



**5<sup>th</sup> INTERNATIONAL SCIENTIFIC CONGRESS  
ALSTRÖM SYNDROME  
October 17-18, 2008  
Padua – Venice, Italy**



**Organizing Committee:** Giovanni Federspil, Pietro Maffei, Jan D. Marshall, Giovanni B. Pozzan, Cesare Scandellari, Nicola Sicolo, Roberto Vettor

**Scientific Committee:** Sebastian Beck, Cathy Carey, Gayle B Collin, Pietro Maffei, Jan D Marshall, Gabriella Milan, Jürgen K. Naggert, Richard Paisey, Roberto Vettor

**Friday, October 17th**

***Padua University, Archivio Antico del Bo, Via 8 Febbraio 2, Padua***

08.30 **Opening Ceremony**  
Cesare Scandellari, Giovanni Federspil, Nicola Sicolo

**OVERVIEW: PHYSICIAN, PARENT, AND RESEARCH PERSPECTIVES**

*Chairman: Phillip Beales, UK*

09.00 **Childhood obesity**  
Claudio Maffeis, University of Verona, Italy

09.30 **Management of Pediatric Patients with Alström Syndrome**  
Cristina Mihai, Spitalul Clinic de Urgenta Constanta, Romania

10.00 **New and emerging phenotypes in Alström Syndrome**  
Jan D. Marshall, The Jackson Laboratory, Bar Harbor, ME USA

10.30 **The Patient & Family Perspective of Living with Alström Syndrome**  
Sandra M. Hubbard-LeBlanc, Alström Syndrome International

11.00-11.30 *Coffee Break*

**CLASSIFICATION AND DIAGNOSTIC ISSUES**

*Chairman: Jan D Marshall, USA*

11.30 **The ciliopathies: a unifying genetic concept for diverse clinical phenotypes**  
Nicolas Katsanis, Johns Hopkins University, USA

12.00 **Genetic testing and new diagnostic criteria of Alström Syndrome**  
Sebastian Beck, Kantonsspital Münsterlingen, Switzerland

12.30 **Microarray Resequencing Chip Application for Detection of Mutations in Rare Inherited Metabolic Diseases**  
R. Köksal Özgül, Hacettepe University, Ankara, Turkey

13.00-14.30 *Lunch*

**DISEASE MANAGEMENT OVERVIEW AND ROUND TABLE**

*Chairman: Roberto Vettor, Italy*

14.30 **Hormonal issues**  
Pietro Maffei, Padua University, Italy

14.45 **Insulin resistance and diabetes in Alstrom syndrome**  
Claudio Pagano, Padua University, Italy

15.00 **Kidney disorders**  
Francesco Scolari, University of Brescia, Italy

15.15 **Cardiac consequences of Alström Syndrome: more questions than answers?**  
Cathy Carey, Torbay Hospital, UK

15.30 **Heart-lung transplantation in a patient with Alström Syndrome**  
Heidi Görler, Hanover Medical School, Germany

15.45 **Interactive Discussion**

16.30-17.00 *Coffee Break*

### **SENSORINEURAL DISORDERS**

*Chairman: Rod Bronson, USA*

17.00 **Electrophysiology and varying degrees of visual function in different phenotypes of Alström Syndrome**  
Sten Andréasson, University of Lund, Sweden

17.30 **Is epilepsy and sleep disordered breathing a contributing factor for learning disabilities in Alström Syndrome?**  
Oswald Hasselmann, Ostschweizer Kinderspital, St. Gallen, Switzerland

18.00 **Understanding developmental profiles that may be present in individuals with Alström Syndrome.**  
Sarah E. Shea, IWK Children's Medical Center, Dalhousie University, Canada

**Saturday, October 18<sup>th</sup>**  
***Istituto Veneto di Scienze, Lettere ed Arti,***  
***Palazzo Cavalli Franchetti, Campo S. Stefano 2945, Venice***

09.15-09.30 **Opening Remarks**  
Caesar Scandellari, Giovanni B. Pozzan

### **FUNCTION OF ALMS1 and ANIMAL MODELS**

*Chairman: Nicolas Katsanis, USA*

09.30 **Use of mouse models to elucidate ALMS1 function**  
Jürgen K. Naggert, The Jackson Laboratory, USA

10.00 **Update on ALMS1 localisation and the search for binding partners**  
Tom Hearn, University Southampton, UK

10.30 **Genotype-phenotype correlations in Alström Syndrome**  
Gayle B. Collin, The Jackson Laboratory, USA

11.00-11.30 *Coffee break*

### **BASIC INVESTIGATION – SECTION 1**

*Chairman: Jürgen K. Naggert, USA*

11.30 **Fibroblast cultures of ALMS patients: from expression profiling to functional studies**  
Gabriella Milan, Padua University, Italy

12.00 **Calcium signalling and cytoskeleton abnormalities**  
Rosario Rizzuto, University of Ferrara, Italy

12.30 **Identification of cytoplasmic material in ALMS neurons, and review of progress in pre-clinical studies for treatment of fibrosis in ALMS**  
Roderick T. Bronson, Harvard Medical School, The Jackson Laboratory, USA

13.00 – 14.30 *Lunch*

### **BASIC INVESTIGATION – SECTION 2**

*Chairman: Gayle B. Collin, USA*

- 14.30 **Alms1 influences on adipose tissue. Studies in Alms1<sup>-/-</sup> mice and in 3T3-L1 cells**  
Francesca Favaretto, Padua University, Italy
- 15.00 **Alström and Bardet-Biedl syndromes; common etiologies, phenotypes and shared therapeutic goals**  
Phillip Beales, Institute of Child Health/Great Ormond Street Hospital for Children, UK
- 15.30-16.30 **Oral Communications**  
*Chairman: Sebastian Beck, Switzerland*
- 16.30 **Closing remarks**  
Cesare Scandellari, Giovanni Federspil, Nicola Siculo