The 10th Annual
International Meeting
of The Velo-Cardio-Facial Syndrome
Educational Foundation, Inc., and the
Fourth International Conference for
22q11.2 Deletions

July 23-25, 2004
Atlanta, Georgia
Sheraton Midtown Atlanta Hotel at Colony Square

Velo-Cardio-Facial Syndrome Educational
Foundation, Inc.

International 22q11.2 Deletion Group

Children's
Healthcare of Atlanta
Table of Contents/Agenda

I. Faculty ................................................................. 1

III. Paper Presentations ................................................. 7

IV. Poster Presentations ................................................ 11

V. Objectives .................................................................. 37

IV. Acknowledgements ..................................................... 39

V. Presentations

Friday, July 23, 2004
*All conference sessions in Ballroom North unless otherwise noted.

7:30 – 9:00  Registration (Grand Ballroom Foyer)
Continental breakfast (Ballroom South)
Options of either “Birds of a Feather” or open seating.
Note: “Birds of a Feather” seating, an opportunity to meet attendees with similar interests, is
open to both professionals and families. Common interest – group signs will be placed on
selected tables.

9:00 – 9:05  Welcome
Jay E. Berkelhamer, M.D., Children’s Healthcare of Atlanta

9:05 - 9:15  Opening Remarks and Welcome
Karen J. Golding-Kushner, Ph.D.
William T. Mahle, M.D.

Session 1:  Plenary Session
Moderator: Bernice Morrow, Ph.D.

9:15 – 9:45  Keynote Address: Molecular Basis for VCFS ............................................. 43
Raju Kucherlapati, Ph.D.

9:45 – 10:05 Epidemiology: The Prevalence of VCFS ................................................ 47
Adolfo Correa, M.D., Ph.D.

10:05 – 10:25 Prevalence of VCFS in Western Sweden ................................................. 55
Solveig Oskarsdottir, M.D.

10:25 – 10:50 A Review of World Recognition of VCFS ............................................ 63
Karen J. Golding-Kushner, Ph.D., Stephan Eliez, M.D., P.D.
(Switzerland), Stephen Russell (Australia),
Antonio Ysunza, M.D., Sc.D. (Mexico), Julie Squair and
Ahmad Al-Khattat, Ph.D. (United Kingdom), Doron Gothelf, M.D.
(Israel)

10:50 – 11:00  Questions and Discussion ................................................................. 67

11:00 – 11:10 Break

Session 2: Molecular Genetics
Moderator: David H. Ledbetter, Ph.D.

11:10 – 11:30 Role of Thx1 in Embryonic Development .................................. 73
Bernice Morrow, Ph.D.

11:30 – 11:50 Mechanisms and Consequences of 22q11.2 Deletions .................... 77
Beverly S. Emanuel, Ph.D.
11:50 – 12:10 COMT: Can Its Role Lead to Effective Treatments? ...........................................87
William Graf, M.D.

12:10 - 12:25 Genetic Association Studies Of Patients With 22q11 Deletion ...............................97
DA Driscoll, J Manson, H Zhao, M Book, EH Zackai, Donna M.
McDonald-McGinn, M.S., C.G.C., Beverly S. Emanuel, Ph.D., J
Catanzaro, Elizabeth Goldmuntz, M.D.

12:25 – 12:45 Questions and Discussion .................................................................................103

12:45 - 1:45 Lunch on your own
Optional “Birds of a Feather” seating available.
(Note: outside food may not be brought into the ballroom. Enjoy “Birds of a
feather” after eating lunch – Ballroom South)

Session 3: Communication Impairment
Moderator: Susan Marks, M.S., C.C.C.
1:50 – 2:10 Speech and Language Issues in 22q11.2 Deletion .............................................109
Cynthia B. Solot

2:10- 2:20 Why Do Children With VCFS Have VPI With or Without Clefts? ....................113
Robert J. Shprintzen, Ph.D.

2:20- 2:40 Assessment of Speech and Language Disorders in VCFS – Special
Considerations ..................................................................................................................117
John E. Riski, Ph.D., F.A.S.H.A., C.C.C-S.

2:40 – 3:00 Treatment of Speech and Language Disorders in VCFS – Special Considerations121
Karen J. Golding-Kushner, Ph.D.

3:00 – 3:15 Questions and Discussion .................................................................................133

3:15 - 3:25 Break

Session 4: Surgery for Speech
Moderator – Eileen Marrinan, M.S., M.P.H.
3:25 – 3:45 Surgery for Velopharyngeal Incompetence ......................................................139
Fernando D. Burstein, M.D.

3:45 – 4:05 Surgical Treatment of VPI in Children with VCFS .........143
J. Kerwin Williams, M.D., Albert Losken, M.D.

4:05 - 4:25 Surgical Outcomes for Management of VPI Associated with VCFS ..............147
Sherard A. Tatum III, M.D., F.A.A.P., F.A.C.S.

4:25 – 4:35 Questions and Discussion .................................................................................153

4:35 – 5:00 Panel Discussion with Audience Interaction – Outcomes of Speech Treatment in
VCFS ..................................................................................................................................157
Moderator: Eileen Marrinan, M.S., M.P.H.
Panel: Fernando D. Burstein, M.D., Karen J. Golding-Kushner, Ph.D., John E.
Riski, Ph.D., F.A.S.H.A., C.C.C-S., Cynthia B. Solot, Sherard A. Tatum III,
M.D., F.A.A.P., F.A.C.S.

Session 5: Parenting: Clinical and Personal Perspectives
Moderator: Dominique Pfeiffer
5:00 – 5:05 Consistent Evaluation And Intervention: A Case Study ...............................................163
Mia De La Rosa-Trujillo, Vidhya Krishnamurthy, Kristy Kuehfuss, Janice
Enriquez, Kiti Freier, Ph.D.

5:05 – 5:10 From A Parent’s Perspective: Coordinating Your Child’s Care .................................169
Dorothea D. Specht

5:10–5:15 Three Generations With DiGeorge: Judy's Journey ..............................................177
Judy Wickun

5:15 – 5:30 The Role of Charities and Support Groups ..................................................183
Julie Wootton.
Saturday, July 24, 2004
*Posters will be displayed all day

6:45  Continental Breakfast Service begins (Ballroom South)
7:00 – 7:50  Registration (Grand Foyer) and CONCURRENT SESSIONS at Breakfast:
(Open and Birds of a Feather seating available in Ballroom South for those not
attending Concurrent sessions)

Session A: Speech and Language Issues (Kennesaw Room) .........................191
  Karen J. Golding-Kushner, Ph.D.

Session B: What’s in a Name? (Peachtree Room) .................................195
  Angelo DiGeorge, M.D., Robert J. Shprintzen, Ph.D., Nathaniel
  H. Robin, M.D.

Session C: Issues in Education (Ballroom North) ...............................199
  Donna Cutler-Landsman, B.S., M.S.

Session D: Leg Pains and Their Management (Sherwood Room) ..........207
  Ahmad Al-Khattat, Ph.D.

Session 6:  Clinical Genetics
  Moderator: Nicole Philip, M.D.

7:55 – 8:15  What is, and What is Not Deletion 22q11.2  .........................215
  Nathaniel H. Robin, M.D.

8:15 – 8:35  Familial Recurrences of VCFS  ........................................223
  Maria Cristina Digilio, M.D.

8:35 – 8:55  Much Ado About 22q ......................................................231
  Donna McDonald-McGinn, M.S., C.G.C.

8:55 – 9:15  Diagnosis, Natural History, Development  .........................239
  Hiroshi Kawame, M.D.

9:15 – 9:30  Questions and Discussion ...............................................245

9:30 – 9:45  Break and Poster Session

Session 7a:  Abstracts: Surgery and Speech
  Moderator: Cynthia B. Solot

9:45 – 10:00  Submucous Cleft Palate: New Perspectives on Old Themes? ....251
10:00 -10:15  Longitudinal Study of Speech and Language in Children with 22q11.2 Deletion257
  Nancy J. Scherer, Ph.D., Linda D’Antonio, Ph.D.
10:15 – 10:30  The 22q11.2 Deletion Syndrome In The Australian Population: Communication
  and Feeding Aspects .................................................................267
  Suzi Drevensek, Alison Purcell, Jan Van Doorn
10:30 – 10:45  Complex Speech Patterns in Children with 22q11.2 Deletion Syndrome -
  Perspectives from the United Kingdom ........................................279
  Louise Mills, Lydia Allen, Marie Pinkstone, Debbie Sell
10:45 – 11:00  Speech Production and Phonological Processing In Children With VCFS....285
  Adriane L. DeMarco, M.A., CCC-SLP, Benjamin R. Munson, Ph.D.
11:00 – 11:10  Questions ........................................................................291

Session 7b:  Abstract Presentations: Psychiatry, Behavior and Neuroscience
  Moderator: Anne S. Bassett, M.D., F.R.C.P.C.

9:45 – 10:00  Basic Timing Mechanism In Velo-Cardio-Facial Syndrome ....297
  M. Debbane, B. Glaser, S. Eliez
10:00 – 10:15  A Cross-Syndromal Comparison of Cognitive Profiles: Velo-Cardio-Facial
  Syndrome (VCFS) and Williams Syndrome (WS) .............................303
  Campbell LE, Stevens AF, Azuma R, Amberly, F, Morris RG,
  Karmiloff-Smith A, Owen MJ, Murphy DGM, Murphy KC
10:15 – 10:30 Behavior Problems In Relation To Intelligence In Children With 22q11 DS, Compared To A Matched Control Group Of Children With Craniofacial Anomalies ................................................................. 313
   Pauline Jansen, Josien Heineman-de Boer, Sasja Duijff, Frits Beemer, Jacob Vorstman
10:30 – 10:45 Neuropsychiatric Aspects And Coping In Familial Cases Of Velocardiofacial Syndrome ................................................................. 319
   Doron Gothelf, Ayala Aviram-Goldring, Merav Burg, Gadi Presburger, Amos Frisch, Elena Michaelovsky, Shlomit Rienstein, Talya Litmanovitz, Alan Apter, Abraham Weizman
10:45 – 11:10 Questions and Discussions ........................................................................ 325

Session 8:  Feeding Disorders
   Moderator: Suzi Drenvsek
11:15 – 11:35 Feeding Disorders: Assessment and Treatment ........................................ 331
   Robert Beecher, M.S., C.C.C.-S.L.P.
11:35 – 11:55 The Myth and Facts About “Swallowing Disorders” in Children with VCFS ................................................................. 335
   Robert J. Shprintzen, Ph.D.
11:55 – 12:00 Questions and Discussion ........................................................................ 341
12:00 – 12:55 CONCURRENT SESSIONS at Lunch:
   (Pick up box lunch in Ballroom South. Teens will not pick up box lunch if attending Session H.) Open seating available in Ballroom South for those not attending a Concurrent Session.)
   Session E: Estate Planning (Sherwood Room) ............................................................ 347
      Keith Hallman, C.F.P., C.L.U., Ch.F.C., R.H.U., R.E.B.C.
   Session F: Clinical and Molecular Genetics (Ballroom North) .................................. 357
      Donna M. McDonald-McGinn, M.S., C.G.C.
   Session G: Ask the Cardiologist (Kennesaw Room) .................................................. 361
      William T. Mahle, M.D.
   Session H: Teen Talk (Workshop for Teens only) (Peachtree Room) ......................... 365
      Alexandra Oppenheimer, M.S.Ed., and Karen Ruckman-Lindsay, B.M., P.D. (Workshop for Teens only with Pizza Party lunch)

Session 9:  Neural Imaging and Behavior
   Moderator: Anne Marie Higgins, M.S., R.N., F.N.P.
1:00 – 1:20 Brain Structure, Connectivity, and Cognitive Function in VCFS .................. 371
      Tony J. Simon, Ph.D.
1:20 – 1:40 Brain Imaging, Development, Cognition, and Memory in VCFS ............... 381
      Stephan Eliez, M.D., P.D.
1:40 – 2:00 Brain Imaging and Other Markers for Mental and Cognitive Function .......... 385
      Wendy R. Kates, Ph.D.
2:00 – 2:20 Cognitive and Behavioral Parameters Associated with VCFS .................. 395
      Kevin M. Antshel, Ph.D.
2:20 – 2:40 Behavioral and Psychological Issues Associated with VCFS: Personality Profiles in Youths with VCFS .................................................. 399
      Ann Swillen, Ph.D.
2:40 – 2:55 The Effect Of Deletion 22q11 On Brain Anatomy: A Structural Imaging Study of Children and Adolescents with Velo-Cardio-Facial Syndrome (VCFS) ...................... 411
      Campbell LE, Stevens AF, Daly EM, Azuma R, Morris RG, Karmiloff-Smith A, Owen MJ, Murphy DGM, Murphy KC
2:55 – 3:10 Questions and Discussion ........................................................................... 421
3:10 – 3:20 Break and Poster Session
Session 10: Psychiatry
Moderator: Ann Swillen, Ph.D.
3:20 – 3:40  Risk Factors for ADHD in VCFS ........................................... 427
         Doron Gothelf, M.D.
3:40 – 4:00  What Can VCFS Tell Us About the Genetics of Schizophrenia? ............ 431
         Kieran C. Murphy, M.D., Ph.D.
4:00 – 4:20  Autism Spectrum Disorders Associated with 22q11 Deletions .................. 441
         Marcia Gerdes, Ph.D.
4:20 – 4:40  Psychiatric Disturbance in Adults with VCFS .................................. 451
         Anne S. Bassett, M.D., F.R.C.P.C.
4:40 – 5:00  Reaction Panel: Questions from the Floor and Discussion .......................... 455
         Kevin M. Antshel, Ph.D., Anne S. Bassett, M.D., F.R.C.P.C., Stephan Eliez,
         M.D., P.D., Doron Gothelf, M.D., William Graf, M.D., Wendy R. Kates, Ph.D.,
         Kieran C. Murphy, M.D., Ph.D, Tony J. Simon, Ph.D., Ann Swillen, Ph.D.
6:00  All-Attendee Optional Dinner – Reservations required (Habersham Room)
Emcee for featured addresses: Julie Wootton
“Footprints of Hope” and Government Recognition of VCFS in Australia
         Raymond Tanner
How to Succeed by Really Trying
         Alexandra Oppenheimer, M.S., M.Ed.
Emcee for concert: Julie Squair
Concert: Karen Ruckman Lindsay, B.M., P.D.
Program:
         Mozart Sonata in C Major, K.330
         Schubert Impromptu No.2 in A flat Major
         Allegretto
         Debussy Preludes
         No. III. La Puerta Del Vino (the Wine Gate)

Sunday, July 25, 2004
6:45  Breakfast Service begins (Ballroom South)
7:00 – 7:55  CONCURRENT SESSIONS at Breakfast:
(Open and Birds of a Feather seating available in Ballroom South for those not
attending Concurrent Sessions)
Session I: Sibling Relationships (Sherwood Room) ........................................ 461
         Solveig Oskarsdottir, M.D., April Elwood, C.C.L.S.
Session J: Tips for Parents of Teens (Kennesaw Room) .................................. 465
         Donna Cutler-Landsman, B.S., M.S. and Sharon Lynn Smith,
         M.Ed, Psy.D.
Session K: Support Groups (Peachtree Room) .............................................. 485
         Maureen Anderson
Session L: Coping with Behavioral and Psychological Issues (Ballroom North) 489
         Stephan Eliez, M.D., P.D., Doron Gothelf, M.D.

Session 11: Education and Living Skills
Moderator: Maureen Anderson
8:00 – 8:20  General Memory Function In Children And Young Adults With 22q11.2
         Deletion Syndrome/Velocardiofacial Syndrome ................................. 495
         Bronwyn Glaser, Verane Braissant, Martin Debbane, Jacqueline
         Delavelle, François Lazeyras, and Stephan Eliez, M.D., P.D.
8:20-8:35  22q11 DS And Nonverbal Learning Disability ................................... 501
         Sasja Duijff, Josien Heineman-De Boer, Frits Beemer, Jacob Vorstman,
         Pauline Jansen
8:35 – 9:00  Voyage into Adulthood .............................................................................507
          Sharon Lynn Smith, M.Ed., Psy.D.
9:00 – 9:25 Impact of VCFS on Learning and School Performance and What Can Be Done ..535
          Donna Cutler-Landsman, B.S., M.S.
9:25 – 9:40 Questions and Discussion ...........................................................................543

Session 12: Cardiology

Moderator: Angelo DiGeorge, M.D.

9:40 – 10:00  Difference in Cardiovascular Outcomes in Patients with VCFS .................549
          William T. Mahle, M.D.
10:00 – 10:20 Approach to Screening for 22q/VCFS in Children with Heart Disease ....553
          Elizabeth Goldmuntz, M.D.
10:20 – 10:40 The Epidemiology of Deletion 22q11.2 in Congenital Heart Disease .......573
          Lorenzo D. Botto, M.D.
10:40 – 11:05 Questions and Discussion ...........................................................................581
11:05 – 11:20 Break

Session 13: Immunology, Medical Emergencies, and Dentistry

Moderator: Lisa Kobrynski, M.D.

11:20 – 11:40 Immunodeficiency with Chromosome 22q11.2 Deletion Syndrome .........587
          Kathleen E. Sullivan, M.D., Ph.D.
11:40 – 12:00 Infections and Immunizations in Children with VCFS .........................595
          Coleen K. Cunningham, M.D.
12:00 – 12:15 Determinants of Growth in Children with a 22q11.2 Deletion ..........599
          Stuart A Weinzierl, Darcy Hayes, Donna M. McDonald-McGinn, M.S.,
          C.G.C., Elaine Zackai, Thomas Moshang
12:15 – 12:35 How to Prepare for Emergencies, .........................................................609
          Edward Stettner, M.D.
12:35 – 12:55 Oral and Dental Issues in 22q11 ...............................................................617
          Gunilla Klingberg, M.D.
12:55 – 1:15 Dental Manifestations in VCFS .................................................................629
          Jack E. Thomas Jr., D.D.S.
1:15 – 1:30 Dental Manifestations of the 22q11.2 Deletion ........................................641
          Schiffman, Kirschner, Bernick, LaRossa, Donna M. McDonald-McGinn,
          M.S., C.G.C., Zackai, Mayro
1:30 – 1:40 Questions and Discussion ...........................................................................647
1:40 Adjourn, Karen J. Golding-Kushner, Ph.D. and William T. Mahle, M.D.

For more information on a variety of CME offerings at Children’s Healthcare of Atlanta, go to
http://www.choa.org/professionals/cme.shtml
Coming soon.... online conference registration!