



Fourth International Conference on Primary Central Hypoventilation Science & Life of CHS



Warsaw, Poland, April 13-15, 2012
Hyatt Regency Warsaw

23 Belwederska Avenue, Warsaw, Poland 00-761

Tel: +48 22 558 1234 Fax: +48 22 558 1235

SCIENTIFIC PROGRAMME

FRIDAY APRIL 13, 2012

7:00	Welcome. Registration
8:45-9:30	4 Workshops. 2 Sessions.
&	
9:45-10:30	<ul style="list-style-type: none">• Tracheostomy Care. Angeles Garcia-Teresa (Spain) and Raquel Porto (Spain)<ul style="list-style-type: none">○ Objectives. To show the different kinds of cannulas, to discuss the care of trach and how to do with problems• Facial Interfaces. Agneta Markstrom (Sweden) and Helena Estevao (Portugal)<ul style="list-style-type: none">○ Objectives. To show a range of nasal, oro-nasal and facial masks, to show how to minimize side effects• Respiratory Pacing. Johannes Schoeber (Germany) and Moritz Rohrbach (Germany)<ul style="list-style-type: none">○ Objectives. To explain the prerequisites of age, airway stability and electrophysiology, to explain pacemaker settings and demonstrate pacemaker EKGs, to discuss regular checkups and on how to carry the external pacemaker• Monitoring. Raffaele Piumelli (Italy), Andreas Pflieger (Austria) and David Kilner (United Kingdom)<ul style="list-style-type: none">○ Objectives. To understand how the devices work, to discuss pitfalls of different types of monitoring, to discuss who, how and when ventilator settings change
(30 min)	Visit of Posters & Exhibition. Coffee break
11:00-12:00	Session 1. Introducing the EUCHS Work Chairing : Marek Migdal (Poland) <ul style="list-style-type: none">• Opening Ceremony<ul style="list-style-type: none">○ Jacek Gralinski, Head of the Polish Advisory Group to the Minister Of Health for Rare Diseases (Poland)○ Mieczyslaw Litwin, Scientific Director of the Children's Memorial Health Institute (Poland)○ Mary Vanderlaan, President of the CCHS Family Association (USA)• Central Hypoventilation Syndromes. What we know and don't know. Ha Trang (France)
(1h30)	Visit of Posters & Exhibition. Lunch
13:30-15:00	Session 2. New insights into PHOX2B gene and function. Chairing: Jeanne Amiel (France) <ul style="list-style-type: none">• Neuronal lesions underlying the respiratory defects of CCHS. Jean-Francois Brunet (France)• PHOX2B and midkine/Alk signaling in the control of sympathetic neuron proliferation and neuroblastoma predisposition. H. Rohrer (Germany)• CCHS: lessons from newborn mice. Jorge Gallego (France)
(30 min)	Visit of Posters & Exhibition (Facilitators: Helena Estevao (Portugal), Raffaele Piumelli (Italy). Coffee break
15:30-17:30	Session 3. From bench to bedside. Chairing : Isabella Ceccherini (Italy) <ul style="list-style-type: none">• CCHS and PHOX2B: almost 10 years. Jeanne Amiel (France)• Germline mosaicism and de novo PHOX2B mutations in CCHS: implications for penetrance and inheritance. Tiziani Bachetti (Italy)• Hints on potential future treatments from other homopolymers associated diseases K. Fischbeck (USA)• Desogestrel. Facts and hope. Thomas Similowski (France)• Keynotes on basic sessions. Isabella Ceccherini (Italy)
19:00-21:00	Gala dinner

SATURDAY APRIL 14, 2012

8:30 -9:30

Session 4. CHS: CCHS and the Others

Chairing: **Andreas Pflieger** (Austria)

- **Putting the Central Hypoventilation Syndromes All Together. Debra Weese-Mayer** (USA)

(30 min)

Visit of Posters & Exhibition (Facilitators: **Angeles Garcia-Teresa** (Spain), **Raquel Porto** (Spain)). Coffee break

10:00-12:00

Session 5. CHS Management.

Chairing : **Giancarlo Ottonello** (Italy)

- **What's new in Ventilatory Support? Matthias Frerick** (Germany)
- **Criteria for Cardiac Pacing in the general population. Katarzyna Bieganowska** (Poland)
- **Round Table: Which Mode of Ventilatory Support for Whom? Whom and when to Cardiac Pace? Can Follow-up be Tailored to Each Patient?** **Giancarlo Ottonello** (Italy)

(1h00)

Visit of Posters & Exhibition. Lunch

13:00-14:00

Session 6. Oral presentations of Abstracts.

Chairing: **Irena Senecic** (Croatia) and **David Kilner** (United Kingdom)

- **In vitro drug treatments reduce the deleterious effects of aggregates containing polyAla expanded PHOX2B proteins.** E. Di Zanni, T. Bachetti, P. Bocca, et al. (Italy) – abstract A4
- **Ocular Anomalies Correlated With Genotype In CCHS.** D. Brémond-Gignac, J. Amiel, H. Trang (France) –abstract B5
- **Bronchoscopic findings and interventions in patients with long-term tracheostomy.** A. Pflieger, E. Eber (Austria) – abstract B10
- **The nationwide epidemiological study of CCHS based on the “Research into Treatment for Specific Child Chronic Diseases” database in Japan.** K. Muramatsu, R. Muramatsu, N. Sawaura, et al (Japan) –abstract B11
- **Diaphragm Pacing in CCHS. The Swedish experience.** M. Winderud, P. Nilsson, A. Jonzon, R. Sindelar (Sweden) –abstract C19
- **Surviving the Night? Independent Living For CCHS Young Adults.** S. S Kun, M. Vanderlaan, T G Keens (USA) – abstract D26
- **A novel alarm device for CCHS patients ventilated during sleep.** F. Brenna, A. Carcano, M. Cavalleri, et al (Italy) – abstract C16
- **Development of a versatile ventilator alarm haptic transmitter to improve safety and comfort of Ondine patients, their parents and their caregivers.** T. Similowski, N. Franckhauser, J.Gonzalez-Bermejo, et al (France) – abstract C17
- **Medical Experience with a Holiday camp in the Swiss Alps for CHS-Patients.** A. Wimmersberger, J. Seiler, D.H. Kinderspitex (Switzerland) F. Morandi et al (Italy) – abstract D27

14:00-16:00

Session 7. Bundles of Care.

Chairing: **Martin Samuels** (United Kingdom)

- **Care Packages across the World (including transition and adult care).**
 - **Kazuhiro Muramatsu** (Japan, Asia)
 - **Vivian Leske** (Argentina, South America)
 - **Iris Perez** (USA, North America)
 - **Marek Migdal** (Poland, Europe)
- **Round Table: How to organize care of CHS, such a rare disease?** **Martin Samuels** (United Kingdom)

(30 min)

Visit of Posters & Exhibition. Coffee break.

16:30-18:00

Session 8. Living with CHS

Chairing: **Mary Vanderlaan** (USA)

- **Life as an Adult. Linda Middleton** (United Kingdom)
- **Impact of CHS on Families. Francesco Morandi** (Italy)

Closing words

- **Central Hypoventilation. What is next?** **Giancarlo Ottonello** (Italy)
- **Future of the CHS Registry and of the CHS Network.** **Ha Trang** (France)

SUNDAY APRIL 15, 2012

8:30 - 11:00

Session 9. Families Session.

Chairing : **Eva Brandenbusch** (Germany)

- **Current Research in CCHS/CHS. An Informal Discussion with Researchers.** **Eva Brandenbusch** (Germany)
- **Family Break-out Sessions**
 - **1. The Young CCHS/CHS Child to age 15: Homecare, School and Lifestyle Issues.** **Alessandro Carcano** (Italy), **Mary Vanderlaan** (USA)
 - **2. Preparing For and Achieving Adult Independence: CCHS/CHS Patients age 15 +.** **Sheila Kun** (USA)



Fourth International Conference on Primary Central Hypoventilation **Science & Life of CHS**



Warsaw, Poland, April 13-15, 2012
Hyatt Regency Warsaw

23 Belwederska Avenue, Warsaw, Poland 00-761

Tel: +48 22 558 1234 Fax: +48 22 558 1235

SPECIAL EVENTS

THURSDAY APRIL 11, 2012

19:00-21:00 **Pre-conference Dinner**
Hyatt Regency Hotel
Q-Club Restaurant

FRIDAY APRIL 12, 2012

19:00-21:00 **Gala Dinner**
Hyatt Regency Hotel
Ballroom

SATURDAY APRIL 13, 2012

18:30 **Guided Tour in Warsaw**
Meeting place : Reception Hall of the Hyatt Regency Hotel

PRACTICAL INFO

**ANY MEDICAL ADVICE ? ANY MEDICAL EMERGENCY ?
ANY ASSISTANCE FOR YOUR CHILD AT THE PLAYGROUND ?**

Emergency Call **112**

Doctors on site Call for advice Dr **Jolanta Kolodziejczyk** and Dr **Anna Gosiewska** are kindly available during conference times
+(48) 603 196 927

PICU Children's Memorial Health Institute
Al. Dzieci Polskich 20
04-730 Warszawa, Poland

Adult ICU