5th 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, Catherine Carey, Gayle B Collin, Pietro Maffei, Jan D Marshall, Gabriella Milan, Jürgen K. Naggert, Richard Paisey, Roberto Vettor

Friday, October 17th – Padua – Archivo Antico del Bo

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

OVERVIEW: PHYSICIAN and PARENTS PERSPECTIVES
Chairman: Jan D. Marshall

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

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

10.00 **Management of pediatric patients with Alström Syndrome**
Cristina Mihai, Spitalul Clinic de Urgenta Constanta, Romania

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 ISSUES
Chairman: Phillip Beales

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 **Emerging, new and borderline phenotypes of Alström Syndrome**
Jan D. Marshall, The Jackson Laboratory, USA

13.00-14.15 Lunch

DISEASE MANAGEMENT OVERVIEW AND ROUND TABLE
Chairman: Roberto Vettor

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

14.30 **Insulin resitance and diabetes in Alstrom syndrome**
Claudio Pagano, Padua Univesity, Italy

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

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 **Pancreatic and kidney transplantation in Alström Syndrome**
Argiris Asderakis, University Hospital of Wales, UK

16.00 **Interactive Discussion**

16.30-17.00 Coffee Break

SENSORINEURAL DISORDERS
Chairman: Richard Paisey

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

17.30 **Hearing and balance disorders in Alström Syndrome: diagnosis and therapy**
Claes Möller, The Swedish Institute for Disability Research, Örebro University Hospital, Sweden

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 18th – Venice – Istituto Veneto di Scienze, Lettere ed Arti

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

FUNCTION OF ALMS1 and ANIMAL MODELS
Chairman: Nicolas Katsanis

09.30 **Overview of *Alms1* mouse models**
Jürgen K. Naggert, The Jackson Laboratory, USA

10.00 **Update on *ALMS1* and basal bodies**
Tom Hearn, University Southampton, UK

10.30 **Update on expression studies of *Alms1***
Gayle B. Collin, The Jackson Laboratory, USA

11.00 Coffee break

BASIC INVESTIGATION – SECTION 1
Chairman: Jürgen K. Naggert

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 **Would antifibrosis drugs help Alström syndrome patients?**
Roderick T. Bronson, Harvard Medical School, The Jackson Laboratory, USA

13.00 –14.30 Lunch

BASIC INVESTIGATION – SECTION 2
Chairman: Gayle B. Collin

14.30 **Alms1 influences on adipose tissue. Studies in Alms1-/- 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 **Oral Communications**

16.30 **Closing remarks**
Cesare Scandellari, Roberto Vettor