

4° HORIZONS FOR DRAVET SYNDROME

**INTERNATIONAL SYMPOSIUM
"DRAVET SYNDROME AND OTHER SODIUM
CHANNEL RELATED ENCEPHALOPATHIES"**

15-16 MARCH 2018, VERONA | PALAZZO DELLA GRAN GUARDIA

info@horizonsdravet.eu · www.horizonsdravet.eu



SCIENTIFIC COMMITTEE

Prof. Renzo Guerrini - Firenze, Italy

Prof. Helen Cross - London, UK

Prof. Bernardo Dalla Bernardina - Verona, Italy

Prof. Rima Nabbout - Paris, France

Dr. Francesca Darra - Verona, Italy

HONORARY PRESIDENT OF SYMPOSIUM

Charlotte Dravet - Marseille, France

ORGANIZING SECRETARIAT

Isabella Brambilla - Verona, Italy

Elisa Giarola - Verona, Italy

Hannah Rawlinson - Verona, Italy

PTS

via Nizza 45, 00198 Roma

Maura Stella

Dear friends and colleagues,

On the occasion of 40 years since Dravet Syndrome was first defined, and 8 years after organizing the first Workshop in Verona, we are very pleased to invite you once again to this magnificent city for the **“Dravet Syndrome and Other Sodium Channel Related Encephalopathies” International Symposium**.

The Symposium consists of two days focusing on scientific research relating to genes SCN1A, SCN2A and SCN8A.

The study of epilepsy and the care of children have changed remarkably in recent years, after the identification of the genetic causes of some epilepsy syndromes. The main epilepsy gene- the sodium channel alpha 1 (SCN1A)- has been linked to Dravet Syndrome, to a number of less severe forms of epilepsy, and to febrile convulsions. However, more than 15 years after the causative role of this gene was identified in these forms, and in spite of the large number of patients identified, the spectrum of clinical manifestations associated with SCN1A mutations continues to be enriched by new phenotypes and only recently has enough evidence been collected to foresee to what extent early clinical and genetic predictors seem to influence prognosis. Thanks to the advent of next-generation sequencing, the process that will eventually lead to fully highlight the phenotypical spectrum, long-term outcome, and role of genetic variation in the epilepsies associated with mutations of the other two main sodium channel genes associated with epilepsy- SCN2A and SACN8A- will hopefully be quicker but is until now nonetheless proving relatively slow.

Even slower, and particularly complex, is the process that has led to the gathering of evidence on the sensitivity of these conditions to medication. It has taken more than 16 years since the first controlled trial demonstrated the efficacy of add-on stiripentol in Dravet syndrome, before new trials to test the efficacy of two different molecules, fenfluramine and cannabidiol, were launched in this same syndrome, and none seems to be on the horizon for the conditions associated with SCN2A- and SCN8A-related epilepsies. In order to address the main clinical, genetic and treatment issues that concern families, the specialists, and basic researchers alike; to explore to what extent disorders arising from mutations in this gene family overlap and differ; to better define the specific burden of comorbidities; and to explore the bases for rational treatment approaches, we have organized a thematic workshop to gather world-leading specialists in Verona to discuss available evidence and perspectives for future developments.

It will be a pleasure to share these two days of scientific research with you.

On behalf of scientific committee

Bernardo Dalla Bernardina, Renzo Guerrini

PROGRAM DAY 1

March 15th, 2018

09.00-09.20

Symposium Opening: Officials' Welcome - Greetings

Federico Sboarina - the Town Mayor
Francesco Cobello - the Managing Director of the General Hospital Board
Nicola Sartor - Rector of Verona University

Chairperson: Bernardo Dalla Bernardina and Helen Cross

09.20-09.40

Introduction: From Dravet Syndrome to Sodium Channel Encephalopathies

Renzo Guerrini

09.40-10.10

Sodium Channel and Human Disease

Jeffrey Noebels

10.10-10.25

Discussion

10.25-10.40

Coffee Break

DRAVET SYNDROME AND OTHER SCN1A RELATED EPILEPSIES

10.40-11.10

SCN1A related phenotypes

Ingrid Scheffer

11.10-11.40

Beyond the epilepsies in SCN1A diseases

Rima Nabbout

11.40-12.00

Discussion

12.00-13.30

Lunch

Chairperson: Rima Nabbout and Tiziana Granata

- 13.30-13.50** **Mechanisms and models**
Massimo Mantegazza
- 13.50-14.10** **Experience on iPSC Cells in Dravet Syndrome**
Vania Broccoli
- 14.10-14.55** **Where are we with treatment options?**
Helen Cross - Renzo Guerrini - Lieven Lagae
- 14.55-15.10** **Discussion**
- 15.10-15.30** **Management of prolonged seizure from prehospital treatment to intensive care unit**
Federico Vigevano
- 15.30-15.45** **Long term outcomes** - Francesca Darra
- 15.45-15.55** **Teenagers - Clinical cases** - Domenica Battaglia
- 15.55-16.05** **Adults - Clinical cases** - Francesca Ragona
- 16.05-16.15** **Marseille Experience** - Pierre Genton
- 16.15-16.30** **Discussion**
- 16.30-16.45** **Coffee Break**
- 16.45-17.05** **1978-2018: A 40 year retrospective view of Dravet Syndrome**
Bernardo Dalla Bernardina and Charlotte Dravet
- 17.05-18.05** **Session with contributions on the topic**
Coordinators: Michelle Bureau and Gaetano Cantalupo
- 18.05-18.25** **Discussion**
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PROGRAM DAY 2

March 16th, 2018

Chairperson: Renzo Guerrini and Jeffrey Noebels

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|--------------------|-------------------------------------------------------|
| 09.00-09.35 | Dravet Syndrome and its mimics Carla Marini |
| 09.35-09.50 | Discussion |

SCN2A

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|----------------------|------------------------------------------------------------------------------------------------------------------------------|
| 09.50 - 10.30 | Phenotypic spectrum of SCN2A related disorders, treatment options and outcomes in Epilepsy and Beyond Markus Wolff |
| 10.30 - 10.45 | Discussion |
| 10.45 - 11.00 | Coffee Break |
| 11.00 - 11.30 | The genetics of SCN2A Johannes Lemke |
| 11.30 - 12.10 | Mechanisms and Models Holger Lerche |
| 12.10 - 12.30 | Discussion |
| 12.30 - 14.00 | Lunch |

SCN8A

Chairperson: Carla Marini and Markus Wolff

- 14.00-14.30** **Phenotypic spectrum of SCN8A related disorders, treatment options and outcomes**
Elena Gardella
- 14.30 -15.00** **The Genetics of SCN8A**
Rikke Steensbjerg Møller
- 15.00 -15.30** **Mechanisms and Models**
Miriam Meisler
- 15.30-15.45** **Discussion**
- 15.45 -16.00** **Coffee Break**
- 16.00-16.30** **Concluding remarks SCN1A - SCN2A - SCN8A**
Bernardo Dalla Bernardina, Charlotte Dravet, Ingrid Scheffer, Renzo Guerrini and Jeffrey Noebels
- 16.30-17.30** **Session with contributions on the topic**
Coordinators: Elena Parrini and Nicola Specchio
- 17.30-17.50** **Discussion**
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FACULTY

| | |
|----------------------------------|----------------------|
| Domenica Battaglia | Roma, Italy |
| Vania Broccoli | Milano, Italy |
| Michelle Bureau | Marseille, France |
| Gaetano Cantalupo | Verona, Italy |
| Helen Cross | London, UK |
| Bernardo Dalla Bernardina | Verona, Italy |
| Francesca Darra | Verona, Italy |
| Charlotte Dravet | Marseille, France |
| Elena Gardella | Dianalund, Denmark |
| Pierre Genton | Marseille, France |
| Tiziana Granata | Milano, Italy |
| Renzo Guerrini | Firenze, Italy |
| Lieven Lagae | Leuven, Belgium |
| Johannes Lemke | Leipzig, Germany |
| Holger Lerche | Tübingen, Germany |
| Massimo Mantegazza | Valbonne, France |
| Carla Marini | Firenze, Italy |
| Miriam Meisler | Ann Arbor, Michigan |
| Rikke Steensbjerre Møller | Dianalund, Denmark |
| Rima Nabbout | Paris, France |
| Jeffrey L. Noebels | Houston, USA |
| Elena Parrini | Firenze, Italy |
| Francesca Ragona | Milano, Italy |
| Ingrid Scheffer | Melbourne, Australia |
| Nicola Specchio | Roma, Italy |
| Federico Vigevano | Roma, Italy |
| Markus Wolff | Tübingen, Germany |

FEE

DAY 1 and DAY 2

Registration Fee includes the following for all delegates:

- admission to all scientific sessions on days registered
- morning and afternoon tea/coffee breaks
- working lunches
- certificate of attendance
- Welcome Reception on March 15th
- ECM accreditation (for Italian delegates only)

CANCELLATIONS

Cancellations must be made in writing to registration@horizonsdravet.eu

On before February registration fee refunded less 60%

| REGISTRATION FEES | EARLY UP TO FEBRUARY 2018 | LATE/LAST MINUTE |
|-------------------------|---------------------------------------------------|---------------------------------------------------|
| | DAY 1 + DAY 2 (included Satellite Simposio DAY 3) | DAY 1 + DAY 2 (included Satellite Simposio DAY 3) |
| REGULAR | 250,00 € | 300,00 € |
| RESIDENT | 100,00 € | 150,00 € |
| FAMILYES / ASSOCIATIONS | 50,00 € | 50,00 € |



Event endorsed by EpiCARE
European Reference Network for Rare and Complex Epilepsies

Organizational secretary



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and Complex Epilepsies

For further information please apply to the Organizing Secretariat



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