

**12<sup>th</sup> Annual International Scientific Meeting**  
**July 7 - 9, 2006, Strasbourg, France**  
**The Velo-Cardio-Facial Syndrome Educational Foundation, Inc.**  
**Hosted by Génération 22 and the VCFSEF European Network**

***Friday Morning, 7 July 2006***

- 9:00 – 9:20: Opening Ceremonies and Introductions  
Dominique Pfeiffer, Présidente, Génération 22, Strasbourg, France  
Stephen Russell, B.A. LL.B, President, VCFSEF  
Karen J. Golding-Kushner, Ph.D., Executive Director, VCFSEF
- 9:20 – 9:35: Keynote Address: Jean-Louis Mandel, Professeur au Collège de France, Directeur de L'IGBMC, Strasbourg, France
- 9:35 – 9:45: A Brief History of VCFS and the Educational Foundation; *Robert J. Shprintzen, Ph.D., Upstate Medical University, Syracuse, NY, USA*
- 9:45 – 10:20: Panel: The European Network of the VCFS Educational Foundation
- 10:20 – 10:45: Recognizing the 22q11 deletion syndrome; *Solveig Oskarsdottir, Göteborg, Sweden*
- 10:40 – 11:00: Clinical findings in neonates with VCFS; M. Cristina DiGilio, M.D., Ospedale Bambino Gesù, Roma, Italia
- 11:00 – 11:10: Surdit  congenitale et deletion 22q11: une deletion peut en cacher une autre.  
*Catherine Blanchet, F. Venail, P. Blanchet, P. Sarda, A.P. Roux, M. Claustre, A. Uziel, M. Mondain, Montpellier, France*
- 11:10 – 11:20: Questions and Discussion
- 11:20 – 11:35: Refreshment Break
- 11:35 – 11:55: Cardiac surgery results in patients with conotruncal heart defects: impact of VCFS; *Bruno Marino, M.D., Ospedale Bambino Gesù, Roma, Italia*
- 11:55 – 12:10: Feeding and weight gain in VCFS: comparisons to normal growth curve is a mistake; *Anne Marie Higgins, R.N., F.N.P., Robert J. Shprintzen, Ph.D., Upstate Medical University, Syracuse, NY, USA*
- 12:10 – 12:30: Panel Discussion and Questions: phenotype to genotype correlation in VCFS and its potential importance to management; *Yves Alembik, Moderator; Nicole Philip, Pierre Sarda, M. Cristina DiGilio*
- 12:30 – 1:45: Lunch with the Experts

***Friday afternoon***

- 1:45 – 2:00: Phonological approach to speech and language therapy for young children with VCFS;

*Virginia L. Dixon-Wood, M.A., University of Florida, Gainesville, FL, USA*

- 2:00 – 2:15: Phonica faces: a tool for providing speech therapy in patients with VCFS; *Antonio Ysunza, M.D., Hospital Gea Gonzales, Mexico City, Mexico*
- 2:15 – 2:30: Application of the sound, shape, and color (SSC) principles for treating VCFS speech disorders; *Ariela Nachmany, Ph.D., Sapir Medical Centre, Kfar Saba, Israel*
- 2:30 – 3:00: Approaches to speech intervention in VCFS: general principles, special “tricks,” and matching techniques to disorders; *Karen J. Golding Kushner, Ph.D., Executive Director, VCFSEF, Private Practice in Speech-Language Pathology, East Brunswick, NJ, USA*
- 3:00 – 3:15: Questions and Discussion
- 3:15 – 3:30: Refreshment Break
- 3:30 – 3:45: Velopharyngeal insufficiency in 22q11.2 deletion syndrome: speech results after surgery; *Nathalie Degardin-Capon, Marie Arnoldi, Sylvie Lejeune, Salima Mahammed, Véronique Martinot-Duquennoy, Philippe Pellerin, Children University Hospital of Lille. France*
- 3:45 – 4:05: Pharyngeal flap surgery in VCFS: outcomes and long-term follow-up in a large sample by a single surgeon. *Sherard A. Tatum III, Upstate Medical University, Syracuse, NY USA*
- 4:05 – 4:30: Panel Discussion: Speech diagnosis, speech therapy, and surgical management; *Sherard A. Tatum, III, M.D., Syracuse, NY, USA; Partice Oger; Noel Garabedian; Judith Hohlfeld; Philippe Pellerin, Karen Golding-Kushner, Ph.D.*
- 4:30 – 4:40: 3D Models of Face Shape in Velo-cardio-facial Syndrome; *Peter Hammond, Judith Allanson, Linda Campbell, Raoul Hennekam, Tim Hutton, Kieran Murphy, Katrina Prescott, Pete Scambler, University College of London, London, UK*
- 4:40 – 4:50: Micro délétion et santé bucco-dentaire: à partir d’une étude clinique; *N. Marchetti, R. Lalonde, D. Droz-Desprez, M. Simonin, C. Strazielle, Nancy, France.*
- 4:50 – 5:00: Micro délétion et santé bucco-dentaire: à partir d’une étude clinique; *Dominique Droz-Desprez, N. Marchetti, N. Beltramo, C. Strazielle, Nancy, France.*
- 5:00 – 5:10: A report from the VCFSEF on recommended diagnostic and treatment protocols
- 5:10 – 5:30: Panel discussion, questions, and comments on the recommendations
- 5:30 – 5:40: VCFSEF Meeting in Australia; *Stephen Russell, Brisbane, Australia*
- 5:45 – 6:30: Annual Business Meeting, VCFSEF

***Saturday Morning, 8 July 2006***

- 9:00 – 9:20: Genetic modifiers and genetic susceptibility in VCFS; *Bernice Morrow, Ph.D., Albert Einstein College of Medicine, Bronx, NY, USA*

- 9:20 – 9:30: Is hyperprolinemia associated with cognitive and/or psychotic symptoms in VCFS patients? An association study; *Dominique Champion, Faculté de Médecine, Rouen, France; E. Bumsel, B. Hecketsweiler, N. Drouin-Garreau, S. Legallic, G. Opolczynski, Th. Frébourg (Rouen); S. Manouvrier-Hanu, C. Fantini (Lille); Th. Van Amelsvoort (Amsterdam); A. Swillen, A. Vogels (Leuven); A. Philippe, D. Heron (Paris); N. Philip, M. Carlier, A. Gerard (Marseille), P. Sarda (Montpellier); Y. Alembik, M. Lacambre (Strasbourg); D. Lacombe (Bordeaux); O. Boespflug-Tanguy (Clermont-Ferrand).*
- 9:30 – 9:40: An analysis of polymorphisms in the remaining PRODH allele, serum Proline levels and brain function measures in individuals with the 22q11.2 deletion syndrome. *J.A.S. Vorstman, A. Willis, C. Kemner, M. De Sain-van der Velden, B. Dorland, F.A. Beemer, R.S. Kahn, D. Valle, B.S. Emanuel, H. van Engeland, Children's Hospital of Philadelphia, Philadelphia, PA, USA.*
- 9:40 – 9:55: The challenges of matching the genetic deletion to the clinical expression in VCFS; *Robert J. Shprintzen, Ph.D., Upstate Medical University, Syracuse, NY, USA*
- 9:55 – 10:05: Questions and Discussion
- 10:05 – 10:25: Behaviors and emotions from childhood to adulthood: implications for psychological treatment in VCFS; *Stephan Eliez, M.D., University of Geneva, Geneva, Switzerland*
- 10:25 – 10:45: Risk factors and developmental trajectories in VCFS; *Doron Gothelf, M.D., Schneider Children's Medical Center, Petah Tiqva, Israel; Stanford University, Stanford, CA, USA*
- 10:45 – 11:00: Questions and Discussion
- 11:00 – 11:15: Refreshment break
- 11:15 – 11:35: Longitudinal data on intelligence in VCFS: from preschool to puberty; *Ann Swillen, Ph.D., Leuven, Belgium*
- 11:35 – 11:55: Multiple related impairments in cognitive processing of objects and space in VCFS; *Tony Simon, Ph.D., University of California, Davis, Sacramento, CA, USA*
- 11:55 – 12:15: Neuropsychiatric and behavioural problems in 100 individuals with 22q11 deletion syndrome; *Lena Niklasson, Peder Rasmussen, Sólveig Óskarsdóttir, Christopher Gillberg, Göteborg University, Göteborg, Sweden*
- 12:15 – 12:30: Questions and Discussion
- 12:30 – 1:30: Lunch with the Experts
- Saturday afternoon**
- 1:30 – 1:40: Carole Bouquet, French film star, will address the meeting.
- 1:40 – 1:50: Living with VCFS in the 21<sup>st</sup> Century; *Raymond Tanner, Adelaide, Australia*

- 1:50 – 2:05: Paths to Success; *Alexandra Oppenheimer, M.Ed., Mamaroneck, NY, USA; Suzi Oppenheimer, New York State Senator, Mamaroneck, NY, USA.*
- 2:05 – 2:15: A personal story from France: Living with someone who has VCFS; *Marie-Christine Della Vedova*
- 2:15 – 2:35: What can functional brain imaging tell us about cognition and emotion in VCFS?; *Wendy Kates, Ph.D., Upstate Medical University, Syracuse, NY, USA*
- 2:35 – 2:45: Social Cognition: In the Eye of the Beholder; *Ryan Murray, Martin Debbané, Bronwyn Glaser, Stephan Eliez, Geneva, Switzerland*
- 2:45 – 3:00: What can face processing tell us about cognitive and social development in VCFS? *Bronwyn Glaser, Martin Debbané, Marie Schaer, Sandra Martinez, Lea Matasci, and Stephan Eliez, Geneva, Switzerland*
- 3:00 – 3:20: Brain connectivity differences in VCFS that might explain cognitive dysfunctions; *Tony Simon, Ph.D., University of California, Davis, Sacramento, CA, USA*
- 3:20 – 3:30: Questions and discussion
- 3:30 – 3:45: Refreshment Break
- 3:45 – 4:05: Structural Imaging of the Brain in VCFS; *David Skuse, Institute of Child Health, London, UK.*
- 4:05 – 4:15: Abnormal cortical folding in the brain of patients with VCFS; *Marie Schaer, Bronwyn Glaser, Stephan Eliez, M.D., University of Geneva, Geneva Switzerland*
- 4:15 – 4:25: Questions and Discussion
- 4:25 – 4:45: Dopaminergic neurotransmission in VCFS before and after dopamine depletion; *Therese van Amelsvoort, M.D., Ph.D., University of Amsterdam, Amsterdam, The Netherlands*
- 4:45 – 5:05: FISHing for the neurobiology of behaviour in VCFS; *Kieran Murphy, M.D., Royal College of Surgeons, Dublin, Ireland.*
- 5:05 – 5:25: Association of the low activity COMT allele with ADD/ADHD and obsessive compulsive disorder in VCFS; *Merav Burg, Elena Michaelovsky, Amos Frisch, Abraham Weizman, Alan Apter, Doron Gothelf, Schneider Children's Medical Center of Israel, Petah Tiqva, Israel.*
- 5:25 – 5:40: Questions and Discussion
- 5:40: Parents meeting: Génération 22.
- 7:00 P.M.: Optional dinner: Restaurant Paul Appel, 6 rue de Palerme, Strasbourg

***Sunday Morning, 9 July 2006***

- 9:00 – 9:15: To recognize or to recollect? Memory processes at work in VCFS; *Martin Debbané, Bronwyn Glaser, Marie Schaer, Stephan Eliez, University of Geneva, Geneva, Switzerland*
- 9:15 – 9:35: Mathematical disabilities in children with Velo-Cardio-Facial Syndrome; *B. De Smedt, A. Swillen, K. Devriendt, J-P Fryns, L. Verschaffel, P. Ghesquière, Leuven, Belgium*
- 9:35 – 9:55: The 22q11.2 deletion in children: high rate of autistic disorders and early onset of psychotic symptoms. *Jacob A.S. Vorstman, Peter Meyer, Monique E.J. Morcus, Sasja N. Duijff, Petra W.J. Klaassen, Josien A. Heineman-de Boer, Frits A. Beemer, Hanna Swaab, Elaine Zackai, Donna M. McDonald-McGinn, Beverly S. Emanuel, René S. Kahn, Herman van Engeland, Children's Hospital of Philadelphia, Philadelphia, PA.*
- 10:05 – 10:35: Educational interventions for children with VCFS; *Donna Cutler-Landsman, Madison, WI, USA*
- 10:35 – 10:50: Questions and Discussion
- 10:50 – 11:05: Refreshment Break
- 11:05 – 11:20: Leg Pains in VCFS; *Ahmad Al-Khattat, M.B.B.Ch., Northampton, U.K.*
- 11:20 – 11:35: Motor development in children with VCFS, part 1, *Katrijn Van Aken, Ann Swillen, Hilde Feys, Tamara Adriaens, Loes Nelissen, Luc Martens, Marc Gewillig, Koen Devriendt, Jean-Pierre Fryns, Leuven, University Hospital Gasthuisberg, Belgium*
- 11:35 – 11:50: Motor development in children with VCFS, part 2; *Katrijn Van Aken, Ann Swillen, Marc Gewillig, Koen Devriendt, Annelies Van Role, Johan Simons, Jean-Pierre Fryns, University Hospital Gasthuisberg, Belgium*
- 11:50 – 12:00: Questions and Discussion
- 12:00 – 12:10: Life with VCFS: Valérie Busserolles Combeau
- 12:10: Conclusion: Closing comments

### **Poster Sessions:**

Social skills, executive function, and obsessional behavior in VCFS; *Arild Berglund, Sissel Iversen, Stavanger University Hospital, Stavanger, Norway*

Cognition and brain in children with VCFS; *L.E. Campbell, A.F. Stevens, R. Azuma, R.G. Morris, D.G.M. Murphy, K.C. Murphy, University of Newcastle, Newcastle, Australia; King's College, London, U.K.; Royal College of Surgeons, Dublin, Ireland*

Short term verbal memory in VCFS. A specific deficit of retention of serial order? *Steve Majerus, Bronwyn Glaser, V erane Braissand, Martial Van der Linden, Stephan Eliez, Universit e de Li ege, Belgique; Universit e de Gen eve, Gen eve, Suisse*

Laterality and cognition in people with genetic disorders: a comparison of VCFS and Williams syndrome. Preliminary results; *Aude G erard-Desplanches (Provence, France), Christine Deruelle (Marseille, France), Silvia Stefanini (Parma, Italy), Nicole Philip (Marseille, France), Virginia Volterra (Rome,*

*Italy), Stefano Vicari (Rome, Italy), Gene Fisch (New York, USA), Michèle Carlier (Provence, France)*

Glottal articulation, obturator and communication; *Björg Solfrid Lia, TAKO Center, Oslo, Norway*

Facial recognition in people with VCFS; *Sandra Martinez, Bronwyn Glaser, Martin Debbané, Stephan Eliez, Geneva, Switzerland*

Summer camping experience with children and adolescents with VCFS at Victory Junction in Randelman, NC; *Paula Goldenberg, M.D., M.S.W., Duke University Medical Center, Durham, NC, USA*

MLPA: A Rapid, Reliable and Sensitive Method for Detection and Analysis of Abnormalities of 22q. Submitted by: *J.A.S. Vorstman, G.R. Jalali, E.F. Rappaport, A.M. Hacker, C. Scott, B.S. Emanuel, Children's Hospital of Philadelphia, Philadelphia, PA, USA.*

Psychiatric Disorders in VCFS: A case report. Béatrice Colomb, Bruno Giordana, Frédérique Feral, Eric Vallier, Dominique Pringuey; Clinique de Psychiatrie et Psychologie Médicale, CHU de Nice. Nice, France.

### **Poster Presentations, Clinic Descriptions**

Traitement et mémoire visuo-spatiale chez les individus atteints du syndrome vélo-cardio-facial : une investigation au travers du Test de rétention visuelle de Benton. *Lea Matasci, Bronwyn Glaser, Martin Debbané, Sandra Martinez, Stephan Eliez, Geneva, Switzerland*

The Stockholm 22q11 Deletion Syndrome Team – a presentation and a report from a parent questionnaire on speech and language and oral motor development; *Liisi Raud Westburg, M.Sc., Gunilla Hinningsson, Ph.D., Karolinska University Hospital, Stockholm, Sweden*

Project for the Formulation of a diagnostic and therapeutic protocol; *Giuletta Angelelli Cafiero, Caterina Cancrini, Rome, Italy*

Presentation du laboratoire de neuroscience et neuroimagerie, Geneva, Switzerland. *Martin Debbané, Stephan Eliez, Bronwyn Glaser, Marie Schaer, Sandra Martinez, Ryan Murray, Léa Matasci*

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