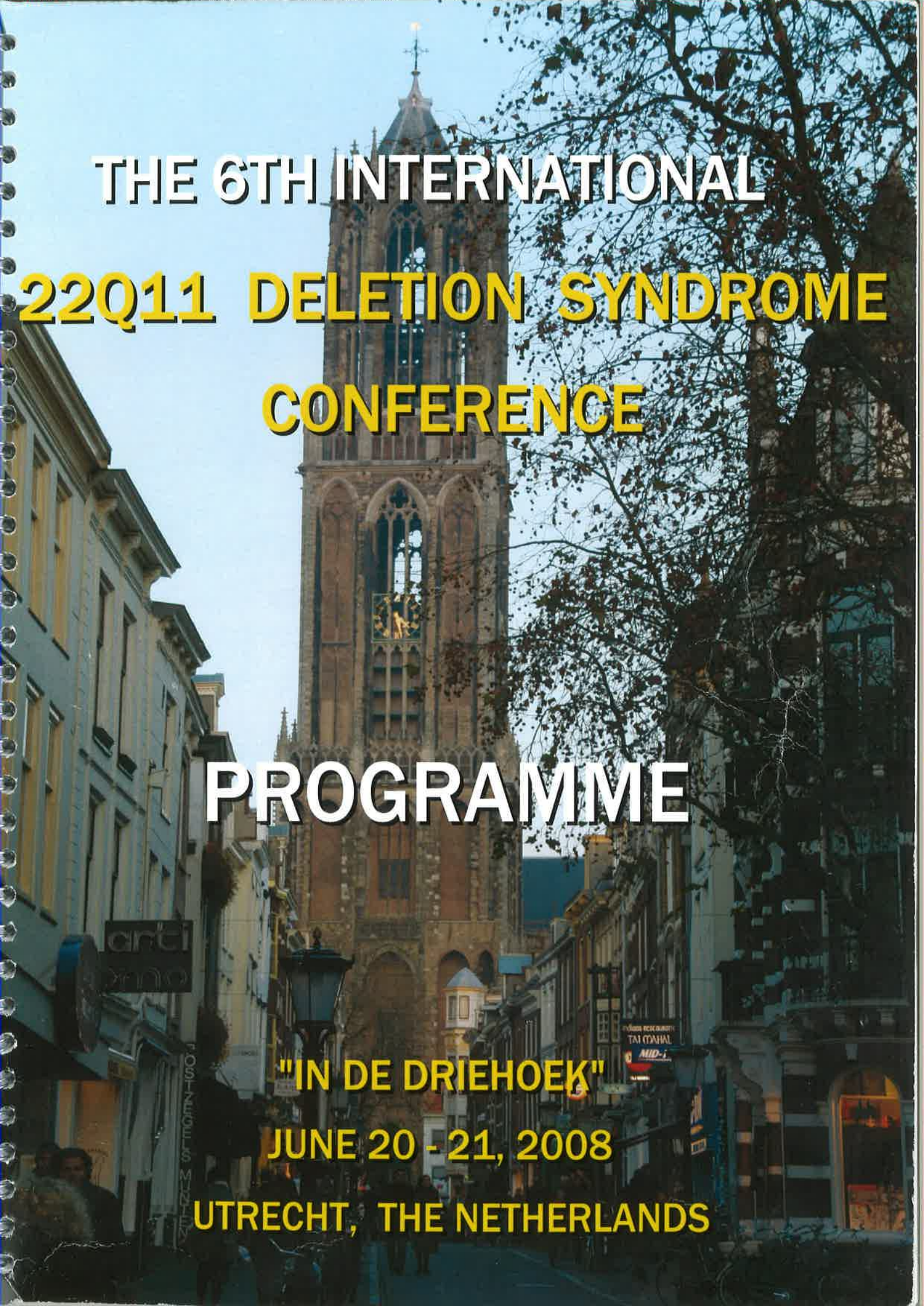

THE 6TH INTERNATIONAL

22Q11 DELETION SYNDROME CONFERENCE

PROGRAMME

"IN DE DRIEHOEK"

JUNE 20 - 21, 2008

UTRECHT, THE NETHERLANDS

Scambler: Tbx1 → Pitx2c; Tbx1 → 6Bx2 → Slit2; FGF8 (Retinoid Signaling)

SCIENTIFIC PROGRAMME

Friday June 20

08.15am	**REGISTRATION**
09.00am	Welcome **– Frits A. Beemer**
09.05am	Opening words **– J.L.L. Kimpen MD PhD, Dean of the Medical Faculty, Vice-chairman Executive Board University Medical Center Utrecht**

GENERAL ASPECTS

Chairman: Koen Deviendt

09.15am	*Key note lecture*: A look at VCFS genetic research from the outside **– Han G. Brunner, Clinical geneticist. Head of the Department of Anthropogenetics, University Medical Centre Nijmegen, The Netherlands.**
10.00am	Prenatal diagnosis of 22q11 microdeletion **– Nicole Philip**
10.15am	Educational strategies for children with 22q11 syndrome **– Donna Cutler-Landsman**
10.30am	Searching for evidence for primary care guidelines for 22q11ds: methods and results of a systematic review **– Paula Goldenberg**
10.45am	Discussion
11.05am	Coffee/tea break and poster viewing

CYTOGENETIC AND MOLECULAR GENETIC ASPECTS

Chairman: Jacob Vorstman

11.30am	*Key note lecture:* Differential gene expression in the 22q11.2 deletion syndrome **- Prof. Peter Scambler, Geneticist, Molecular Medicine Unit. Institute of Child Health London, UK**
12.15pm	Atypical 22q11.2 deletions – possible genotype-phenotype correlations **– Donna McDonald-McGinn**
12.30pm	Detailed analysis of 22q11.2 with a high density mlpa probe set **– Beverly Emanuel**
12.45pm	High resolution technology reveals additional layers of genomic complexity in 22q11ds **– Alexander Urban**
01.00pm	Identification of genetic modifiers of 22q11DS in humans and mice **– Bernice Morrow**
01.15pm	Discussion
01.35pm	Lunch and poster viewing

PSYCHIATRIC ASPECTS
Chairman: Anne Bassett

02.15pm	*Key note lecture:* Behaviour and Psychopathology Associated with Specific Genetic Disorders **- Patrick Bolton, Professor of Child & Adolescent Psychiatry, Institute of Psychiatry, University of London, UK.**
03.00pm	Association of the pik4ca schizophrenia-susceptibility gene in adults with the 22q11.2 deletion syndrome **– Jacob Vorstman**
03.15pm	22q11.2 deletion syndrome: dementia and low intellectual functioning **– Laurens Evers**
03.30pm	Profile of psychotic symptoms in 22q11DS **– Stephan Eliez**
03.45pm	Discussion
04.05pm	Coffee/tea break and poster viewing

CLINICAL ASPECTS I
Chairman: Gerben Sinnema

04.30pm	Intellectual abilities and developmental delay in 3½ year old children with 22q11DS **– Sasja Duijff**
04.45pm	Prevalence and severity of attentional problems in 22q11ds children **– Eva Chow**
05.00pm	The motor performance of 22q11del children and age- and IQ-matched controls **- Katrijn van Aken**
05.15pm	Behavior in young children with 22q11DS **– Petra Klaassen**
05.30pm	Discussion
07:00pm	Welcome at the 'Academiegebouw' (University building); drinks
08.00pm	(optional) social dinner (Senate's room)

Saturday June 21

IMAGING AND METABOLISM
Chairman: Therese van Amelsvoort

08.30am	Key note lecture: 22q11 genes interactions for risk and protection for schizophrenia and cognitive function **- Lucas Kempf, Clinical Research Fellow in the Genes, Cognition and Psychosis Program, National Institute of Mental Health, Bethesda, Maryland, USA**
09.15am	Disrupted dopaminergic neurotransmission in 22q11 deletion syndrome **– Erik Boot**
09.30am	Striatal d2 receptor binding in 22q11 deletion syndrome **– Erik Boot**
09.45am	Dynamic of cortial changes in 22q11 deletion syndrome: a cross-sectional and longitudinal MRI study **– Marie Schaer**
10.00am	White matter microstructure in children with velocardiofacial syndrome **- Frederick Sundram**

10.15am Discussion

10.35am Coffee/tea break and poster viewing

CLINICAL ASPECTS II
Chairman: Donna McDonald-McGinn

5 minutes

11.00am Cognitive development, psychosocial adjustment and needs in non-psychiatric adolescents with 22q11.2 DS
– **Ann Swillen** – Leuven

11.15am Social cognitive skills in young adults with 22q11.2 deletion syndrome
– **Linda Campbell** – Newcastle, Australia

11.30am Sudden and premature death in adults with 22q11.2 deletion syndrome
– **Anne Bassett** – Toronto

11.45am Discussion

12.05am Lunch and poster viewing
Optional: Presentation of the Consortium study by **Bernice Morrow**

CLINICAL ASPECTS III
Chairman: Donna McDonald-McGinn

→ reunite in session room picture + tour

01.00pm Clinical findings in neonates with deletion 22q11.2 (DiGeorge/velo-cardio-facial syndrome
– **Silvia Macerola**

01.15pm Treatment of VPI in patients with 22q11 deletion syndrome: the Utrecht experience
- **Aebele Mink van der Molen** – Utrecht

01.30pm Surgical management of velopharyngeal insufficiency in 22q11.2 deletion syndrome
- **Josine Widdershoven** – Utrecht

DUPLICATIONS
Chairman: Frits Beemer

02.05pm Phenotype-genotype correlations in two families with a duplication on 22q11.2
– **Emma van Daalen**

02.20pm Microduplication 22q11.2: clinical and molecular cytogenetic analysis of five patients
– **Marie-France Portnoï**

02.35pm Deletions, duplications and mutations in 22q11.2: clinical and molecular characterization of a de novo atypical duplication
– **Damian Heine-Suñer**

02:50pm Discussion

03.10pm Short break

INTERNATIONAL GUIDELINES

03.20pm Introduction
03.30pm General discussion

05.00pm End of the meeting; closing remarks