Scientific Programme

DAY 1: MONDAY, JULY 10, 2006

8:15 am  Welcome and Introduction, Nicole Philip

> BASIC SCIENCE RESEARCH

8:30 am  Mouse models of 22q11 deletion, Bernice Morrow
9:05 am  Behavioral disorders in mouse and humans are linked to Tbx1 haploinsufficiency, Elizabeth Lindsay
9:40 am  Discussion
9:50 am  The role of TBX1 in myogenic progenitor cells in pharyngeal mesoderm, Robert Kelly
10:00 am  Microarray analysis of 22q11 genes during mouse development, Francesca Amati
10:10 am  Microarray analysis to identify downstream effects of Tbx1 mutation, Peter Scambler
10:20 am  Discussion
10:30 am  COFFEE BREAK

> 22Q11.2 DELETIONS AND COPY NUMBER CHANGES

10:45 am  Cytogenetic mechanisms of the deletion, Beverly Emanuel
10:55 am  Multiplex Ligation-dependent Probe Amplification (MLPA) in patients with 22q11.2 deletion-like features, Jacob Vorstman
11:05 am  Array CGH in patients with 22q11.2 deletion-like features, Koen Devriendt
11:15 am  Non deletion patients with "22q11 deletion syndrome", Peter Scambler
11:25 am  Patients with smaller deletions, Donna McDonald-McGinn
11:35 am  Another 22q11.2 microdeletion, Nicole Philip
11:45 am  Discussion

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

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

01:30 pm  The 22q11.2 deletion in children: autism and psychosis, Jacob Vorstman
01:45 pm  Neuropsychiatric and behavioural problems in 100 individuals with 22q11 deletion syndrome, Lena Niklasson
02:00 pm  Psychiatric illness and outcome in adults with 22q11.2 Deletion Syndrome, Anne Bassett
02:30 pm  Discussion

> BRAIN AND COGNITION

02:45 pm  Neurocognition and functioning in adults with 22q11.2 Deletion Syndrome, Eva Chow
03:05 pm  Longitudinal data in cognition in del(22q11, Ann Swillen
03:25 pm  Cognition and brain in children with 22q11.2 deletion syndrome, Kieran Murphy
03:35 pm  Discussion
03:50 pm  Neural correlates of psychotic symptoms in 22q11 deletion syndrome, Martin Debbane
04:05 pm  fISHing for the neurobiology of behaviour, Kieran Murphy
04:20 pm  Is hyperprolinemia associated with cognitive and/or psychotic symptoms in VCFS patients?, Thierry Frebourg
04:30 pm  Discussion
04:40 pm  COFFEE BREAK

> LANGUAGE/SPEECH/OTORHINOLARYNGOLOGIC MANIFESTATIONS/FEEDING DISORDERS

04:55 pm  Language and narrative abilities in the 22q11.2 Deletion, Christina Persson
05:05 pm  Reflections on the assessment and management of VPI in patients with a 22q11.2 deletion, Debbie Sell
05:15 pm  Videofluoroscopic analysis of swallowing difficulties in children with 22q11.2 microdeletion syndrome, Alexandra Kemp
05:25 pm  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  Discussion

05:45 pm  ADJOURN DAY 1
Scientific Programme

DAY 2: TUESDAY, JULY 11, 2006

> CLINICAL CHALLENGES: IDENTIFICATION, ASSESSMENT AND TREATMENT

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

GROUP 3
Led by: Ann Swinnen, Jacob Vortman, 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  • Group I - Discussion - 15 minutes
02:30 pm  • Group II - Discussion - 15 minutes
03:00 pm  • Group III - Discussion - 15 minutes
03:30 pm  • Group IV - Discussion - 15 minutes

04:00 pm COFFEE BREAK

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

> SUMMARY STATEMENTS

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