



ISTITUTO DI RICOVERO E CURA  
A CARATTERE SCIENTIFICO

**WORKSHOP ON**

***ENDOCRINE INVOLVEMENT  
IN DEVELOPMENTAL SYNDROMES***

*Rome, April 21-22, 2008*

*Venue: The Workshop will take place in Rome, Italy  
at the "Pontificia Università Urbaniana"  
Via Urbano VIII, 16 – Rome*

*Dear Friends,*

*We are pleased to welcome you all to Rome to attend the Workshop on Endocrine Involvement in Developmental Syndromes. This Workshop is the follow-up of the previous Workshop on Congenital Endocrinopathies, held in Genoa last year. We will have the privilege of hosting outstanding experts, who have accepted our invitation to jointly discuss issues including genetics, endocrine system, and developmental syndromes. We are confident that the workshop will offer a precious occasion for a thorough discussion among endocrinologists, genetists, researchers, and clinicians with the aim of better understanding the pathophysiology of rare diseases.*

*We hope that the quality of the scientific programme, together with a pleasant social programme will make this workshop a successful event.*

*We look forward to seeing you all in Rome*

*Gian Franco Bottazzo*

*Marco Cappa*

*Under the Patronage of:*

**S.I.E.**        *Società Italiana di Endocrinologia (Italian Society of Endocrinology)*  
**S.I.E.D.P.**   *Società Italiana di Endocrinologia Pediatrica (I.S.P.E.D. Italian Society of Pediatric Endocrinology)*  
**A.M.E.**       *Associazione Medici Ospedalieri (Association of Hospital Endocrinologists)*

### **Honour Committee**

Giuseppe Profiti: President, “Bambino Gesù” Children’s Hospital  
Gian Franco Bottazzo: Scientific Director, “Bambino Gesù” Children’s Hospital  
Tommaso Langiano: Medical Director, “Bambino Gesù” Children’s Hospital

### **Scientific Planning Committee**

Serge Amselem, Créteil, France  
Gian Franco Bottazzo, Rome, Italy  
Marco Cappa, Rome, Italy  
Steven D. Chernausek, Oklahoma, USA  
George Chrousos, Athens, Greece  
Stefano Cianfarani, Rome, Italy  
Lucia Ghizzoni, Parma, Italy  
Sandro Loche, Cagliari, Italy  
Renata Lorini, Genoa, Italy  
Mohamad Maghnie, Genoa, Italy  
Martin O. Savage, London, UK  
Alberto G. Ugazio, Rome, Italy

### **Local Scientific Secretariat**

Carla Bizzarri  
Armando Grossi  
Paolo Ciampalini  
Mariacristina Digilio  
Romana Marini  
Patrizia Patera  
Riccardo Schiaffini  
Grazia Maria Ubertini

### **Local Organizing Committee**

Gian Franco Bottazzo (Chair)  
Marco Cappa (co-Chair)  
Paola Cambiaso  
*Bambino Gesù Children’s Hospital, IRCCS, Rome, Italy*

### **Faculty**

S. Amselem (Créteil, France), E. Arbustini (Pavia, Italy), E. Bertini (Rome, Italy), R. Bocciardi (Genoa, Italy), G. F. Bottazzo (Rome, Italy), S. A. Camper (Ann Arbor, USA), M. Cappa (Rome, Italy), L. Cavallo (Bari, Italy), S. D. Chernausek (Oklahoma, USA), G. Chrousos (Athens, Greece), S. Cianfarani (Rome, Italy), A. Cicognani (Bologna, Italy), F. Cucca (Sassari, Italy), B. Dallapiccola (Rome, Italy), M. T. Dattani (London, UK), N. De Roux (Paris, France), C. L. Deal (Montreal, Canada), T. Eggermann (Aachen, Germany), S. Farooqi (Cambridge, UK), A. L. Fowden (Cambridge, UK), A. Garg (Dallas, USA), L. Ghizzoni (Parma, Italy), F. Guerrieri (Rome, Italy), O. Hiort (Luebeck, Germany), K. Hussain (London, UK), S. Loche (Cagliari, Italy), R. Lorini (Genoa, Italy), M. Maghnie (Genoa, Italy), K. M. Main (Copenhagen, Denmark), L. Mazzanti (Bologna, Italy), A. Musarò (Rome, Italy), G. Neri (Rome, Italy), A. M. Pasquino (Rome, Italy), P. Pozzilli (Rome, Italy), A. Riccio (Caserta, Italy), M. O. Savage (London, UK), C. Sultan (Montpelier, France), A. G. Ugazio (Rome, Italy), R. Weksberg (Toronto, Canada), O. Zuffardi (Pavia, Italy)

### **Local Organising Secretariat**

*Coordinamento Amministrativo Manifestazioni Scientifiche*  
*Sig.ra M.T. Caldaretti*  
*Piazza S. Onofrio, 4 – 00165 Rome*  
*Tel: +39.06.68592290 – Fax: + 39.06.68592443*  
*E-mail: congressi@opbg.net*

## **SCIENTIFIC PROGRAMME**

**Monday – April 21, 2008**

8.00 Registration

10.00 Welcome

### **Beyond Mendelian inheritance**

Chairs: *G.F. Bottazzo (Rome, Italy)*, *G. Chrousos (Athens, Greece)*

10.30 Imprinted genes and growth regulation

*R. Weksberg (Toronto, Canada)*

11.00 Genetic Imprinting: The paradigm of Prader-Willi and Angelman syndromes - *F. Gurrieri (Rome, Italy)*

11.30 Epimutations in human disease. The Beckwith-Wiedemann syndrome example - *A. Riccio (Caserta, Italy)*

12.00 Mitochondrial DNA and related syndromes

*E. Bertini (Rome, Italy)*

12.30 Lunch

### **Disruption of signalling processes and early origin of hypothalamic-pituitary diseases**

Chairs: *M.O. Savage (London, UK)*, *L. Cavallo (Bari, Italy)*

14.30 Neuroendocrine signals and molecular mechanisms of pituitary organogenesis

*S. A. Camper (Ann Arbor, USA)*

15.00 Roles of transcription factors in midline CNS and pituitary defects - *M.T. Dattani (London, UK)*

15.30 Developmental abnormalities of the posterior pituitary gland  
*M. Maghnie (Genoa, Italy)*

16.00 Genome-wide array: beyond mental retardation

*O. Zuffardi (Pavia, Italy)*

16.30 Coffee break

### **Growth and Growth Hormone/IGFI Signalling**

Chairs: *S. Loche (Cagliari, Italy)*, *S. Cianfarani (Rome, Italy)*

17.00 Hormonal regulation of fetal growth

*A. L. Fowden (Cambridge, UK)*

17.30 GH resistant syndromes and IGF-I resistant syndromes: phenotypic aspects – *M. O. Savage (London, UK)*

18.00 GH resistant syndromes: long-term follow up

*S. D. Chernausek (Oklahoma, USA)*

18.30 Developmental syndromes: GH deficiency and treatment

*L. Mazzanti (Bologna, Italy)*

19.00 Epigenetic regulation of growth: lessons from Silver-Russell syndrome *T. Eggemann (Aachen, Germany)*

## Thursday – April 22, 2008

8.30 Lecture

Chair: *A. G. Ugazio (Rome, Italy)*

### **Muscle involvement and IGF-I signalling in genetic disorders: new therapeutic approaches**

*A. Musarò (Rome, Italy)*

### **Overgrowth syndromes**

Chairs: *A. Cicognani (Bologna, Italy)*, *S. Amselem (Créteil, France)*

9.00 Classification - *G. Neri (Rome, Italy)*

9.30 The C-type natriuretic peptide and overgrowth – *R. Bocciardi (Genoa, Italy)*

10.00 Gs Alpha hyperactivity and overgrowth – *C. Sultan (Montpelier, France)*

10.30 Marfan syndrome - *E. Arbustini (Pavia, Italy)*

11.00 Coffee break

### **Diabetes and obesity**

Chairs: *S. D. Chernausek (Oklahoma, USA)*, *R. Lorini (Genoa, Italy)*

11.30 Generalized lipodystrophy - *A. Garg (Dallas, USA)*

12.00 Hyperinsulinism in developmental syndromes

*K. Hussain (London, UK)*

12.30 Extreme phenotype of human obesity and insulin resistance

*S. Farooqi (Cambridge, UK)*

13.00 Mixture of Type1 and Type2 diabetes, Double diabetes

*P. Pozzilli (Rome, Italy)*

13.30 Lunch

### **Genes and the hypothalamic- pituitary – gonadal axis**

Chairs: *L. Ghizzoni (Parma, Italy)*, *A. M. Pasquino (Rome, Italy)*

14.30 Novel genes regulating puberty onset and gonadal function

*N. de Roux (Paris, France)*

15.00 Intersex disorders in developmental syndromes

*O. Hiort (Luebeck, Germany)*

15.30 New concepts in testicular development and descent

*K. M. Main (Copenhagen, Denmark)*

16.00 Genetic basis of developmental anomalies in

Turner's syndrome - *C. L. Deal (Montreal, Canada)*

16.30 Coffee break

17.00 Lecture

Chair: *B. Dallapiccola (Rome, Italy)*

### **Genome-wide scan in complex genetic diseases**

*F. Cucca (Sassari, Italy)*

17.45 **Closing remarks: G. F. Bottazzo and M. Cappa**

## GENERAL INFORMATION

### REGISTRATION FEES

The registration fee is:

- €250.00 for physicians, surgeons, and biologists
- €100.00 for trainees

and it includes:

- participation in the scientific sessions
- congress kit – coffee breaks, lunches
- certificate of attendance

### MODALITIES OF PAYMENT

#### Bank transfer to:

“Ospedale Pediatrico Bambino Gesù”, Roma  
Banca di Roma, Ag 61, account n. 000000350036  
IBAN code: IT75N03002033610000003500/36; SWIFT Code: BROMITR1061  
or  
Credit Card (VISA, Carta Si, Diners, Mastercard)

### HOTEL ACCOMMODATION

To get detailed information regarding Hotel accommodation please contact the  
**Organizing Secretariat** at the following address: [congressi@opbg.net](mailto:congressi@opbg.net).

Please, book your hotel with convenient advance!! Rome is a very busy and touristic city.

*For further information, please contact:*  
*Local Organising Secretariat*  
*Coordinamento Amministrativo Manifestazioni Scientifiche*  
*Piazza S. Onofrio, 4 – 00165 Rome*  
*Tel: +39.06.68592290 – Fax: + 39.06.68592443*  
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**ENDOCRINE INVOLVEMENT IN DEVELOPMENTAL SYNDROMES**  
**Rome, April, 21<sup>th</sup>-22<sup>th</sup>, 2008**

*REGISTRATION FORM (Please write in capital letters)*

NAME AND SURNAME \_\_\_\_\_

INSTITUTE \_\_\_\_\_ POSITION \_\_\_\_\_

ADDRESS \_\_\_\_\_ CITY \_\_\_\_\_

ZIP \_\_\_\_\_ TEL \_\_\_\_\_ FAX \_\_\_\_\_ E-MAIL \_\_\_\_\_

**MODALITIES OF PAYMENT:**

Bank transfer to Ospedale Bambino Gesù

Credit Card:       VISA       CARTA SI'       DINERS       MASTERCARD

Expiry date: \_\_\_\_\_ Card holder name: \_\_\_\_\_

Credit card N: \_\_\_\_\_ SIGNATURE \_\_\_\_\_

**TO BE SENT BY FAX TO: +39-06-68.59.24.43**

**Personal data privacy protection**

I acknowledge that I have been informed by the Bambino Gesù Hospital on the content of art. 13 of Legislative Decree n. 196/2003 about privacy protection. I give my consent for the treatment of my personal data for the purpose indicated by the notice, and limited to what is provided by Law.

Your personal information will also be used to keep you informed about Bambino Gesù Children's Hospital's congress activities and initiatives via newsletters and briefing notes.

DATE \_\_\_\_\_ SIGNATURE \_\_\_\_\_

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INFORMATION ON PERSONAL DATA PROCESSING CARRIED OUT BY THE BAMBINO GESÙ CHILDREN'S HOSPITAL, BASED ON WHAT PROVIDED IN ART. 13 OF LEGISLATIVE DECREE N. 196/2003 CONCERNING PERSONAL DATA PROTECTION AND PRIVACY.

The Bambino Gesù Children's Hospital would like to inform you that legislative Decree N. 196 of June the 30th ("Code about personal data protection") sets a series of regulations to assure personal data protection.

The Hospital, in accordance with the mentioned decree, will process all personal data you provide upon registration to the course, only with the purpose of managing all aspects (accounting, didactic, etc.) of the relation with you.

Based on art. 13 of the above mentioned decree, the Hospital is bound to provide you with the following information:

The data you provide will be handled by persons expressly in charge (Hospital employees or collaborators). Data will be disclosed to:

- other individuals, in accordance with the provisions in force;
- other individuals for the supply of services of various kinds (accounting mailing, bank services, etc.)
- only upon specific request, to the judicial authority;

These data will not be diffused.

Data subjects will be entitled to exercise their rights in compliance with what provided in art 7 of the above mentioned legislative decree, i.e. they will be entitled to access their own data, to request their updating, amendment, integration, or deletion, if data are incomplete, erroneous, or have been collected against the law. Moreover, data subjects will be entitled to prevent the processing of their own data, for legitimate reasons. Such requests should be addressed to "Ospedale Pediatrico Bambino Gesù, Coordinamento Amministrativo Manifestazioni Scientifiche, Piazza S. Onofrio 4 – 00165 Roma.