



# VCFSEF NEWS

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## EXECUTIVE DIRECTOR'S REPORT

**Karen Golding-Kushner, Ph.D.**

Knowledge is Hope! Thanks to the vision and enthusiasm of our President, Nancy Robbins and the efforts of many others we have a slogan and a new logo. We've embossed the slogan on wristbands and hope you will wear them proudly to promote awareness of VCFS/22q11.2 DS and to help fulfill our Mission as an Educational Foundation. Each wristband comes with a card explaining the syndrome and includes our website (on the card and on the inside of the band). None of this would have happened without the input and work of our lay and professional members working together.

We have now turned our attention to planning our 11<sup>th</sup> Annual International Scientific Conference, which will be held in Syracuse July 29-31. Registration forms are going in the mail every day and hotel rooms are going fast. Our theme this year is *Intervention*, so look for a lot of practical information. We are introducing some new features, such as a pre-Conference "primer" for those new to talks on speech, brain imaging, and genetics. We are repeating some of our previous interactive features, such as Breakfasts and Lunch with the Experts. The informal meal sessions will enable lay and professional participants to discuss clinical issues related to speech, genetics, foot pain, and others, and also to engage in dialogue on topics such as "Telling your child with VCFS about VCFS," "Psychosocial aspects of VCFS," and "Planning for your child's adult life." We are

organizing formal and informal opportunities for teens and young adults with VCFS to get together and form connections and have many surprises planned for the Kid's Club. Needless to say, the program will also include State of the Art presentations from clinicians and researchers from around the world, presentations by parents, and a lot of time for questions and discussion. You can read more about the meeting elsewhere in this newsletter, and on our website at [www.vcfsef.org](http://www.vcfsef.org).

We are also working on informational brochures and would like to see one in the office of every pediatrician we can find. Volunteers are writing the brochure but printing, obtaining mailing lists and labels, and postage are not inexpensive!

All of these activities require funding and we have been very fortunate to have seen a dramatic increase in donations to support these initiatives in the last few months. Payment of dues also helps to offset our costs, so if you have not yet paid your 2004-05 dues, please do so! Membership dues for 2005-06 are due on June 1.

See you in Syracuse!

Karen J. Golding-Kushner, Ph.D.  
Executive Director

### VELO CARDIO FACIAL SYNDROME EDUCATIONAL FOUNDATION, INC

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#### SAVE THE DATES!

...for the International Scientific Conferences

- **2005:**  
July 28-30, 2005  
Syracuse, NY
- **2006:**  
July 7-9, 2006  
Strasbourg, France
- **2006:**  
November 2-4, 2006  
Brisbane, Australia

# 11<sup>TH</sup> ANNUAL INTERNATIONAL SCIENTIFIC MEETING OF THE VCFS EDUCATIONAL FOUNDATION SYRACUSE, NEW YORK USA JULY 29-31, 2005

The 11<sup>th</sup> Annual International Scientific Meeting of the VCFS Educational Foundation, Inc. will be held at the Sheraton University Hotel and Conference Center in Syracuse, New York from July 29 through 31. Many exciting event have been planned for the program that runs from midday Friday through noon Sunday. Several new features for the meeting have been added to favorite components that are regular features of the meeting. We will once again be holding speech, leg pain, and feeding clinics throughout the meeting. There will also be a Kids' Club adjacent to the meeting room where you can leave your children with our volunteers for parts of the day when you wish to be in the meeting. Entertainment is planned for your kids, including visits from small animals from the Rosamond Gifford Zoo, magicians, and a dazzling display of bubbles and comedy. The annual dinner (separate fee) will be held Friday evening, and on Saturday, there is an optional event at the Museum of Science and Technology that will include demonstrations just for kids and many hands-on exhibits for all ages. The museum also houses a domed Imax theatre, and a planetarium. Included in the museum event will be catering by the world famous Dinosaur Barbeque. Transportation will be provided to and from the museum. Located in trendy Armory Square, a great place to explore in the summer with its many interesting restaurants and shops. On Sunday, we will plan an optional outing to the Rosamond Gifford Zoo for those who are interested. Teen forums and social events will also be scheduled.

The meeting will feature faculty from all over the world, and will return to our usual format of substantial time for questions and interaction with the present-

ers. Sessions will cover all aspects of VCFS, from speech to psychiatry, palate to feet, and there will be a special emphasis on treatment, both present and what to expect in the future. Every session will be followed by extended periods for questions. A new feature will be the ability for people to submit questions ahead of time to be answered by a panel of experts. We will also add small discussion sessions with faculty members at breakfasts and lunches, giving you an opportunity for small group discussions of topics of interest. Prior to the official start of the meeting, we will hold three "primer" sessions on speech, genetics, and brain imaging where small groups will be able to learn the basics in order to enhance their understanding of the papers on the regular program to follow. As always, continental breakfast and refreshments are included in the registration fee, and lunch on Saturday is also included. CMEs will be offered for physicians by Upstate Medical University, and CEUs for speech-language pathologists and audiologists will also be offered by the Communication Disorder Unit of Upstate Medical University.

The Sheraton University Hotel and Conference Center is a beautiful and comfortable venue for the meeting and it is filling fast, so we recommend you register as soon as possible. A pre-registration form for the meeting is in this Newsletter, and hotel reservations can be made by calling the Sheraton directly at 1-800-395-2105. Make sure to mention that you will be attending the VCFSEF meeting for the special room rate. You may also find all available information about the meeting on our web site at [www.vcfsef.org](http://www.vcfsef.org).

See you in Syracuse!

## Call for Newsletter Submissions

The VCFS Educational Foundation wishes to reflect the views and experiences of a wide variety of its lay and professional members. Please contact the Editor, Eileen Marrinan at 315-464-6580 or via email to [marrinae@upstate.edu](mailto:marrinae@upstate.edu) with your ideas or submissions.

*Look on the web site for translations into Arabic, Hebrew and Spanish.*

## New Support Groups are Forming.....

### Dallas -

Contact: Keri Alexander  
411 Fall Creek Drive  
Richardson, TX 75080  
972-889-1103

### New Jersey -

Contact: Yolanda Ortiz at  
[PaulW28165@aol.com](mailto:PaulW28165@aol.com)

## VCFS EDUCATIONAL FOUNDATION, INC. BOARD FOR 2004-05

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### Nominees for VCFSEF, Inc. Board of Directors

President Nancy Robbins and Board Members Wendy Kates PhD. (Professional) and Fred Berg (Lay) will complete their terms of office at the conclusion of our conference this summer in Syracuse, New York. The Board of Directors approved the following slate, submitted by the nominating committee:

President (2005-2006): Stephen Russell, Brisbane, Queensland, Australia. Steve is the VCFS Educational Foundation Regional Director for Australia / Asia, and has been an active participant in past VCFSEF meetings. He is also President of the VCFS Foundation (Old) Inc.

Board Member (Lay) (2005-2008): Dominique Pfeiffer, Strasbourg, France. Dominique is President of Generation 22, the VCFS Family Support Group in France.

Board Member (Professional) (2005-2008): Nathaniel Robin M.D., Birmingham, Alabama. Dr. Robin is a clinical geneticist, and an expert on VCFS.

Elections will take place at the VCFSEF business meeting, to be held on Friday, July 29, 2005 at the annual VCFSEF meeting in Syracuse. All members of the VCFSEF who are at the meeting are eligible to vote. We hope to see you at the annual business meeting in Syracuse!

# ATTENTION DEFICIT HYPERACTIVITY DISORDER (ADHD) IN VCFS

**Doron Gothelf, M.D.**

**Child Psychiatrist, Middle East Regional Director and Professional Council Member of the VCFES Educational Foundation**

ADHD is a common and underdiagnosed disorder in VCFS. About 40%-60% of children with VCFS fulfill the diagnostic criteria of ADHD. It is important to screen children with VCFS for the existence of ADHD as it, as will be described below, is a treatable disorder. When not treated ADHD impedes the school and occupational performance of subjects with VCFS and negatively affects their behavior.

## Diagnostic Criteria

There are three types of ADHD: 1. where inattention is the main problem; 2. where hyperactivity and impulsivity are the main problems; 3. where there are problems of both. Symptoms of inattention are more common in VCFS than hyperactive symptoms.

A child is diagnosed with ADHD when he has at least six symptoms of inattention and/or at least 6 symptoms of hyperactivity and impulsivity. The list of these symptoms is presented in the table below.

## How diagnosis is made

Symptoms of Inattention	Symptoms of Hyperactivity and Impulsivity
1. Makes careless mistakes in schoolwork and other activities.	1. Fidgets or squirms in seat.
2. Has difficulty sustaining attention.	2. Often leaves seat in classroom.
3. Often does not seem to be listening.	3. Runs about.
4. Finds it hard to follow instructions.	4. Has difficulty playing quietly.
5. Has difficulty with organization (such as getting prepared for school).	5. Acts as if "driven by a motor."
6. Avoids or dislikes activities that require mental effort such as homework.	6. Talks excessively.
7. Often loses things.	7. Blurts out answers before questions have been completed.
8. Easily distracted.	8. Has difficulty awaiting turn.
9. Forgetful.	9. Bursts into conversations or games.

Diagnosis and treatment of VCFS is conducted by a child psychiatrist or child neurologist. When the child is having other emotional and behavioral problems besides ADHD, evaluation by a child psychiatrist is recommended. For diagnosing ADHD the clinician checks for the presence of the above mentioned symptoms. The diagnosis is based on reports from both the child's parents and teacher. The teacher's report is essential for the diagnosis as he closely observes the child in the setting where the symptoms of ADHD appear most, e.g., where sustained attention and well-controlled behavior are required. In addition, teachers have good perspective for what is the expected span of attention and behavior of the child in his level of development. It is recommended the teacher complete a written report about the child and that the parents and teacher complete a standardized questionnaire for the evaluation of ADHD symptoms such as the Conners' Rating Scale. This questionnaire is helpful in making the diagnosis, and later, for quantifying the effectiveness of the treatment. Some clinicians use a computer test such as the TOVA or Conners' CPT as an aid in confirming the diagnosis of ADHD. Our experience is that children with VCFS frequently fail to follow the computer exam instructions, rendering the computer assessment invalid.

## Cases that seem like ADHD but are not ADHD

Like all children with developmental disabilities, children with VCFS should be evaluated according to their mental age, not their chronological age. The mental age of children with VCFS is frequently 2 to 4 years less than their chronological age. Expectations regarding the time span they can concentrate, their ability to organize their activities, etc. should be gauged accordingly. We frequently encountered cases of children with VCFS who study in the mainstream who were inattentive in class because the school program was too difficult for their academic level. Thus, it is important to distinguish whether the child has a learning disability, ADHD, or the combination of the two.

Children with VCFS may suffer from psychiatric disorders that also manifest in inattention or hyperactivity. These conditions include depression, mania, anxiety disorders, and psychosis. The child psychiatrist evaluates the child for the presence of these disorders which can mimic ADHD or be comorbid with ADHD. One of the main features that helps distinguish ADHD from other psychiatric conditions is that ADHD symptoms, per definition, begin during preschool years and are a continuous problem. In contrast, if depression causes the inattention, it begins with the depressive

*(Continued on page 5)*

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episode and subsides when the depression subsides.

### What are the risk factors for ADHD in VCFS?

ADHD is 4 to 8 times more common in boys than in girls with VCFS. Similar to typical children, there is an increased risk of developing ADHD if another family member has ADHD. We found that the rate of ADHD in first degree relatives of children with VCFS and ADHD was 4 times higher than in the relatives of VCFS children without ADHD.

The presence of more developmental problems (such as pregnancy and labor complications, delayed developmental milestones, and lower IQ) and the severity of medical comorbidities (such as the cardiac and palate anomalies) were not found to increase the risk of developing ADHD in children with VCFS. Thus it seems likely that ADHD in VCFS is the result of gene(s) from the chromosome 22 deleted region interacting with modifying genes located elsewhere in the genome. These genes are yet to be found.

### Treatment

In contrast with psychiatric disorders such as anxiety disorder and depression that can be treated equally effectively with medications and psychotherapy, the treatment of ADHD requires medications. Many studies in typical children and also some studies in children with developmental disabilities have shown the robust therapeutic effect and safety of medications in the treatment of ADHD including ritalin, concerta, stratera, and aderall.

Unfortunately only one study was conducted on ADHD treatment in VCFS. In this study we prescribed a low dose of Ritalin (0.3 mg per kg body weight) to children and adolescents with VCFS. The treatment significantly improved the ADHD symptoms in 9 of the 12 children (75%). The average duration of the Ritalin effect was 3.2 hours. The side effects recorded were similar to those described in typical children and included: poor appetite (the child always regained appetite later in the day), irritability (this often occurs as a "rebound" when the medication effect wears off), sadness, and stomachaches. In all cases the side effects were mild so that

none of the children discontinued the medication due to a side effect. None of the treated children developed psychotic or manic symptoms. This preliminary study suggests that low dose of Ritalin is safe and effective in children with VCFS. However, to confirm this finding larger longitudinal placebo controlled studies are required.

In addition to medication treatment, psychosocial treatments that are often helpful in the treatment of ADHD are group therapy for improving social skills and parent guidance on implementing behavioral programs (also frequently conducted in groups). For more information on behavioral management and groups for treatment of ADHD see the reference list below.

### Course

Since about half of children with VCFS have predominantly inattentive symptoms, the ADHD is frequently not diagnosed. Those with marked hyperactivity symptoms are usually referred for evaluation and treatment early, during the preschool years. There are no specific data on the course of ADHD in VCFS, but the general trend for typical children is that the symptoms, especially the hyperactive symptoms, become more moderate beginning in late adolescence or early adulthood. However, most children with ADHD continue to have various degrees of symptoms into adulthood, in which case continued treatment is recommended and is effective. Future studies are needed to explore the longitudinal course of ADHD in individual with VCFS.

### References

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3. Barkley RA: *Taking Charge of ADHD: The Complete Authoritative Guide for Parents*. 1995; New York: Guilford Press.
4. Barkley RA, Benton CM: *Your Defiant Child: Eight steps to Better Behavior*. 1998; New York: Guilford Press.
5. Website: [www.adhd.com](http://www.adhd.com)

## Exceptional Parent Article

Raymond Tanner, *Footprints of Hope* author, shared his personal story of living with VCFS and the experience of his son who is also diagnosed with the syndrome in the April, 2005 issue of *Exceptional Parent*. Visit the magazine's web site at [www.eparent.com](http://www.eparent.com) for more details.

## FREQUENTLY ASKED QUESTIONS

### Dr. Robert Shprintzen

**Question:** *I have a 17-year-old daughter with VCFS. I am trying to get information about young adults with VCFS. I have been told I should apply for Social Security Disability for her when she turns 18. She has a lot of learning problems, anxiety and depression. She also seems to be missing "connections" - she doesn't grasp that there are consequences for everything she does and will sometimes do something dangerous without thinking about what could happen. I am not sure if she will be able to work a regular job, and if she can, I don't think she will be able to handle a full-time job. Do you know where I can get more information?*

**Response:** Many adults with VCFS are able to get Social Security benefits, but often times it takes a lot of documentation to explain how this syndrome impacts job functioning. Social Security looks at several areas in determining whether a person can be gainfully employed. In order to qualify under the neurocognitive area, a person must demonstrate severe deficits in cognition, attention, language, self direction, daily living skills and/or memory. Social Security uses specific criteria to assess whether an applicant's problems are severe enough to prevent him/her from being gainfully employed. It would be very helpful to do some research before applying, so that you have all the documentation/testing that you need to support your claim. A letter explaining how these deficits impact your daughter, along with specific examples, would help the Determination Board understand the day to day difficulties she faces. I would also recommend supplying the Social Security Office with relevant articles about

VCFS—especially those that relate to learning, cognition, and psychiatric difficulties. In addition, contact your local Department of Vocational Rehabilitation and ask for an assessment. This evaluation can be helpful in highlighting areas of weakness and how these deficits relate to job performance. Finally, contact your state office that serves the developmentally disabled population and see if your daughter can qualify for those services. This can help expedite the application for Social Security.

**Question:** *Do people with VCFS live a normal life span?*

**Response:** Calculating life span for a specific condition is a little tricky, but let us answer the question initially this way. People with VCFS who reach adult life probably do live a normal life span, although there are no statistics to demonstrate the actual longevity. There are documented cases of people with VCFS in their late 60s who were alive and well and expected to live many years more. There is plenty of pedigree information that points to people in their 70s, as well. Having said this, longevity is often calculated by looking at entire populations of people with a specific condition and taking the average length of their survival. Because there are some babies with VCFS who do not survive infancy because of severe heart anomalies, and others who might succumb to infections if their immune systems are deficient (although this is rare these days), these cases are calculated into the average. Therefore, if there were such a calculation for VCFS, it might show reduced longevity. However, if we eliminate such cases of dangerously ill infants, life expectancy is

probably within the normal range for the large majority of people with VCFS assuming they receive sound medical care.

**Question:** *My son is three months old and was born with a tetralogy of Fallot. FISH was done and confirmed VCFS when the tetralogy was detected. He had heart surgery and has done well. There is no cleft palate and he seems to have a very mild case otherwise. If his case is mild, what are the chances of him having significant learning problems or mental illness?*

**Response:** We are glad that your son has done well and that his surgery has been successful. When you say that he does not have a cleft palate and that his case is mild, you need to be cautious in this type of interpretation. Based on your question, you are clearly aware that many of the clinical features are behavioral, such as learning disorders, certain psychological issues, and even the potential for mental problems. It is obviously far too early at three months to estimate the expression of the syndrome because many of the traits that concern you would not be apparent at this age. In addition, speech and language have not begun to develop, nor have you had an opportunity to see if he will manifest any type of immune disorder, thyroid problems, dental problems, or other issues that develop with age in VCFS. Therefore, it would be in your best interest to continue careful follow-up using the Specialist Fact Sheet from our web site as a guide. Certain traits are dependent on age, and others may simply not be evident at this time. In other words, it is important to pay attention to the natural history of

*(Continued on page 7)*

## FAQs

*(Continued from page 6)*

VCFS so that you can check for abnormalities that might develop rather than assuming that they will not. Finally, there does not seem to be a correlation between specific sets of anomalies in terms of predictions for others. The presence or absence of congenital heart disease does not seem to predict the presence or absence of severe learning impairments or developing psychiatric problems. Therefore, the best advice is always to anticipate the need for evaluations in a timely manner.

**Question:** *My 14-year-old son with VCFS has shown some inappropriate sexual behaviors in the presence of his peers. He doesn't seem to be embarrassed by these. He has made some inappropriate comments in public and even touched some girls where he shouldn't have. This is clearly not malicious. He just doesn't seem to understand that it is wrong. Is this common in VCFS?*

**Response:** We have encountered several cases of inappropriate sexual behavior in adolescents with VCFS. As you may know, VCFS can be associated with psychiatric problems. We therefore recommend that everyone with VCFS be evaluated by a child psychiatrist every 1 to 2 years. The behavior you are describing may be a symptom of a psychiatric problem. Such behavior can occur in adolescents with VCFS and inappropriate sexual behavior can be one of the symptoms. However, depending on the degree of cognitive impairment, this may be symptomatic of developmental delay and the impulsivity that can accompany it. It is recommended to refer your son for evaluation by a child psychiatrist that is familiar with working with adolescents with developmental disabilities. In case mania or other psychiatric problems are in operation, there are efficient medications that can help. We have also found that sexual-social education groups for adolescents and their parents can be very effective. with VCFS.

## 2006 VCFSEF Conference in Strasbourg

The next international VCFSEF conference will be held July 7, 8 and 9, 2006 in the beautiful city of Strasbourg. Strasbourg is a city rooted in tradition, where its art, architecture and history make it one of France's "must-see" visitor destinations. The Rhine River encircles the medieval city center where visitors can admire the awe-inspiring cathedral, picturesque timber-facade houses and quaint shops. Relying heavily on its local products, Strasbourg's regional cuisine and wine are world renowned and can be enjoyed in the city's traditional restaurants as well as in its local "winstubs." Strasbourg therefore promises to be a lovely setting for next year's conference. The conference presentations will be held in the European Pole of Economy building which is fully-equipped for any audio-visual requirements, including simultaneous translation services that will be offered in both French and English. Reduced rates for accommodations in Strasbourg are currently being researched. A portion of the conference costs will be paid for with the assistance of local subsidies. Further details regarding conference registration will be available in the coming months. In the meantime, please save the date! For more information, you may contact Dominique Pfeiffer, president of the French association GENERATION 22, at [dom.pfeiffer@wanadoo.fr](mailto:dom.pfeiffer@wanadoo.fr)

## RISK BEHAVIORS

Wendy Kates PhD.

Upstate Medical University Hospital      Syracuse, NY

During the past decade, clinicians and researchers who work with families of children with VCFS have learned that children with VCFS often develop behavioral difficulties during childhood. Compared to typically developing children of similar age, children with VCFS exhibit a higher incidence of attention deficit hyperactivity disorder and phobias (primarily fears of dark and of noises). Children may also display mood swings, irritability, and social withdrawal. Usually, with some guidance from professionals, families are able to manage these difficulties as their children develop.

As most families of children with VCFS are aware, however, some children develop more severe psychiatric disorders as they approach adulthood. Some symptoms of these disorders can appear during adolescence. Researchers are currently trying to identify factors that will help us understand which children are at risk for developing severe psychiatric disorder. Hopefully, within the next decade, we will develop a better understanding of the specific precursors of psychiatric illness in children with VCFS. We anticipate that this knowledge will allow us to provide early mental health intervention to the children who may be at greatest risk. Meanwhile, many families would like to know what behaviors or symptoms they should be "on the look-out for" now.

We have listed below several changes in behavior that might warrant a psychological or psychiatric consultation. These include:

- Increase in anxiety or depression, associated with:
  - Regressive behaviors
  - Clingy behaviors
  - Change in appetite or sleep patterns
  - Increase in acting out behaviors
  - Decreased interest in previous activities
  - Sudden decline in academic performance
- Increase in intense irritability or mood swings
- Increase in voluntary social withdrawal and social isolation
- Significant decrease in personal hygiene
- Increase in odd, unusual or disorganized thoughts or behaviors

Only a minority of children with VCFS develops severe psychiatric illness, and for those that do, scientists are currently investigating strategies for effective treatment. Hopefully, continued collaboration between parents, clinicians and researchers will enable us to acquire the knowledge we need to enhance the quality of the lives of all individuals with VCFS.

The VCFS Educational Foundation relies on dues and contributions to fulfill its mission to disseminate educational information about the syndrome. We gratefully acknowledge the following individuals for their donations, which were made since our last newsletter.

### In Honor of Allison Robbins

Kevin & Amy Bell  
Lynn & Bob Cohen  
Judith & Henry Felber  
Marc Labgold  
David E Levanson  
Joel & Monika Levanson  
Suzanne & Mark Pearson  
Nancy & John Robbins (with 2:1 matching gift from Fannie Mae Foundation)  
Tem Weisman

### In Memory of Hangal Kusumaker

Stuart Kushner

### In Memory of Donald Anderson

Kenneth Balagur  
Dr. & Mrs. Wilbur Blackey  
Brian & Lois Callahan  
Mr & Mrs. Joe Carr  
Brian & Julie Cooper  
Denis & Donna Drapea  
Barbie & James Doherty  
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Manuel & Kathleen Mendonca and sons  
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Amy Johnson  
Diane Kelly  
Michael & Sandra Lynch  
Norraine Wylonis

### In Memory of Myrtle Moor Setzer

Lenore Cohen

## VELO-CARDIO-FACIAL SYNDROME – A MODEL FOR UNDERSTANDING MICRODELETION DISORDERS

Review by Kieran C Murphy April 2005

Editors: Kieran C. Murphy and Peter J. Scambler  
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In July 2005, Cambridge University Press will publish one of the most comprehensive books on Velo-cardio-facial syndrome (VCFS) ever published. Everyone who has had contact with someone with VCFS knows that affected individuals often have a multitude of physical and psychological problems associated with this condition. This often causes great problems for health-care professionals who may not be aware of the existence of such a disorder and, even if they are aware, their knowledge is usually confined to the area of the body covered by their own discipline. Over the years, Pete Scambler and I have had many conversations both with parents and professionals about these issues and, arising out of these discussions, we decided to publish a book on VCFS with contributions from many of the leading international authorities on VCFS.

The following is an extract from our Forward: "VCFS patients may present at many different clinics given the protean nature of the condition – over 100 different manifestations have been described in the literature. This book attempts to summarise the rapid progress that has recently been made in understanding and treating people with VCFS. We hope that publication of this book will be useful for several reasons: (1) professionals studying or treating one aspect of VCFS are often relatively unaware of the involvement of other systems and this book will assist them in obtaining a more holistic view of people with VCFS (2) VCFS may be seen as a paradigm for other less common microdeletion disorders

and experience with VCFS may help to direct research and treatment strategies across a range of other microdeletion disorders (3) while this book emphasises the clinical issues relevant to VCFS, it also reflects the increasing recognition that an understanding of relatively rare disorders such as VCFS can tell us much about more common conditions, such as predisposition to psychiatric illness (4) the study of the embryological basis for the structural malformations observed in VCFS is helping to uncover some basic mechanisms of developmental biology."

In Chapter 1, Bob Shprintzen gives an historical introduction to VCFS and describes in detail how recognition of VCFS as a specific syndrome progressed from the initial description in 1955 by Sedlackova to the present day. In Chapter 2, Katrina Prescott and Pete Scambler outline the amazing developments that have taken place in our understanding of the molecular genetics of VCFS over the past decade or so. The major systems affected in VCFS are then discussed in subsequent chapters; Bruno Marino and colleagues discuss cardiovascular disease, Richard Kirschner outlines palatal and velopharyngeal impairments while Koen Devriendt and colleagues detail the nephro-urologic, gastrointestinal and ophthalmic findings. In view of the importance of immunodeficiency in VCFS, Kathleen Sullivan gives a detailed account of the problems facing people with VCFS with extremely helpful practical guidelines for both professionals, people with VCFS and their families.

The brain is very commonly affected in VCFS and we have devoted three chapters to the brain in this volume. Angela Stevens and I discuss behavioural and psychiatric disorder in both children and adults with VCFS with practical guidelines for treatment, Linda Campbell and Ann Swillen give a very comprehensive account of cognitive function in VCFS explaining how information is processed in the brain of people with the deletion while Stephan Eliez and Therese van Amelsvoort outline how examining the brain using MRI scanning has increased our understanding of the structure and function of the brain in people with VCFS. In the final chapters, Karen Golding-Kushner writes about speech and language disorders, Donna McDonald-McGinn and Elaine Zackai discuss the issues that occur in genetic counselling for people with VCFS and their families and provide helpful practical guidelines for professionals and affected families. Finally, because every VCFS person is part of a family, Julie Squair gives a very frank and heart-felt account of the impact that a diagnosis of VCFS can have on the immediate and extended family.

There has been considerable excitement at how rapidly our understanding of VCFS has evolved, particularly over the past decade and Pete and I have sought to convey this excitement in this book. You will have to judge for yourselves whether or not we have been successful!

## LETTER FROM THE PRESIDENT

As President, one of my primary duties is to help plan the annual conference. Because I am physically located in VA, planning a meeting in Syracuse could have been quite challenging without the assistance of several Foundation members located in Syracuse. Dr. Shprintzen, Ann Marie Higgins, Eileen Marrinan, and Beth Wallace have gone above and beyond the call of duty in helping me with this huge task. They have taken care of arranging the local accommodations, organizing the Kids' Club, planning social events and securing entertainment. Each week we meet via conference call, and I am so amazed by the energy and enthusiasm they all have! Everyone is working so hard to make this meeting a success. I am very grateful to have their support!

This year we are adding some new things to the program to include: a primer session for new attendees, relevant books for sale, entertainment at the Friday night dinner and resources for attendees to take home, just to name a few. We have also built in more time for questions and answers.

Because the "Syracuse team" is taking care of so much of the planning, I have had more time to devote to some projects I have wanted to see come to life. With the help of a few Foundation members, especially Lisa Jennings and Maureen Anderson, we came up with a slogan "Knowledge is Hope" and have ordered blue awareness wristbands displaying the slogan on the outside with the Foundation website on the inside of the band. The bands have been going quickly and we have already had to place a second order to replenish our supply of child-sized bands. In the meantime, Kelvin updated our website so wristbands can be ordered online easily. He is also creating a Donation button on the homepage so that people can easily make a contribution to the Foundation. We have just finalized a Foundation logo incorporating the slogan "Knowledge is Hope" with a design that reflects our international mission. You have already seen the logo at the top of this newsletter and you will soon see the logo on our website and on other Foundation materials. Finally, we have drafted an informational brochure that we are now editing. We hope to have the brochure completed and available at the conference and would like to see it in the office of every pediatrician in the country! This will be the first of a series of brochures that will be distributed internationally to doctors, educational facilities and specialists. The first brochure will be a basic overview of VCFS to be given to anyone who is not familiar with the syndrome. Subsequent brochures will be specialty based – such as speech, cardiology, psychiatry, etc. The Amanda McPherson Foundation has generously donated money to make this project possible. We are very appreciative of their continued support.

While I was a bit hesitant about accepting the position of President, I am thrilled to have the opportunity to be able to give something back to the Foundation, which has been such a source of information and support to me and my family. I look forward to seeing you all in July!

Nancy Robbins

Wife to John and mom to Allison (10) with VCFS and Steven (12)

### Knowledge Is Hope Bracelets

Promote VCFS awareness by purchasing "Knowledge is Hope" wristbands, available in Adult size (8 1/4") and Child size (7 1/4"). The wristbands are made of 100% silicone and read **KNOWLEDGE IS HOPE** on the outside of the band, with [www.vcfsef.org](http://www.vcfsef.org) on the inside.

**Each wristband is \$1.50 US and shipping is FREE!!!** Due to shipping costs, we suggest a minimum order of 5 wristbands for international purchases.

You can purchase your bracelets directly from the foundation's web site at <http://vcfsef.org/Donations/wrist.html>. Online payments are processed through PayPal.

For questions regarding wrist bands, contact Nancy Robbins at [nirobbins@aol.com](mailto:nirobbins@aol.com).



## UPDATE FROM AUSTRALIA

Well, the Aussies have been busy!

We in the Queensland Foundation have been extremely busy with our two major projects. The first is the establishment of the first ever multi-disciplinary VCFS Clinic in Australia – at the Mater Children’s Hospital in Brisbane. And the second, of course, is the planning for the conference here in Brisbane from 2 to 4 November, 2006.

On top of this, we continue with our usual work, keeping VCFS on the map here. We are busy planning to attend a number of conferences here in Australia, not only to raise the profile of VCFS in the medical and therapy professions, but also (shamelessly) to promote the 2006 VCFS Conference. We will be at the World Congress of Neurology, Sydney, 5 to 11 November, 2005; the Society for the Study of Behavioural Phenotypes Conference, Cairns, 6 to 8 October, 2005 and the Conference for the International Association for Paediatric Dentistry, Sydney, 31 October to 5 November, 2005.

All of this work has required our Foundation to take a very important step. We have hired a full time executive. Our Executive Officer is Ms Joanna Friesen. Jo’s contact details are:-

Ms. Jo Friesen  
Executive Officer  
VCFS Foundation (Qld) Inc

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Australia

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The planning for our annual fundraiser – the Faces of Sunshine Ball – is well underway. The ball this year is on 1 October, 2005 and promises to be a great night. We are looking forward to beating last year’s fundraising effort – and we have every reason to expect we will!!!

Which brings me back to our two major projects. In my last update, I mentioned that we were aiming to open our multi-disciplinary clinic by the 2006 conference, but we were hoping to announce the opening of the clinic this year. Well, we are almost there. In fact, we have set a date of 1 July, 2005 to open our doors. Where? At one of Australia’s best children’s hospitals, The Mater Children’s Hospital, Brisbane.

The Mater is uniquely placed to provide the holistic service which VCFS kids need. It has a wealth of key professionals in cardiac, speech and language and paediatric care, and in child and mental health. Professor Brett McDermott, head of the Mater Child and Youth Mental Health Service has established a pre-eminent clinical service for the care of young people with significant emotional and behavioural difficulties and complex mental health disorders. Brett and his team are committed to establishing the VCFS Clinic at the Mater as a Centre of Excellence with the help of the Queensland Foundation, and our overseas partners.

The VCFS Centre will eventually be housed in its own building at the Mater. It is planned as a stand-alone facility, with a significant identity. We will create a domestic, creative, strengths-based Centre that facilitates early intervention and an approach that optimises the potential of each child. The

traditional health-care environment will not be emphasised. As of March 2005, an independent estimate of indicative project costs for the Centre was AU\$2.7 million, within a solution that included the Mater Health Services donating appropriate land on the Mater Health Campus.

Because VCFS is not well known in Australia, our Foundation has recognised the need to seek help in establishing and running the clinic from the world’s experts in VCFS. We have commenced discussions to establish the Brisbane VCFS Clinic as part of an international consortium of centres of excellence for VCFS around the world and hope to develop this exciting concept over the coming months. This ambitious project underscores the value of the VCFS Educational Foundation Inc – all of our work in Australia has been inspired and supported by various officers of the Foundation. We simply couldn’t have achieved what we have, and what we hope to achieve, without the support from our friends all over the world – all of whom are associated with the Foundation.

I will keep you all informed as we go.

Our second major project – the VCFS Conference in Brisbane from 2 to 4 November, 2006 – is well in hand. Our present status is:-

1. We already have commitments from a substantial international faculty. One of the suggestions for Brisbane 2006 (an excellent one, I think) was to ask each overseas speaker/faculty member to partner a local speaker/faculty member. We will be implementing this

*(Continued on page 12)*

## NEW IMMUNOLOGIC FINDINGS MAY EXPLAIN THE BASIS FOR LOW THYROID FUNCTION

Joseph A. Bellanti M.D.  
Georgetown University Medical Center , Washington D.C.

Scientists at Georgetown University Medical Center studying 3 young pre-adolescent females with VCFS and hypothyroidism, found the presence of autoimmune thyroid antibodies in the sera of each of the children. Although hypothyroidism has been occasionally detected in patients with VCFS/DGS and has recently been reported in a Tbx1 mouse model of VCFS/DGS, the precise basis for the low thyroid function is unknown. One theory ascribes the condition to downstream molecular effects of altered TBX1 dosage in human embryonic development resulting in malformation of the thyroid gland. In their study of the children at the Georgetown University Medical Center Department of Pediatrics International Immunology Center, Dr. Julia Mendez de Inocencio and Dr. Joseph A. Bellanti initially suspected hypothyroidism in their patients by mild elevations of blood lipids and slight increases in liver enzymes which are sometimes seen in patients with low

thyroid function. Reasoning from the observation that the onset of the condition had begun during the prepubertal period of each patient and not an earlier age which would be more in keeping with a gene-related developmental defect, the two doctors asked if the cause could be more likely related to an injurious attack of the patients' immune systems directed against their own thyroid tissues, a condition referred to as autoimmune thyroiditis or Hashimoto's disease, which can be diagnosed by the finding of elevated serum anti-thyroid antibodies. The occurrence of other autoimmune diseases, such as JRA, ITP, AHA, IBD, has been observed with an overall frequency of 9% and was reported at last year's meeting by Dr. Kathleen Sullivan. The occurrence of autoimmune thyroiditis, however, has not been previously reported in the published literature.

The results of this preliminary study

will be reported at the 2005 VCFS Conference in Syracuse and is in keeping with the theme of this year's conference on INTERVENTION and its focus on treatments illustrates how research data may eventually result in treatment options. Since the behavioral symptoms may not only be seen in VCFS but also may accompany hypothyroidism, it is critically important that a correct diagnosis be made. Children with VCFS presenting with symptoms of hypothyroidism, i.e, sluggishness, behavioral disturbances unusual weight gain, should be first evaluated for a thyroid condition before reaching adolescence and before referral for psychological or behavioral assessment. This is not only essential for making the correct diagnosis but also crucial in prescribing the correct treatment of thyroid administration and avoiding unnecessary use of psychotropic drugs and their associated potential adverse effects.

## AUSTRALIA

*(Continued from page 11)*

- suggestion.
2. Similarly, many local faculty have confirmed their speaking engagements.
  3. There will be many more local speakers and attendees from the Brisbane Clinic. Our first specialists' evening for the Clinic will be held in the next two weeks.
  4. Our venue and dates are of course confirmed - Carlton Crest Hotel, Ann Street, Brisbane, 2 to 4 November, 2006.

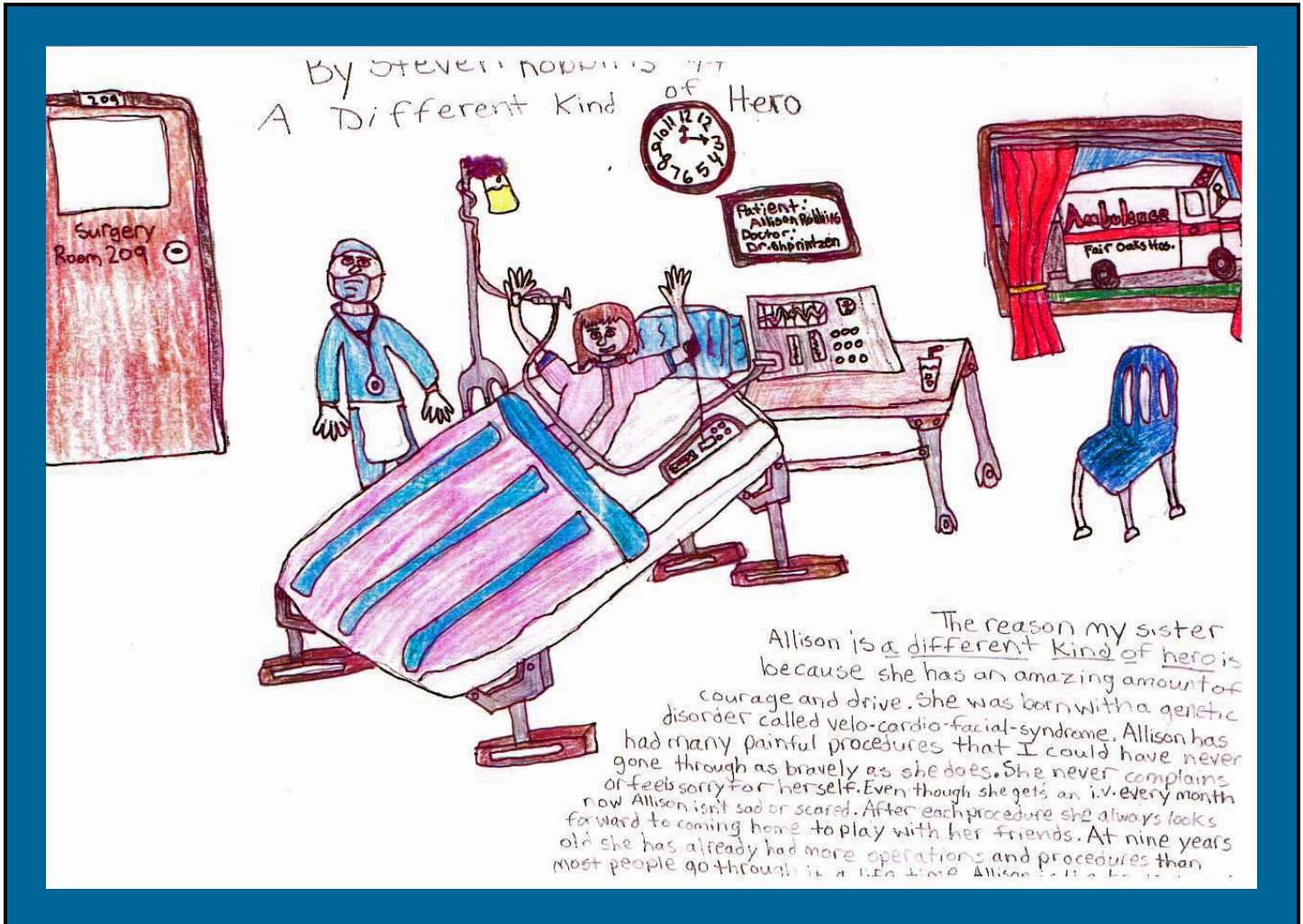
5. We will be in Syracuse with another promotion team (headed up by Jo Friesen) with literature and promotional material. This will definitely include firm travel and accommodation packages with prices.

I look forward to seeing you all in Syracuse in July.

Steve Russell  
President  
VCFS Foundation (Old) Inc  
Regional Director  
Australia/Asia

## A DIFFERENT KIND OF HERO

Steven Robbins, age 12 years, submitted this drawing and story to the National PTA Reflections Program – an arts recognition and achievement program for students. The theme was “A Different Kind of Hero”. Steven is the son of John and Nancy Robbins and brother to Allison. His mother, Nancy is the current president of the VCFS Educational Foundation.



*“The reason my sister Allison is a different kind of hero is because she has an amazing amount of courage and drive. She was born with a genetic disorder called velo-cardio-facial syndrome. Allison has had many painful procedures that I could have never gone through as bravely as she does. She never complains or feels sorry for herself. Even though she gets an IV every month now Allison isn't sad or scared. After each procedure she always looks forward to coming home to play with her friends. At nine years old she has already had more operations and procedures than most people go through in a life time. Allison is the bravest girl I know and has taught me to overcome my fears.”*

Steven Robbins



# 11<sup>th</sup> Annual International Scientific Meeting

## The Velo-Cardio-Facial Syndrome Educational Foundation, Inc.

### Sheraton University Hotel and Conference Center

### Syracuse, NY, July 29-31, 2005

Jointly sponsored by The Velo-Cardio-Facial Syndrome Educational Foundation, Inc. and The Office of Continuing Medical Education, College of Medicine at SUNY Upstate Medical University.

This activity has been planned and implemented in accordance with the Essential Areas and policies of the Accreditation Council for Continuing Medical Education through the joint sponsorship of SUNY Upstate Medical University and Velo-Cardio-Facial Syndrome Educational Foundation, Inc. SUNY Upstate Medical University is accredited by the ACCME to provide continuing medical education for physicians.

SUNY Upstate Medical University designates this continuing medical education activity for a maximum of 18.50 category 1 credits toward the AMA Physician's Recognition Award. Each physician should claim only those credits that he/she actually spent in the activity.

The SUNY Upstate Medical University Communication Disorder Unit is approved by the Continuing Education Board of the American Speech-Language-Hearing Association (ASHA) to provide continuing education activities in speech-language pathology and audiology. This program is offered for up to 1.8 CEU's (intermediate level; professional area). ASHA approval of CE provider status does not imply endorsement of course content, specific products or clinical procedures.

#### Friday, July 29, 2005 – Pre-meeting Primer Sessions

- 11:00 A.M.: Speech and language development and speech problems in VCFS – Eileen Marrinan
- 11:00 A.M.: Genetics for nongeneticists – Robert J. Shprintzen
- 11:00 A.M.: Brain Imaging in VCFS – Wendy Kates
- 11:00 A.M.: Teen Get-Together 1: Making Plans, Alexandra Oppenheimer, M.Ed.

#### 12:00 Noon – Opening Remarks

Welcome from the VCFSEF – Karen J. Golding-Kushner, Ph.D., Executive Director

Nancy Robbins, President

Welcome from Upstate Medical University

#### Moderator: Susan Marks, Childrens Hospital of Wisconsin, Milwaukee, WI, USA

#### Speech Disorders and Their Treatment:

- 12:10 – 12:30: *Velopharyngeal Function in VCFS: What Advanced Diagnostic Techniques Have Taught Us*, Robert J. Shprintzen, Upstate Medical University, Syracuse, NY.
- 12:30 – 12:45: *Asymmetry of the Pharynx, Larynx, and Palate*, Burke Chegar, Upstate Medical University, Syracuse, NY
- 12:45 – 12:55: *Muscle Volume of the Palate in VCFS*, Robert Brown, Wendy Kates, Robert J. Shprintzen, Upstate Medical University, Syracuse, NY
- 12:55 – 1:10: *Facial Nerve Function in Individuals with VCFS and Facial Asymmetry*, Antonio Ysunza, Hospital Gea Gonzales, Mexico City, Mexico
- 1:10 – 1:30: *Surgical Management of Hypernasal Speech in VCFS*, Sherard A. Tatum III, Upstate Medical University, Syracuse, NY
- 1:30 – 1:45: Questions and Comments
- 1:45 – 2:15: *The Computer in the Speech Clinic for: Therapy, Productivity, and Resource*, John E. Riski, Ph.D., Children's Health-care of Atlanta, Atlanta, GA

- 1:45 – 2:15: *The Computer in the Speech Clinic for: Therapy, Productivity, and Resource*, John E. Riski, Ph.D., Children's Health-care of Atlanta, Atlanta, GA
- 2:15 – 2:40: *Speech Therapy to Correct Articulation Impairment in VCFS*, Karen J. Golding-Kushner, Ph.D., East Brunswick, NJ
- 2:40 – 2:55: Questions and Comments
- 2:55 – 3:05: Refreshments

**Moderator: Shane Zim, University of Southern California, Los Angeles, CA**

**Molecular Genetics and Developmental Biology Presentations:**

- 3:05 – 3:30: *Genetic Control of Development in VCFS*, Bernice Morrow, Albert Einstein College of Medicine, Bronx, NY
- 3:30 – 3:45: *Interactive Partners for Tbx1 in Skeletal Muscle Development*, Evan Braunstein, Albert Einstein College of Medicine, Bronx, NY
- 3:45 – 3:55: Questions and Comments

**Pediatric Issues, Moderator: Anne Marie Higgins, Upstate Medical University, Syracuse, NY**

- 3:55 – 4:15: *A Primer on the Immune System: When and How to Look for Problems*; Joseph Domachowske, Upstate Medical University, Syracuse, NY
- 4:15 – 4:40: *Self-Hypnosis to Reduce Anxiety in Patients with VCFS*, Ran Anbar, M.D., Kim Hummell, C.S.W., Upstate Medical University, Syracuse, NY
- 4:40 – 4:50: Questions and Comments
- 4:50 – 5:00: **VCFS Around the World**  
*Developments in Australia*, Stephen Russell, Brisbane, Queensland, Australia  
*Developments in Argentina*, Antonio Ysunza, Mexico City, Mexico
- 5:00 – 6:30: Business meeting
- 6:30: **Annual VCFS Dinner (optional)**

**Saturday, July 30, 2005**

**Breakfast with the experts**

- 7:30 – 8:45 A.M.: Psychosexual aspects of VCFS, Doron Gothelf, Merav Burg, Schneider Children's Hospital, Petah Tiqwa, Israel  
 Leg Pains, Discussant to be announced  
 Feeding Issues: Bob Beecher, Jill Merrow

Poster Session: *Single Case Study of Metyrosine Treatment for Schizophrenia in VCFS*, Martin Debbané, Bronwyn Glaser, Stephan Eliez, Geneva, Switzerland

*Brain Imaging in VCFS: where will the studies lead us?*

**Moderator: Doron Gothelf, Schneider Children's Medical Center, Petah Tiqwa, Israel and Stanford University, Stanford, CA**

- 9:00 – 9:30: *Brain Structure and Function in VCFS: Does COMT Make a Difference?* Wendy Kates, Upstate Medical University, Syracuse, NY, USA
- 9:30 – 9:45: *Multiple Attention system Impairments in Children with VCFS*, Tony J. Simon, S. Ferrante, V. Nguyen, H. Ferrante, Donna McDonald-McGinn, Elaine Zackai, J. Bish
- 9:45 – 10:00: *Brain Structure, connectivity and function changes and their relation to*

- 9:30 – 9:45: *Multiple Attention system Impairments in Children with VCFS*; Tony J. Simon, S. Ferrante, V. Nguyen, H. Ferrante, Donna McDonald-McGinn, Elaine Zackai, J. Bish
- 9:45 – 10:00: *Brain Structure, connectivity and function changes and their relation to cognitive impairments in VCFS*; Tony J. Simon, J. Bish, L. Ding, Machado, Nguyen, Donna McDonald-McGinn, Elaine Zackai, Gee, University of California, Davis, Sacramento, CA
- 10:00 – 10:15: *Polymicrogyria in VCFS*; Nat Robin, University of Alabama, Birmingham, Birmingham, AL
- 10:15 – 10:30: Questions and Comments
- 10:30 – 10:45: Refreshments

### ***COMT and VCFS and Treatment Effects***

**Moderator: Tony Simon, University of California, Davis, Sacramento, CA**

- 10:45 – 11:00: *The Effect of Genetic Variation in COMT on Risk for Psychiatric Disease Neural Function*; Andreas Meyer-Lindenberg, National Institute of Mental Health, Cognition and Psychosis Program, Bethesda, MD
- 11:00 – 11:15: *COMT Polymorphisms and Language Impairment in VCFS*; Eileen Marrinan, Upstate Medical University, Syracuse, NY
- 11:15 – 11:30: *Inconsistent Evidence for an Effect of COMT Polymorphisms on Brain Morphometry and Cognition in VCFS*; Bronwyn Glaser, Martin Debbané, Christine Hinard, Michael Morris, Sophie Dahoun, Stylianos Antonarakis, Stephan Eliez, University of Geneva School of Medicine, Geneva, Switzerland.
- 11:30 – 11:50: *Drugs in the Management of Behavior in VCFS*; Wanda Fremont, M.D., Upstate Medical University, Syracuse, NY
- 11:50 – 12:05: Questions and Comments
- 12:05 – 1:30: Lunch with the Experts**
- Early Speech Development: Karen Golding-Kushner, Susan Marks, Eileen Marrinan, John Riski
- Genetic Implications: Nat Robin, Robert Shprintzen, Bernice Morrow
- Chronic Illness and Its Impact on Families, David Keith, Anne Marie Higgins
- 12:05 – 1:30:** Teen (High School) and Young Adult Pizza Lunch, Alexandra Oppenheimer, M.Ed., Karen Lindsay

**Saturday P.M., Moderator: Wendy Kates, Syracuse, NY**

### **Psychiatric and Behavioral Treatment of VCFS**

- 1:30 – 1:55: *Mentoring Program as an Effective Psychosocial Intervention in VCFS*; Merav Burg, Tamar Steinberg, Schneider Children's Hospital, Petah Tiqwa, Israel Doron Gothelf, Schneider Children's Hospital, Petah Tiqwa, Israel and Stanford University, Stanford, CA
- 1:55 – 2:20: *Longitudinal Study of Behavior in VCFS*; Doron Gothelf, Stanford University and Schneider Children's Medical Ctr., Petah Tiqwa, Israel; Allan L. Reiss, Stanford University, Stanford, CA
- 2:20 – 2:40: *Social Skills Training & VCFS: The Moderating Influence of ADHD*; Kevin Antshel, Ph.D., Upstate Medical University, Syracuse, NY
- 2:40 – 2:55: Questions
- 2:55 – 3:05: Refreshments

### **Neuropsychological and Psychiatric Findings in VCFS**

- 3:05 – 3:30: *Relative Strengths and Weaknesses in Visuo-Spatial Abilities in Individuals with VCFS*; Bronwyn Glaser, Martin Debbané, Stephan Eliez, University of Geneva School of Medicine, Geneva, Switzerland
- 3:30 – 3:50: *Cognitive and Psychosocial Aspects of Children with VCFS*; Neil Nicoll, Belinda Barton, Meredith Wilson, The Children's Hospital at Westmead, Westmead, NSW, Australia
- 3:50 – 4:05: Questions

**Chronic Problems in VCFS****Moderator: Nancy Robbins, Oakton, VA**

- 4:05 – 4:20: *Autoimmune Thyroiditis as a Previously Unrecognized Cause of Hypothyroidism in VCFS*; JI Mendez-Inocencio, MB Bueso, JA Bellanti, Georgetown Univ. Med. Ctr., Washington, D.C.
- 4:20 – 4:35: *Use of Intravenous Immunoglobulin (IVIg) Therapy in Patients with VCFS: Replacement or Immunomodulatory Effects?* JI Mendez-Inocencio, JA Bellanti, Georgetown University Medical Center, Washington, D.C.
- 4:35 – 4:45: *A Perspective on Leg Pain*, Marc Heatherington,
- 4:45 – 5:00: *Annual Meetings in 2006: An International Affair*  
Stephen Russell, Brisbane Australia  
Dominique Pfeiffer, Strasbourg, France
- 6:00: **Museum of Science and Technology and Dinosaur BBQ (optional)**

**Sunday, July 31, 2005****7:30 – 8:45: Breakfast with the Experts**

Speech therapy for school age children and later, K Golding-Kushner, S Marks, E Marrinan, J Riski  
 Planning for Your Child's Adult Life, Donna Landsman, Jeff Landsman, Madison, WI  
 Telling Your Child with VCFS about VCFS, Nat Robin

**7:30 – 8:45: Teen (High School) and Young Adult Breakfast, Alexandra Oppenheimer, Karen Lindsay****Cardiology, Molecular Genetics, Gene Interactions: Moderator – Nat Robin**

- 9:00 – 9:30: *VCFS: The Heart of the Matter*, Roger Ruckman, Children's National Medical Center, Washington, D.C.
- 9:30 – 9:45: *Alteration in neural crest cell function by inactivation of Tbx1 in mouse models*, Vimla Aggarwal, Albert Einstein College of Medicine, Bronx, NY
- 9:45 – 10:00: *Identification of genes in the genetic pathway downstream of Tbx1 by microarray analysis*, Jun Liao, Albert Einstein College of Medicine, Bronx, NY
- 10:00 – 10:15: *Understanding the basis for the 22q11 deletion*, Melanie Babcock, Albert Einstein College of Medicine, Bronx, NY
- 10:15 – 10:30: Questions and Comments
- 10:30 – 10:45: Refreshments

**Developmental and Educational Issues****Moderator: Maureen Anderson; Salem, NH, USA**

- 10:45 – 11:00: *Developmental Milestones in VCFS*; Nuria Abdul Sabur, Nancy Roizen, M.D., Upstate Medical University, Syracuse, NY
- 11:00 – 12:00: *Education, Advocacy, and Wrights Law: helping your child achieve*; Donna Landsman, M.S., Madison, Wisconsin, USA; Judy Gaughran, Boston, MA; Yolanda Ortiz, Cedar Grove, NJ; Debbie Lightfoot, Syracuse, NY
- 12:00 noon: Questions and Comments

**Adjournment**

Target Audience (CME): All specialties treating children or adults with VCFS, including otolaryngology, psychiatry, pediatrics, neurology, radiology, GI, pathology, genetics, all pediatric subspecialties.

Non-physician target audiences: speech-language pathologists and audiologists, psychologists, neuropsychologists, social workers, genetic counselors, dental specialists, nurses, physical therapists, occupational therapists, others involved in developmental disorders or the care of children with multiple anomalies, parents of children with VCFS, and individuals with VCFS.