

THE FIFTH INTERNATIONAL 22Q11.2 DELETION SYNDROME CONFERENCE



MARSEILLES, FRANCE
JULY 10 - 11, 2006

SCIENTIFIC ORGANIZATION

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ESPACE ETHIQUE MÉDITERRANÉEN
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FINAL PROGRAMME / PROGRAMME FINAL

www.22q11deletion.mcocongres.com

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Scientific Programme

DAY 1: MONDAY, JULY 10, 2006

8:15 am Welcome and Introduction. **Nicole Philip**

> BASIC SCIENCE RESEARCH

8:30 am μ Mouse models of 22q11 deletion. **Bernice Morrow**

9:05 am μ Behavioral disorders in mouse and humans are linked to Tbx1 haploinsufficiency. **Elizabeth Lindsay**

9:40 am μ Discussion

9:50 am μ The role of TBX1 in myogenic progenitor cells in pharyngeal mesoderm. **Robert Kelly**

10:00 am μ Microarray analysis of 22q11 genes during mouse development. **Francesca Amati**

10:10 am μ Microarray analysis to identify downstream effects of Tbx1 mutation. **Peter Scambler**

10:20 am μ Discussion

10:30 am **COFFEE BREAK**

> 22Q11.2 DELETIONS AND COPY NUMBER CHANGES

10:45 am μ Cytogenetic mechanisms of the deletion. **Beverly Emanuel**

10:55 am μ Multiplex Ligation-dependent Probe Amplification (MLPA) in patients with 22q11.2 deletion-like features. **Jacob Vorstman**

11:05 am μ Array CGH in patients with 22q11.2 deletion-like features. **Koen Devriendt**

11:15 am μ Non-deletion patients with "22q11 deletion syndrome". **Peter Scambler**

11:25 am μ Patients with smaller deletions. **Donna McDonald-McGinn**

11:35 am μ Another 22q11.2 microdeletion. **Nicole Philip**

11:45 am μ Discussion

12:00 am **LUNCH AND POSTER VIEWING** (authors present from 01:00 -01:30 pm)

> PSYCHIATRIC AND BEHAVIORAL ASPECTS OF THE 22Q11.2 DELETION ACROSS THE LIFESPAN

01:30 pm μ The 22q11.2 deletion in children: autism and psychosis. **Jacob Vorstman**

01:45 pm μ Neuropsychiatric and behavioural problems in 100 individuals with 22q11 deletion syndrome. **Lena Niklasson**

02:00 pm μ Psychiatric illness and outcome in adults with 22q11.2 Deletion Syndrome. **Anne Bassett**

02:30 pm μ Discussion

> BRAIN AND COGNITION

02:45 pm μ Neurocognition and functioning in adults with 22q11.2 Deletion Syndrome. **Eva Chow**

03:05 pm μ Longitudinal data in cognition in del22q11. **Ann Swillen**

03:25 pm μ Cognition and brain in children with 22q11.2 deletion syndrome. **Kieran Murphy**

03:35 pm μ Discussion

03:50 pm μ Neural correlates of psychotic symptoms in 22q11 deletion syndrome. **Martin Debbané**

04:05 pm μ FISHing for the neurobiology of behaviour. **Kieran Murphy**

04:20 pm μ Is hyperproliferation associated with cognitive and/or psychotic symptoms in VCFS patients? **Thierry Frebourg**

04:30 pm μ Discussion

04:40 pm **COFFEE BREAK**

> LANGUAGE/SPEECH/OTORHINOLARYNGOLOGIC MANIFESTATIONS/FEEDING DISORDERS

04:55 pm μ Language and narrative abilities in the 22q11.2 Deletion. **Christina Persson**

05:05 pm μ Reflections on the assessment and management of VPI in patients with a 22q11.2 deletion. **Debbie Sell**

05:15 pm μ Videofluoroscopic analysis of swallowing difficulties in children with 22q11.2 microdeletion syndrome. **Alexandra Kemp**

05:25 pm μ High incidence of oto-rhino-laryngeal disorders, speech and mental delay related to neonatal brainstem dysfunction in children with 22q11 deletion syndrome. **Véronique Abadie**

05:35 pm μ Discussion

05:45 pm **ADJOURN DAY 1**

Scientific Programme

DAY 2 : TUESDAY, JULY 11, 2006

> CLINICAL CHALLENGES: IDENTIFICATION, ASSESSMENT AND TREATMENT

- 08:00 am n Morbidity and mortality associated with the 22q11.2 deletion. **Donna McDonald-McGinn**
- 08:20 am n The challenge of the palate – assessment, interventions, and outcomes. **Brian Sommerlad**
- 08:40 am n Unusual clinical findings. **Cristina Digilio**
- 09:00 am n Discussion
- 09:10 am n Facial characteristics in children with the 22q11.2 deletion - help with identification. **Solveig Oskarsdottir**
- 09:20 am n Computerized technology as an adjunct to the gestalt. **Peter Hammond**
- 09:30 am n Well care for teens with the 22q11.2 deletion. **Paula Goldenberg**
- 09:40 am n Multidisciplinary clinics in the setting of restricted funding. **Alex Habel**
- 09:50 am n Discussion

10:00 am COFFEE BREAK

- 10:30 am **LAUNCHING THE BREAKOUT – GOALS:**
establishing clinical practice guidelines, determining prevalence,
and increasing awareness of the syndrome
Anne Bassett, Nicole Philip, Donna McDonald-McGinn

- Ⓜ Immunology
- Ⓜ Endocrinology
- Ⓜ Cardiology
- Ⓜ General Pediatric Concerns

- Ⓜ Palatal Interventions / Speech / Language
- Ⓜ Gastroenterology / Feeding Disorders

- Ⓜ Psychology/Psychiatry /Child Development/
Behavior / Education
- Ⓜ Adult Screening

- Ⓜ Dedicated Clinics
- Ⓜ Prevalence Studies
- Ⓜ Continuing Education

GROUP 1

Led by:
**Bruno Marino, Solveig Oskarsdottir,
Fritz Beemer and Donna McDonald-McGinn**

GROUP 2

Led by:
**Brian Sommerlad, Debbie Sell,
Christina Persson and Cristina Digilio**

GROUP 3

Led by:
**Ann Swillen, Jacob Vorstman,
Peter Scambler and Anne Bassett**

GROUP 4

Led by:
**Alex Habel, Paula Goldenberg,
Koen Devriendt and Nicole Philip**

10:30 am LUNCH AND POSTER VIEWING (authors present from 01:30 - 2:00 pm)

> REASSEMBLE FOR GROUP PRESENTATIONS

- 02:00 pm n **Group I** - Discussion - 15 minutes
- 02:30 pm n **Group II** - Discussion - 15 minutes
- 03:00 pm n **Group III** - Discussion - 15 minutes
- 03:30 pm n **Group IV** - Discussion - 15 minutes

04:00 pm COFFEE BREAK

- 04:15 pm n What do the parents want and need? **Julie Wootton, Max Appeal**
- 04:40 pm n Discussion

> SUMMARY STATEMENTS

- 04:55 pm n **Group Leaders**
- 05:25 pm n **Closing Statements. Nicole Philip and Anne Bassett** - (Next meeting: Toronto in 2008)
- 05:45 pm **ADJOURN**