, IGBMC - Illkirch, Institut De Myologie - Paris

- **P-019 Mutant CUG RNA Chronically Activates PKR Pathway Via Stimulating NADPH Oxidase Activity to Induce Skeletal Muscle Degeneration in Myotonic Dystrophy Type 1**
Xiang FANG, Rui GAO, Yong-ping LIU, Robert G. SMITH, Annalisa BOTTA, Guiseppe NOVELLI, Tetsuo ASHIZAWA, Nicolas CHARLET BERGUERAND, Denis FURLING, Partha SARKAR
University Of Texas Medical Branch - Galveston, Biopathology Department - -, Biopathology Department - Rome, University Of Florida - Gainesville, IGBMC - Illkirch, Institut De Myologie - Paris

- **P-003 Epiretinal Membrane: A Treatable Cause of Visual Disability in Myotonic Dystrophy Type 1**
Richard ROXBURGH, Hannah M. Kersten BOPTOM, Nicholas CHILD, Philip J. POLKINGHORNE, Chris FRAMPTON, Helen V. DANESH-MEYER
Department Of Neurology, Auckland City Hospital - Auckland, Department Of Ophthalmology, University Of Auckland - Auckland, Department Of Psychological Medicine, University Of Otago - Christchurch

12:00 **P1.2 Tissue specific disease and cell/animal models**
Chairpersons: Mani S. MAHADEVAN - Charlottesville, Va, Usa, Nicolas SERGEANT - Lille

- **P-020 Oligodendroglioma in Patient with Myotonic Dystrophy Type 1**
Vidosava RAKOCEVIC STOJANOVIC, Valerija DOBRICIC, Vesna RALIC, Milena MILICEV, M MILICEVIC, Danica GRUJICIC, Stojan PERIC
Neurology Clinic, Clinical Center Of Serbia, School Of Medicine, University Of Belgrade, Belgrade, Serbia - Belgrade

- **P-021 Synaptic Abnormalities in the Brain of DMSXL Mice are Not Associated with Neurodevelopmental Deficits or Structural Abnormalities**
Oscar HERNANDEZ-HERNANDEZ, Geraldine SICOT, Diana Mihaela DINCA, Aline HUGUET, Annie NICOLE, Luc BUÉE, Arnaud MUNNICH, Nicolas SERGEANT, Frank ANGESTEIN, Genevieve GOURDON, Mario GOMES-PEREIRA
Inserm U781, Hôpital Necker Enfants Malades - Paris, Inserm U837-1, Alzheimer And Tauopathies, Université Lille Nord De France - Paris, Laboratory For Non-Invasive Brain Imaging, Leibniz Institute For Neurobiology - Magdeburg

- **P-022 MSH3 Overexpression in Pancreas Destabilizes CTG Triplet Repeat in DM1 Mice Carrying 55 or ~500 CTG Repeats**
Stéphanie TOMÉ, Annie NICOLE, Laurent FOIRY, Aline HUGUET, Ian HOLT, Glenn E. MORRIS, Genevieve GOURDON
Inserm U781, Hôpital Necker Enfants Malades - Paris, Wolfson Center For Inherited Neuromuscular Disease - Oswestry, Shropshire, Wolfson Center For Inherited Neuromuscular Disease, RJAH Orthopaedic Hospital - Oswestry, Shropshire

- **P-023 Mutational dynamics of the CTG repeat in buccal cells reveals cheek swabs as an alternative source of DNA for genotyping in myotonic dystrophy type 1**
Eyleen CORRALES, Melissa VÁSQUEZ, Baili ZHANG, Carolina SANTAMARÍA, Patricia CUENCA, Mario SIRITO, Ralf KRAHE, Darren MONCKTON, Fernando MORALES
Universidad De Costa Rica - San José, University Of Texas MD Anderson Cancer Center - Houston, TX, University Of Glasgow - Glasgow

- **P-024 Dynamic Changes of Intranuclear Foci in Proliferating and Non-proliferating DM1 Neural Cells**
Guangbin XIA, Apoorva MOHAN, SH SUBRAMONY, Maurice SWANSON, Tetsuo ASHIZAWA
University Of Florida - Gainesville

- **P-025 Neonatal DMSXL mice carrying >1000CTG reveal severe molecular and morphological alterations in the**

diaphragm
Fadia MEDJA, Aline HUGUET, Annie NICOLE, Lise MICHEL, Arnaud MUNNICH, Genevieve GOURDON
Inserm U781 - Paris, Université Paris Descartes-Sorbonne Paris Cité, Institut Imagine - Paris

- ### P-026 The RNA-binding Protein Staufen1 Inhibits Myogenic Differentiation Via a c-Myc Dependent Pathway
Tara Ellen CRAWFORD, Aymeric RAVEL-CHAPUIS, Marie-Laure BLAIS-CRÉPEAU, Guy BELANGER, Bernard Jean JASMIN
Department Of Cellular And Molecular Medicine, Faculty Of Medicine, University Of Ottawa - Ottawa

- ### P-027 RAB3A and SYN1 Protein Abnormalities in DM1 Brain are Not Mediated by Missplicing or Developmental Delays
Diana Mihaela DINCA, Geraldine SICOT, Aline HUGUET, Annie NICOLE, Arnaud MUNNICH, Genevieve GOURDON, Mario GOMES-PEREIRA
Inserm U781, Hôpital Necker Enfants Malades - Paris

- ### P-028 Combining MBNL1 Loss and Gain of CELF1 to Model DM1 in Mice
Ginny MORRISS, Thomas COOPER
Baylor College Of Medicine - Houston

- ### P-029 Expression profile of RNA-binding proteins in several DM1 mouse muscles.
Aymeric RAVEL-CHAPUIS, Guy BELANGER, Tara Ellen CRAWFORD, Jocelyn COTE, Bernard Jean JASMIN
University Of Ottawa - Ottawa

- ### P-030 Characterization of MBNL1 Dependent Splicing Responses in a Tissue Culture DM Model
Stacey WAGNER, Riti GUPTA, Dylan FARNSWORTH, Andy BERGLUND
University Of Oregon - Eugene

- ### P-031 MBNL Compound Loss-of-function Models for Myotonic Dystrophy
Moyi LI, Kuang-Yung LEE, Mini MANCHANDA, Ranjan BATRA, Konstantinos CHARIZANIS, Apoorva MOHAN, Sonisha A. WARREN, Dustin FINN, Hannah HONG, Hassan ASHRAF, Hideko KASAHARA, Christopher M. CHAMBERLAIN, Laura P. W RANUM, Maurice SWANSON
Department Of Molecular Genetics And Microbiology And The Genetics Institute,University Of Florida, College Of Medicine - Gainesville FL, Department Of Physiology And Functional Genomics,University Of Florida, College Of Medicine - Gainesville FL

- ### P-032 Increased Phosphorylation of eIF2alpha and Reduced Levels of CDK2 Contribute to Premature Growth Arrest in DM1 Lens Cells.
Jeremy RHODES, Sarah RUSSELL, Michael WORMSTONE
University Of East Anglia - Norwich

14:00-14:30 S3.1 Overview Lectures
Chairpersons: Shoichi ISHIURA - Tokyo, Derick G. WANSINK - Nijmegen

14:00-14:30 ### S3.1 The multi-facets of DM
Genevieve GOURDON, Lise MICHEL, Aline HUGUET, Annie NICOLE, Fadia MEDJA, Arnaud MUNNICH
Inserm, U781 - Paris

14:30-16:00 Tissue specific mechanisms
Chairpersons: Shoichi ISHIURA - Tokyo, Derick G. WANSINK - Nijmegen

14:30-14:45 ### O-13 SCN5A Splicing Alteration in Heart of Myotonic Dystrophy Patients
Fernande FREYERMUTH, Chantal SELLIER, Christelle THIBAULT, Thomas ZIMMER, Didier AUBOEUF, Eric WANG, Vincent NAVRATIL, Denis FURLING, Masanori TAKAHASHI, Nicolas CHARLET BERGUERAND
IGBMC - Illkirch, Institut Für Physiologie Kollegiengasse - JENA, Centre Léon Bérard - Lyon, Koch Institute For Integrative Cancer Research - Cambridge, PRABI - Villeurbanne, Institut De Myologie - Paris, University Of Osaka - Osaka

14:45-15:00 ### O-14 Implication of BIN1 in Myotonic Dystrophy
Michel NEY, Denis FURLING, Nicolas CHARLET BERGUERAND
IGBMC - Illkirch-Graffenstaden, Institut De Myologie - Paris Cedex 13

15:00-15:15 ### O-15 Early Abnormalities in the DMSXL Mouse Model for DM1: a Model of the Congenital Form?
Lise MICHEL, Aline HUGUET, Annie NICOLE, Fadia MEDJA, Arnaud MUNNICH, Genevieve GOURDON
Inserm U781 - Hôpital Necker Enfants Malades - Paris

15:15-15:30 ### O-16 Phenotype-genotype Correlation in Congenital Myotonic Dystrophy
Masayuki NAKAMORI, Kohei HAMANAKA, Mitsuru FURUTA, Hideki MOCHIZUKI, Yukiko HAYASHI, Ichizo NISHINO, Masanori TAKAHASHI
Department Of Neurology, Osaka University Graduate School Of Medicine - Suita, Osaka, National Institute Of Neuroscience, National Center Of Neurology And Psychiatry - Kodaira, Tokyo

15:30-15:45 ### O-17 Altered Splicing of Cardiac Sodium Channel Might Be Responsible for Cardiac Conduction Defects in Myotonic Dystrophy
Yosuke KOKUNAI, Hideki ITOH, Yoshihiro KINO, Moyi LI, Masayuki NAKAMORI, Takashi KIMURA, Tsuyoshi MATSUMURA, Harutoshi FUJIMURA, Nobuyuki NUKINA, Hideki MOCHIZUKI, Saburo SAKODA, Minoru HORIE, Shouichi ICHIURA, Keiji IMOTO, Maurice SWANSON, Nicolas CHARLET BERGUERAND, Masanori TAKAHASHI
Osaka University Graduate School Of Medicine - Suita, Shiga University Of Medical Science - Ohtsu, Juntendou University - Tokyo, University Of Florida College Of Medicine - Gainesville, Hyogo Medical College - Nishinomiya, National Hospital Organization Toneyama Hospital - Toyonaka, University Of Tokyo - Tokyo, National Institute For Physiological Sciences - Okazaki, Institut De Génétique Et De Biologie Moléculaire Et Cellulaire (IGBMC) - Illkirch

15:45-16:00 ### O-18 The distribution of splicing defects in the Myotonic Dystrophy type 1 brain
Takashi KIMURA, Koichi SUENAGA, Masayuki NAKAMORI, Tsuyoshi MATSUMURA, Masanori TAKAHASHI, Harutoshi FUJIMURA, Kenji JINNAI, Hiroo YOSHIKAWA
Division Of Neurology, Hyogo College Of Medicine - Nishinomiya, Hyogo, Department Of Neurology, Osaka University Graduate School Of Medicine - Suita, Osaka, Department Of Neurology, NHO Toneyama Hospital - Toyonaka, Osaka, Department Of Neurology, NHO Hyogo-Chuo

Hospital - Sanda, Hyogo

16:30-18:00 **Tissue specific mechanisms**
Chairpersons: Shoichi ISHIURA - Tokyo, Derick G. WANSINK - Nijmegen

16:30-16:45 **O-19 Genome Wide Identification of Aberrant Alternative Splicing Events in Myotonic Dystrophy Type 2**

Alessandra PERFETTI, Simona GRECO, Pasquale FASANARO, Enrico BUGIARDINI, Rosanna CARDANI, Jose Manuel GARCIA MANTEIGA, Michela RIBA, Elia STUPKA, Giovanni MEOLA, Fabio MARTELLI

IRCCS Policlinico San Donato - San Donato Milanese (Milan), IRCCS Fondazione Santa Lucia - Rome, San Raffaele Scientific Institute - Milan

16:45-17:00 **O-20 Contribution of Dystrophin Alternative Mis-splicing to DM Muscle Dysfunction**

Frédérique RAU, Laetitia RAMANOUDJAME, Ludovic ARANDEL, ARNAUD JOLLET, Cécile PECCATE, Stéphanie LORAIN, Edor KABASHI, Valérie ALLAMAND, Joelle MARIE, Denis FURLING

UPMC Univ Paris 06, UM 76, Institut De Myologie And Inserm, U974 And CNRS, UMR7215 - Paris, Inserm, UMR_S975, CRICM, F-75013, UPMC Univ Paris 06, UMR_S975, F-75013, Paris, France ; CNRS UMR 7225 - Paris

17:00-17:15 **O-21 Two Enhancers Control Expression of Drosophila Muscleblind in the Somatic Musculature and in the Nervous System**

Ariadna BARGIELA, M. Beatriz LLAMUSI, Estefania CERRO, Rubén D. ARTERO

University Of Valencia - Burjassot, Valencia, University Of Valencia - Valencia

17:15-17:30 **O-22 Contribution of Endogenous Tau Loss of Expression on Behaviour, Splicing and Biochemical Patterns in SXL Transgenic Mouse Model of Myotonic Dystrophy.**

Fco. Jose FERNANDEZ-GOMEZ, Céline CARPENTIER, Sabiha EDDARKAOUI, Mario GOMES-PEREIRA, Claire-Marie DHAENENS, SEBASTIEN CARRIER, HELENE OBRIOT, Aline HUGUET, Susanna SCHRAEN-MASCHKE, DAVID BLUM, Luc BUÉE, Marie-laure CAILLET-BOUDIN, Genevieve GOURDON, Nicolas SERGEANT

Inserm UMR 837, Alzheimer & Tauopathies, Jean-Pierre Aubert Research Centre, Faculty Of Medicine, IMPRT - Lille, Inserm U781, Faculté De Medecine Paris Descartes - Paris

17:30-17:45 **O-23 Continuing somatic expansion of the CTG repeat in myotonic dystrophy type 1 patients contributes to the age of onset of symptoms**

Melissa VÁSQUEZ, Rebeca VINDAS, Eyleen CORRALES, Baili ZHANG, Carolina SANTAMARÍA, Patricia CUENCA, Gerardo DEL VALLE, Roberto BRIAN, Mauricio SITTENFELD, Mario SIRITO, Ralf KRAHE, Darren MONCKTON, Fernando MORALES

Instituto De Investigaciones En Salud (INISA)-Universidad De Costa Rica (UCR) - San José, Department Of Genetics, University Of Texas MD Anderson Cancer Center - Houston, TX, Laboratorio De Neurofisiología (Neurolab) - San José, Servicio De Neurología, Hospital Nacional De Niños - San José, Servicio De Neurología, Hospital San Juan De Dios - San José, Institute Of Molecular, Cell And Systems Biology, College Of Medical, Veterinary And Life Sciences, University Of Glasgow - Glasgow

17:45-18:00 **O-24 Identification of Molecular Pathways Misregulated by CUGBP1 and ZNF9 in Myotonic Dystrophies Type 1 and Type 2**

Lubov TIMCHENKO, Kyle LEWIS, Zachary ZALEWSKI, Karlie JONES, David NELSON, Nikolai TIMCHENKO, Christina WEI

Baylor College Of Medicine - Houston

Friday, October 18

08:00-08:30 **S4.1 Overview Lectures**
Chairpersons: Giovanni MEOLA - San Donato Milanese (mi), Richard-Thomas MOXLEY, III - Rochester, Ny

08:00-08:30 **S4.1 New insights into adult and childhood DM1 phenotype, cardio-respiratory prognostic considerations, and outcome measures**

Bruno EYMARD, Guillaume BASSEZ, D. DUBOC, D. ANNANE, D. ORLIKOWSKI, H. PRIGENT, delphine HERON, Aurelia JACQUETTE, Nathalie ANGEARD, M. DOUNIOL, D. COHEN, Helene RADVANYI, C. BOILEAU, Jean - yves HOGREL, Luc J. HÉBERT, Jack PUYMIRAT,

Salpêtrière Hospital, Myology Institute - Paris, Henri Mondor Hospital, Histology Department - Creteil, Raymond Poincaré Hospital, Respiratory Intensive Care Department - Garches, Salpêtrière Hospital, Genetic Department - Paris, Salpêtrière Hospital, Child Psychiatry Department - Paris, Ambroise Paré Hospital, Molecular Genetic Department - Boulogne, Laval University Quebec - Quebec, French DM Clinical - -

08:30-10:00 **Clinical measures and biomarkers**
Chairpersons: Giovanni MEOLA - San Donato Milanese (mi), Richard-Thomas MOXLEY, III - Rochester, Ny

08:30-08:45 **O-25 Histological and Magnetic Resonance Imaging Findings of Vastus Lateralis in Myotonic Dystrophies: Do they Match Up?**

Carla MERKEL, Mike P. WATTJES, Jens REIMANN, Cornelia KORNBLUM

Department Of Neurology, University Of Bonn - Bonn, Department Of Radiology, VU University Medical Center - Amsterdam

08:45-09:00 **O-26 Myotonic Dystrophies - Disorders of the Central Nervous System**

Vidosava RAKOCEVIC STOJANOVIC, Stojan PERIC, Aleksandra PAVLOVIC, Leposava BRAJKOVIC, Massimo FILIPPI, Vladimir KOSTIC

Neurology Clinic, Clinical Center Of Serbia, School Of Medicine, University Of Belgrade, Belgrade, Serbia - Belgrade, Clinical Center Of Serbia, School Of Medicine, University Of Belgrade, Belgrade, Serbia - Belgrade, Center For Nuclear Medicine, Clinical Center Of Serbia, School Of Medicine, University Of Belgrade, Belgrade, Serbia - Belgrade, San Raffaele Scientific Institute, Vita-Salute San Raffaele University, Milan, Italy - Belgrade

09:00-09:15 **O-27 Natural History of Skeletal Muscle Decline in Myotonic Dystrophy Type 1: a Retrospective Study in 204 Cases**

Jack PUYMIRAT, Louise COSSETTE, Jean Pierre BOUCHARD, Guillaume BASSEZ

CHU Quebec - Quebec, Enfant Jesus Hospital - Quebec, CHU Henri Mondor - Creteil

09:15-09:30 **O-28 Increased Brain Biomarker Plasmatic Concentration in Myotonic Dystrophy Type I**

Susanna SCHRAEN-MASCHKE, Florence RICHARD, Patrick GELÉ, Claire-Marie DHAENENS, Vercruysse OLIVIER, Amélie LABUDECK,

Bernard SABLONNIERE, Sabiha EDDARKAOUI, Marie-laure CAILLET-BOUDIN, Vermersch PATRICK, Arnaud LACOUR, Luc BUÉE, Nicolas SERGEANT

Inserm UMR 837, Alzheimer & Tauopathies, Jean-Pierre Aubert Research Centre, Faculty Of Medicine, IMPRT - Lille, University Of Lille II - Lille, Department Of Neurology - Lille

09:30-09:45 **O-29 Seeking for Convenient Outcome Measures for the Characterization of the Neuromuscular Function in Dm1 Patients**

Jean - yves HOGREL, Valérie DECOSTRE, Gwenn OLLIVIER, Jack PUYMIRAT, Luc J. HÉBERT, Anthnoy BÉHIN, Tanya STOJKOVIC, Pascal LAFORÊT, Laurent SERVAIS, Bruno EYMARD, Guillaume BASSEZ

Institut De Myologie - Paris, Université De Laval - Quebec, National Defense Of Canada - Quebec

09:45-10:00 **O-30 Which Myotonic Dystrophy Type 1 Adult Patients Require Cardiology Referral?**

William GROH, Robert M. PASCUZZI

Indiana University School Of Medicine - Indianapolis

10:30-12:00 Clinical measures and biomarkers

Chairpersons: Giovanni MEOLA - San Donato Milanese (mi), Richard-Thomas MOXLEY, III - Rochester, Ny

10:30-10:45 **O-31 Challenges Related to the Selection of Outcome Measures in the Context of a Rare and Slowly Progressive Disease: Conclusions From the Outcome Measures in Myotonic Dystrophy (OMMYD-2) Meeting**

Cynthia GAGNON, Giovanni MEOLA, Luc J. HÉBERT, Luc LABERGE, Chad HEATWOLE, Mario LEONE

Groupe De Recherche Interdisciplinaire Sur Les Maladies Neuromusculaires/Université De Sherbrooke - Jonquière, University Of Milan - Milan, Université Laval/Garnison De Valcartier - Quebec, Écobes/Cégep De Jonquière - Jonquière, University Of Rochester Medical Center - Rochester, Université Du Québec à Chicoutimi - Chicoutimi

10:45-11:00 **O-32 Effect of mild and adult DM1 phenotype on neuropsychological functioning**

Louis RICHER, Stéphane JEAN, Cynthia GAGNON

Département Des Sciences De La Santé, Université Du Québec à Chicoutimi - Chicoutimi, Québec, Programme Santé Mentale, Centre De Santé Et De Services Sociaux De Chicoutimi - Chicoutimi, Québec, École De Réadaptation, Faculté De Médecine Et Des Sciences De La Santé, Université De Sherbrooke - Sherbrooke, Québec

11:00-11:15 **O-33 Cardiometabolic Risk Factors in Patients with Myotonic Dystrophy Type 1**

Patricia BLACKBURN, Diane BRISSON, Luigi BOUCHARD, Cynthia GAGNON

Department Of Health Sciences, Université Du Québec à Chicoutimi - Saguenay, ECOGENE-21 Laboratory And Lipid Clinic, Chicoutimi Hospital - Saguenay, Department Of Biochemistry, Université De Sherbrooke - Sherbrooke, Faculty Of Medicine And Health Sciences, Université De Sherbrooke, Groupe De Recherche Interdisciplinaire Sur Les Maladies Neuromusculaires (GRIMN), Neuromuscular Clinic, Centre De Santé Et De Services Sociaux De Jonquière - Saguenay

11:15-11:30 **O-34 ACTN3 Genotypes and Muscular Performance in DM1 Patients: Preliminary Results**

Mario LEONE, Anne-marie FORTIN, Patrick DESJARDINS, Luc J. HÉBERT, Élise DUCHESNE, Cynthia GAGNON, Hung Tien BUI

Université Du Québec à Chicoutimi - Saguenay, Université De Laval - Quebec, Université De Sherbrooke - Sherbrooke

11:30-11:45 **O-35 A Clinical Study of Myotonic Dystrophy Type-1 (DM1) Patients' Perception and Prioritization of Cognitive Symptoms**

Chad HEATWOLE, Nicholas JOHNSON, Elizabeth LUEBBE, Nuran DILEK, William MARTENS, Charles THORNTON, Richard-Thomas MOXLEY, III

The Department Of Neurology, University Of Rochester - Rochester, NY, The Department Of Neurology, University Of Utah - Salt Lake City, Utah

11:45-12:00 **O-36 Finding Predictors for Cardiac Conduction Abnormalities in DM1. A 33 Yrs. Prospective Study in a Cohort of 102 DM1 Patients with Normal ECG at Baseline**

Giovanni ANTONINI, ELISABETTA BUCCI, Erica GABRIELE, Alessandra FRATTARI, Loretta LICCHELLI, Nicola VANACORE, Marco TESTA

Dept. Od Neurosciences, Mental Health And Sensory Organs (NESMOS). University Of Rome - Rome, Cardiologic Institute. Ospedale Sant'Andrea - Rome, Ospedale Sant'Andrea - Rome, National Institute Of Health - Rome

12:00 P2.1 Clinical measures and biomarkers

Chairpersons: Gerardo GUTIÉRREZ - San Sebastián De Los Reyes, Chad HEATWOLE - Rochester

- **P-033 Self-questionnaire is Effective for Screening of Patients with Myotonic Dystrophy**

Tsuyoshi MATSUMURA, Takashi KIMURA, Yosuke KOKUNAI, Masayuki NAKAMORI, Katsuhisa OGATA, Harutoshi FUJIMURA, Masanori TAKAHASHI, Hideki MOCHIZUKI, Saburo SAKODA

Department Of Neurology, National Hospital Organization Toneyama National Hospital - Toyonaka, Department Of Internal Medicine, Division Of Neurology, Hyogo Medical College Of Medicine - Nishinomiya, Department Of Neurology, Osaka University Graduate School Of Medicine - Suita, Department Of Neurology, National Hospital Organization Higashi-Saitama Hospital - Hasuda

- **P-034 Obesitas Surgery in DM1 - a Case Report.**

Karin HAKANSSON

Neuromuscular Centre Sahlgrenska University Hospital - Gothenburg

- **P-035 Dental Health Condition is Closely Associated with Upper Limb Motor Function in Myotonic Dystrophy Type 1**

Hiroto TAKADA, Yuka TAKEUCHI, Junko SATO, Takayuki HANADA, Mao KUDO, Kazuya ONO, Yumiko OGASAWARA, Hiroko TAKAYA

Aomori Hospital, National Hospital Organization - Aomori

- **P-036 Polyneuropathy and Myotonic Dystophy Type1: Primary Involvement of Nerves or Incidental Coexistence?**

Marcella MASCIULLO, Anna MODONI, Marco LUIGETTI, Maria Laura Ester BIANCHI, Mauro LOMONACO, Massimo SANTORO, Gabriella SILVESTRI

IRCCS San Raffaele Pisana - Rome, Institute Of Neurology, Catholic University Of Sacred Heart - Rome, Don Carlo Gnocchi Foundation - Milan

- **P-037 Daytime Sleepiness and Fatigue Symptoms in Myotonic Dystrophy Patients: a 9-year Longitudinal Study**

Luc LABERGE, Benjamin GALLAIS, Cynthia GAGNON

ÉCOBES - Recherche Et Transfert - Jonquière, École De Réadaptation, FMSS, Université De Sherbrooke - Sherbrooke

- **P-038 Standardized Quantitative Motor Assessment in Patients with Myotonic Dystrophy Type 1: the Use of Sensitive Measures to Characterize the Disease Progression**

  Luc J. HÉBERT, Joanne SAULNIER, Jack PUYMIRAT

  Centre For Interdisciplinary Research In Rehabilitation And Social Integration - Quebec City, Institut De Réadaptation En Déficience Physique De Québec - Quebec City, Human Genetic Research Unit, Centre Hospitalier De L'Université Laval - Quebec City

- **P-039 Experienced Fatigue in Myotonic Dystrophy Type 1 (DM1) is Associated with Muscle Impairment and Depression**

  Stefan WINBLAD, Christopher LINDBERG

  Department Of Psychology, University Of Gothenburg - Gothenburg, Neuromuscular Center - Gothenburg

- **P-040 Validation of Sensitivity and Specificity of a New Diagnostic Certified Assay to Better Characterize the CCTG Number Repeats in DM2**

  Fortunata LOMBARDI, Rea VALAPERTA, Rosanna CARDANI, Enrico BUGIARDINI, Federica SCHIAVON, Elena COSTA, Giovanni MEOLA

  Research Laboratories - Molecular Biology, IRCCS Policlinico San Donato, Milan, Italy. - Milano, Department Of Neurology, Stroke Unit And Centre For Neuromuscular Disease, IRCCS Policlinico San Donato, Milan, Italy. - Milano, Experteam S.r.l - Venezia

- **P-041 Elevated miRNA Levels in Serum of Myotonic Dystrophy Patients Relate to Disease Progress**

  Andrie KOUTSOULIDOU, Tassos KYRIAKIDES, Yiolanda CHRISTOU, Eleni ZAMBA PAPANICOLAOU, Leonidas PHYLACTOU

  Department Of Molecular Genetics, Function & Therapy, The Cyprus Institute Of Neurology & Genetics - Nicosia, Yale Center For Analytical Sciences At Yale School Of Public Health, University Of Yale - New Haven, Neurology Clinic D, The Cyprus Institute Of Neurology & Genetics - Nicosia

- **P-042 The Quebec's Founder Effect Has Little Influence on the Phenotype of Patients with Myotonic Dystrophy Type 1 (DM1).**

  Louise COSSETTE, Celine DOGAN, Jean MATHIEU, Guillaume BASSEZ, Jack PUYMIRAT

  CHU Quebec - Quebec, CHU Henri Mondor - Creteil, Centre De Santé Et De Services Sociaux De Jonquière - Jonquière

- **P-043 The Usefulness and Limitations of Repeat-primed PCR in Myotonic Dystrophy Type 1 and 2 Molecular Testing**

  Jan RADVANSZKY, Andrej FICEK, Ludevit KADASI

  Institute Of Molecular Physiology And Genetics, Slovak Academy Of Sciences - Bratislava, Comenius University, Faculty Of Natural Sciences, Department Of Molecular Biology - Bratislava

- **P-044 Chronic pain in persons with adult and mild phenotypes of Myotonic Dystrophy type 1**

  Maud-Christine CHOUINARD, Marye-Ève ST-GELAIS, Danielle POIRIER, Mélissa LAVOIE, Cynthia GAGNON

  Université Du Québec à Chicoutimi - Chicoutimi (Québec), Université De Sherbrooke - Campus Saguenay - Chicoutimi, Québec, Groupe De Recherche Interdisciplinaire Sur Les Maladies Neuromusculaires - Jonquière (Québec)

- **P-045 Patient-reported Weakness, Myotonia and Swallowing Difficulties in the UK Myotonic Dystrophy Patient Registry.**

  Antonio ATALAIA, Libby WOOD, Ros QUINLIVAN, Richard ORRELL, Richard PETTY, Margaret PHILLIPS, Mark ROBERTS, Mark ROGERS, Michael ROSE , Cheryl LONGMAN, Darren MONCKTON, David HILTON-JONES, Chris TURNER, Hanns LOCHMÜLLER

  Newcastle University - Newcastle , Great Ormond Street Hospital - London, University College London - London, Southern General Hospital - Glasgow, Royal Derby Hospital - Derby, Salford Royal - Manchester , University Hospital Of Wales - Cardiff, Kings College London - London, Institute Of Molecular Cell And Systems Biology - Glasgow, John Radcliffe Hospital - Oxford , University College Hospital - London

- **P-046 Non-invasive Ventilation in Myotonic Dystrophy Type 1 (DM1): Tolerance, Compliance and Impact on Quality of Life**

  Elisa FALCIER, Elisa DE MATTIA, Barbara GARABELLI, Marino IATOMASI, Ksenija GORNI, Fabrizio RAO, Elisabetta ROMA, Paola Francesca CASTELLOTTI, Valeria Ada SANSONE

  Centro Clinico NEMO, Fondazione Serena - Pneumology Unit, University Of Milan - Milan, Centro Clinico NEMO, Fondazione Serena - Neurology Unit, University Of Milan - Milan

- **P-047 Nutritional Intake of Patients with Myotonic Dystrophy Type 1**

  Mélissa BRIEN, Patricia BLACKBURN, Maud-Christine CHOUINARD, Cynthia GAGNON

  Department Of Health Sciences, Université Du Québec à Chicoutimi - Saguenay, Faculty Of Medicine And Health Sciences, Université De Sherbrooke, Groupe De Recherche Interdisciplinaire Sur Les Maladies Neuromusculaires (GRIMN), Neuromuscular Clinic, Centre De Santé Et De Services Sociaux De Jonquière - Saguenay

- **P-048 A preliminary prevalence estimate of DM1 and DM2 in the Lazio region, Italy.**

  Nicola VANACORE, Emanuele RASTELLI, ELISABETTA BUCCI, Maria Laura Ester BIANCHI, CHIARA TERRACCIANO, MANLIO GIACANELLI, Antonio PETRUCCI, Sandro COSTANZI-PORRINI, Gabriella SILVESTRI, Giovanni ANTONINI, Roberto MASSA

  National Institute Of Health - Rome, TOR VERGATA UNIVERSITY - Rome, LA SAPIENZA UNIVERSITY - Rome, CATTOLICA UNIVERSITY - Rome, S.CAMILLO-FORLANINI HOSPITAL - Rome

- **P-049 The Symptomatic Impact of Childhood and Congenital Onset Myotonic Dystrophy Type-1 (DM1): a Multi-national, Cross Sectional Study**

  Nicholas JOHNSON, Craig CAMPBELL, Anne-berit EKSTROEM, Heather ADAMS, Nuran DILEK, Eileen EASTWOOD, Elizabeth LUEBBE, James HILBERT, Richard-Thomas MOXLEY, III, Chad HEATWOLE

  The Department Of Neurology, The University Of Utah - Salt Lake City, The Department Of Paediatrics, London Health Science Centre - London, Ontario, Department Of Paediatrics, Northern Älvsborg County Hospital - Trollhättan, Department Of Neurology, University Of Rochester - Rochester, New York

- **P-050 The High Competing Risk of Non-cancer Mortality Obscures Cancer Burden in Patients with Myotonic Dystrophy**

  Shahinaz GADALLA, Ruth PFEIFFER, Sigurdur KRISTINSSON, Magnus BJÖRKHOLM, James HILBERT, Richard-Thomas MOXLEY, III, Ola LANDGREN, Mark GREENE

Division Of Cancer Epidemiology And Genetics, National Cancer Institute - Rockville, Maryland, Department Of Medicine, Division Of Hematology, - Stockholm, Department Of Neurology, Neuromuscular Disease Center, - Rochester, NY, Metabolism Branch, National Cancer Institute - Bethesda, Maryland

- **P-051 Gender-related Differences in Myotonic Dystrophy Type 1: the DM-Scope Registry Observational Study**

  Guillaume BASSEZ, Celine DOGAN, Marie DE ANTONIO, Dalil HAMROUN, Romain GHERARDI, Bruno EYMARD, Jack PUYMIRAT, French - Quebec MYOTONIC DYSTROPHY CLINICAL NETWORK

  Henri Mondor University Hospital - Creteil, Montpellier University Hospital - Montpellier , Pitié-Salpêtrière University Hospital - Paris, Laval University Hospital - Quebec

- **P-052 Cognitive Imparment in DM1: a Longitudinal Study**

  Adolfo LÓPEZ DE MUNAIN, Sara LÓPEZ , Arantza ALDALUR, Roberto FERNÁNDEZ, Miren ZULAIKA, Miren MANEIRO, José Ignacio EMPARANZA, Andone SISTIAGA

  Hospital Universitario Donostia - San Sebastián, Universidad Autonoma De Barcelona Student - Barcelona, Universidad Del País Vasco Student - San Sebastián, Instituto De Investigación Biodonostia - San Sebastián, Universidad Del País Vasco - San Sebastián

- **P-053 Long-term Outcomes of a Series of Childhood-onset Form of DM1 Patients**

  Aurelia JACQUETTE, Jenifer FABRE-TESTE, Helene RADVANYI, Nathalie ANGEARD, Bruno EYMARD, delphine HERON

  Departement De Genetique, Myology Institute, Pitie-Salpetriere Hospital - Paris, Departement De Genetique, Pitie-Salpetriere Hospital - Paris, Laboratoire De Biologie Moleculaire, Ambroise Pare Hospital - Boulogne Billancourt, Myology Institute, Pitie-Salpetriere Hospital - Paris

- **P-054 Neuromuscular Transmission Defects in Myotonic Dystrophy Type 1: a Neurophysiological Study.**

  Antonio PETRUCCI, Ludovico LISPI, Sandro COSTANZI-PORRINI, Angela ROSINI, MANLIO GIACANELLI, Emanuele RASTELLI, Roberto MASSA

  Neuromuscular And Neurological Rare Diseases Center, San Camillo Hospital - Rome, Neuromuscular Center, Dept. Of Systems Medicine, Tor Vergata University Of Rome - Rome

- **P-055 Peculiar signs and symptoms of Myotonic Dystrophy MD1 affected members in a Hungarian family**

  Zsuzsanna HARTYANI, Piroska IMRE, Timea ENGLERT, Ilona PATAKY, Gabor KISS, Jakab GEISZT

  Department Of Earth And Environmental Sciences - Veszpém, Department Of Neurology - Veszpém, Department Of Psychiatry - Veszprem, Institute Of Psychology - Budapest, Department Of Neurology - Budapest, Fourmed Medical - Veszprem

- **P-056 A Ten-year Follow-up Study on Muscle Strength and Motor Function in Children, Adolescents and Young Adults with Myotonic Dystrophy Type 1 in Southern and Western Sweden**

  Anne-berit EKSTROEM, Anna-Karin KROKSMARK

  Queen Silvia Children's Hospital - Gothenburg

- **P-057 Analysis of unexpanded and intermediate CTG polymorphisms at the DMPK gene in Mexican population and in Native Amerindian Groups**

  Nadia Mireya MURILLO, Luz MÁRQUEZ, Lorena OROZCO, Rocío GÓMEZ, Elvia MENDOZA, Yessica Sarai TAPIA, Norberto LEYVA, Bulmaro CISNEROS, Oscar HERNÁNDEZ, Jonathan Javier MAGAÑA

  Department Of Genetics, National Rehabilitation Institute - Mexico, Medicine Genomic National Institute - Mexico, Department Of Toxicology, CINVESTAV-IPN - Mexico, Department Of Genetics And Molecular Biology, CINVESTAV-IPN - Mexico

12:00 **P2.2 Therapeutic development in model system**
Chairpersons: Ralf KRAHE - Houston, Benedikt SCHOSER - Munich

- **P-058 Balance Exercise Programme in Myotonic Dystrophy Type 1: Evaluation of a Single-subject Experimental Study**

  Elisabet HAMMARÉN, Christopher LINDBERG, Gunilla KJELLBY

  Sahlgrenska University Hospital - Göteborg

- **P-059 Role of BRUNO-3, CUGBP1 Orthologue, in a Drosophila Model of Myotonic Dystrophy Type 1**

  Lucie PICCHIO, Yoan RENAUD, Emilie PLANTIÉ, Krzysztof JAGLA

  GReD, INSERM 1103, CNRS 6293 - Clermont-FerrandL

- **P-060 Manumycin a Corrects Aberrant Splicing of Clcn1 in Myotonic Dystrophy Type 1 (DM1) Mice**

  Kosuke OANA, Shoichi ISHIURA

  University Of Tokyo - Tokyo

- **P-061 High-throughput Screen for Pharmacological Compounds That Target CUG-rich RNA Foci in Myotonic Dystrophy**

  Pascal CHARTRAND, Emmanuelle QUERIDO, Catherine MÉNARD, Jean-François MERCIER

  Université De Montréal - Montréal

- **P-062 Evaluating the Effects of CELF1 Deficiency in a Mouse Model of RNA Toxicity**

  Yun Kyoung KIM, Mahua MANDAL, Ramesh S YADAVA, Mani S. MAHADEVAN

  Univerity Of Virginia - Charlottesville

- **P-063 Muscle and Brain-specific Inducible Mouse Models of DM2**

  John CLEARY, Jamie MARGOLIS, Tao ZU, Tammy REID, Yuanjing LIU, Christopher M. CHAMBERLAIN, Yuan-Lin KANG, Laura P. W RANUM

  Center For NeuroGenetics, College Of Medicine, University Of Florida - Gainesville, FL , Department Of Genetics, Cell Biology And Development, University Of Minnesota - Minneapolis, MN

- **P-064 Observations on Oligonucleotide Based Therapy for Myotonic Dystrophy**

  Rebecca MOORE, Ami KETLEY, Paulina POWALOWSKA, Christopher J. HAYES, DAVID BROOK

  University Of Nottingham - Nottingham

- **P-065 High Content Screening Using Hes Derived Mesenchymal Stem Cells Carrying the Myotonic Dystrophy**

Type 1 Mutation

Yves MAURY, Marc LECHUGA, Benjamin BRINON, Julien CÔME, Pauline POYDENOT, Morgane GAUTHIER, Stephane NEDELEC, Camille LECUYER, Geneviève PIÉTU, Marc PESCHANSKI, Cécile MARTINAT

CECS/I-Stem, AFM - Evry, Institut De La Vision - Paris, INSERM UMR 861, I-Stem AFM - Evry

- ### P-066 GSK3 Inhibition is a Novel Therapeutic Approach to Suppress Progression of Myotonic Dystrophy Type 1

Christina WEI, Karlie JONES, Polina IAKOVA, James KILLIAN, Nikolai TIMCHENKO, Lubov TIMCHENKO

Baylor College Of Medicine - Houston

- ### P-067 Metformin control of multiple alternative RNA splicing opens new therapeutic potentials

Sandrine BAGHDOYAN, Delphine LAUSTRIAT, Jacqueline GIDE, Laetitia BARRAULT, Alban VIGNAUD, Aline HUGUET, Genevieve GOURDON, Guillaume CHARPENTIER, Denis FURLING, Guillaume BASSEZ, Marc PESCHANSKI

INSERM/UEVE UMR 861, I-STEM - Evry, Genethon - Evry, Inserm U781 - Paris, CERITD - Evry, Institut De Myologie, UMR7215 - Paris, GH Henri Mondor - Creteil

- ### P-068 Systemic delivery of an RNAi therapy improves the phenotype of the HSALR mouse model of DM1

Darren R. BISSETT, Carrie A. STOLTZMAN, Ewa STEPNIAK-KONIECZNA, Charles THORNTON, Joel R. CHAMBERLAIN

University Of Washington School Of Medicine - Seattle, WA, University Of Rochester School Of Medicine And Dentistry - Rochester, NY

12:00 P2.3 Therapeutic trials

Chairpersons: DAVID BROOK - Nottingham, Virginia ARECHAVALA - Barakaldo

- ### P-069 Prevalence and Clinical Correlates of Sleep Disordered Breathing in Myotonic Dystrophy Type 1 and 2

Maria Laura Ester BIANCHI, Anna LOSURDO, Chiara DI BLASI, Massimo SANTORO, Marcella MASCIULLO, Venanzio VALENZA, Antonello DAMIANI, Giacomo DELLA MARCA, Gabriella SILVESTRI

UCSC - Rome, Fondazione Don Gnocchi - Rome, IRCCS San Raffaele Pisana - Rome, UILDM Sezione Laziale - Rome

- ### P-070 Effects of a Physical Exercise Programme in Adults with Myotonic Dystrophy Type 1 – a One-year Follow-up Study with Per-protocol Analysis

Marie KIERKEGAARD

Department Of Physical Therapy, Karolinska University Hospital - 171 76 Stockholm

- ### P-071 Quantitative Muscle Strength Impairments at the Ankle in Myotonic Dystrophy Type 1 (DM1) Patients: a Five-year Follow-up

Luc J. HÉBERT, Joanne SAULNIER, Jean-Francois REMEC, Christophe VIAL, Jack PUYMIRAT

Centre For Interdisciplinary Research In Rehabilitation And Social Integration - Quebec City, Institut De Réadaptation En Déficience Physique De Québec - Quebec City, Service De Neuro-rééducation, Hôpital Pierre Werteimer Groupement Hospitalier Est - Lyon, Department Of Electro-neurophysiology And Muscular Pathology, Hôpital Pierre Werteimer Groupement Hospitalier Est - Lyon, Human Genetic Research Unit, Centre Hospitalier De L'Université Laval - Quebec City

- ### P-072 The Italian Registry for Myotonic Dystrophy Type 1 and 2: Ready to Start

Barbara FOSSATI, Giovanni MEOLA

IRCCS Policlinico San Donato - San Donato Milanese (MI)

- ### P-073 UK Myotonic Dystrophy Patient Registry: Developing Standards of Care and Clinical Research

Chris TURNER, David HILTON-JONES, Libby WOOD, Ros QUINLIVAN, Richard ORRELL, Richard PETTY, Mark ROBERTS, Mark ROGERS, Michael ROSE , Cheryl LONGMAN, Margaret PHILLIPS, Darren MONCKTON, Hanns LOCHMÜLLER

University College Hospital - London, John Radcliffe Hospital - Oxford , Newcastle University - Newcastle , Great Ormond Street Hospital - London, Institute Of Neurology - London, Southern General Hospital - Glasgow, University Hospital Of Wales - Cardiff, Salford Royal - Manchester , Kings College London - London, Division Of Rehabilitation Medicine, - Derby, Institute Of Molecular Cell And Systems Biology - Glasgow

- ### P-074 A Roman Network for the Myotonic Dystrophies: Construction of a DM1/DM2 Clinical and Genetic Database in a Large Population

Emanuele RASTELLI, ELISABETTA BUCCI, Maria Laura Ester BIANCHI, CHIARA TERRACCIANO, Nicola VANACORE, MANLIO GIACANELLI, Antonio PETRUCCI, Sandro COSTANZI-PORRINI, Gabriella SILVESTRI, Giovanni ANTONINI, Roberto MASSA, on behalf of THE ROMAN NETWORK OF MYOTONIC DYSTROPHIES

TOR VERGATA UNIVERSITY - Rome, LA SAPIENZA UNIVERSITY - Rome, CATTOLICA UNIVERSITY - Rome, National Institute Of Health - Rome, S.CAMILLO-FORLANINI HOSPITAL - Rome

- ### P-075 Impact of an 8-week Periodized Combined Training Program in DM1 Patients: a Pilot Study

Mario LEONE, Anne-marie FORTIN, Patrick DESJARDINS, Luc J. HÉBERT, Cynthia GAGNON

Université Du Québec à Chicoutimi - Saguenay, Université De Laval - Quebec, Université De Sherbrooke - Sherbrooke

- ### P-076 Assessment of Gait Instability in Myotonic Dystrophy Type 1

Valentina GATTI, Marco GUALANDRIS, Fatmira BESHIRI, Valentina MORETTINI, Cristina GRANDI, Valentina RE, Ksenija GORNI, Nadia CELLOTTO, Valeria Ada SANSONE

Centro Clinico NEMO, Fondazione Serena Onlus - University Of Milan - Milan

- ### P-077 Magnetic Resonance Identifies Muscle Wasting Associated with Fatty Infiltration and Depletion of Contraction Over a 60-month Period in Type 1 Myotonic Dystrophy

Eric LAROSE, Karine BIBEAU, Luc J. HÉBERT, Jack PUYMIRAT

Université Laval - Quebec, Institut Universitaire De Cardiologie Et De Pneumologie De Québec - Quebec

12:00 P2.4 Ethical-Legal-Social Issues

Chairpersons: Cynthia GAGNON - Jonquière

- ### P-078 Myotonic Dystrophy Guidelines and the Critical Importance of Resourcing and Support

Petty RICHARD, Rahman MONIKA, McKeown ANNE, Robb YVONNE, McLeish LORNA, Gregory HELEN, Wilcox ALISON, Longman CHERYL

Department Of Neurology - Glasgow, West Of Scotland Regional Genetics Service - Glasgow, South East Scotland Regional Genetics Service -

Edinburgh, East Of Scotland Genetics Service - Dundee, North Scotland Genetic Service - Aberdeen

- **P-083 Patient Education Strategies Used by Nurses to Support Self-management of People with Myotonic Dystrophy Type 1**
  Maud-Christine CHOUINARD, Mélissa LAVOIE, Chantale SIMARD, Frances GALLAGHER, Louise-Catherine DE JORDY, Céline NEPTON, Julie BOUCHARD, Cynthia GAGNON
  Université Du Québec à Chicoutimi - Chicoutimi, Québec, Université De Sherbrooke - Sherbrooke, Québec, Université De Sherbrooke - Sherbrooke, Université De Sherbrooke - Camous Saguenay - Jonquière, Québec

- **P-079 Assessing Quality of Care in Myotonic Dystrophy: the Scottish Experience**
  Cheryl LONGMAN, Yvonne ROBB, Helen GREGORY, Lorna MCLEISH, Alison WILCOX, Anne MCKEOWN, Monika RAHMAN, Barry KOURYASYS, Karyn ROBERTSON, Maria FARRUGIA, Anne-Marie TAYLOR, Maureen EDWARDS, Richard PETTY
  West Of Scotland Regional Genetics Service - Glasgow, South East Scotland Regional Genetics Service - Edinburgh, North Scotland Genetic Service - Aberdeen, East Of Scotland Genetics Service - Dundee, Scottish Muscle Network - Glasgow, Institute Of Neurology - Glasgow

- **P-080 The Challenge of Increasing Attendance at Myotonic Dystrophy Clinics**
  Irene PARSONS, Yvonne ROBB, Maureen EDWARDS, Helen GREGORY, Lorna MCLEISH, Alison WILCOX, Maria FARRUGIA, Richard PETTY, Cheryl LONGMAN
  West Of Scotland Regional Genetics Service - Glasgow, South East Scotland Regional Genetics Service - Edinburgh, North Scotland Genetic Service - Aberdeen, East Of Scotland Genetics Service - Dundee, Institute Of Neurology - Glasgow

- **P-081 Do Guidelines Guide the Generalist? the Scottish Experience**
  Yvonne ROBB, Helen GREGORY, Lorna MCLEISH, Maria FARRUGIA, Richard PETTY, Cheryl LONGMAN
  South East Scotland Regional Genetics Service - Edinburgh, North Scotland Genetic Service - Aberdeen, East Of Scotland Genetics Service - Dundee, Institute Of Neurology - Glasgow, West Of Scotland Regional Genetics Service - Glasgow

- **P-082 The Scottish Myotonic Dystrophy Database: What Can It Tell Us?**
  Mark HAMILTON, Cheryl LONGMAN, Anne MCKEOWN, Yvonne ROBB, Lorna MCLEISH, Helen GREGORY, Catherine MCWILLIAM , Monika RAHMAN, Anne-Marie TAYLOR, Alison WILCOX, Barry KOURYASYS, Karyn ROBERTSON, Maria FARRUGIA, Richard PETTY, Maureen EDWARDS
  West Of Scotland Regional Genetics Service, Southern General Hospital - Glasgow, South East Scotland Regional Genetics Service - Edinburgh, East Of Scotland Genetics Service - Dundee, North Scotland Genetic Service - Aberdeen, Scottish Muscle Network - Glasgow, Institute Of Neurology - Glasgow

- **P-084 Myotonic Dystrophy Type 1 and 2: Time-lag to Diagnosis in Italy**
  Alice ZANOLINI, Barbara FOSSATI, Valeria Ada SANSONE, Giovanni MEOLA
  Dept. Of Neurology, IRCCS Policlinico San Donato, Università Degli Studi Di Milano - Milan, Centro Clinico NEMO, Fondazione Serena - Milan

- **P-085 Healthcare Organization Disparities Across Neuromuscular Clinics in the Province of Quebec (Canada) for DD1 Population: Observation and Challenges**
  Annie PLOURDE, Maud-Christine CHOUINARD, Mélissa LAVOIE, Danielle MALTAIS, Julie LÉTOURNEAU, Cynthia GAGNON
  GRIMN - Groupe De Recherche Interdisciplinaire Sur Les Maladies Neuromusculaires - Jonquiere (Quebec), UQAC - Université Du Québec à Chicoutimi - Chicoutimi (Quebec), Université De Sherbrooke, Site Saguenay - Jonquiere (Quebec)

- **P-086 Factors Influencing the Effectiveness of Educational Interventions by Nurses for People with Myotonic Dystrophy Type 1(DM1): an Ecological Approach**
  Mélissa LAVOIE, Frances GALLAGHER, Maud-Christine CHOUINARD, Chantale SIMARD, Julie BOUCHARD, Louise-Catherine DE JORDY, Cynthia GAGNON
  Université De Sherbrooke - Campus Saguenay - Chicoutimi (Québec), Université De Sherbrooke - Sherbrooke (Québec), Université Du Québec à Chicoutimi - Chicoutimi (Québec), Groupe De Recherche Interdisciplinaire Sur Les Maladies Neuromusculaires - Jonquière (Québec)

- **P-087 Referral Criteria for Occupational Therapy Services Related to Independent Housing Among DM1 Patients**
  Kateri RAYMOND, Louis-Pierre AUGER, Sabrina ST-ONGE, Marie-France CORMIER, Christine VACHON, Cynthia GAGNON
  Université De Sherbrooke - Jonquière, Université De Sherbrooke - Sherbrooke, Université De Sherbrooke - Saguenay

- **P-088 Resonance and Myotonic Dystrophy Type 1**
  Claire-cecile MICHON
  Université Paris 8 - 93200 Saint-Denis

- **P-089 Illuminating Loss - an Artistic Study of Siblings Affected by Myotonic Dystrophy Type 1**
  Jacqueline DONACHIE
  University Of Northumbria - Newcastle Upon Tyne

15:00-15:30 **S5.1 Overview Lectures**
Chairpersons: Denis FURLING - Paris, Laura P. W RANUM - Gainesville

15:00-15:30 **S5.1 Multiple models, diverse approaches and progress on many fronts**
Thomas COOPER
Baylor College Of Medicine - Houston

15:30-17:00 **Therapeutic development in model system**
Chairpersons: Denis FURLING - Paris, Laura P. W RANUM - Gainesville

15:30-15:45 **O-37 Age of Onset of RNA Toxicity Influences Phenotypic Severity: Evidence from an Inducible Mouse Model of Myotonic Dystrophy (DM1)**
Jordan GLADMAN, Mahua MANDAL, Varadamurthy SRINIVASAN, Mani S. MAHADEVAN
Department Of Pathology, University Of Virginia - Charlottesville , Parexel International - Bethesda

15:45-16:00 **O-38 Shorter CAG Morpholino Oligo is More Effective to Reverse the Function of MBNL1.**
Kanako NAGANO, Shoichi ISHIURA
The University Of Tokyo - Meguro-Ku, Tokyo

16:00-16:15 **O-39 Inhibition of the Tweak/fn14 Pathway Leads to Improved Survival and Functional Outcomes in a Mouse Model of Myotonic Dystrophy Type 1**

Mani S. MAHADEVAN, Erin P FOFF, Ramesh S YADAVA, Qing YU, Jordan GLADMAN, Carla D FRENZEL, Timothy ZHENG

University Of Virginia-Department Of Pathology - Charlottesville, Virginia, Univeristy Of Virginia-Department Of Neurology - Charlottesville, Virginia, Biogen-Idec - Cambridge, MA

16:15-16:30 **O-40 Rapid in Vivo Detection of Therapeutic Drug Effects in a Novel Therapy Reporter Mouse Model of DM1**

Thurman WHEELER, Soumya MITRA, Andrew SOROKA, Frank C. BENNETT, Thomas FOSTER

Massachusetts General Hospital - Boston, University Of Rochester Medical Center - Rochester, Isis Pharmaceuticals Inc. - Carlsbad

16:30-16:45 **O-41 New Human Muscle Cell Models for DM**

Ludovic ARANDEL, Coralie SAINT-JEAN, Arnaud KLEIN, ARNAUD JOLLET, Frédérique VOVARD, Kamel MAMNCHAOUI, Jack PUYMIRAT, Vincent MOULY, Denis FURLING

UPMC UM76, Institut De Myologie - Paris, UPMC Univ. Paris 6, Um76 / Inserm, U974 / CNRS, Umr7215 - Paris, Département De Génétique Humaine - Quebec

16:45-17:00 **O-42 High Content Imaging Screens Using Myotonic Dystrophy Cell Lines Identify Small Molecules That Remove Nuclear Foci**

Ami KETLEY, Catherine Z. CHEN, Xin LI, Sukrat ARYA, Thelma ROBINSON, Javier GRANADOS-RIVERON, Inyang UDOSEN, Glenn E. MORRIS, Ian HOLT, Denis FURLING, Soraya CHAOUCH, Ben HAWORTH, Noel SOUTHALL, Paul SHINN, Wei ZHENG, Christopher P. AUSTIN, Christopher J. HAYES, DAVID BROOK

University Of Nottingham - Nottingham, National Center For Advancing Translational Sciences - Bethesda, Wolfson Centre For Inherited Neuromuscular Disease - Keele, Institut De Myologie And Inserm - Paris, Molecular Devices - Wokingham

17:30-19:30 Therapeutic development in model system

Chairpersons: Denis FURLING - Paris, Laura P. W RANUM - Gainesville

17:30-17:45 **O-43 Development of Small Molecules Able to Lessen Myotonic Dystrophy Disease Traits After in Vivo Drosophila Screening Candidate Identification**

Irma GARCÍA ALCOVER, Jordi COLONQUES BELLMUNT, Juan Manuel FERNÁNDEZ COSTA, Isabel CAMPILLO NUEVO, Aida TOMÁS PRADO, Mari Carmen ALVAREZ, Manuel PÉREZ ALONSO, Arturo LÓPEZ CASTEL

Valentia BioPharma SL - Paterna

17:45-18:00 **O-44 Systemic Delivery of an Antisense Oligonucleotide (ASO) Targeting Dmpk RNA Improves the Phenotype of DMSXL Mice**

Dominic JAUVIN, Jessina CHRETIEN, Sanjay PANDEY, Laurie MARTINEAU, Guillaume BASSEZ, Aline HUGUET, Robert A MCLEOD, Genevieve GOURDON, Thurman WHEELER, Charles THORNTON, Frank C. BENNETT, Jack PUYMIRAT

CHU Quebec - Quebec, Isis Pharmaceuticals Inc. - Carlsbad, CHU Henri Mondor - Creteil, INSERM U781, Institut Imagine - Paris, University Of Rochester Medical Center - Rochester

18:00-18:15 **O-45 Small molecule inhibitor of MBNL1 induces Myotonic Dystrophy type 1 pre-mRNA splicing defects**

Ewa STEPNIAK-KONIECZNA, Jessica L. CHILDS-DISNEY, Tuan TRAN, Ilyas YILDIRIM, HaJeung PARK, Catherine Z. CHEN, Jason HOSKINS, Noel SOUTHALL, Juan J. MARUGAN, Samarjit PATNAIK, Wei ZHENG, Christopher P. AUSTIN, George C. SCHATZ, Charles THORNTON, Matthew D. DISNEY, Krzysztof SOBCZAK

Department Of Gene Expression, Institute Of Molecular Biology And Biotechnology, Adam Mickiewicz University - Poznan, Poland, Department Of Chemistry, The Scripps Research Institute, Scripps Florida - Jupiter, Department Of Chemistry, Northwestern University - Evanston, Illinois , NIH Chemical Genomics Center, National Center For Advancing Translational Sciences, National Institutes Of Health, Bethesda - Bethesda, Maryland , Department Of Neurology, School Of Medicine And Dentistry, University Of Rochester - Rochester, New York , Department Of Chemistry, Northwestern University - Evanston, Illinois

18:15-18:30 **O-46 Identification of New Candidate Genes Involved in Myotonic Dystrophy Type 1: From Drosophila Model to DM1 Cases in Human**

Emilie PLANTIÉ, Lucie PICCHIO, Yoan RENAUD, Krzysztof JAGLA

GReD, CNRS UMR 6293, INSERM U1103, Clermont-Ferrand, France - CLERMONT-FERRAND

18:30-18:45 **O-47 Assessment of Tricyclo-DNA Antisens Oligonucleotide to Target Nuclear CUG-expanded RNA**

Arnaud KLEIN, Ludovic ARANDEL, Christian LEUMANN, Luis GARCIA, Denis FURLING

UPMC Univ Paris 06, UM 76 ; Institut De Myologie ; Inserm, U974 And CNRS, UMR7215 - Paris, Department Of Chemistry & Biochemistry - Bern, Inflammation & Therapeutiques - Saint Quentin En Yvelines

18:45-19:00 **O-48 Very short oligonucleotides composed of locked nucleic acids as potential therapeutic molecules in Myotonic Dystrophy**

Agnieszka WOJTKOWIAK-SZLACHCIC, Katarzyna KSIAZEK, Lukasz SZNAJDER, Agnieszka PIASECKA, Krzysztof SOBCZAK

Department Of Gene Expression, Institute Of Molecular Biology And Biotechnology, Adam Mickiewicz University, - Poznan, Poland

19:00-19:15 **O-49 Development of a gene therapy strategy to target the CUG repeats and restore MBNL activity**

Denis FURLING, Ludovic ARANDEL, Fco. Jose FERNANDEZ-GOMEZ, ARNAUD JOLLET, Claire-Marie DHAENENS, Arnaud FERRY, Sabiha EDDARKAOUI, Geraldine SICOT, Mario GOMES-PEREIRA, Genevieve GOURDON, Frédérique RAU, Helene TRAN, Camille LEMERCIER, Morvane COLLIN, Luc BUÉE, Arnaud KLEIN, Marie-laure CAILLET-BOUDIN, Joelle MARIE, Nicolas SERGEANT

UPMC UM76, Institut De Myologie - Paris, Inserm UMR 837, Alzheimer & Tauopathies, Jean-Pierre Aubert Research Centre, Faculty Of Medicine, IMPRT - Lille, Inserm U781 - Paris

19:15-19:30 **O-50 CNBP Knock-out Mouse Model for Myotonic Dystrophy Type 2 (DM2)**

Mario SIRITO, L. Clifton STEPHENS, Bjarne UDD, Ralf KRAHE

University Of Texas MD Anderson Cancer Center - Houston, Texas, University Of Texas MD Anderson Cancer Center - Houston, Tampere University Hospital And Medical School - Tampere

Saturday, October 19

08:00-08:30 S6.1 Overview Lectures
Chairpersons: Frank C. BENNETT - Carlsbad, Virginia ARECHAVALA - Barakaldo

08:00-08:30 **S6.1 Trial Readiness and Implementation**

Charles THORNTON

University Of Rochester Medical Center - Rochester

08:30-10:00 Therapeutic trials
Chairpersons: Frank C. BENNETT - Carlsbad, Virginia ARECHAVALA - Barakaldo

08:30-08:45 **O-51 Efficacy and Cellular Uptake of Antisense Oligonucleotides to Treat Myotonic Dystrophy Type 1**

Anchel GONZALEZ, S MULDERS, Julia KRANZEN, Jeroen VAN DE GIESSEN , Huib CROES, Ingeborg VAN KESSEL, Walther VAN DEN BROEK, Nicole DATSON, Judith VAN DEUTEKOM, Be WIERINGA, Derick G. WANSINK

Prosensa Therapeutics B.V. - Leiden, Department Of Cell Biology, Nijmegen Centre For Molecular Life Sciences, Radboud University Nijmegen Medical Centre - Nijmegen

08:45-09:00 **O-52 Strategies in Antisense Oligo Treatment of Muscle and Brain in DM1**

S MULDERS, Walther VAN DEN BROEK, B AGUILERA, Jeroen VAN DE GIESSEN , CE VAN DER ZEE , Nicole DATSON, Be WIERINGA, Judith VAN DEUTEKOM, Derick G. WANSINK

Prosensa Therapeutics B.V. - Leiden, Department Of Cell Biology, Nijmegen Centre For Molecular Life Sciences, Radboud University Nijmegen Medical Centre - Nijmegen

09:00-09:15 **O-53 The Ultrasound-enhanced Delivery of Morpholino: Therapeutic Trial of Myotonia with Antisense Oligonucleotides**

Michinori KOEBIS, Shoichi ISHIURA

University Of Tokyo - Tokyo

09:15-09:30 **O-54 Pre-clinical Characterization of Generation 2.5 (gen 2.5) Antisense Oligonucleotides (asos) Targeting DMPK in Mice and Cynomolgus Monkeys for the Treatment of DM1**

Sanjay PANDEY, Thurman WHEELER, Samantha JUSTICE, Aneeza SALIM, Husam YOUNIS, Susan FREIER, Charles THORNTON, Frank C. BENNETT, Robert MACLEOD

Isis Pharmaceuticals Inc. - Carlsbad, University Of Rochester - New York

09:30-09:45 **O-55 Potential Therapeutics for Myotonic Dystrophy: Pentamidine and Analogs as Inhibitors of Transcription**

Leslie COONROD, Masayuki NAKAMORI, Wenli WANG, Samuel CARRELL, Cameron HILTON, Micah BODNER, Ruth SIBONI, Aaron DOCTER, Michael HALEY, Charles THORNTON, Andy BERGLUND

Institute Of Molecular Biology, University Of Oregon - Eugene, Oregon, Department Of Neurology, Osaka University Graduate School Of Medicine - Suita, Osaka, Department Of Neurology, University Of Rochester - Rochester, New York, Department Of Chemistry, University Of Oregon - Eugene, Oregon

09:45-10:00 **O-56 Actinomycin D Inhibits Transcription of CUG RNA in a DM1 Model**

Ruth SIBONI, Matthew TANNER, Masayuki NAKAMORI, Leslie COONROD, John BERGLUND

Institute Of Molecular Biology, University Of Oregon - Eugene , University Of Osaka - Osaka

10:30-11:00 S7.1 Overview Lectures
Chairpersons: Tetsuo ASHIZAWA - Gainesville, Chad HEATWOLE - Rochester

10:30-11:00 **S7.1 Myotonic dystrophy type 1 as a model of organized complexity**

Cynthia GAGNON

Université De Sherbrooke - Jonquière

11:00-12:00 Ethical, legal and social issues
Chairpersons: Tetsuo ASHIZAWA - Gainesville, Chad HEATWOLE - Rochester

11:00-11:15 **O-57 Assessment of Occupational Performance with the Evaluation Instrument Assessment of Motor and Process Skills (AMPS) in Persons with Myotonic Dystrophy Type 1.**

Ulrika EDOFSSON, Isabelle OTTENVALL HAMMAR

Neuromuscular Centre Sahlgrenska University Hospital - Gothenburg, Sahlgrenska University Hospital - Gothenburg

11:15-11:30 **O-58 Implementation of a nursing case management in Myotonic Dystrophy type 1 patients**

Maud-Christine CHOUINARD, Mélissa LAVOIE, Danielle MALTAIS, Cynthia GAGNON

Université Du Québec à Chicoutimi - Chicoutimi (Québec), Université De Sherbrooke - Campus Saguenay - Chicoutimi, Québec, Groupe De Recherche Interdisciplinaire Sur Les Maladies Neuromusculaires - Jonquière (Québec)

11:30-11:45 **O-59 Cognitive Impairments and Their Effect Upon Everyday Life in Non Congenital Forms of Myotonic Dystrophy**

Margaret PHILLIPS, Amy BANKS

University Of Nottingham - Derby, University Of Nottingham - Nottingham

11:45-12:00 **O-60 Optimistic: Observational Prolonged Trial in Myotonic Dystrophy Type 1 to Improve Quality of Life-Standards, a Target Identification Collaboration**

Benedikt SCHOSER, Libby WOOD, Hans KNOOP, Jeffrey GLENNON, Arend HEERSCHAP, Tom HESKES, Michael TRENELL, Guillaume BASSEZ, Darren MONCKTON, Shaun TREWEEK, Hanns LOCHMÜLLER, Baziel VAN ENGELEN

Ludwig Maximilians University Munich - Munich, University Of Newcastle - Newcastle Upon Tyne, Radbound University Medical Centre - Nijmegen, Organisation: Radbound University Medical Centre - Nijmegen, L'Assistance Publique ♦ Hôpitaux De Paris - Paris, University Of Glasgow - Glasgow, University Of Dundee - Dundee

14:00-14:30 S.8.1 Overview Lecture: Basic science highlights
Chairpersons: Tetsuo ASHIZAWA - Gainesville, Mari Carmen ALVAREZ - Valencia

14:00-14:30 **S8.1 Basic Science Highlights**

Darren MONCKTON

University Of Glasgow - Glasgow

14:30-15:00 **S.8.2 Overview Lecture: Clinical science highlights**

Chairpersons: Tetsuo ASHIZAWA - Gainesville, Mari Carmen ALVAREZ - Valencia

14:30-15:00 **S8.2 Clinical Science Highlights**

Mark ROGERS

All Wales Medical Genetics Service - Cardiff

15:00-15:30 **S.8.3 Overview Lecture: Theraphy: current and future**

Chairpersons: Tetsuo ASHIZAWA - Gainesville, Mari Carmen ALVAREZ - Valencia

15:00-15:30 **S8.3 Theraphy: Current and Future**

Bruce WENTWORTH

Genzyme, A Sanofi Company - Framingham

16:45 **Social and Organizational Issues**

Chairpersons: Maurice SWANSON - Gainesville, Jon Andoni URTIZBEREA - Hendaye