



Ospedale Pediatrico
Bambino Gesù

ISTITUTO DI RICOVERO E CURA
A CARATTERE SCIENTIFICO

DELETION 22q11.2

Third International Meeting

Rome, Italy
June 7-8, 2002

Pontificia Università Urbaniana
Auditorium « Giovanni Paolo II »



del(22)

SCIENTIFIC PROGRAMME

Friday, June 7, 2002

8:00 Conference Registration

8:30 Welcome to participants
Francesco Silvano,
Gian Franco Bottazzo (Rome)

Introduction
Bruno Marino, Alberto G. Ugazio,
Bruno Dallapiccola (Rome)

8:45 **Round Table: Historical Overview
and Clinical Findings**
Angelo DiGeorge (Philadelphia)
Robert Shprintzen (Syracuse, NY)
Kazuo Momma (Tokyo)
John Opitz (Salt Lake City)
Donna McDonald-McGinn (Philadelphia)

10:15 **Immunodeficiency**
Chairmen: *Franco Pandolfi,*
Paolo Rossi (Rome)

Immunodeficiency: clinical aspects
Solveig Oskarsdottir (Goteborg)

New insights in immunodeficiency
Kathleen Sullivan (Philadelphia)

11:00 Break and Poster Exhibition

11:30 **Palatal / Craniofacial Anomalies**
Chairmen: *Robert Shprintzen*
(Syracuse, NY),
Ferruccio De Stefano (Rome)

Palatal anomalies
Robert Shprintzen (Syracuse, NY)

Craniofacial anomalies
Nicole Philip (Marseille)

Oral communications

Speech outcome after cleft palate repair in patients with a chromosome 22q11.2 deletion

Kirschner, Solot, McDonald-McGinn, Randall, Zackai, La Rossa (Philadelphia)

Aberrant carotid arteries associated with 22q11.2 deletions: influence on surgical management of VPI

Kirschner, Solot, Slemp, McDonald-McGinn, Emanuel, Zackai, La Rossa (Philadelphia)

12:35 Feeding / Growth

Chairmen: *Manuel A. Castello, Marcello Orzalesi (Rome)*

Feeding anomalies

Donna McDonald-McGinn (Philadelphia)

Growth

M. Cristina Digilio (Rome)

13:15 Lunch and Poster Exhibition

14:30 Cardiac Anomalies

Chairmen: *Kazuo Momma (Tokyo), Giuseppe De Simone (Rome)*

Neural crest

Adriana C. Gittenberger-de Groot (Leiden)

Types – Clinical

Elizabeth Goldmuntz (Philadelphia)

Surgery

Roberto Di Donato (Rome)

Follow up

Kazuo Momma (Tokyo)

Oral communications

Can we predict 22q11 status of fetuses with tetralogy of Fallot?

Boudjemline, Fermont, Le Bidois, Villain, Sidi, Bonnet (Paris)

⑥ Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: a North - Italian collaborative study
Memo, Bottigelli, Capelli, Cocchi, Donadio, Garavelli, Magnani, Mainini, Selicorni (Treviso, Milano, Bologna, Parma, Reggio Emilia)

⑥ Phenotypic variability in patients with conotruncal diseases and del 22q11.2
Prandstraller, Cocchi, Capelli, Canzi, Mazzoni, Capucci, Galdi, Trabanelli, Baroncini, Calzolari, Picchio, Salvioli (Bologna, Imola, Ferrara)

Prenatal diagnosis of the 22q11.2 deletion by routine fetal ultrasonographic examination across Europe
Stoll, Clementi and EUROSCAN Study Group (Strasbourg and Padova)

Saturday, June 8, 2002

8:30 Aspects of Development (1)

Chairmen: *Ann Swillen (Leuven), Stefano Vicari (Rome)*

Neuropsychological findings in children
Donna McDonald-Mc Ginn (Philadelphia)

Behavioral phenotype of children
Ann Swillen (Leuven)

Neuropsychological and behavioral phenotype findings in adults
Anne Bassett, Eva Chow (Toronto)

Holistic molecular genetic medicine in del 22q11.2
Yuriko Tatsuguchi (Tokyo)

Oral communications (Language)

Language and brain function in children with deletion 22q11.2 (del22q11)
Eliez, Liron, Glaser, Reiss (Geneva)

Impact of parental origin on language abilities in children with deletion 22q11.2
Glaser, Mumme, Blasey, Antonarakis, Reiss, Eliez (Geneva)

A study of verbal expression in a group of children with the 22q11 deletion syndrome
Persson, Johansson, Oskarsdottir, Soderpalm (Goteborg)

Longitudinal study of language development in the 22q deletion syndrome
Solot, Gerdes, Tonnesen, Miao, McDonald-McGinn, Zackai, Wang (Philadelphia)

Efficacy of use of sign language with preschool children with 22q11.2 deletion
Solot, Gerdes, Wang, McDonald-McGinn, Zackai (Philadelphia)

10:30 Break and Poster Exhibition

10:50 **Aspects of Development (2)**

Oral communications (Neuropsychology)

Brain anatomy in psychotic and non-psychotic adults with chromosome 22q11 deletion
van Amelsvoort, Daly, Robertson, Henry, Owen, Murphy, Murphy (London)

Identifying risk factors for schizophrenia in children with deletion 22q11.2 syndrome
Kates, Pearlson (Baltimore)

Frequency of psychiatric disorders in children with deletion 22q11.2 compared to cognitively matched controls
Feinstein, Blasey, Reiss, Eliez (Geneva)

11:15 **Ear-Audiological-Ophthalmological Anomalies / Del22 Related Syndromes**

Chairmen: *Karlene Coleman (Atlanta), Pierpaolo Mastroiacovo (Rome)*

Atlanta July 23-25, 2002

Ear-Audiological-Ophthalmological
Robert Shprintzen (Syracuse, NY)

Del 22 Related Syndromes
M. Cristina Digilio (Rome)

12:00 **Round Table "Parent Support Group"**

Chairmen: Donna McDonald-McGinn
(Philadelphia)

✓ Karlene Coleman (Atlanta)

✓ Robert Shprintzen (Syracuse, NY)

✓ Solveig Oskarsdottir, Carina Hvalstedt
(Goteborg)

✓ Nicole Philip (Marseille)

✓ Ann Swillen (Leuven)

13:00 Lunch and Poster Exhibition

14:15 **Molecular Genetics**

Chairmen: Bruno Dallapiccola (Rome),
Peter Scambler (London)

Molecular aspects

Beverly Emanuel (Philadelphia)

The role of TBX1

Peter Scambler (London)

Animal Models

Antonio Baldini (Houston)

New perspectives

Giuseppe Novelli (Rome)

Oral communications

DGCR6, another DiGeorge gene that is
involved in cardiovascular development
Hierck, Poelmann, Gittenberger-de Groot
(Leiden)

Altered TBX1 dosage causes
ear malformations in mice

Liao, Funke, Adams, Van De Water,
Epstein, Morrow (New York)

16:00 Closing Remarks

Bruno Dallapiccola (Rome)

L. Quere - May 1 - Family Day
3 activities + poster
- Newsletter - only 3 months