IDMC-9

INTERNATIONAL MYOTONIC DYSTROPHY CONSORTIUM MEETING

16-19 OCTOBER 2013 KURSAAL CENTER **DONOSTIA - SAN SEBASTIAN** SPAIN

Program & Contributions

www.idmc-9.org

Index

Title:
IDMC-9. INTERNATIONAL MYOTONIC DYSTROPHY CONSORTIUM MEETING

Edited by:
BioDonostia and Ilundain Fundazioa

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Lankor, S.L.

D.L.: SS 1349-2013

Welcome to IDMC-9

Welcome to the **9th International Myotonic Dystrophy Consortium (IDMC-9) Meeting** which will be held in San Sebastián, Basque Country, Spain in October 2013. The IDMC is an informal group of clinicians and research scientists who have a common interest in understanding the clinical and molecular events leading to myotonic dystrophy and to develop effective therapeutic strategies for this disease.

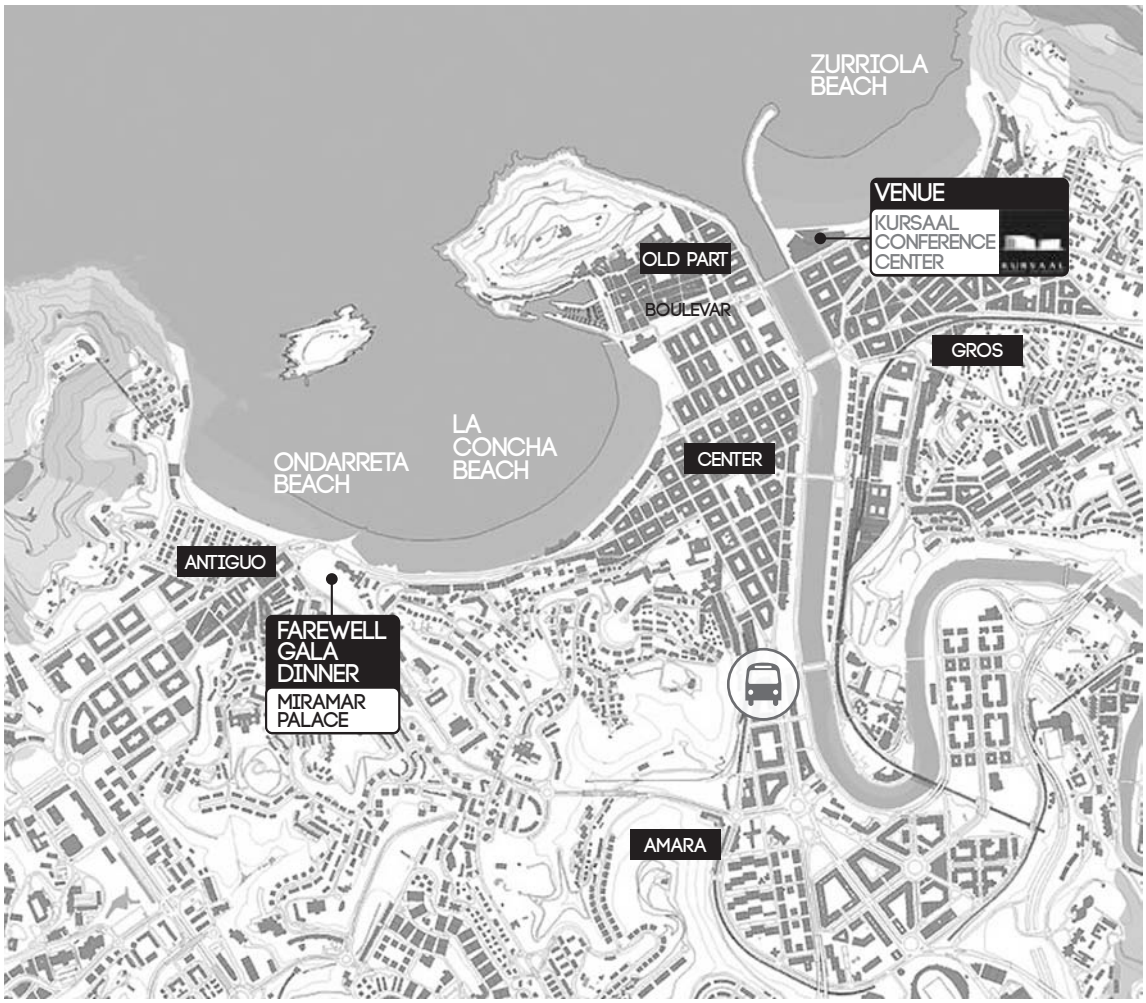
The **IDMC-9** meeting will be a great opportunity to provide a forum for the latest scientific developments on the disease with a special emphasis in Pathogenesis and Treatments. We are living a challenging time for the myotonic dystrophy research around the world. Thus, we are confident that 2013, declared in Spain as the Year for Rare Diseases, will contribute to launch this international event, and hopefully will be remembered as a special milestone in the battle against myotonic dystrophy.

Looking forward for seeing you in San Sebastian in 2013.

Adolfo LÓPEZ DE MUNAIN MD PhD
Co-Chairmen
IDMC-9 Organizing Committee

Jon Andoni URTIZBEREA MD
Co-Chairmen
IDMC-9 Organizing Committee

Map of the city



VENUE	FAREWELL GALA DINNER
Kursaal Conference Center Avenida de Zurriola, 1	Miramar Palace Paseo de Miraconcha, 48

How to move around the city

San Sebastián is a small and friendly city, divided into little areas called **Gros** (Kursaal Center, Zurriola beach...), **Parte Vieja** (Old part), el **Antiguo** (Ondarreta beach, University...), el **Centro** (city centre: La Concha, the Cathedral...), **Amara**, as an example.

Main areas of the city can be walked or cycled easily by bike lanes, but if you want to, you can take city buses.

BUSES

It is recommended to take the Boulevard as the main stop to get to Kursaal, since most of the bus stops are located there.

Tickets are 1,60€ per ride and they are taken directly from the driver; otherwise you can buy the "San Sebastián Card 5" at the Tourist Office (Boulervard, 8) for 15€: 12 trips, valid for 5 days.
Bus stops-hotels:

Hotel NH Aranzazu
At the back of the main entrance.
Bus numbers 5 (every 6-8 min.) and 25 (every 20 min.).
Stop at the Boulevard. Duration: 14 minutes.

Hotel Codina
At the same side of the main entrance.
Bus numbers 5 (every 6-8 min.) and 25 (every 20 min.).
Stop at the Boulevard. Duration: 11 minutes.

Hotel Monte Ulia
At the same side of the main entrance.
Bus number 13 (every 6-10 min.) and 24 (every 30 min.).
Stop at Zurriola beach (same street as Kursaal is). Duration: 7 minutes.

Hotel Palacio de Aiete
At the back of the main entrance.
Bus numbers 19 and G1 (every 30 min.).
Stop bus number 19, Urbieta 6. Stop bus number G1, Okendo, Duration: 20 minutes.

Hotel Astoria
In front of the main entrance.
Bus number 28 (every 5-8 min.).
Stop at the Boulevard. Duration: 10 minutes.

+info: www.dbus.es/en

TAXIS

Taxis do not pick up passengers in the street. You should go to a taxi stop (Boulevard) or call to a radio taxi Company.

Vallina: +34.943 40 40 40 | Taxi Donosti +34.943 46 46

BIKES

The city is perfectly suited for cycling with more than 30 km of bike lanes painted red on the streets. Local name for bike lanes is "bide-gorri", that literally means red path. You can pick a free map at the Tourist Office (Boulevard, 8) with the paths marked in red and some addresses for bike rental companies.

Committees

Organizing Committee

Chairs

Adolfo LÓPEZ DE MUNAIN MD PhD
Head of Neuroscience Area, Institute Biodonostia, San Sebastián, Spain; Neuroscience Department. University of Basque Country, San Sebastián, Spain.

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Neuromuscular Unit, Hôpital Marin, Hendaye-Hôspitaux de Paris, France.

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Members

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Ana AIASTUI PhD
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Administrative Office

Leyre CURTO ARZAC
Ilundain Foundation for Research in Neuromuscular Disorders, San Sebastián, Spain.

Local Scientific Committee

Chairs

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Neuromuscular Unit, Hôpital Marin, Hendaye-Hôspitaux de Paris, France.

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Neuromuscular Disorders Unit. Neurology Department. Hospital Donostia, San Sebastián, Spain.

Prof. Rubén ARTERO
Department of Genetics, University of Valencia, Valencia, Spain.

Prof. Manuel PÉREZ-ALONSO
Head of Department of Genetics, University of Valencia, Valencia, Spain.

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Committees

International Scientific Committee

Members

Virginia ARECHAVALA-GOMEZA
Biocruces Health Research Institute, Barakaldo, Spain.

Tetsuo ASHIZAWA
University of Florida, United States.

David BROOK
University of Nottingham, United Kingdom.

Claude BOURLIER
DM Support group, France.

Margaret BOWLER
DM Support group, United Kingdom.

John DAY
University of Minnesota, United States.

Bruno EYMARD
Université Pierre et Marie Curie, Paris, France.

Geneviève GOURDON
Université Paris-Descartes, Paris, France.

Tiemo GRIMM
University of Würzburg, Germany.

Shoichi ISHIURA
University of Tokyo, Japan.

Ralf KRAHE
University of Texas, Houston, United States.

Shannon M. LORD (†)
DM Foundation.

Don MACKENZIE
Marigold Foundation.

Mani MAHADEVAN
University of Virginia, United States.

Giovanni MEOLA
University of Milan, Italy.

Darren MONCKTON
University of Glasgow, United Kingdom.

Richard MOXLEY
University of Rochester, United States.

Nakaaki OHSAWA
University of Tokyo, Japan.

Christopher PEARSON
University of Toronto, Canada.

Jack PUYMIRAT
Université Laval, Quebec, Canada.

Laura RANUM
University of Minnesota, Minneapolis, United States.

Mark ROGERS
University of Wales, United Kingdom.

Benedikt SCHOSER
University of Munich, Germany.

Nicolas SERGEANT
Université de Lille, Lille, France.

Maurice SWANSON
University of Florida, United States.

Charles THORNTON
University of Rochester, United States.

Bjarne UDD
University of Helsinki, Finland.

Berend WIERINGA
University of Nijmegen, The Netherlands.

Organized by

BioDonostia
Instituto de Investigación Sanitaria

ILUNDAIN
fundazioa

Collaborators

AFM TÉLÉTHON
CURE THROUGH INNOVATION

Center For Neuro Genetics UF

The Marigold Foundation
Creating Hope & Opportunity

MDA Muscular Dystrophy Association
Fighting Muscle Disease

MYOTONIC DYSTROPHY FOUNDATION

MD Myotonic Dystrophy SUPPORT GROUP

SENC

genzyme
A SANOFI COMPANY

EUSKO JAURLARITZA
GOBIERNO VASCO
HEZKUNTZA, HIZKUNTZA POLITIKA ETA KULTURA SAILA
DEPARTAMENTO DE EDUCACIÓN, POLÍTICA LINGÜÍSTICA Y CULTURA

Gipuzkoako Foru Aldundia
Berrikuntzako, Landa Garapeneko eta Turismoko Departamentua
Departamento de Innovación, Desarrollo Rural y Turismo

Donostia San Sebastián
TOURISM & CONVENTION BUREAU

KURSAAL
DONOSTIA - SAN SEBASTIAN

Technical Secretariat

Lankor
kongresu eta ekitaldiak
congresos y eventos

Declaration of Scientific and Health Care Interest Basque Government. Department of Health

EUSKO JAURLARITZA
OSASUN SAILA
OsasunSailburuordetza
Plangintza, Antolamendu eta Ebaluazio Sanitarioko Zuzendaritza

GOBIERNO VASCO
DEPARTAMENTO DE SALUD
Viceconsejería de Salud
Dirección dePlanificación, Ordenación y Evaluación Sanitaria

EKINTZA ZIENTIFIKOETARAKO INTERES SANITARIOAREN ONARPENA
RECONOCIMIENTO DE INTERÉS SANITARIO PARA ACTOS CIENTÍFICOS

Eusko Jaurlaritzako Ezagutzaren Gestioko eta Ebaluazioko Zuzendaritzak, 2006ko maiatzaren 16ko Aginduan, hots, Euskal Herriko Autonomia Erkidegoan izango diren ekintza zientifikoak osasunerako interesgarritzat jotzea arauten duen Aginduan, erabakitakoari jarraituz eta bertan ezarritako baldintzak bete egiten direla kontuan izanik, "***9 INTERNATIONAL MYOTONIC DISTROPHY CONSORTIUM***" izeneko zientzi bilkura interesgarritzat jotzen du. Izan ere, "***Instituto de Investigación Sanitaria Biodonostia***" izeneko elkarteak antolatu du eta Donostian 2013ko urriaren 16tik 19era izango da.

La Dirección de Gestión del Conocimiento y Evaluación del Departamento de Sanidad y Consumo del Gobierno Vasco, en base a lo dispuesto en la Orden de 16 de mayo de 2006 por la que se regula el reconocimiento de interés sanitario para actos de carácter científico que tengan lugar en la Comunidad Autónoma del País Vasco, y considerando que cumple los requisitos establecidos, concede este Reconocimiento al acto "***9 INTERNATIONAL MYOTONIC DISTROPHY CONSORTIUM***" organizado por "***Instituto de Investigación Sanitaria Biodonostia***" a celebrar en Donostia-San Sebastian los dias 16 al 19 de octubre de 2013.

Vitoria-Gasteiz, 2013ko irailaren 13.

Mª Luisa Arteagoitia Gonzalez
PLANGINTZA, ANTOLAMENDU ETA EBALUAZIO SANITARIOKO ZUZENDARIA
DIRECTORA DE PLANIFICACIÓN, ORDENACIÓN Y EVALUACIÓN SANITARIA

EUSKO JAURLARITZA
GOBIERNO VASCO
DEPARTAMENTO DE SALUD

Invited Speakers

Keynote Lectures Speakers

Ángel RAYA
Institute for Bioengineering
Barcelona, SPAIN

Bé WIERINGA
Nijmegen Centre for Molecular Life Sciences
Nijmegen, THE NETHERLANDS

Overview Lectures Speakers

Rubén ARTERO
University of Valencia
Burjasot - SPAIN

Thomas COOPER
Baylor College of Medicine
Houston - UNITED STATES

Bruno EYMARD
Institut de Myologie Pitié Salpêtrière
Paris - FRANCE

Cynthia GAGNON
Université de Sherbrooke / Grimn
Jonquière - CANADA

Genevieve GOURDON
INSERM U781, Institut Imagine
Paris - FRANCE

Charles THORNTON
University of Rochester Medical Center
Rochester - UNITED STATES

Chairs

Mari Carmen ÁLVAREZ
Valentia Biopharma,
Valencia, Spain.

Virginia ARECHAVALA-GOMEZA
Biocruces Health Research Institute,
Barakaldo, Spain.

Tetsuo ASHIZAWA
University of Florida,
Gainesville, United States.

Franck C BENNETT
Isis Pharmaceuticals,
Carlsbad, United States.

David BROOK
University of Nottingham,
Nottingham, United Kingdom.

Nicolas CHARLET
Institut de Génétique et de Biologie Moléculaire et Cellulaire, Illkirch - France.

Denis FURLING
UPMC UM76, Institut Myologie,
Paris - France.

Cynthia GAGNON
Université de Sherbrooke / Grimn,
Jonquière, Canada.

Gerardo GUTIÉRREZ
Hospital Universitario Infanta Sofía,
San Sebastián de los Reyes, Spain.

Chad HEATWOLE
University of Rochester,
Rochester, United States.

Shoichi ISHIURA
University of Tokyo,
Tokyo, Japan.

Ralf KRAHE
University of Texas MD Anderson Cancer Center, Houston, United States.

Mani S. MAHADEVAN
University of Virginia,
Charlottesville, United States.

Giovanni MEOLA
University of Milan - IRCCS Policlinico San Donato, San Donato, Italy.

Richard-Thomas MOXLEY, III
University of Rochester Medical Center,
Rochester, United States.

Christopher PEARSON
University of Toronto,
Toronto, Canada.

Manuel PÉREZ-ALONSO
Universitat de Valencia,
Valencia, Spain.

Laura RANUM
University of Florida Center for Neurogenetics,
Gainesville, United States.

Benedikt SCHOSER
Friedrich-Baur-Institute, LMU,
Munich, Germany.

Nicolas SERGEANT
INSERM UMR 837, Alzheimer & Tauopathies,
Lille, France.

Lubov TIMCHENKO
Baylor College of Medicine,
Houston, United States.

Eric WANG
Massachusetts Institute of Technology,
Boston, United States.

Derick G. WANSINK
NCMLS, Dept. Cell Biology,
Nijmegen, The Netherlands.

General Information

Meeting Venue:
Palacio de Congresos y Auditorio Kursaal
Avenida de Zurriola, 1
20002 Donostia-San Sebastián
Tel: (+34) 943-003000

Registration:
The Registration desk is located outside of the Chamber Hall. The Registration desk will open Wednesday, October 16, at 15:00. The desk will remain open for the duration of the conference (excluding lunch time).

Oral Presentations:
Each speaker will have a 10 minutes period for slide presentations with an additional five minutes for discussion. The program is packed and the session chairs will stick rigorously to the schedule. Speakers will have an opportunity to review their presentations in the **Speaker Room**.

Posters:
Posters should be attached on their boards on Wednesday, October 16, from 6.00 to 8.30PM or Thursday, October 17, from 8.00 to 10.00AM and removed on Friday, October 18, by 7.30PM (Basque Hall; Floor -1).

Exhibition Desks:
Opened during Conference opening hours, at Kursaal Center (outside of the Chamber Hall), from Wednesday, October 16, to Saturday, October 19.

Meal Functions:
Welcome cocktail, lunches, coffee-breaks and Farewell dinner are included in Scientists' and Young Scientists' fee.

Association, patient and private individual fee includes welcome cocktail, lunches and coffee-breaks.

Please, refer to the agenda for the various locations.

If anyone has any special need (dietary) should inform the Secretariat about their preferences, if they had not done it previously. They will find a card in their identification badge to be placed at the table at dinner time.

Social events:
Welcome cocktail: it will be held on Wednesday, October 16, at 7.45 PM in Kursaal (Banquet Hall).

Sightseeing walking tour around San Sebastián: Thursday, October 17 at 6.00 PM (depart from Kursaal Center). Duration expected: 2.5 hours.

Farewell dinner: it will take place on Saturday, October 19, at 8.00 PM in Miramar Palace (no transportation arranged).

Language:
The official language of the IDMC-9 is English.

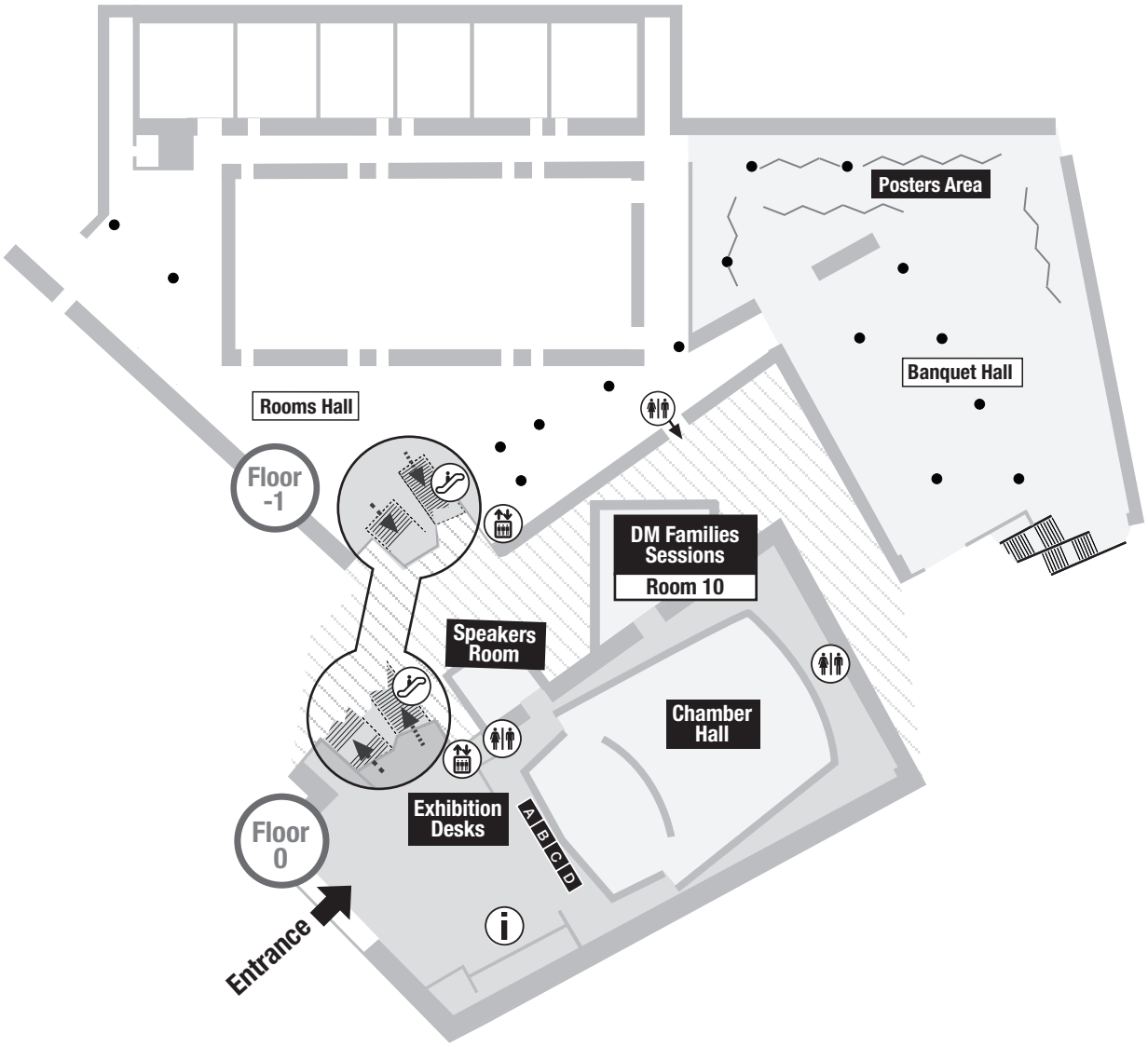
Internet:
Free Wi-Fi access will be available at Kursaal Center.
User: idmc Password: 2013

Badge:
All participants in the Conference must wear the badges visible at all times, to access Kursaal Center and Social events.

Satellite Meetings:
Wednesday, October 16, 2013, 08.00 AM-04.00 PM
Outcome Measures for Myotonic Dystrophy Organizer: Cynthia Gagnon

Map of the main venue

Kursaal Conference Centre



LEGEND		ASSOCIATIONS	
ⓘ	Secretariat	A	AFM
🚻	Toilet	B	MDSG
	Elevator	C	MDF
	Escalator	D	MDA

Social Activities

Wednesday, Oct. 16
7:45PM

Welcome Reception
Kursaal Conference Center

No transportation arranged

The Welcome Reception will take place at the Chamber Hall in Kursaal Conference Center.

A cocktail will be served after the event and Kandakidetza Choir performance (official choir of OSAKIDETZA, Basque Health Service) at the Banquet Hall (Floor -1).

Thursday, Oct. 17
6.00 - 8.30PM

Sightseeing Tour
San Sebastián

The groups will departure from Kursaal

Sightseeing Tour around San Sebastián admiring the breathtaking views from the Concha bay, the Romantic Area, Old Town and so on.

Saturday, Oct. 19
8.30 - 11.30PM

Farewell Dinner
Miramar Palace*

No transportation arranged

At the end of the 19th century, the regent Queen María Cristina ordered to build this palace in order to accommodate the Court during summer. The location of the royal palace could not be better: on the hillock ending the Loreto pick, which separates La Concha and Ondarreta beaches and offering splendid views over the bay and the city.

Farewell dinner, livened up by “Jazzte Borrazte Band”.

* Address: Paseo de Miraconcha, 48. 20007 Donostia-San Sebastián

Program at a glance

Wednesday, October 16

15.00 - 17.00	Registration on site of IDMC-9
17.00 - 17.30	Opening ceremony
17.10 - 17.30	Welcome address **Jon Andoni URTIZBEREA**, Paris-Hendaye **Adolfo LÓPEZ DE MUNAIN**, San Sebastián
17.30 - 19.30	**Session 1 - Keynote lectures** Chairs: Eric WANG, Boston and Genevieve GOURDON, París
17.30 - 18.45	**S1.1 Molecular/cellular mechanisms in repeat expansion disorders** Bé WIERINGA, Nijmegen Centre for Molecular Life Sciences - Nijmegen, The Netherlands
18.45 - 19.30	**S1.2 IPS cells in biology an disease** Angel RAYA, Institute for Bioengineering - Catalonia, Spain
19.30 - 21.00	**Musical entertainment** Kantakidetza Choir Welcome cocktail

Program at a glance

Thursday, October 17

08.00 - 12.00	**Session 2 - Disease mechanisms** Chairs: Nicolas CHARLET-BERGUERAND, Paris and Christopher PEARSON, Toronto
08.00 - 08.30	**S2.1 Overview lecture** **RNA mediated toxicity in DM1** Rubén 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** **The multi-facets of DM** 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 - 20.30	Sightseeing tour in San Sebastián

Program at a glance
Friday, October 18

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** **New insights into adult and childhood DM1 phenotype, cardio-respiratory prognostic considerations, and outcome measures** 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 - San Sebastián de los Reyes, Chad 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, Barakaldo 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, United States
15.30 - 17.00	**S5.2 Platform presentations**
17.00 - 17.30	Coffee break
17.30 - 19.30	**S5.3 Platform presentations**

Program at a glance

Saturday, October 19

08.00 - 10.00	**Session 6 – Therapeutic trials** Chairs: Frank BENNET, Carlsbad and Virginia ARECHAVALA-GOMEZA, Barakaldo
08.00 - 08.30	**S6.1 Overview lecture** **Trial readiness and implementation** Charles THORNTON, Rochester, United States
08.30 - 10.00	**S6.2 Platform presentations**
10.00 - 10.30	Coffee break
10.30 - 12.00	**Session 7 – Ethical, legal and social issues** Chairs: Tee ASHIZAWA, Gainesville and Chad HEATWOLE, Rochester
10.30 - 11.00	**S7.1 Overview lecture** **Myotonic dystrophy type 1 as a model of organized complexity** Cynthia GAGNON, Jonquière
11.00 - 12.00	**S7.2 Platform presentations**
12.00 - 14.00	Lunch at the Kursaal

Program at a glance

Saturday, October 19

DM PATIENTS AND FAMILIES SESSION

14.00 - 16.30	**Session 8- Interactive session with patients & family groups** Chairs: Tee ASHIZAWA, Gainsville, FL, United States and Mª Carmen ÁLVAREZ, Valencia, Spain
14.00 - 14.30	**S8.1 Basic science highlights** Darren MONCKTON, Glasgow, United Kingdom
14.30 - 15.00	**S8.2 Clinical science lighlights** Mark ROGERS, Wales, United Kingdom
15.00 - 15.30	**S8.3 Therapy: current and future** Bruce WENTWORTH, Framingham, United States
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.8.4.3 Other Associations (tentatively French, Italian and German speaking associates)
16.45 - 18.40	**Session 9- Social and organizational issues** Chairs: Jon Andoni URTIZBEREA, Hendaya, France and Maury SWANSON, Gainesville, FL, United States
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 - 18.40	**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 Annoncement for the next IDMC Meeting location End of conference
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

Full Program
Thursday, October 17

08.00 - 12.00	**Session 2 - Disease mechanisms** Chairs: Nicolas CHARLET-BERGUERAND, Paris and Christopher PEARSON, Toronto
08.00 - 08.30	**S2.1 Overview lecture**
	RNA mediated toxicity in DM1 **Rubén ARTERO** Fundación de Investigación Sanitaria INCLIVA. Departamento de Genética, Universidad de Valencia - Burjasot (Valencia)
08.30 - 10.00	**S2.2 Platform presentations**
08:30 - 08:45	O-01 **Expanded (CUG)n RNA Expression as a Variable in DM1 Patients and Disease Models** **Anke GUDDE et al.** UMC St. Radboud - Nijmegen, Netherlands
08:45 - 09:00	O-02 **Correlation Between Muscle Histopathology Impairment and Spliceopathy in Myotonic Dystrophies** **Rosanna CARDANI et al.** IRCCS-Policlinico Sandonato, Milan, Italy
09:00 - 09:15	O-03 **CELF1 regulates SERCA1 splicing via PMA pathway** **Yimeng ZHAO et al.** The University of Tokyo - Tokyo, Japan
09:15 - 09:30	O-04 **Modifications to Toxic CUG RNAs Induce Structural Stability and Rescue mis-splicing in Myotonic Dystrophy** **Elaine DELORIMIER et al.** University of Oregon - Eugene, United States
09:30 - 09:45	O-05 **Nkx2-5, a Modifier and Marker of RNA Toxicity in Skeletal Muscle in Myotonic Dystrophy** **Ramesh S YADAVA et al.** University of Virginia - Charlottesville, United States
09:45 - 10:00	O-06 **Minigene Tau E1-E13 : a New Tool to Develop DM1 Transgenic Mice with an Associated Tauopathy** **Nicolas SERGEANT et al.** INSERM UMR 837, Alzheimer & Tauopathies - Lille, France
10.00 - 10.30	Coffee break and poster viewing
10.30 - 12.00	**S2.3 Platform presentations**
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 et al.** Instituto de Investigaciones en Salud, Universidad de Costa Rica - San José, Costa Rica
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 et al.** University of Glasgow - Glasgow, United Kingdom
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 et al.** University of Glasgow - Glasgow, United Kingdom
11:15 - 11:30	O-10 **Aberrant Methylation Spreading in DM1-affected Human Embryonic Stem Cells** **Rachel EIGES et al.** Shaare Zedek Medical Center - Jerusalem, Israel
11:30 - 11:45	O-11 **Ran Proteins From Intronic Cctg Expansions in DM2 Patient Brains** **Tao ZU et al.** University of Florida - Gainesville, United States
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 et al.** University of Glasgow - Glasgow, United Kingdom

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12.00 - 14.00	Lunch **P1 - Poster viewing with presenters**
	P1.1 Disease Mechanisms Chairpersons: Manuel PÉREZ ALONSO - Valencia, Lubov TIMCHENKO - Houston
P-001	**Alternative Splicing Alterations of Ca^2+ Handling Genes are Associated with Ca^2+ Signal Dysregulation in DM1 and DM2 Myotubes** **Massimo SANTORO et al.** Fondazione Don Gnocchi - Milan, Italy
P-002	**Antioxidative Enzymes Activities in Patients with Myotonic Dystrophy Type 1** **Vidosava RAKOCEVIC STOJANOVIC et al.** Neurology Clinic, Clinical Center of Serbia, School of Medicine, University of Belgrade - Belgrade, Serbia and Montenegro
P-003	**Epiretinal Membrane: A Treatable Cause of Visual Disability in Myotonic Dystrophy Type 1** **Richard ROXBURGH et al.** Auckland City Hospital - Auckland, New Zealand
P-004	**Aberrant Splicing of Annexin Vii by PTB Impairs Muscle Cell Differentiation in Myotonic Dystrophy Type 1** **Richard PELLETIER et al.** CHU Quebec - Quebec, Canada
P-005	**Rbfox1 Cooperates with MBNL1 to Regulate Muscle-specific Splicing Events Altered in Myotonic Dystrophy** **Roscoe KLINCK et al.** University of Sherbrooke - Sherbrooke, Canada
P-006	**Sodium Channel Gene Mutation: a Possible Role in Myotonic Dystrophy Type 2 with Severe Myotonia** **Enrico BUGIARDINI et al.** IRCCS Policlinico San Donato- University of Milan - San Donato Milanese, Italy
P-007	**DMPK, Hexokinase Ii and SRC Form a Multimeric Complex on the Mitochondrial Outer Membrane, That Prevents Ros-induced Apoptosis** **Sergio SALVATORI et al.** University of Padova - Padova, Italy
P-008	**Gangliosides Composition in Patients Affected by Myotonic Dystrophy Type 1 and Type 2** **Rea VALAPERTA et al.** IRCCS Policlinico San Donato - Milan, Italy
P-009	**Roles of MBLN1 and/or MBLN2 on Tau Exon 2 Splicing in Physiological and Myotonic Dystrophy** **Céline CARPENTIER et al.** INSERM U837 Alzheimer & Tauopathies - Lille, France
P-010	**Regulation of Staufen1 during skeletal muscle development, differentiation, and regeneration** **Aymeric RAVEL-CHAPUIS et al.** University of Ottawa - Ottawa, Canada
P-011	**Re-expression of PKM2 in Type 1 Myofibers Correlates with Altered Glucose Metabolism in Myotonic Dystrophy** **Zhihua GAO et al.** Baylor College of Medicine - Houston, United States
P-012	**Early Senescence of DM1 Myoblasts is Mediated by a Cugexp-rna/ros Pathway** **Coralie SAINT-JEAN et al.** Institute of Myology - Paris, France
P-013	**Splicing of ABLIM1 is Aberrant in DM1 Patients** **Natsumi OHSAWA et al.** University of Tokyo - Tokyo, Japan
P-014	**Staufen1 as a Splicing Modulator: Implications for DM1 Therapy?** **Emma BONDY-CHORNEY et al.** University of Ottawa - Ottawa, Canada
P-015	**Characterization of Complex Variant Expanded DMPK Alleles in Italian Patients with Myotonic Dystrophy Type 1** **Giulia ROSSI et al.** Tor Vergata University of Rome - Roma, Italy
P-016	**Biomolecules Modeling Implied in Myotonic Dystrophy Type 1 for Structure Based Drug Design** **Alex LOPEZ et al.** Institut Quimic de Sarria - Ramon Llull University - Barcelona, Spain

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P-017	**Antagonism Between MBNL and CELF Proteins in the Nucleus and Cytoplasm** Eric WANG et al. Massachusetts Institute of Technology - Boston, United States
P-018	**CUG RNA Suppresses PGC-1alpha Activity and Perturbs Metabolic Homeostasis Via Activating DNa Damage Response ATM Signaling in DM1** Xiang FANG et al. University of Texas Medical Branch - Galveston, United States
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 et al. University of Texas Medical Branch - Galveston, United States
	P1.2 Tissue specific disease and cell/animal models Chairpersons: Mani S. MAHADEVAN - Charlottesville, United States, Nicolas SERGEANT - Lille
P-020	**Oligodendroglioma in Patient with Myotonic Dystrophy Type 1** Vidosava RAKOCEVIC STOJANOVIC et al. Neurology Clinic, Clinical Center of Serbia, School of Medicine, University of Belgrade - Belgrade, Serbia and Montenegro
P-021	**Synaptic Abnormalities in the Brain of DMSXL Mice are Not Associated with Neurodevelopmental Deficits or Structural Abnormalities** Oscar HERNANDEZ-HERNANDEZ et al. Instituto Nacional de Rehabilitación - Mexico City, Mexico
P-022	**MSH3 Overexpression in Pancreas Destabilizes CTG Triplet Repeat in DM1 Mice Carrying 55 or ~500 CTG Repeats** Stéphanie TOMÉ et al. INSERM U781 - Paris, France
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 et al. Universidad de Costa Rica - San José, Costa Rica
P-024	**Dynamic Changes of Intranuclear Foci in Proliferating and Non-proliferating DM1 Neural Cells** Guangbin XIA et al. University of Florida - Gainiesville, United States
P-025	**Neonatal DMSXL mice carrying >1000CTG reveal severe molecular and morphological alterations in the diaphragm** Fadia MEDJA et al. INSERM U781 - Paris, France
P-026	**The RNA-binding Protein Staufen1 Inhibits Myogenic Differentiation Via a c-Myc Dependent Pathway** Tara Ellen CRAWFORD et al. Department of Cellular And Molecular Medicine, Faculty of Medicine, University of Ottawa - Ottawa, Canada
P-027	**RAB3A and SYN1 Protein Abnormalities in DM1 Brain are Not Mediated by Missplicing or Developmental Delays** Diana Mihaela DINCA et al. INSERM U781 - Paris, France
P-028	**Combining MBNL1 Loss and Gain of CELF1 to Model DM1 in Mice** Ginny MORRISS et al. Baylor College of Medicine - Houston, United States
P-029	**Expression profile of RNA-binding proteins in several DM1 mouse muscles** Aymeric RAVEL-CHAPUIS et al. University of Ottawa - Ottawa, Canada
P-030	**Characterization of MBNL1 Dependent Splicing Responses in a Tissue Culture DM Model** Stacey WAGNER et al. University of Oregon - Eugene, United States
P-031	**MBNL Compound Loss-of-function Models for Myotonic Dystrophy** Moyi LI et al. University of Florida - Gainesville, United States
P-032	**Increased Phosphorylation of eIF2alpha and Reduced Levels of CDK2 Contribute to Premature Growth Arrest in DM1 Lens Cells.** Jeremy RHODES et al. University of East Anglia - Norwich, United Kingdom

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14.00 - 18.00	**Session 3 - Tissue specific mechanisms** Chairpersons: Shoichi ISHIURA - Tokyo, Derick G. WANSINK - Nijmegen
14.00 - 14.30	**S3.1 Overview lecture**
	The multi-facets of DM **Genevieve GOURDON et al.** INSERM U781, Institut Imagine - Paris, France
14.30 - 16.00	**S3.2 Platform presentations**
14.30 - 14.45	**O-13 SCN5A Splicing Alteration in Heart of Myotonic Dystrophy Patients** **Fernande FREYERMUTH et al.** IGBMC - Illkirch, France
14.45 - 15.00	**O-14 Implication of BIN1 in Myotonic Dystrophy** **Michel NEY et al.** IGBMC - Illkirch, France
15.00 - 15.15	**O-15 Early Abnormalities in the DMSXL Mouse Model for DM1: a Model of the Congenital Form?** **Lise MICHEL et al.** INSERM U781 - Paris, France
15.15 - 15.30	**O-16 Phenotype-genotype Correlation in Congenital Myotonic Dystrophy** **Masayuki NAKAMORI et al.** Osaka University - Osaka, Japan
15.30 - 15.45	**O-17 Altered Splicing of Cardiac Sodium Channel Might Be Responsible for Cardiac Conduction Defects in Myotonic Dystrophy** **Yosuke KOKUNAI et al.** Osaka University Graduate School of Medicine - Suita, Japan
15.45 - 16.00	**O-18 The distribution of splicing defects in the Myotonic Dystrophy Type 1 brain** **Takashi KIMURA et al.** Hyogo College of Medicine - Nishinomiya, Japan
16.00 - 16.30	Coffee break and poster viewing
16.30 - 18.00	**S3.3 Platform presentations**
16.30 - 16.45	**O-19 Genome Wide Identification of Aberrant Alternative Splicing Events in Myotonic Dystrophy Type 2** **Alessandra PERFETTI et al.** IRCCS-policlinico San Donato - San Donato Milanese, Italy
16.45 - 17.00	**O-20 Contribution of Dystrophin Alternative Mis-splicing to DM Muscle Dysfunction** **Frédérique RAU et al.** UPMC Univ Paris 06, UM 76, Institut de Myologie and INSERM, U974 and CNRS, UMR7215 - Paris, France
17.00 - 17.15	**O-21 Two Enhancers Control Expression of Drosophila Muscleblind in the Somatic Musculature and in the Nervous System** **Ariadna BARGIELA et al.** Universidad de Valencia - Valencia, Spain
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** **Francisco Jose FERNANDEZ-GOMEZ et al.** INSERM U837 - Lille, France
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 et al.** Universidad de Costa Rica - San José, Costa Rica
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 et al.** Baylor College of Medicine - Houston, United States

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08.00 - 12.00	**Session 4 - Clinical measures and biomarkers** Chairpersons: Giovanni MEOLA - San Donato Milanese, Richard-Thomas MOXLEY, III - Rochester, Ny
08.00 - 08.30	**S4.1 Overview lecture:**
	New insights into adult and childhood DM1 phenotype, cardio-respiratory prognostic considerations, and outcome measure **Bruno EYMARD et al.** Institut de Myologie Pitié Salpêtrière - Paris, France
08.30 - 10.00	**S4.2 Platform presentations**
08.30 - 08.45	O-25 **Histological and Magnetic Resonance Imaging Findings of Vastus Lateralis in Myotonic Dystrophies: Do they Match Up?** **Carla MERKEL et al.** Department of Neurology - Bonn, Germany
08.45 - 09.00	O-26 **Myotonic Dystrophies - Disorders of the Central Nervous System** **Vidosava RAKOCEVIC STOJANOVIC et al.** Neurology Clinic, Clinical Center of Serbia, School of Medicine, University of Belgrade - Belgrade, Serbia And Montenegro
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 et al.** University Laval - Quebec, Canada
09.15 - 09.30	O-28 **Increased Brain Biomarker Plasmatic Concentration in Myotonic Dystrophy Type I** **Susanna SCHRAEN-MASCHKE et al.** INSERM UMR 837, Alzheimer & Tauopathies - Lille, France
09.30 - 09.45	O-29 **Seeking for Convenient Outcome Measures for the Characterization of the Neuromuscular Function in Dm1 Patients** **Jean-Yves HOGREL et al.** Institut de Myologie - Paris, France
09.45 - 10.00	O-30 **Which Myotonic Dystrophy Type 1 Adult Patients Require Cardiology Referral?** **William GROH et al.** Indiana University School of Medicine - Indianapolis, United States
10.00 - 10.30	Coffee break and poster viewing
10.30 - 12.00	**S4.3 Platform presentations**
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 et al.** Université de Sherbrooke / Grimn - Jonquière, Canada
10.45 - 11.00	O-32 **Effect of mild and adult DM1 phenotype on neuropsychological functioning** **Louis RICHER et al.** Université du Québec à Chicoutimi - Chicoutimi, Canada
11.00 - 11.15	O-33 **Cardiometabolic Risk Factors in Patients with Myotonic Dystrophy Type 1** **Patricia BLACKBURN et al.** Université du Québec à Chicoutimi - Saguenay, Canada
11.15 - 11.30	O-34 **ACTN3 Genotypes and Muscular Performance in DM1 Patients: Preliminary Results** **Mario LEONE et al.** University of Quebec in Chicoutimi - Saguenay, Canada
11.30 - 11.45	O-35 **A Clinical Study of Myotonic Dystrophy Type-1 (DM1) Patients' Perception and Prioritization of Cognitive Symptoms** **Chad HEATWOLE et al.** University of Rochester - Rochester, United States
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 et al.** University of Rome "La Sapienza" - Roma, Italy

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12.00 - 15.00	Lunch **P2 - Poster viewing with presenters**
	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 et al. National Hospital Organization Toneyama National Hospital - Toyonaka, Japan
P-034	**Obesitas Surgery in DM1 - a Case Report** Karin HAKANSSON Sahlgrenska University Hospital - Gothenburg, Sweden
P-035	**Dental Health Condition is Closely Associated with Upper Limb Motor Function in Myotonic Dystrophy Type 1** Hiroto TAKADA et al. Aomori Hospital, Nho - Aomori, Japan
P-036	**Polyneuropathy and Myotonic Dystophy Type1: Primary Involvement of Nerves or Incidental Coexistence?** Marcella MASCIULLO et al. Catholic University of Sacred Heart - Rome, Italy
P-037	**Daytime Sleepiness and Fatigue Symptoms in Myotonic Dystrophy Patients: a 9-year Longitudinal Study** Luc LABERGE et al. ÉCOBES - Recherche et Transfert - Jonquière, Canada
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 et al. Laval University - Québec, Canada
P-039	**Experienced Fatigue in Myotonic Dystrophy Type 1 (DM1) is Associated with Muscle Impairment and Depression** Stefan WINBLAD et al. University of Gothenburg - Gothenburg, Sweden
P-040	**Validation of Sensitivity and Specificity of a New Diagnostic Certified Assay to Better Characterize the CCTG Number Repeats in DM2** Fortunata LOMBARDI et al. Research Laboratories - Molecular Biology, IRCCS Policlinico San Donato - Milan, Italy
P-041	**Elevated miRNA Levels in Serum of Myotonic Dystrophy Patients Relate to Disease Progress** Andrie KOUTSOULIDOU et al. The Cyprus Institute of Neurology and Genetics - Nicosia, Cyprus
P-042	**The Quebec's Founder Effect Has Little Influence on the Phenotype of Patients with Myotonic Dystrophy Type 1 (DM1)** Louise COSSETTE et al. CHU Quebec - Quebec, Canada
P-043	**The Usefulness and Limitations of Repeat-primed PCR in Myotonic Dystrophy Type 1 and 2 Molecular Testing** Jan RADVANSZKY et al. Institute of Molecular Physiology And Genetics, Slovak Academy of Sciences - Bratislava, Slovakia
P-044	**Chronic pain in persons with adult and mild phenotypes of Myotonic Dystrophy Type 1** Maud-Christine CHOUINARD et al. Université du Québec à Chicoutimi - Chicoutimi, Canada
P-045	**Patient-reported Weakness, Myotonia and Swallowing Difficulties in the UK Myotonic Dystrophy Patient Registry** Antonio ATALAIA et al. Newcastle University - Newcastle, United Kingdom
P-046	**Non-invasive Ventilation in Myotonic Dystrophy Type 1 (DM1): Tolerance, Compliance and Impact on Quality of Life** Elisa FALCIER et al. Centro Clinico NEMO, Fondazione Serena - Pneumology Unit, University of Milan - Milan, Italia
P-047	**Nutritional Intake of Patients with Myotonic Dystrophy Type 1** Mélissa BRIEN et al. Department of Health Sciences, Université du Québec à Chicoutimi - Saguenay, Canada

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P-048	**A preliminary prevalence estimate of DM1 and DM2 in the Lazio region, Italy** Nicola VANACORE et al. National Institute of Health - Rome, Italy
P-049	**The Symptomatic Impact of Childhood and Congenital Onset Myotonic Dystrophy Type-1 (DM1): a Multi-national, Cross Sectional Study** Nicholas JOHNSON et al. University of Utah - Salt Lake City, United States
P-050	**The High Competing Risk of Non-cancer Mortality Obscures Cancer Burden in Patients with Myotonic Dystrophy** Shahinaz GADALLA et al. National Cancer Institute, National Institute of Health - Rockville, Maryland, United States
P-051	**Gender-related Differences in Myotonic Dystrophy Type 1: the DM-Scope Registry Observational Study** Guillaume BASSEZ et al. Henri Mondor Hospital - Creteil, France
P-052	**Cognitive Imparment in DM1: a Longitudinal Study** Adolfo LÓPEZ DE MUNAIN et al. Biodonostia - San Sebastián, Spain
P-053	**Long-term Outcomes of a Series of Childhood-onset Form of DM1 Patients** Aurelia JACQUETTE et al. Departement de Genetique, Hopital de la Pitie Salpetriere - Paris, France
P-054	**Neuromuscular Transmission Defects in Myotonic Dystrophy Type 1: a Neurophysiological Study** Antonio PETRUCCI et al. Neuromuscular and Neurological Rare Diseases Center, San Camillo Hospital - Rome, Italy
P-055	**Peculiar signs and symptoms of Myotonia Dystrophica MD1 affected members in a Hungarian family** Zsuzsanna HARTYANI et al. University of Pannonia, Veszprem, Hungary - Veszprem, Hungary
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 et al. Queen Silvia Children's Hospital - Gothenburg, Sweden
P-057	**Analysis of unexpanded and intermediate CTG polymorphisms at the DMPK gene in Mexican population and in Native Amerindian Groups** Nadia Mireya MURILLO et al. National Rehabilitation Institute - Mexico, Mexico
	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 et al. Sahlgrenska University Hospital - Gothenburg, Sweden
P-059	**Role of BRUNO-3, CUGBP1 Orthologue, in a Drosophila Model of Myotonic Dystrophy Type 1** Lucie PICCHIO et al. Gred - Clermont-Ferrand, France
P-060	**Manumycin a Corrects Aberrant Splicing of Clcn1 in Myotonic Dystrophy Type 1 (DM1) Mice** Kosuke OANA et al. University of Tokyo - Tokyo, Japan
P-061	**High-throughput Screen for Pharmacological Compounds That Target CUG-rich RNA Foci in Myotonic Dystrophy** Pascal CHARTRAND et al. Université de Montréal - Montréal, Canada
P-062	**Evaluating the Effects of CELF1 Deficiency in a Mouse Model of RNA Toxicity** Yun Kyoung KIM et al. University of Virginia - Charlottesville, United States
P-063	**Muscle and Brain-specific Inducible Mouse Models of DM2** John CLEARY et al. University of Florida - Gainesville, United States
P-064	**Observations on Oligonucleotide Based Therapy for Myotonic Dystrophy** Rebecca MOORE et al. The University of Nottingham - Nottingham, United Kingdom

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P-065	**High Content Screening Using Hes Derived Mesenchymal Stem Cells Carrying the Myotonic Dystrophy Type 1 Mutation** **Yves MAURY et al.** ISTEM / CECS - Evry, France
P-066	**GSK3 Inhibition is a Novel Therapeutic Approach to Suppress Progression of Myotonic Dystrophy Type 1** **Christina WEI et al.** Baylor College of Medicine - Houston, United States
P-067	**Metformin control of multiple alternative RNA splicing opens new therapeutic potentials** **Sandrine BAGHDOYAN et al.** INSERM/UEVE UMR 861, I-STEM - Evry, France
P-068	**Systemic delivery of an RNAi therapy improves the phenotype of the HSALR mouse model of DM1** **Darren R. BISSETT et al.** University of Washington School of Medicine - Seattle, United States
P-069	**Prevalence and Clinical Correlates of Sleep Disordered Breathing in Myotonic Dystrophy Type 1 and 2** **Maria Laura Ester BIANCHI et al.** UCSC - Rome, Italy
	P2.3 Therapeutic trials Chairpersons: David BROOK - Nottingham, Virginia ARECHAVALA-GOMEZA - Barakaldo
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** Karolinska Institutet / Karolinska University Hospital - Stockholm, Sweden
P-071	**Quantitative Muscle Strength Impairments at the Ankle in Myotonic Dystrophy Type 1 (DM1) Patients: a Five-year Follow-up** **Luc J. HÉBERT et al.** Laval University - Québec, Canada
P-072	**The Italian Registry for Myotonic Dystrophy Type 1 and 2: Ready to Start** **Barbara FOSSATI et al.** IRCCS Policlinico San Donato - San Donato Milanese, Italy
P-073	**Uk Myotonic Dystrophy Patient Registry: Developing Standards of Care and Clinical Research** **Chris TURNER et al.** National Hospital for Neurology and Neurosurgery, London , United Kingdom
P-074	**A Roman Network for the Myotonic Dystrophies: Construction of a DM1/DM2 Clinical and Genetic Database in a Large Population** **Emanuele RASTELLI et al.** Tor Vergata University of Rome - Rome, Italy
P-075	**Impact of an 8-week Periodized Combined Training Program in DM1 Patients: a Pilot Study** **Mario LEONE et al.** University of Quebec in Chicoutimi - Saguenay, Canada
P-076	**Assessment of Gait Instability in Myotonic Dystrophy Type 1** **Valentina GATTI et al.** Centro Clinico NEMO, Fondazione Serena Onlus - Milan, Italy
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 et al.** University Laval - Quebec, Canada
	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 et al.** Southern General Hospital - Glasgow, United Kingdom
P-079	**Assessing Quality of Care in Myotonic Dystrophy: the Scottish Experience** **Cheryl LONGMAN et al.** NHS - Glasgow, United Kingdom

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P-080	**The Challenge of Increasing Attendance at Myotonic Dystrophy Clinics** **Irene PARSONS et al.** West of Scotland Regional Genetics Service - Glasgow, United Kingdom
P-081	**Do Guidelines Guide the Generalist? the Scottish Experience** **Yvonne ROBB et al.** National Health Service - Edinburgh, United Kingdom
P-082	**The Scottish Myotonic Dystrophy Database: What Can It Tell Us?** **Mark HAMILTON et al.** National Health Service - Glasgow, United Kingdom
P-083	**Patient Education Strategies Used by Nurses to Support Self-management of People with Myotonic Dystrophy Type 1** **Maud-Christine CHOUINARD et al.** Université du Québec à Chicoutimi - Chicoutimi, Canada
P-084	**Myotonic Dystrophy Type 1 and 2: Time-lag to Diagnosis in Italy** **Alice ZANOLINI et al.** IRCCS Policlinico San Donato - Università Degli Studi di Milano - Milan, Italy
P-085	**Healthcare Organization Disparities Across Neuromuscular Clinics in the Province of Quebec (Canada) for DD1 Population: Observation and Challenges** **Annie PLOURDE et al.** Grimn - Jonquière, Canada
P-086	**Factors Influencing the Effectiveness of Educational Interventions by Nurses for People with Myotonic Dystrophy Type 1(DM1): an Ecological Approach** **Mélissa LAVOIE et al.** Université de Sherbrooke - Sherbrooke, Canada
P-087	**Referral Criteria for Occupational Therapy Services Related to Independent Housing Among DM1 Patients** **Kateri RAYMOND et al.** Université de Sherbrooke - Jonquière, Canada
P-088	**Resonance and Myotonic Dystrophy Type 1** **Claire-Cecile MICHON** AFM-Telethon - Evry, France
P-089	**Illuminating Loss - an Artistic Study of Siblings Affected by Myotonic Dystrophy Type 1** **Jacqueline DONACHIE** University of Northumbria - Newcastle, United Kingdom
15.00 - 19.30	**Session 5 - Therapeutic development in model system** Chairpersons: Denis FURLING - Paris, Laura P.W RANUM - Gainesville
15.00 - 15.30	**S5.1 Overview lecture**
	Multiple models, diverse approaches and progress on many fronts **Thomas COOPER** Baylor College of Medicine - Houston, United States
15.30 - 17.00	**S5.2 Platform presentations**
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 et al.** University of Virginia - Charlottesville, United States
15.45 - 16.00	**O-38 Shorter CAG Morpholino Oligo is More Effective to Reverse the Function of MBNL1** **Kanako NAGANO et al.** The University of Tokyo - Tokyo, Japan
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 et al.** University of Virginia - Charlottesville, United States
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 et al.** Massachusetts General Hospital - Boston, United States

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16.30 - 16.45	**O-41 New Human Muscle Cell Models for DM** **Ludovic ARANDEL et al.** UPMC UM76, Institut de Myologie - Paris, France
16.45 - 17.00	**O-42 High Content Imaging Screens Using Myotonic Dystrophy Cell Lines Identify Small Molecules That Remove Nuclear Foci** **Ami KETLEY et al.** University of Nottingham - Nottingham, United Kingdom
17.00 - 17.30	Coffee break and poster viewing
17.30 - 19.30	**S5.3 Platform presentations**
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 et al.** Valentia BioPharma SL - Paterna, Spain
17.45 - 18.00	**O-44 Systemic Delivery of an Antisense Oligonucleotide (ASO) Targeting DMPK RNA Improves the Phenotype of DMSXL Mice** **Dominic JAUVIN et al.** Centre Hospitalier Universitaire de Québec - Québec, Canada
18.00 - 18.15	**O-45 Small molecule inhibitor of MBNL1 induces Myotonic Dystrophy Type 1 pre-mRNA splicing defects** **Ewa STEPNIAK-KONIECZNA et al.** Adam Mickiewicz University - Poznan, Poland
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 et al.** Gred - Clermont-Ferrand, France
18.30 - 18.45	**O-47 Assessment of Tricyclo-DNA Antisens Oligonucleotide to Target Nuclear CUG-expanded RNA** **Arnaud KLEIN et al.** Um76 - Therapie des Maladies du Muscle Strié - Paris, France
18.45 - 19.00	**O-48 Very short oligonucleotides composed of locked nucleic acids as potential therapeutic molecules in Myotonic Dystrophy** **Agnieszka WOJTKOWIAK-SZLACHCIC et al.** Department of Gene Expression, Institute of Molecular Biology and Biotechnology, Adam Mickiewicz University, 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 et al.** UPMC UM76, Institut de Myologie - Paris, France
19.15 - 19.30	**O-50 CNBP Knock-out Mouse Model for Myotonic Dystrophy Type 2 (DM2)** **Mario SIRITO et al.** University of Texas MD Anderson Cancer Center - Houston, United States

Full Program
Saturday, October 19

08.00 - 10.00	**Session 6 - Therapeutic trials** Chairpersons: Frank C. BENNETT - Carlsbad, Virginia ARECHAVALA-GOMEZA - Barakaldo
08.00 - 08.30	**S6.1 Overview lecture**
	Trial Readiness and Implementation **Charles THORNTON** University of Rochester Medical Center - Rochester, United States
08.30 - 10.00	**S6.2 Platform presentations**
08.30 - 08.45	**O-51 Efficacy and Cellular Uptake of Antisense Oligonucleotides to Treat Myotonic Dystrophy Type 1** **Anchel GONZALEZ et al.** Prosensa Therapeutics B.V. - Leiden, Netherlands
08.45 - 09.00	**O-52 Strategies in Antisense Oligo Treatment of Muscle and Brain in DM1** **S. MULDERS et al.** Prosensa - Leiden, Netherlands
09.00 - 09.15	**O-53 The Ultrasound-enhanced Delivery of Morpholino: Therapeutic Trial of Myotonia with Antisense Oligonucleotides** **Michinori KOEBIS et al.** The University of Tokyo - Tokyo, Japan
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 et al.** Isis Pharmaceuticals, Inc. - Carlsbad, United States
09.30 - 09.45	**O-55 Potential Therapeutics for Myotonic Dystrophy: Pentamidine and Analogs as Inhibitors of Transcription** **Leslie COONROD et al.** University of Oregon - Eugene, United States
09.45 - 10.00	**O-56 Actinomycin D Inhibits Transcription of CUG RNA in a DM1 Model** **Ruth SIBONI et al.** University of Oregon - Eugene, United States
10.00 - 10.30	Coffee break and poster viewing
10.30 - 12.00	**Session 7 - Ethical, legal and social issues** Chairpersons: Tetsuo ASHIZAWA - Gainesville, Chad HEATWOLE - Rochester
10.30 - 11.00	**S7.1 Overview lecture**
	Myotonic dystrophy Type 1 as a model of organized complexity **Cynthia GAGNON et al.** Université de Sherbrooke / Grimn - Jonquière, Canada
11.00 - 12.00	**S7.2 Platform presentations**
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 et al.** Sahlgrenska University Hospital - Gothenburg, Sweden
11.15 - 11.30	**O-58 Implementation of a nursing case management in Myotonic Dystrophy Type 1 patients** **Maud-Christine CHOUINARD et al.** Université du Québec à Chicoutimi - Chicoutimi, Canada
11.30 - 11.45	**O-59 Cognitive Impairments and Their Effect Upon Everyday Life in Non Congenital Forms of Myotonic Dystrophy** **Margaret PHILLIPS et al.** University of Nottingham - Derby, United Kingdom
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 et al.** Friedrich-Baur-Institute, LMU Munich - Munich, Germany
12.00 - 14.00	Lunch

PLATFORM and POSTER PRESENTATIONS

The Platform and Poster presentation descriptions (pages 33 through 223) have been removed from this document to honor confidentiality requirements

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