Program

Program IDMC-14

TUE 9 April

14:30-16:30 Registration
16:30-19:00 **Opening**
Chairs: Baziel van Engelen/Rick Wansink
16:30-16:45 Welcome Address - *Rick Wansink*
16:45-17:15 History of DM: past, present, future
Prof. dr. Bé Wieringa, Radboudumc, Nijmegen, NL
17:15-17:30 Musical Intermezzo Q lite
17:30-18:00 History of Neurology
Dr. Peter Koehler, MUMC+, Maastricht, NL
18:00-18:15 Musical Intermezzo Q lite
18:15-18:45 Lessons Learned from DMD for DM
Prof. dr. Annemieke, Aartsma-Rus, LUMC, Leiden, NL
18:45-19:00 Closing & housekeeping remarks - *Rick Wansink*

19:00-20:30 **Welcome Reception**

WED 10 April

08:00-08:30 Registration
08:30-10:00 **Session 1a: Pathogenic Mechanisms I**
Chairs: Mario Gomes Pereira/Darren Monckton
08:30-09:00 Why do CTG18.1 expansions in TCF4 not cause myotonic dystrophy type 3 (or do they)?
Prof. dr. Alice Davidson, University College, London, UK
09:00-09:15 Smooth muscle specific loss of MBNL1 and MBNL2 causes muscle hypercontraction and delayed gastrointestinal motility
Janel AM Peterson
09:15-09:30 Investigating global and cell type-specific transcriptomic dysregulation in the DM1 brain
Emily E. Davey
09:30-09:45 Isolating the role of myotonia in DM1 pathogenesis through a novel myotonia resistant mouse model
Matthew Sipple
09:45-10:00 Differentiation shifts from a reversible to an irreversible heterochromatin state at the DM1 locus
Rachel Eiges
10:00-10:30 **Speed Dating**
Chair: Peter-Bram 't Hoen

10:30-11:00 **Coffee break with refreshments and games**

11:00-12:15 **Session 1b: Pathogenic Mechanisms II**
Chairs: Analisa Botta/Krzysztof Sobczak

11:00-11:30 Primate-specific factors in disease-associated somatic repeat instability: RPA and Alt-RPA suppress and drive expansions
Prof. dr. Christopher Pearson, Hospital for Sick Children, Toronto, Canada

11:30-11:45 Actin cystoskeleton abnormalities triggered by toxic CUG RNA in DM1 brain cells
Paul Magneron

11:45-12:00 Expression levels of core spliceosomal proteins modulate the MBNL-mediated spliceopathy in myotonic dystrophy
Jiss Louis

12:00-12:15 Sense and antisense RAN proteins in DM1 brain and skeletal muscle
Eduardo Rijos

12:15-12:30 **Flash Posters (2.5 min. each)**
Chair: Renée Buurman - Raaijmakers

Quantification of DMPK transcript modulation by antisense oligonucleotides using digital PCR and in-cell western techniques
Andrea López-Martinez

Congenital and Childhood Myotonic Dystrophy Health Index (CCMDHI): Italian validation of a disease specific measure of perceived burden
Susanna Pozzi

Endogenous modulation of MBNL1 expression via RNA activation (RNAa) as a novel therapeutic approach towards myotonic dystrophy type 1 (DM1)
Nikola Musiala-Kierklo

Physiological routine blood test fluctuations in myotonic dystrophy type 1 (DM1): implications for safety monitoring in clinical trials
Elena Aragona

Comprehensive transcriptome analysis of the liver from patients with myotonic dystrophy type 1
Aono Fukumoto

12:30-13:30 **Lunch** (*with posters*)

13:30-14:30 **Poster Session A** (uneven numbers)

14:30-15:30 **Session: Patient Engagement**
Chairs: Hilde Braakman/Renée Buurman - Raaijmakers

14:30-14:35 Introduction: experiences in the clinic - *Hilde Braakman*

14:35-14:40 Viewpoint from a partner & mother - *Cathy Gibson*

14:40-14:50 What role can patients and patients' caregivers play - *Peter Ashley*

14:50-15:00 What role can patient organizations play - *Alain Geille*

15:00-15:15 Patient participation in research - *Charlotte van Esch*

15:15-15:30 Open discussion - *Hilde Braakman/Renée Buurman - Raaijmakers*

15:30-16:00 **Session 2a: Biomarkers and Outcome Measures I**
Chairs: Elise Duchesne/Fernando Morales

15:30-16:00 Clinimetrics in DM1
Prof. dr. Ingemar Merkies, MUMC+
Maastricht, The Netherlands

16:00-16:30 **Coffee break with refreshments and games**

16:30-17:30 **Session 2b: Biomarkers and Outcome Measures II**
Chairs: Elise Duchesne/Fernando Morales

16:30-16:45 A central role for disordered renal metabolism in myotonic dystrophy type 1
Preeti Kumari

16:45-17:00 Large-scale proteomics profiling of peripheral blood of DM1 patients identifies biomarkers for disease severity and physical activity
Daniël van As

17:00-17:15 The Splice Index as a prognostic biomarker of strength and function in myotonic dystrophy type 1
Melissa A. Hale

17:15-17:30 Strength training improves transcriptomic alterations in DM1 patients
Cécilia Légaré

17:30-18:40 **Session 3: Clinical Manifestation and Quality of Life**
Chair: Masanori Takahashi

17:30-17:45 Costs and healthcare resource utilization evaluation in myotonic dystrophy type 1: Results from the real-world CARE-DM1 Study
Johanna Hamel

17:45-18:00 Mortality rate and predictors of death in the DM1 population, a registry-based study
Guillaume Bassez

18:00-18:25 Women Health in DM1
Cynthia Gagnon
Enhancing Dysphagia Assessment in DM1: Identifying Promising PROMs and Key Symptomatic Insights
Claudia Côté

18:25-18:40 The Myotonic Dystrophy Health Index: a valid measure to detect disease burden changes over time
Carola Ferrari Aggradi

THU 11 April

08:00-08:30 Registration

08:30-10:00 **Session 4: Cell and Animal Models**
Chairs: Geneviève Gourdon/Rachel Eiges

08:30-08:45 Natural antioxidants reduce oxidative stress and the toxic effects of RNA-CUG(exp) in an inducible glial myotonic dystrophy type 1 cell model
Fernando Morales

08:45-09:00 3D skeletal muscle constructs from human pluripotent stem cells for myotonic dystrophy Type 1 modeling
Lise Morizur

09:00-09:15 Development of a new inducible mouse model for myotonic dystrophy
Alain Sureau

09:15-09:30 Brain and muscle phenotypes in a novel BAC transgenic mouse model of myotonic dystrophy type 2
Tala Ortiz

09:30-09:45 Modeling neurodevelopmental defects in congenital dystrophy using forebrain organoids
Thiéry De Serres-Bérard

09:45-10:00 DM1 in mice with expanded repeats in Dmpk
Zhenzhi Tang

10:00-10:30 **Fringe Session I**
Chair: Rick Wansink
Deciphering RNA foci: Unveiling the hallmark of myotonic dystrophy
David Brook and Cameron Niaz

10:30-11:00 **Coffee break with refreshments and games**

11:00-12:15 **Session 5: Clinical Trials and Trial Design**
Chairs: Charles Thornton/Guillaume Bassez
11:00-11:15 Avidity trial
Dr. Nick Johnson MD, Virginia Commonwealth, University, Richmond, VA, USA
11:15-11:30 AMO trial
Prof. dr. Hanns Lochmüller, Univ. Ottawa, Ottawa, Canada
11:30-11:45 Initial data from the ACHIEVE trial of DYNE-101 in adults with myotonic dystrophy type 1 (DM1)
Baziel van Engelen
11:45-12:00 Efficacy and safety of metformin on mobility and strength in myotonic dystrophy type 1: the Metmyd study demographics, baseline data and lessons learnt
Roberto Massa
12:00-12:15 Erythromycin for myotonic dystrophy type 1: a multicenter, randomized, double-blind, placebo-controlled, phase 2 trial
Masayuki Nakamori

12:15-12:30 **Flash Posters (2.5 min. each)**
Chair: Anne Bruijnes

Galectin-3 as potential biomarker of cardiac conduction disorders in myotonic dystrophy type 1
Vukan Ivanovic
Masseter muscle size as a proxy of disease severity in myotonic dystrophy type 2
Diana A. Madrid
Modified polycyclic compounds as a new class of DM1 small molecule therapeutics
Jesus Frias
Deciphering the consequences of SORBS1 mis-splicing in myotonic dystrophy type 1
Morgan Gazzola
Characterization of sleep phenotypes and underlying mechanisms in mouse models of myotonic dystrophy type 1
Juan D. Arboleda

12:30-13:30 **Lunch** (with posters)

13:30-14:30 Poster Session (even numbers)

14:45-19:30 **Walking Tour Nijmegen + Drinks at De Hemel**

FRI 12 April

08:00-08:30 Registration

08:30-10:00 **Session 6: Therapeutic Strategies and Drug Development**
Chairs: Eric Wang/Cécile Martinat

08:30-08:45 Repeat length and genetic background as driving forces for disease heterogeneity and therapeutic response in myotonic dystrophy type 1
Najoua El Boujnouni

08:45-09:00 Primary pericytes as muscle progenitors in cell therapy for myotonic dystrophy
Renée Raaijmakers

09:00-09:15 Use of lipophilic-conjugation to deliver antimiR-23b into skeletal muscle and nervous system with a dual therapeutic approach to DM1
Beatriz Llamusi

09:15-09:30 Therapeutic tuning of MBNL1 expression in myotonic dystrophy
Ewa Stępniak-Konieczna

09:30-09:45 Treatment of a severe DM1 mouse model with verapamil, amlodipine, and ranolazine
Lily Cisco

09:45-10:00 Early signs of efficacy: Using an EEV-PMO(CAG) to identify the first markers of rescue in HSA^{LR} mice
Emma N. Shea

10:00-10:30 **Fringe Session II**
Chair: Karin Faber
Myotonic dystrophy in promising times of RNA therapeutics, mind the illness!
Baziel van Engelen

10:30-11:00 **Coffee break with refreshments and games**

11:00-12:00 **Session 7: Biomarkers and Outcome Measures III**
Chair: Cynthia Gagnon

11:00-11:15 Exploring the role of serum NFL and IL-6 as biomarkers for CNS affection in DM1
Joana Garmendia

11:15-11:30 Muscle MRI in DM1: long term follow-up analysis
Matteo Garibaldi

11:30-11:45 Beyond traditional tests: Video-based metrics for advancing precision in movement analysis
Tina Duong

11:45-12:00 Splicing alterations are common among muscular dystrophies – what is different in myotonic dystrophy?
Vanessa Todorrow

12:00-12:30 **Making a Career in DM**
Chair: Peter-Bram 't Hoen
with Melissa Hale and Benedikt Schoser

12:30-13:30 **Lunch** (with posters)

13:30-14:15 **Session 8: Children with DM**
Chair: Anne-Berit Ekström

13:30-13:45 Splicing dysregulation in adults and children with myotonic dystrophy type 1 is associated with physical function
Julia M. Hartman

13:45-14:00 Understanding swallowing, mastication and speech difficulties in children with myotonic dystrophy type 1: Insights from clinical and ultrasound assessments
Saskia Scholten

14:00-14:15 Theory of mind – Virtual reality training in children with DM1
Nathalie Angeard

14:15-15:00 **Fringe Session III**
Chair: Hans van Bokhoven
The impact of artificial intelligence on research and clinical practice in DM

14:45-15:15 **Session 9 Clinical Manifestation and Quality of life II**
Chair: Anne Bruijnes

14:45-15:00 Palliative care guidelines for patients with myotonic dystrophy type 1
Derek Willis

15:00-15:15 A cognitive and social portrait of adults presenting the infantile DM1 phenotype
Benjamin Gallais

15:15-15:45 **Coffee break with refreshments and games**

15:45-16:30 **Session 10: Clinical Management and Rehabilitation**
Chair: Benedikt Schoser

15:45-16:00 Respiratory muscle strength and respiratory endurance training in myotonic dystrophy type 1: results from a controlled, randomized, three-arm interventional study
Stephan Wenninger

16:00-16:15 Surface electromyography thresholds as a measure for performance fatigability during incremental cycling in patients with myotonic dystrophy type 1
Nicoline Vloet

16:15-16:30 Substantial improvement of shoulder function with a new physiotherapy approach in children with myotonic dystrophy
Maaike Pelsma

16:30-17:00 **Awards and Closing Remarks**
Rick Wansink

17:45-23:00 **Gala dinner at Fort Lent**

SAT 13 April

Family day: more information Spierziekten Nederland: Bijeenkomst