

Treatment As We Move Into A New Decade

The Seventh Biennial International 22q11.2 Deletion Syndrome
Conference

29th and 30th July 2010



One Step Beyond

10th Max Appeal Annual Conference

30th and 31st July 2010



22Q

The International 22q11.2
Deletion Syndrome Foundation, Inc.



JAMES TUDOR
FOUNDATION



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**“TREATMENT AS WE MOVE INTO A NEW DECADE”
THE SEVENTH BIENNIAL INTERNATIONAL 22q11.2 DELETION
SYNDROME CONFERENCE**

**RICOH CENTER, COVENTRY, ENGLAND
JULY 29 – 31, 2010**

PRELIMINARY PROGRAMME

Thursday, July 28th

- 8:00 am Conference Registration
- 8:45 am Welcome and Conference Opening
 – *Julie Wooten and Dinakanthe Kumararatne*
- 9:00 am Meeting Dedication and Presentation of Angelo DiGeorge Inaugural Medal
 – *Donna McDonald-McGinn, Bruno Marino, Julie Wooten and
 Christopher DiGeorge*

Session I – *TBX1* and Cardiovascular Development

- Chairs:** *Donna McDonald-McGinn and Bruno Marino*
- 9:15 am *Angelo DiGeorge Inaugural Lecture*
 Pathways Affected By Loss of *TBX1* – *Peter Scambler*
- 9:45 am Cardiac Stem Cell Homeostasis – *Antonio Baldini*
- 10:15 am Discussion
- 10:30 am **Coffee Break**

Session II - General Aspects 1

- Chairs:** *Peter Scambler and Antonio Baldini*
- 11:00 am Update on Cardiologic Associations, Treatments, and Outcomes - *Bruno Marino*
- 11:15 am Can 22q11.2 Deletions be Detected by Clinical Assessment of an Individual with
 Congenital Cardiac Malformation? – *P. Agreggaard*

- 11:19 am Asymmetric Crying Facies: Implications for the General Population – *Oksana Jackson*
- 11:26 am Ophthalmologic Findings: Why an Eye Exam is Important – *Brian Forbes*
- 11:33 am **Discussion**
- 11:43 am Scoliosis as a Major Complication in Adolescence – *John Dormans*
- 11:50 am “Safe and Sustainable” – Collaborative Cardiac Services – Implications for the United Kingdom and Beyond - *Julie Wooten*
- 12:00 pm **Current Results from the International Consortium:** Genetic Modifiers of Intracardiac Defects in Humans with a 22q11.2 Deletion – *Bernice Morrow*
- 12:15 pm **Discussion**
- 12:30 pm **Lunch**

Session III – General Aspects 2

Chairs: *Kathleen Sullivan and Bernice Morrow*

- 1:30 pm Approach to Endocrine Management: The South American and Asian Experiences – *Nancy Unanue (Chile) and Hiroshi Kawame (Japan)*
- 1:45 pm Somatic Growth and the Development of a Diagnosis Specific Growth Chart – *Alex Habel*
- 1:52 pm Growth Studies from the South of France – *Nicole Philip*
- 1:59 **Discussion**
- 2:09 pm Secondary Immunologic Consequences – *Kathleen Sullivan*
- 2:16 pm Immunologic Abnormalities in Adults – *Aron Hilmarsson*
- 2:23 pm Most Patients Are Not Tested For Immune Deficiency – A Survey in the West Midlands Region 2005 – 2009 – *S. Hackett*
- 2:27 pm Longitudinal Analysis of T Lymphocyte Subpopulations – *Torstein Overland*
- 2:31 pm Evaluation of the Immune Function in a Man with a Familial 22q11.2 Deletion at 35 Years of Age – *Ania Manson*
- 2:35 pm Spry1 and Spry2 Deletions Result in a Spectrum of Developmental Defects typical of the 22q11.2 deletion - *M. Albert Basson*
- 2:45 pm **Discussion**
- 3:00 pm **Afternoon Tea**

Session IV – General Aspects 3

Chairs: *Nicole Philip and Solveig Oskarsdottir*

- 3:00 pm Prevalence, “B to D’s” and Dups: Everything But the Kitchen Sink – *Donna McDonald-McGinn*
- 3:15 pm Unusual 22q11 Reciprocal Rearrangement in a Phenotypically Normal Mother of a Child with a 22q11.2 Deletion – *Luis Fernandez*
- 3:19 pm Results, Evaluation and Comparison of Different Methodological Approaches for the Study of the 22q11.2 Region – *D. Heine-Suner*
- 3:23 pm **Discussion**
- 3:33 pm Gastrointestinal Manifestations – *Maria Mascarenhas*
- 3:40 pm Feeding Problems: Optimizing Evaluation and Management, Can SAD & RP Help? – *Peggy Eicher*
- 3:47 pm Challenging Commonly Held Views of Feeding Difficulties in Infants and Children – *Alex Forsyth*
- 3:54 pm **Discussion**
- 4:04 pm Bleeding Manifestations – *Michelle Lambert*
- 4:11 pm Predicting 22q11.2 Deletion Syndrome: A Novel Method Using Routine Full Blood Count – *N. Naqvi*
- 4:18 pm **Discussion**
- 4:30 pm **Adjourn**
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Friday, July 30th

Session VI – Speech and Palate

Chairs: *Richard Kirschner and Brian Sommerlad*

- 8:00 am Speech Outcome Following Anatomical Muscle Dissection Velar Repair of the Submucous Cleft Palate – *Ali Ghanem*
- 8:07 am Velopharyngeal Dysfunction: A Longitudinal Study of Functional Outcome and Preoperative Prognostic Factors – *N.E. Spruijt*
- 8:14 am Speech Outcome: Is There an Optimal Age for Velopharyngeal Dysfunction Repair? – *Cindy Solot*
- 8:21 am Speech in Adults – *Cristina Persson*

- 8:28 am Candidate Gene Approach to Identify Modifiers of the Palatal Phenotype – *Justine Widdershoven*
- 8:32 am **Panel:**
 “Velopharyngeal Dysfunction Repair Before or after School Age - That is the Question” and “Are Pre-Operative MRA’s Really Necessary?” – *Richard Kirschner (7), Brian Sommerlad (7), Christina Persson, Debbie Sell, Cindy Solot, (whomever else you had in mind)*
- 9:00 am **Discussion**
- 9:15 am **Coffee Break**

Session VI – Cognitive Development, Language, and Neuro-Behavior 1

Chairs: *Anne Bassett and Ann Swillen*

- 9:45 am Developmental Trajectories of Fronto-Executive Functions – *S. A. Howley*
- 9:49 am Language of Emotion in Older Children and Autistic Spectrum Disorder – *A. Mayne*
- 9:53 am Autistic Spectrum Disorders and Gender Differences in Young People – *D. Robertson*
- 9:57 am Psychiatric Screens and Gender Differences in Children and Adolescents – *C.M. Murphy*
- 10:01 am Neuropsychological Characteristics of Children and Adolescents – *Clare Jacobson*
- 10:04 am Parental Perception of Sleep Behavior and Sleep Disorders in Children and Their Siblings - *Ann Swillen*
- 10:11 am **Discussion**
- 10:21 am Can Clustering Analyses be Used as a Tool to Identify Sub Groups and People with the Disorder? – *B. Sinderberry*
- 10:25 am Psychiatric and Behavioral Characteristics: An Irish Population – *S.E. Prasad*
- 10:32 am Developmental Aspects of Cognition and Psychiatric Symptoms in Youngsters: Preliminary Data From an Ongoing Longitudinal Study – *Jacob Vorstman*
- 10:39 am Mental Health from Childhood to Adult Age: A Prospective Longitudinal Study of 70 Individuals – *Ylva Liander Ankarcrona*
- 11:03 am Evolution and Predictive Value of Attenuated Negative Symptoms: A 3 Year Longitudinal Study – *Maude Schneider*

- 11:07 am **Discussion**
- 11:17 am Childhood Behavior Problems and Prodromal Symptoms in Schizotypal Personality Disorder – *D.I. Shapiro*
- 11:24 am Computerized Neuropsychological Battery with Children, Adolescents, and Young Adults - *Paula Goldenberg*
- 11:28 am Age-Dependent Alterations in Spatial Memory, Synaptic Plasticity, and Calcium Dynamics in a Mouse Model of 22q11.2 Deletion Syndrome – *Laurie Earls*
- 11:35 am *tbx1* Heterozygosity Impairs Hippocampus-Dependent Reference Memory in Mice – *Noboru Hiroi*
- 11:42 am Learning, and Memory Impairments in Mice Deleted for the DiGeorge/VCFS Syndrome Region – *Elizabeth Illingworth*
- 11:57 am **Discussion**
- 12:15 pm **Lunch**

Session VII – Neuro-Behavior 2, Imaging, Outcomes and Treatment

Chairs: *Jacob Vorstman and Tony Simon*

- 1:00 pm Brain, Behavior and Overcoming Challenges in Adults – *Anne Bassett*
- 1:07 pm Reality Monitoring Processing: An fMRI Investigation – *Annalaura Lagioia*
- 1:11 pm Posterior Brain Volumetric Reductions in Children – *Eva Chow*
- 1:14 pm Differential Activation of the Fusiform Gyrus by Category – *Bronwyn Glaser*
- 1:18 pm Neuroanatomical Predictors of Prodromal Symptoms of Psychosis: A 3 Year Follow-up – *Wendy Kates*
- 1:25 pm Can Multi-Modal Imaging Offer Unique Insight – *E. O’Hanlon*
- 1:29 pm **Discussion**
- 1:39 pm Early Intervention Strategies Specific for Delays in Emergence of Language – *Cynthia Solot*
- 1:43 pm Evidenced-Based Computerized Intervention Enhances Working Memory Skills - *Edward Moss*
- 1:50 pm Clinical Screening for Developmental Disabilities – *O. Ousley*
- 1:54 pm Setting the Stage for Success: Effective Strategies to Enhance Social Skills – *Barb Haas-Givler*

- 2:01 pm What's in a Name? Symptoms Versus Causes in the Diagnostic Age: Why Schools Frequently Don't Appreciate the Importance of the Underlying Etiology – *Brenda Finucane*
- 2:07 pm Visuospatial Cognitive Deficits - *Tony Simon*
- 2:22 pm Analysis of CNV's – *Beverly Emanuel*
- 2:30 pm **Discussion**
- 2:45 pm **Afternoon Tea**

Session VIII – Prenatal Diagnosis and Transition to Adulthood

Chairs: Deborah Driscoll and Nancy Unanue

- 3:15 pm Prenatal and Preimplantation Genetic Diagnosis – *Deborah Driscoll*
- 3:30 pm **Panel:**
 Transitions to Adulthood – *Anne Bassett, Deborah Driscoll, Brenda Finucane Nancy Unanue, Donna Cutler-Landsman, and Ed Moss*
- 4:00 pm **Discussion**

Session VIII – Multidisciplinary Clinics and International Collaborations

Chairs: Dinakanthe Kumararatne, Hiroshi Kawame and Alex Habel

- 4:15 pm Specialized Multi-Disciplinary Clinic – *Nicola Herberholz*
- 4:19 pm To Screen or Not to Screen: That is the Question – *Annique Hogan*
- 4:23 pm Report on UK Guidelines – *Alex Habel*
- 4:33 pm Summation on International Consortium Guidelines and “Where Do We Go From Here?” – *Anne Bassett and Donna McDonald-McGinn*
- 4:43 pm Establishing Standards for Clinical Centers of Excellence: Is it Worth the Effort? – *Marilyn Cohen*
- 4:53 pm Empowering Parents and Building Partnerships – *Michelle Breedlove*
- 5:00 pm **Discussion**
- 5:30 pm Closing Remarks: See You in 2012! – *Julie Wooten and ?*
- Adjourn**

Appeal!

by DiGeorge Syndrome/VCF5/22q11.2 deletion

Who are we?

What we do!



Max Appeal is run almost entirely by an active and dedicated team of volunteers and its trustees. It is funded by donations from families, organised fundraising events and other activities.

Support:

Max Appeal was established to offer support to families because this is a life changing event for everyone. Babies frequently require a lot of medical intervention and, when they get home, lots of extra support and dedication from their parents to help them grow into independent young people, and ultimately we hope, adults with families of their own.



Who was Max?

Max was born on 5th November 1998. He died of overwhelming septicaemia when he was four months old due to complex heart defects combined with severe immunity problems. Max was a resilient and beautiful baby who endured ceaseless pain during his short life.

Max Appeal is establishing a national protocol for the care of people with 22q11.2 deletion syndromes. This will materially improve the quality of life for all our families. (Cost: £50,000)

We hold an annual conference/ open day: past venues include Centre for Life in Newcastle Upon Tyne, National Space Centre in Leicester and Bristol Zoo Pavilion. (Cost: £10,000)

Our newsletter keeps parents and professionals up to date with information and has family stories to share. (Cost: £3,000)

We organise weekend breaks to provide children with exciting challenges and a unique opportunity to meet others with 22 Deletion. (Cost: £1,000)

We run a national freephone helpline: 0800 389 1049, which is manned by trained volunteers. (Cost: £500)

We make grants towards travel costs to parents who may live many hours drive from the hospital where their child is being treated. (Cost: £100)

We provide a comprehensive information pack and handbook for a family of a newly diagnosed person. (Cost: £10)



about us

What's Max Appeal all about?

ies affected by DiGeorge syndrome,

varied group of conditions. Babies and complex heart defects requiring surgery, low calcium which can cause dangerous arrhythmia which can be life threatening but usually resolved with time off school; speech and language abnormalities; and specific learning difficulties and adults develop schizophrenia or other problems.

me and Velo-Crdio-Facial syndrome: is caused by a '22q11.2 deletion' which is a sort of chromosome abnormality. 22q deletion affects as many as 1 in 10,000 people. It is the most common chromosome abnormality. As an inherited condition, it is a fault of the chromosome, not a phenomenon, or just plain bad luck!

Our Aims

MaxAppeal aims to improve the quality of life of children, young people and adults with 22q Deletion by raising awareness of this common chromosome defect and to bring about permanent improvements in the social and medical care of people with 22q deletion.

Think Big:

Max Appeal is a small charity but is the largest voluntary group for this condition in the world and helps hundreds of families every year. 22q Deletion is being more frequently recognised and diagnosed, it is essential that Max Appeal continues to grow to meet the needs of this increasing population of people.

National Protocol

Max Appeal is establishing a national protocol for the care of people with 22q11.2 deletion syndromes from birth to adulthood. This is will materially improve the quality of life for all our families in a real and practical way. We work continuously with educators, healthcare professionals and other charities.



See you next summer in Edinburgh

Details to follow in the autumn on

www.22qdeletion.com
and
www.maxappeal.org.uk

