

European MH Group 2018 Meeting Ferrara, Italy

On behalf of the board of directors of the European Malignant Hyperthermia Group (EMHG) and the members of the local organizing committee, we are very pleased to welcome you to the 37th annual meeting of the European Malignant Hyperthermia Group which will take place in Ferrara, Italy on May 17th - 19th 2018.

The congress will be held in the center of the city of Ferrara.

We will certainly try to continue the trend of successful past annual meetings and hope to further contribute to the spirit of friendship and collaboration as known since the first meetings of our group. Indeed the EMHG always stood for close collaborations, an increasingly important asset in facilitating an integrated approach to bridge the gap between the bench and bedside in the ryanodinopathy field.

We look forward to welcoming you and spending a great time in Ferrara.

Sman Zers Isaa Sont

Susan Treves

Francesco Zorzato

SCIENTIFIC PROGRAM

European MH Group 2018 Meeting Ferrara, Italy

Date: 17-19 May 2018

Venue: Palazzo Roverella, Corso Giovecca 47, 44121 Ferrara, Italy

Thursday, May 17th 2018 Conference Room Hotel Europa

15:00-18:00 Malignant Hyperthermia Group Executive Committee Meeting.

15:30-18:00 For other interested participants, visit to the Castle Cavallini Sgarbi exhibit or Jewish museum.

18:00-19:15 Genetics workshop, Conference Room Hotel Europa

19:00-20:00 Welcome reception and meeting registration at the Hotel Europa

20:00-22:00 Dinner for all participants

Friday, May18th 2018 Palazzo Roverella Meeting Room

8:00-8:30 Later registration at Palazzo Rov	erella
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8:30- 8:35 Welcome / Introduction

- 8:40-9:10 Guest Speaker: Prof. Andrew R. Marks, Chairman Department of Physiology, Center for Molecular Cardiology, Columbia University, New York, N.Y, U.S.A. What the high resolution structure reveals about RyR1 regulation
- 9:10-10:30 <u>Scientific session 1</u>: Clinical issues and case reports (Chaired by Dr. Sheila Riazi and Prof. Philip Hopkins).
- 9:10- 9:30 Essam (Ali) Aboelsaod (Leeds, U.K.): Ambiguous anaesthetic history and the susceptibility to malignant hyperthermia.
- 9:30- 9:50 Anne- Frederique Dalmas (Lille, France): Masseter muscle spasm in a cesarean section.
- 9:50-10:10 Martina Klincova (Ostrava, Czech Republic): Malignant Hyperthermia in the Czech Republic our practice end experience.
- 10:10-10:30 Claudia Piccinini (Varese, Italy): A case of Malignant Hyperthermia in a patient with Tourette syndrome.

10:30-11:00 Coffee break

- 11:00-12:30 Scientific session 2: Variants in genes encoding components of the excitation-contraction coupling macromolecular complex (ECC-MC) and neuromuscular disorders (Chaired by Prof. Vincenzo Sorrentino and Dr. Erik-Jan Kamsteeg)
- 11:00-11:20 Violeta Glauber (Tel Aviv, Israel): Molecular analysis of patients with Malignant Hyperthermia.
- 11:20- 11:40 Catherine Daly (Leeds, U.K.): A new old method for confirming whole exome NGS findings.
- 11:40-12:00 Carlos Ibarra (Toronto, Canada): Survey on associated symptoms and morbidities in carriers of *RYR1* genetic variants.
- 12:00- 12:20 Kathryn Stowell (Palmerston, New Zealand): The EMHG RYR1 variant list
- 12:20- 12:40 Asensio Gonzalez (Basel, Switzerland): Filtering exome variants with phenotype segregation and muscle expression in an MHS family

12:40-14:00 Lunch break Palazzo Roverella

- 14:00-14:30 Guest speaker: Prof. C. Reggiani Department of Biomedical Sciences, University of Padova, Italy. Do mitochondria contribute to Malignant Hyperthermia (MH) crisis? A study of murine models.
- 14:30-16:00 Scientific session 3: Phenotypic consequences of mutations in *RYR1* and other genes of ECC-MC (Chaired by Prof. Feliciano Protasi and Prof. Heinz Jungbluth)
- 14:30- 14:50 Heinz Jungbluth (London, U.K.): Malignant hyperthermia (MH) as a multifactorial event new insights into a life threatening anaesthesia complication.
- 14:50-15:10 Carlos Ibarra (Tornoto, Canada): Comparison of the North American caffeine halothane contracture test and the Japanese calcium-induced calcium release test for diagnosis of Malignant Hyperthermia susceptibility.
- 15:10- 15:30 Nicol Voermans (Nijmegen, The Netherlands): The histopathological spectrum of Malignant Hyperthermia and non-anaesthetic rhabdomyolysis due to *RYR1* mutations.
- 15:30- 15:50 Feliciano Protasi (Chieti, Italy): Link between Malignant Hyperthermia and environmental exertional heat stroke: just an hypothesis?

15:50-16:10 Coffee break

16:10 -17:30 Scientific session 4: Experimental studies on muscle and non-muscle cells from patients and animal models (Chaired by Prof. Paul Allen and Prof. Albert Urwyler)

16:10-16:30 Leon Chang (Leeds, U.K.): Halothane exposure reduces oxidative capacity in RYR1 G2435R knock-in mice. 16:30- 16:50 Vikas Kaura (Leeds, U.K.): Characterization of a novel knock-in mouse model of the human Malignant Hyperthermia G2434R RYR1 variant. 16:50-17:10 Albert Urwyler (Basel, Switzerland): Importance of RyR1 in thermogenesis of mammals apart from MH? 17:10-17:30 Carolina Figueroa (Chicago, U.S.A.): Abnormal calcium signaling in MH susceptible patients with halothane hypersensitivity. 17:30-19:30 Walking tour of Ferrara 19:30-20:00 Apéro at Palazzo Roverella 20:00-23:00 Gala Dinner at Palazzo Roverella Saturday, May 19th 2018 Palazzo Roverella Meeting Room 8:30-10:10 Scientific session 5: Diagnosis and treatment(s) of ryanodinopathies and management of MH patients (Chaired by Dr. Oliver Bandschapp and Dr. Stephan Johannsen) 8:30-8:50 Anne- Frederique Dalmas (Lille, France): Improvement of reconstitution of intravenous dantrolene (Dantrium®IV). 8:50-9:10 Carla Scattolini (Siena, Italy): A case of mis-diagnosed MHS during heart transplant. 9:10-9:30 Enrico Pierantozzi (Siena, Italy): 20 years of MH diagnosis in Siena. 9:30-9:50 Bastian Börge (Leipzig, Germany): Survey about malignant hyperthermia events in German intensive care units 9:50-10:10 Florence Julien-Marsollier (Paris, France): Management of Malignant Hyperthermia in France: current organization and future development. 10:10-10:40 Coffee break 10:40-11:00 Board Meeting 11:00-12:00 Annual EMHG meeting **12:00-12:30** Awards Ceremony

12:30

End of the meeting

12:30-13:30 Lunch at Hotel Europa