



# VCFSEF NEWS

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## LIVING WITH VCFS: MY STORY

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I did not ask to be born with VCFS. In fact, countless times I have despised the syndrome as if it were a terminal illness. Even today, although I have overcome many obstacles there is always a part of me who wishes I could be "normal."

As a young child, I was very shy, clingy, and rarely wanted to leave my mother's side, even to walk. Walking was always very painful and I tired easily. Growing up, I was often lonely even though I had friends because I lacked self-esteem that many youngsters are plentiful in. It was not until college, and I found the guidance and comfort of my best friend and old college roommate, Angie that I was truly happy and comfortable in social situations. My friends in college taught me how to love myself, and that my idea of normality doesn't exist, which in turn has helped me to accept my having VCFS, and become at peace with it.

As a school-aged youngster and later an adolescent, around middle school I began to have difficulties in academia, especially in the areas of math and science. Of course, back then my parents had a very vague picture of VCFS and what sort of problems I would encounter both physically and emotionally. Middle school was as for most adolescents, extremely difficult and I faced my first rounds of teachers who quite frankly didn't care two cents about whether I passed or failed. Little did I know this would follow me through all the way to earning my Baccalaureate degree in Music at the University of Southern Maine.

What made all the difference in the world, however, was my family. They have endlessly supported me, endured many emotional break-downs, and of course many triumphs as well. My parents allowed me to enroll in ballet class over 10 years ago, and that has given me an outlet for my emotions, and often times pure joy. After forcing me to attend summer camp at age 13, little did Mom and Dad know I would be a counselor throughout my years of college. They have allowed me to try and explore different avenues in order to find my forte in life. Piano lessons, flute lessons, summer basketball, music camp etc. And every time I started to feel normal and be a kid again, it seemed as if I landed in the hospital again, for yet another surgery.

I am not trying to scare parents of young children with VCFS. I am simply being as honest as possible.  
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...for the International Scientific Conferences

**Info on our web site:**

- **2006:** July 7-9, 2006  
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- **2006:** November 2-4, 2006  
Brisbane, Australia

## MY STORY ...

*(Continued from page 1)*

ble, and give you hope that your child can succeed if you give them the endless support and love my parents, family, and friends have shown me. A member of the University of Southern Maine School of music class of 2006, I will graduate as a Russell Scholar and Linguistics Minor as I receive my BA in music this spring. Although a graduating senior, my fears for the future are still evident and strong, but at all costs, I have hope. Hope that I will obtain my master's in child life and be a therapist to young cancer patients, hope in having a successful marriage just like my own parents, and maybe even children someday. My struggles in life due to VCFS have been far more plentiful than triumphs, but for every struggle I have, my next triumph will be ten times greater, in turn making me a better and stronger person in the end.

## LIVING WITH VCFS: A PARENT'S STORY

**Elliott Epstein**

When my daughter, Ariel, was born on July 25, 1982, I was the first person besides the obstetrician to hold her in my arms. A program at our local hospital, one designed to give fathers a greater role in the birthing process, allowed me to be present in the delivery room. In fact, the physician handed Ariel to me even before my wife, Ellen, had the chance to see or hold her. The wave of emotion, a mixture of awe and joy, which swept over me at that moment was unlike anything I have ever experienced. Ariel was my first biological child, and, through her, the miracle of birth became a reality for me instead of just a cliché.

If I had a mental picture then of what Ariel would be like as she grew, it was probably no more than a series of Hallmark greeting-card images – of a smiling, gapped tooth girl in a lacey party dress, a willowy teenager in a fashionable prom outfit, a graceful, confident young woman in a graduation cap and gown. I had no inkling, of course, that Ariel had VCFS (a diagnosis that would not be made for another 14 years) or of the daunting challenges that she, Ellen and I would face as a result of that condition. Hallmark does not make cards for VCFS moments. Perhaps it was just as well that I had no such foreknowledge, because it might have led to an early sense of hopelessness and would not have prepared me in any event for the challenges ahead.

VCFS manifests itself differently in every person whose 22d chromosome carries the genetic deletion, and the severity of its symptoms can vary widely. Ariel is considered one of the "lucky" ones. Unlike many with VCFS, she has no life-threatening cardiac problems and functions at a high intellectual level. Her major physical problems have been corrected by skilled surgeons at Boston's Children's Hospital: pharyngeal flap surgery at age 13, which eliminated most of her nasality, and lumbar-fusion surgery a little

more than a year ago, which checked the deterioration of, and partly corrected, her scoliosis.

Fortunate as Ariel may be to have only a mild form of VCFS, it has led to a variety of physical, cognitive and emotional problems and limitations. The broad-ranging effects of the syndrome have made her life a constant struggle, and the social and emotional impact has been profound. Individually and in combination, they sapped her self-confidence at an early age, and rebuilding that confidence has been a slow and arduous process.

Because of her non-verbal disability, Ariel tends to "think out loud," verbalizing her thoughts over and over again in order to process them, a mannerism which puts many listeners off. Her anxiety causes her to worry excessively about making new acquaintances, about adapting to novel situations and about her future in general; consequently her first (although fortunately not always her last) instinct is to cling to the familiar and avoid change. When under stress, she sometimes acts awkwardly or inappropriately in social settings. Her self-esteem is fragile, and she needs a good deal of reassurance and support. She does not "roll with the punches," and setbacks and disappointments have often sent her into an emotional tailspin. Her learning disabilities, coupled with low energy and frequent illness, have raised the cost, both emotional and physical, of academic success.

Yet for all these problems, Ariel has a tenacious will to succeed and a remarkable track record of success. Through courage and perseverance, she has accomplished a great deal in the face of adversity. Her mother and I have watched with pride as she has repeatedly set and attained ambitious goals that would have been

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### 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.*

## A PARENT'S STORY ...

*(Continued from page 2)*

beyond the reach of most others laboring under similar handicaps. Again and again, she has rebounded from failure, overcome her fears and pushed forward into risky territory. Her constant theme is her desire to have a "normal" life. To her that means an advanced academic degree, a career, marriage and children. If the past is any indication, I believe she will achieve her goals.

Ariel has demonstrated many times that she can accomplish what she sets her mind to. In high school, she graduated in the top 10% of her class. Despite physical limitations, she enrolled in a local dance program, studied ballet and modern dance, and participated in recitals for over 10 years. She is a year away from completing her bachelor's degree at the University of Southern Maine's School of Music, a rigorous five-year program taught by conservatory-trained musicians who are far more interested in producing a crop of talented performers than in nurturing students with learning disabilities. She has achieved considerable mastery over her chosen instrument, the flute, and has performed in numerous band concerts, ensembles and individual recitals. During the summers, she has worked as a counselor and volunteer at various camps, including her favorite, Camp Sunshine, which serves critically ill children and their families. Indeed, her current plan is to work as a child-life specialist or recreational therapist with young cancer patients.

Ariel has had some singular advantages in her struggle to cope with VCFS. She has had a stable home life (Ellen and I having been married and resident in the same community for over 25 years), a close-knit and loving extended family, lots of educational and cultural opportunities, and, above all, the most devoted mother one could ask for. Ellen has single-mindedly dedicated herself to our daughter's welfare. She has brought to bear her own considerable experience in education and social work, combined with her natural and intuitive understanding of children, to see that Ariel's needs are met. She has spent countless hours nursing her through illness, counseling her in emotional crisis, helping her with schoolwork, advocating for her with school and health-care providers and generally organizing the logistical details of her life. Ellen has also educated herself about the various aspects of VCFS and related health issues and used that knowledge to insure that Ariel receives appropriate support and assistance.

There is no parents' handbook for raising a VCFS child. No one can provide you instructions on how to handle the intense pain of witnessing your child confront rejection, failure, frustration and despair. No one can supply you with the just the right soothing words to answer your child's questions, "Why me? Why can't I be normal? Why can't I be like everyone else?" No one can give you the wisdom to decide when to intervene to assist, protect or advocate for your child – whether at home, in school, on the playground, in the workplace, in the bureaucracy – and when to let her sort things out on her own. It's all a matter of trial and error,

and I can say, for my part, that I have had a lot more misses than hits on my scorecard.

It is not just that the challenges are intrinsically hard. I have also had to overcome many of my own inadequacies in order to try to become part of the solution instead part of the problem. By some cosmic irony, my own personality, style and approach are, in many significant ways, incompatible with Ariel's and antithetical to her needs. A few examples will suffice to illustrate the point.

I keep a tight rein on my feelings, and, while fairly articulate at expressing ideas, am very poor at expressing emotions. In the same vein, I view problems rationally and have little tolerance for those who approach things in an irrational fashion. I am impatient with conversation that does not get directly to the point. I do not put much stock in praise or the external trappings of success, believing instead that a good job is its own reward. I admire independence and self-reliance in people and become irritated with those who make constant demands on myself or others. This outlook and behavior, although not without its positive aspects, has proven far too rigid, intolerant and insensitive for my daughter's needs.

By contrast, Ariel is a bundle of raw emotions. She is voluble and talks constantly and repetitively about her thoughts and feelings. She is very unsure of herself, constantly checking with others to see if she is doing the right thing, turning to others for help at the first sign of difficulty, and seeking compliments and expressions of approval for the purpose of validating her self-worth.

When I balk at engaging repeatedly in the same conversation with Ariel, when I fail to commiserate with her during moments of self-pity, when I refuse to offer her a compliment for a trivial accomplishment, she typically becomes irritated with me and accuses me of "not caring" about her or "not listening" to her. As she becomes more upset with me, I become more irritated with her, and the sparks soon fly. My lack of patience and sensitivity must seem all the more inexcusable to her in light of her mother's patient and understanding approach. As Ariel once said to me in a moment of pithy clarity. "Mom listens to me. You just tell me to suck it up!"

Perhaps such interactions are not all that unusual in father-daughter relationships, but their importance seems much more heightened in the context of VCFS, because the needs of a "special needs" child are particularly acute and there is, therefore, less room for parental error. Over the years, I have tried to moderate my instinctive, but wrong-headed, approach, and, for the most part, Ariel and I have reached an accommodation. I try to do a better job of listening and not being judgmental, of de-emphasizing our differences, and of finding common ground (our love of music and quirky humor, for instance).

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# SIBLINGS OF CHILDREN WITH SPECIAL NEEDS: WHAT DO THEY EXPERIENCE AND WHAT ARE THEIR NEEDS?

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Siblings play an important role in the lives of their brothers and sisters with special needs. Like their parents, the siblings tend to experience concerns and distress. Therefore it seems that the siblings need to be guided and supported, but unfortunately their needs are often overlooked by professionals as well as parents and other family members.

The purpose of this article is to describe the common issues that siblings of children with disabilities are coping with, and to provide some guidelines for clinicians and parents.

## Research Findings

Research findings indicate that there are positive aspects in being the sibling of individuals with a developmental disability. Siblings of individuals with developmental disability tend to be empathic, altruistic, more tolerant to children with special needs, and more mature and responsible. They also have a greater sense of closeness to family, tend to be proud of their disabled siblings' achievements and are more appreciative of their own good health and talents. Some of them develop greater leadership skills, especially in areas where sensitivity to human needs and rights are important.

However, they also experience more distress during family conflicts, perceive their siblings as receiving unequal treatment from parents, and have higher rates of emotional and behavioral problems. There are tremendous concerns and difficulties that non-disabled sibling experience in daily lives.

Listed below are some of the common attitudes and emotions that siblings may experience;

- Fear that they might develop the same disability themselves or that it might be contagious. Older siblings may appre-

hend that in the future they will give birth to children with the same syndrome, or with other developmental disability.

- Guilt feelings that arouse while confronting the sibling's medical problems and other difficulties. Some of them are concerned 'why is it him that has the syndrome and not me?'
- Shame in the unusual physical appearance and inappropriate behavior of the sibling. They may resent having to integrate the sibling with a disability into the neighborhood peer group and consequently would avoid hanging out with their disabled sibling or would avoid inviting friends to their homes.
  - Experience or perceive peer rejection because of having a sibling with special needs.
- Anger, jealousy and feeling rejected by the parents are often experienced due the intensive time and attention the parents invest in the handicapped sibling. These negative feelings may provoke guilt- 'How can I be angry and jealous of my disabled brother, while he is the one that suffers/ rejected/ sad?'
- Some children are motivated to achieve in order to 'make up for' a brother or sister inabilities. They would try to compensate the parents by excelling in school and sports, by being very compliant, and by assuming more responsibility at

Siblings of individuals with developmental disability tend to be empathic, altruistic, more tolerant to children with special needs, and more mature and responsible.

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home. In these ways they are trying to ease and please their parents.

All this being said, it is important to remember that each child's reaction to having a sibling with special needs is different, and varies depending on his or her age, developmental level and subjective interpretation of the sibling's handicap. The perceptions that typically developing sibling has towards their special brother or sister are not likely to be static, but rather tend to change over time. With time they usually develop skills to cope with the challenges of growing in a family with a child that has special needs.

### Age-Specific Issues

#### Preschoolers (before age 5 years):

Preschoolers are cognitively limited to fully understand the special needs of their sibling, but they notice that 'something is different'. They often try to teach the disabled sibling the 'right' behavior. They are not judgmental yet, and tend to enjoy their interactions with their siblings, without being embarrassed or concerned.

#### Elementary School age (6-12 years):

At this age there tend to be increased awareness to differences between people, and as a result, elementary school children are very aware of their sibling special characteristics. They can now better understand the special needs of their sibling, and the causes for their difficulties, when it is explained to them in simplified terms. Openly discussing the syndrome cause and its manifestations significantly with the siblings reduces his fears and allow them to raise their concerns.

#### Adolescents (13-18 years):

Adolescent siblings are able to fully understand the meaning of the syndrome and its associated disabilities, and are expecting more elaborate explanation. They tend to ask many questions regarding their disabled sibling; sometimes the questions are provocative and incisive ('Is he going to get married?', 'Who is going to take care of him after you will be gone?', 'Will I have children with the same syndrome?'). They tend to be in a conflict between investing in their own developmental tasks and their commitment and responsibility to the disabled sibling. They might want to spend more time with peers or partners, but find it hard to refuse the siblings request to join them. Often they hesitate to ask for some financial support from

the parents knowing that this money is needed for the special care of their sibling. Many of them are concerned about how the people they socialize with, date and later marry will accept their brother or sister with a disability.

#### What can the parents do? / How can we help them?

First, it is important to provide the sibling with age appropriate information regarding his/her sibling's nature of disability and its treatment. Often, simply knowing the facts about the syndrome and its characteristics reduces fears, embarrassment and uncertainty and correct for false perceptions.

#### Siblings support groups

Special support groups for siblings of disabled children (such as SibNet or SibKits are very helpful for obtaining relevant knowledge and emotional support. One supportive resource specifically designed for siblings of children with disabilities, known as "Sibshops", was developed by Meyer, Vadasy, and Fewell in response to what they viewed as a lack of resources provided for siblings of persons with disabilities. The goals of the Sibshops were: (1) to give siblings a chance to meet other siblings in a comfortable atmosphere; (2) to have the chance to openly discuss shared troubles and delights of their experiences; (3) to learn how others handle the difficult situations involving their siblings; (4) to become educated on the medical conditions and handicaps of their siblings; and (5) to be able to share with peers who understand and have similar worries. The authors maintain that the meetings are not to be regarded as group therapy nor to provide intense counseling. Rather, the authors see Sibshops as a flexible curriculum for workshops in which the primary goal is to bring siblings together to share with one another and provide support and understanding in a way that only other siblings of children with disabilities can. Meyer and his colleague observed that siblings need to have clear explanations about their brother/sister disabilities, and freedom to express their emotions about their sibling. As parents you may find it very worthwhile to help your children join a group such as Sibshops, because interactions with peers that deal with similar issues provide needed opportunities to share joys and concerns. (see <http://www.thearc.org/siblingsupport/sibshops-about> for more information).

Parents and other family members are the main important source for support. It is therefore essential to keep open communication between parents and children, to dedi-

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cate time to ask them about their needs, and to encourage them to express their ambivalent thoughts and feelings. It is good to spend some one-on-one time with each one of the children, to do something fun together. These interactions convey a message from the parents to the sibling that they care for him as an individual, and that his or her experience and needs are not less important.

In addition, the expectations from the typically developing sibling should not be overwhelming; the responsibilities should be appropriate to his age. Parents must know that sometimes typically developing brothers and sisters react to their siblings' disability by setting unrealistically high expectations for themselves. It is crucial that parents will prepare their typically developing children for the challenges and difficulties in the future. Parents of adolescents should reassure them that they are making plans for the future of their children with special needs. They should also take into account the adolescents' considerations in planning for the future of the disabled child. The message that should be conveyed to the adolescent is that his or her type of future involvement with their sibling is their choice.

The resources listed in this article will help lead to the development of healthy sibling coping strategies, as well as simple, open family discussions of the issues facing the family. It is important to remember that a birth of a child a genetic syndrome, has a profound effect on the whole family, and coping with the new situation involves the siblings, as well as the parents. Coping with a family member that has special needs is challenging. It can be hard and frustrating. The family system of a person with disabilities will experience many challenges, but it will also experience many rewards. Our clinical experience shows that many families of children with VCFS are dealing with those challenges in an admirable way. More often than not warm, caring, relationships are formed between the siblings, and the whole family wins a unique, consolidating experience.

### Recommended Reading :

1. **Living with a brother or sister with special needs. A Book for Sibs.** By Donale Meyer and Patricia Vadasy. Seattle: University of Washington Press (1996).
2. **Sibshops: A handbook for implementing workshops for siblings of children with special needs.** Handouts by D.J. Meyer, P.F. Vadasy and R.R. Fewell. Seattle: University of Washington Press (1985).
3. **Siblings Without Rivalry: How to Help Your Children Live Together So You can Live Too.** A book by Adele Fabar and Elaine Mazlich (1998).
4. **How To Talk So Kids Will Listen & Listen So Kids Will Talk.** A book by Adele Fabar and Dlainé Mzalich (1991).
5. **Ordinary families, special children: a systems approach to childhood disability.** M. Seligman & R.B. Darling . New York: The Guilford Press (1997).

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1. Davis, E. (2004) **Siblings of persons with disabilities: A narrative analysis of the specialized sibling bond.** Kalamazoo College, Kalamazoo MI.
2. Powell, T. H., & Ogle, P. A. (1985). **Brothers & sisters: A special part of exceptional families.** Baltimore: Paul H. Brookes Publishing.
3. Meyer, D. J., Vadasy P. F., & Fewell, R. R. (1985). **Living with a brother or sister with special needs : A book for sibs.** Seattle: University of Washington Press.
4. [www.nichcy.org](http://www.nichcy.org)
5. [www.thearc.org/siblingsupport/](http://www.thearc.org/siblingsupport/)

### About Merav Burg, M.A.

I am a psychologist at the Schneider Children's Medical Center in Israel and I am currently working on my Ph.D in psychology at Bar Ilan University. My research is on social cognition and social behavior in children with VCFS.

### About Elizabeth Davis, B.A.

I began my research on the topic of siblings of persons with disabilities about two years ago as a psychology student who is also a sibling of a young woman with VCFS. I was frustrated by the dire lack of information about siblings of persons with disabilities and decided to address the need for literature in an undergraduate thesis. In this thesis I interviewed 16 siblings of persons with disabilities and reported in great length their responses; in the future I hope to continue this important work as the field of psychology needs more research on these sibling experiences.

## AROUND THE WORLD NOTES

### Continental Europe

By Stephan Eliez, MD, Reg Dir

On September 24, 2005, the French and Swiss associations (*Generation 22* and *Connect22*) held a joint parent meeting focusing on language and communication difficulties in the syndrome, as well as ways to tailor the French and Swiss special education systems to our children's needs. Our speakers were: Yves Alembic, a pediatrician in Strasbourg who often diagnoses and treats children with VCFS; Dr Marianne Till, who spoke about immunity problems in the syndrome; and Marielle Lacroix, a speech and language therapist who spoke about her experiences working with preschool age children with VCFS. Two experts on the French and Swiss special education systems were also at the meeting to present ideas for learning strategies and ways to adapt school programs to the needs of affected children. Parents told us that they much appreciated having a day devoted to such pertinent topics. Further, it is a pleasure to see our recently founded Swiss parent association, *Connect22*, grow with the years. Based on the success of the meeting, there will be a session devoted to school interventions at the Strasbourg conference

The European branch of the Educational Foundation has been hard at work planning for the 2006 conference in Strasbourg, France. Bob Shprintzen, Karen Golding-Kushner, Dominique Pfeiffer of *Generation 22*, the French parent association, and Stephan Eliez have set the scientific program, and conference preparations are coming together. As the first EF conference in central Europe, the event in Strasbourg will have the usual informative EF structure with parent clinics, along with a special French touch. It is the first time the conference has been accessible to many European parents, so we hope that families from the rest of the world will rally to make it an international affair.

### Denmark on the VCFS World Map

By Maria Boers, SLP, MA  
Copenhagen Cleft Palate Centre, Denmark

In May 2005 professionals from all over Denmark were invited to the first Danish 22q11 meeting for professionals at Copenhagen Cleft Palate Centre. At the meeting a 22q11 working group was established. The working group consists of a cardiologist, a neuropsychiatrist, a

neuropsychologist, an oculist, an orthodontist, a pediatric neurologist and a speech pathologist. We are trying to establish a general evaluation and treatment plan for all children diagnosed with VCFS in Denmark. We have also discussed how to coordinate and centralize the treatment of VCFS in Denmark.

Furthermore we are trying to establish a Danish Association of 22q11 Deletions for Professionals and motivate the parent group to establish a parent association for VCFS.

Regarding the parent group a meeting was held in April 2005 in West Denmark and in October we have another parent meeting in East Denmark at Copenhagen Cleft Palate Centre.

1st of August 2005 a research project ('VCF-Syndrome in Focus') was initialized in Copenhagen Cleft Palate Centre. The study is a retrospective study and the purpose of the study is to improve diagnosing persons with VCFS. The method is to review charts of children born 1990-2000 and registered in Copenhagen Cleft Palate Centre. There will be about 900 charts of children with different kinds of clefts and VPI. Furthermore we will evaluate the speech and language abilities and initiate speech therapy in our centre.

Other research projects will be planned in the near future among the other professionals in the 22q11 working group in Denmark.

### Latin America

By Antonio Ysunza, MS, ScD, Reg Dir

Greetings from Latin America to everyone in the VCFS Educational Foundation.

A support group has been founded in Argentina. This is the first support group I know in Latin America. They are a very enthusiastic group of families. They can be contacted at:

[jeseverio@vcfs.com.ar](mailto:jeseverio@vcfs.com.ar)

[www.vcfs.com.ar](http://www.vcfs.com.ar).

For clinical information concerning VCFS in Latin America, I provide my own e mail again:

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## AROUND THE WORLD...

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### **Australia & Pacific Rim** **By Steve Russell, President** **VCFS Foundation of Queensland** **VCFS Educational Foundation Inc.**

Well, what a six months this has been!

Things continue to go very well (if a little bit hectic!) here in Australia. We were very pleased that three members of the Management Committee of the VCFS Foundation of Queensland were able to attend the annual meeting of the Foundation in Syracuse, New York in July. We hope that those of you who attended picked up the Information Packs for the annual meeting in Brisbane in 2006. Since returning, Kath and I and the other members of the Committee were thrown into preparations for our annual Faces of Sunshine Charity Fundraising Ball.

I am very pleased to report that the Ball was a stunning success. Over 300 people attended the Ball and the Foundation attracted all of its old corporate sponsors and recruited one new one. Gross receipts from the evening were over AU\$ 100,000.00! A truly remarkable effort, and our thanks go to all the hardworking members of the management committee who helped to make the night such a wonderful success.

I must also record here my thanks to Professor Brett McDermott and his team from the Mater. Not only did the Mater people attend the ball, and not only did Brett speak (his usual entertaining and insightful presentation), but we continue to make wonderful progress with the VCFS Centre at the Mater Children's Hospital at Brisbane.

We are also very pleased that the Queensland Minister for Health was amongst the patrons at the ball - it's amazing what a one-way conversation with a microphone can do to raise the awareness of our political leaders with respect to the VCFS movement.

Finally, a big thank you to Mr Raymond Tanner who travelled with his lovely wife Ruth from Adelaide to attend the ball and to make a wonderful speech and presentation. Raymond is truly an inspiration and was a very big part of the success of the ball. With all of that going for us, how could we fail?!

As I have just mentioned, we have continued to work on the establishment of the VCFS Centre at the Mater Children's Hospital in Brisbane. Staff are being recruited. Our Developmental Paediatrician is Doctor Honey Heussler who visited with Doctor Shprintzen and his team at Upstate Medical University in New York in the last few weeks. There are plans for Brett McDermott to visit soon and I should also mention that Ms Erica Lee, the manager of the VCFS Centre, attended the meeting with us in July, 2005. We will be having a "hard" (official) opening of the centre before Christmas. Look out on the website ([www.vcfs.com.au](http://www.vcfs.com.au)).

The Foundation continues its work in generally raising the awareness of the community about VCFS. The old saying is true - the harder you work the luckier you get. We were very pleased to have attracted the interest of Suncorp-Metway Limited, a local bank, and Suncorp has already held a charity day to benefit the VCFS Foundation of Queensland - all from a couple of beers at the ball with a Suncorp executive!

## NOTE FROM PAST PRESIDENT

### **Nancy Robbins**

Thank you so much to the VCFSEF Board for allowing me the privilege of serving as President this past year. It was such a great honor to be able to give something back to an organization that has been so helpful in my personal life dealing with the challenges of having a child with VCFS. The conference was a great success with much credit to the local team in Syracuse who handled all of the local arrangements. Everyone involved with the child care did such a great job. There were lots of activities and special surprises that seemed to keep everyone enter-

tained. Many parents commented that their kids had a great time and it was nice that they were able to listen to the talks and not have to worry about their children. As with the past conferences that I have attended I thought that the speakers offered a great balance in presenting to professionals and lay people at the same time. I love that our conferences allow our speakers to present this way. I got feedback from several presenters that they really liked having the opportunity to talk one-on-one with fami-

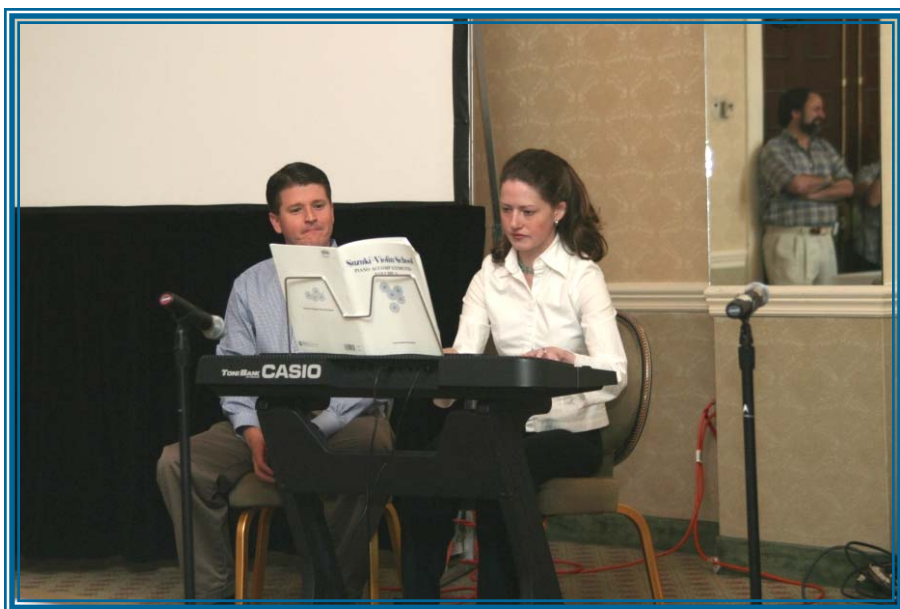
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## MUSICAL PERFORMANCE INSPIRES BENEFIT EVENT

Amy Dotson

I am aunt to sweet six-year old Cassie Southwick who has been diagnosed with 22q11 deletion syndrome (VCFS/DiGeorge). I am a senior at the Hartt School of Music in Hartford Connecticut and a member of SAI, Eta Mu Fraternity. This past July, I attended the Velo-Cardio-Facial Syndrome Educational Foundation Conference in Syracuse with Cassie's family. Karen Ruckman Lindsey, a pianist, and a young violin student, performed together at the meeting. I met Karen and her husband the night before the performance at the Dinosaur BBQ held at the Museum of Science and Technology. I was inspired, as you can see from the following:

The sisters of Sigma Alpha Iota, Eta Mu chapter, proudly announce that the 5th annual Benefit for Life will be held in honor of the Velo-cardio-facial Syndrome Educational Foundation, Inc. SAI, a professional music fraternity for women,



is committed to strengthening our ties with the community and reaching outside of our walls to help our neighbors. A large part of this mission is brought to life through our annual concert gala, the Benefit for Life. On this special evening, SAI sisters from the Hartt School will be presenting a formal recital for our family, friends and community. There is no ticket price, but 100% of the donations collected at the door will go directly to the VCFSEF. The Benefit will be held on March 3, 2006 at 7pm in the University of Hartford's Lincoln Theater, West Hartford, Ct. We would be honored to have you and your families in attendance. If you would like more information please go to our website [uhaweb.hartford.edu/sai](http://uhaweb.hartford.edu/sai) or contact Amy Dotson at [dotson@hartford.edu](mailto:dotson@hartford.edu), 339-234-0210.

*Karen Ruckman-Lindsay performing, assisted by her husband. (photo by Kelvin Ringold)*

## PRESIDENT'S REPORT...

*(Continued from page 8)*

lies and to interact with the kids. Parents also said that they thought it was so valuable to be able to talk with the "experts" in an informal setting.

Now that the conference is over, I am concentrating on completing the VCFS informational brochure. It is in it's final draft stage! It's going to be a huge project to get them distributed around the world so If anyone is willing to help, please let me know by emailing me at [nirrobbins@aol.com](mailto:nirrobbins@aol.com).

My other big project is the Knowledge is Hope awareness wristbands. To date, we have distributed more than 7000 wristbands across the world - And we have only just begun! We really need everyone to commit to buying some to give them to doctors,

therapists, teachers etc. If everyone gave out 25 (or more) wristbands we would make a huge impact. Someone recently purchased the bands to use at their wedding in honor of a family member with the syndrome. Karen Golding-Kushner used them as favors at her son's Bar Mitzvah. Fred Berg had a booth at a festival in his town and sold the bands. These are just a few examples of ways to increase awareness. I have received notes from people saying they are going to put them in their holiday cards this year to help spread the word. I hope everyone will get involved and buy some wristbands to sell or give. Together we will increase awareness!

Thank you again to the VCFSEF for allowing me to be part of this incredible organization!

# NOVEL RESEARCH FINDINGS ON GENETIC RISK FACTORS IN VCFS

Doron Gothelf, M.D., Stanford University and Tel Aviv University, Middle East Regional Director and Professional Council Member of the VCFES Educational Foundation

Stephan Eliez, M.D., Ph.D., Department of Psychiatry, University of Geneva School of Medicine, Geneva, Switzerland, European Regional Director, the VCFES Educational Foundation

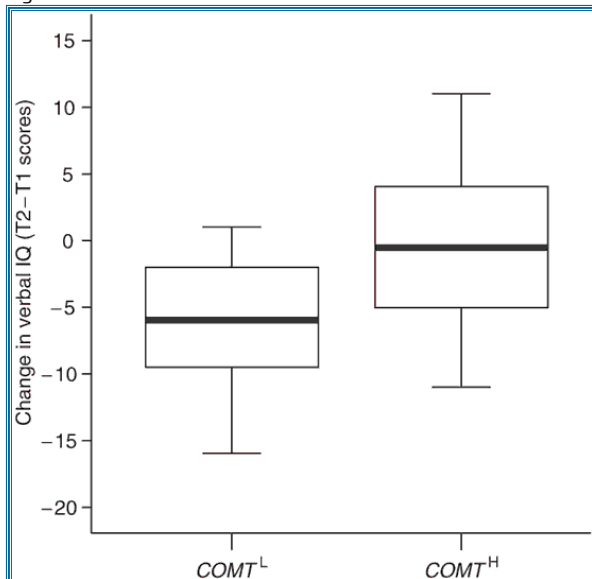
Allan L. Reiss, M.D. Director of the Center for Interdisciplinary Brain Sciences Research (CIBSR) and the Behavioral Neurogenetics Research Center, Stanford University Medical Center

A study that has just been published in the November issue of Nature Neuroscience has identified a variant of the *COMT* gene as a risk factor for a more rapid decline of prefrontal volume, slower development of language abilities, and higher risk for psychosis in adolescents with VCFS.

The Study was conducted by Drs. Doron Gothelf, Stephan Eliez, Allan L. Reiss and colleagues at the Stanford Center for Interdisciplinary Brain Sciences Research (CIBSR) and in collaboration with scientists from the Geneva University.

Twenty-four subjects with VCFS and 23 IQ-matched control subjects were evaluated first at an average age of 13 years and re-evaluated about 5 years later. At first evaluation none of the subjects had a psychotic disorder. Yet, at second evaluation seven subjects (29%) with VCFS developed psychotic disorders. As can be seen in Figure 1 the evolution of psychotic symptoms was highly correlated to a slower development of verbal cognitive skills.

Figure 1



Legend to Figure 1: Box plots show the range of values for each group, as well as the 25th, 50th (dark bar), and 75th percentiles. Significant longitudinal effect ( $P < 0.05$ ) of *COMT* genotype on change in Verbal IQ in subjects with 22q11.2DS.

The *COMT* gene is located within the 22-chromosome deletion region. Thus subjects with VCFS carry only one copy of the gene,

and not two copies, as it is in healthy subjects. The *COMT* enzyme that is coded by the *COMT* gene is essential for the disposal of dopamine especially in the prefrontal cortex. The gene has two natural variations that generate two versions of *COMT* enzyme: one with high activity ( $COMT^H$  or Val) and one with low activity ( $COMT^L$  or Met).

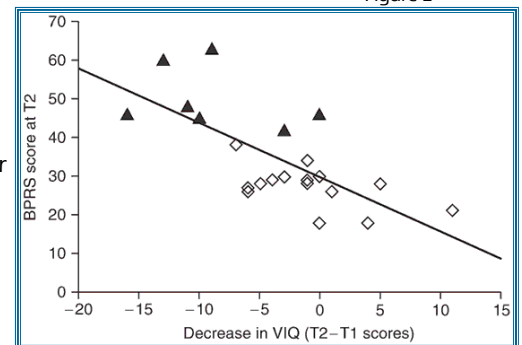
The study found that VCFS subjects with the  $COMT^L$  variant had a slower cognitive development in comparison to VCFS subjects with the  $COMT^H$  variant (Figure 2). The  $COMT^L$  variant was also associated with emergence of psychotic symptoms and with a more robust decline of gray matter volume of the prefrontal cortex. The results of the study suggest that subjects with VCFS and the  $COMT^L$  variant do not seem to have enough *COMT* activity required for disposal of dopamine. It is probable that these subjects have high dopamine levels in the prefrontal cortex that interfere with cognitive function and increase the risk for psychosis.

It is important to emphasize that these are preliminary results and not all subjects with the  $COMT^L$  variant develop psychotic disorders. In the general population as well as in VCFS about half of the subjects carry the  $COMT^L$  variant and many of them are developing well and are not having psychotic symptoms. Thus we recommend at this stage to conduct the test only for research purposes. However, we do hope that as we will learn more about additional genetic and environmental risk factors related to VCFS, we will be able in the future to detect children with VCFS at risk for psychosis and provide them with more specific and more efficient treatments.

We would like to thank the wonderful children and their families for their continual support and participation in the study. We would also like to thank the Northern-California Parent Association and Pam Hunter and Carrie Heran for their collaboration and help.

We would like to thank the Northern-California Parent Association and Pam Hunter and Carrie Heran for their collaboration and help.

Figure 2



Legend to Figure 2: Correlation between change in Verbal IQ and Brief Psychiatric Rating Scale in subjects with 22q11.2DS ( $r = 0.71$ ,  $P < 0.0001$ ). Dark triangle subjects with psychosis; white diamond, nonpsychotic subjects.

## ONGOING RESEARCH STUDIES

*As published on our web site.*

**Notice:** The VCFS Educational Foundation encourages research that will advance our knowledge. We do not sponsor or endorse these research projects, but post requests for participation from time to time as a service to those conducting research.

The following research studies are posted on the VCFSEF web site. For more information on these studies, please visit the web site at [www.vcfsef.org/Links.htm](http://www.vcfsef.org/Links.htm) (and see bottom of page)

– **Study of Polymicrogyria and 22q11 deletion**

This study is searching for genes that are involved in brain development. We are currently enrolling individuals in our research who have a chromosome 22q11 deletion and polymicrogyria. Polymicrogyria is diagnosed by brain imaging (MRI or CT) which shows many more and smaller folds than usual in the brain.

– **Taylor Request for Parental Participation**

This project seeks to study parents' experiences with the diagnosis of their child with a genetic condition, and focuses on the specific information parents were provided during the diagnostic process, and the information they would have liked to receive.

– **MRI Study Of Geneitics and Brain Functioning In Adults With VCFS**

Adults with VCFS appear to be at increased risk for developing psychiatric disorders as adults. An important goal of this study is to understand *how* the gene-brain-behavior relations in VCFS may lead to these difficulties. Valuable information is acquired about one's brain's functioning with functional magnetic resonance imaging (fMRI).

## A PARENT'S STORY...

*(Continued from page 3)*

My increasing awareness of Ariel's needs has also opened my eyes to the needs of others. Although an attorney by occupation, I teach a U.S. history class at a local community college at night. Twenty or even ten years ago, I might have insisted on treating all students alike, imposing uniformly rigorous standards on them irrespective of their different learning styles or disabilities. Now, I make a point of trying to accommodate students, even when they do not formally seek such accommodations, by teaching in ways that are best suited to their individual needs. My students do not realize it, but they have Ariel to thank for that.

When Ariel was born 23 years ago, Ellen and I were traveling in "terra incognita." We knew nothing of VCFS, and it would be many more years before we understood that VCFS was the unifying explanation for the bewildering variety of physical and emotional problems our daughter exhibited. Even after Ariel was diagnosed with VCFS, we were still dealing with a silhouette rather than a clear, detailed picture. Medical science's understanding of the syndrome – its causes, its characteristics, its short-term and long-term effects, the modalities of treatment – has evolved

slowly over time and in a fragmented way. (Each conference we have attended has shed more light on the subject for us, though sometimes too late to be of help for Ariel). Moreover, the medical issues have paled in comparison to the emotional dilemmas we have faced at each step of the way.

For 23 years, then, we have been seeking solutions for the problems caused by VCFS, often without a clear path to follow. However, we have never lost hope, and, for those with young children afflicted by VCFS, there is every reason to hope. The medical know-how for treating at least some of the symptoms of VCFS is improving all the time. Of even greater importance is the persistence and resilience of the human spirit. Inside every child with VCFS is the will to succeed. If that willpower can be harnessed to the love, guidance and support of parents, family, friends and teachers, then a child with VCFS can, indeed, enjoy a full and worthwhile life. We have certainly seen our daughter triumph over adversity and grow into a lovely and accomplished young woman. She, in turn, hopes to build a career by helping other youngsters struggling against even greater odds. There is, indeed, light at the end of the tunnel.

## RESEARCH UPDATES

Wendy Kates, PhD

Associate Professor, Psychiatry & Behavioral Sciences Department

Upstate Medical University Hospital, Syracuse, NY

### Cognitive Development

- 1) Debbane, M., Glaser, B., Gex-Fabry, M., Eliez, S. (2005) Temporal perception in velo-cardio-facial syndrome. *Neuropsychologia*, Vol 43: 1754-1762

This study investigated the ability to perceive time, based on the ability to accurately judge and reproduce the timing sequence of auditory stimuli and visual stimuli. Individuals with VCFS were less accurate than control subjects in both judging and accurately reproducing the timing of the stimuli presented. The authors attribute these deficits in time perception to the well-documented alterations in neural circuits that include the frontal lobe, basal ganglia and cerebellum.

- 2) [Simon TJ](#), [Bearden CE](#), [McGinn DM](#), [Zackai E](#). (2005) Visuospatial and numerical cognitive deficits in children with chromosome 22q11.2 deletion syndrome. *Cortex*, Vol.41: 145-155.

This study investigated visuospatial and numerical cognitive deficits in VCFS. Relative to typically developing controls, children with VCFS exhibited deficits in visual attentional orienting, visual enumeration, and judgment of relative numerical magnitude. The authors attribute these deficits to previously described deficits in posterior parietal dysfunction.

- 3) [Bish JP](#), [Ferrante SM](#), [McDonald-McGinn D](#), [Zackai E](#), [Simon TJ](#). (2005) Maladaptive conflict monitoring as evidence for executive dysfunction in children with chromosome 22q11.2 deletion syndrome. *Developmental Science*, Vol. 8: 36-43.

This study investigated aspects of executive function in VCFS. The authors administered an attentional task in which children were required to maintain attention in the presence of distractors. Children with VCFS demonstrated greater difficulty than controls in locating and processing target items in the presence of distractors, suggesting that they have difficulty monitoring and adapting to conflicting information in their environment. This is associated with difficulty shifting attention.

- 4) [Sobin C](#), [Kiley-Brabeck K](#), [Karayiorgou M](#) (2005) Lower prepulse inhibition in children with the 22q11 deletion syndrome. *American Journal of Psychiatry*, Vol.162: 1090-1099.

This study investigated startle responses in VCFS. Previous research has demonstrated that if typical individuals are exposed to a relatively soft sound just prior to exposure to a loud sound, they show an increased ability to inhibit a startle response (as measured by an eye blink). Individuals with schizophrenia (but not VCFS) have shown deficits in prepulse inhibition. Relative to their siblings, children with VCFS exhibited a decreased ability to inhibit the startle response. The authors conclude that deficits in prepulse inhibition, coupled with other behavioral and cognitive deficits, may constitute a predictor of risk for severe psychiatric disorder.

- 5) [Sobin C](#), [Kiley-Brabeck K](#), [Karayiorgou M](#). (2005) Associations between prepulse inhibition and executive visual attention in children with the 22q11 deletion syndrome. *Molecular Psychiatry*, Vol. 10: 553-562.

This study found the inability to inhibit the startle response in VCFS was associated with impaired performance on tasks of visual attention. The authors suggest that these corresponding deficits are due to a disruption in the brain circuit consisting of the prefrontal cortex and the basal ganglia (a group of structures located in the middle of the brain).

(Continued on page 13)

## RESEARCH...

(Continued from page 12)

- 6) [Lajiness-O'Neill RR](#), [Beaulieu I](#), [Titus JB](#), [Asamoah A](#), [Bigler ED](#), [Bawle EV](#), [Pollack R](#). (2005) Memory and learning in children with 22q11.2 deletion syndrome: evidence for ventral and dorsal stream disruption? *Journal of Child Neuropsychology* Vol. 11: 55-71.

This study examined memory functioning in children and adolescents VCFS. Overall, children with VCFS exhibited relative strengths in verbal as opposed to nonverbal memory. Relative to their siblings, children with VCFS displayed deficits in verbal working memory, facial memory, and recall of information after a delay. However, the authors found that some of the memory deficits in children with VCFS were similar to those of children with autism, suggesting that some deficits are not due to the 22q11.2 microdeletion per se.

### Brain Development

- 7) [Simon TJ](#), [Ding L](#), [Bish JP](#), [McDonald-McGinn DM](#), [Zackai EH](#), [Gee J](#). (2005) Volumetric, connective, and morphologic changes in the brains of children with chromosome 22q11.2 deletion syndrome: an integrative study. *NeuroImage*, Vol 25: 169-180.

This study used multiple imaging modalities to demonstrate morphological changes and posterior displacement in the corpus callosum in children with VCFS. Insofar as the corpus callosum consists of nerve fibers spanning the left and right hemisphere of the brain, the authors suggest that posterior displacement of the corpus callosum could affect communication of information to the parietal lobe. The parietal lobe underlies visuospatial, numerical and attention abilities, all known to be impaired in VCFS.

- 8) [Antshel KM](#), [Conchelos J](#), [Lanzetta G](#), [Fremont W](#), [Kates WR](#). (2005) Behavior and corpus callosum morphology relationships in velocardiofacial syndrome (22q11.2 deletion syndrome). *Psychiatry Research: Neuroimaging Section*, Vol. 138: 235-245.

This study used structural brain imaging to investigate the size of the corpus callosum in VCFS. Relative to controls, the corpus callosum in children with VCFS was significantly larger. Notably, children with VCFS and ADHD exhibited smaller areas of the corpus callosum than children with VCFS-only (although still larger than that of controls). Posterior regions of the corpus callosum were more aberrant than anterior regions, which is consistent with the alterations in parietal lobe volume that have been described in children with this disorder.

### Behavioral and Psychiatric Issues

- 9) [Baker KD](#), [Skuse DH](#). (2005) Adolescents and young adults with 22q11 deletion syndrome: psychopathology in an at-risk group. *British Journal of Psychiatry*, Vol. 186, 115-120.

(Continued on page 14)

## 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)

## RESEARCH...

(Continued from page 13)

This study administered a semi-structured psychiatric interview to 25 adolescents and young adults with VCFS and an IQ- and age-matched group of controls. Youth and adults with VCFS had a significantly higher incidence of ADHD, anxiety disorders and mood disorders. Moreover, 36% of participants with VCFS reported rapid emotional swings, and 48% reported psychosis-like phenomenon. Participants with VCFS also reported higher levels of social and occupational dysfunction relative to controls.

- 10) [Bearden CE](#), [Jawad AF](#), [Lynch DR](#), [Monterosso JR](#), [Sokol S](#), [McDonald-McGinn DM](#), [Saitta SC](#), [Harris SE](#), [Moss E](#), [Wang PP](#), [Zackai E](#), [Emanuel BS](#), [Simon TJ](#). (2005) Effects of COMT genotype on behavioral symptomatology in the 22q11.2 Deletion Syndrome. *Journal of Child Neuropsychology* Vol. 11: 109-117.

The catechol-O-methyltransferase (COMT) gene is located with the 22q11.2 deleted region, and is responsible for the breakdown of the neurotransmitter, dopamine. This study examined the effect of variation of the COMT gene on behavioral symptomatology in VCFS. Children who had one copy of the Met variant of the gene displayed significantly less behavioral symptoms than children who had one copy of the Val variant of the gene. These findings suggest that specific variants of the COMT gene may confer risk for psychiatric disorder in VCFS.

### DONATIONS

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.

The total in gifts received from April through September 2005 was \$11,355. Thank you to all of our generous donors and to those who solicited their support.

**In honor of Adam Shprintzen receiving his Master's Degree**  
Stuart Kushner &  
Karen Golding-Kushner

**In honor of Allison Robbins**  
Lisa Arthur  
Robert Dillaway  
Mr & Mrs Jack Jaffe  
Dr. & Mrs. Steven Lipsius  
Gordon and Whit Robbins

Nancy Singer & Joel Platt

**In honor of Monika Levanson's Birthday for Allison Robbins**  
Sonia & Steve Thacher

**In honor of Dr. Robert J. Shprintzen**  
Susan & Greg Knapp

**In honor of Leor Kushner becoming Bar Mitzvah**  
Rosyln Kravitz  
Stuart Kushner &  
Karen Golding Kushner  
Leor Eitan Kushner

**In honor of Meryl Phipps**  
Margaret Hale

**In honor of Monica Jennings**  
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Marguerite Fasino  
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Kathleen Jennings  
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Deborah Downey

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Carrol Magee &  
The Collins Family

**In Memory of Norma Moody**  
Carrie & Colin Heran

**In Memory of Paul Allen**  
Carrie & Colin Heran

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## EXECUTIVE DIRECTOR'S REPORT

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

Time marches on and, although our 11th Annual International Scientific Meeting is over, we continue to benefit from the knowledge we gained during our weekend in Syracuse. We welcomed over 300 participants from all over the world and heard presentations from over 40 international experts. For the first time, we offered primer sessions in speech and language, brain imaging, and genetics immediately before the official opening of the meeting. During these hour-long workshops, participants gained an understanding of specialized vocabulary and concepts that enhanced their appreciation of the scientific talks that followed. The formal presentations and round table discussions at breakfast and lunch were of outstanding quality and appreciated by our professional and lay attendees alike. The childcare arrangements were more extensive than at any previous meeting and the children's programming was superb. We are extremely grateful to the local arrangements committee- Anne Marie Higgins, Bob Shprintzen, Kelvin Ringold, Beth Wallace, and Nancy Robbins- for the outstanding job they did in organizing the program and social events.

We welcome new Board members who were elected at the annual business meeting. Our President, a familiar name and face to anyone who has attended any of our meetings or read any of these newsletters for the last few years, is Stephen Russell, the VCFSEF Regional Director for Australia and the Pacific Rim, and Director of the VCFS Educational Foundation of Queensland. He brings a wealth of experience to the post, and his unique perspective from "Down Under." Nathaniel Robin, MD, is Associate Professor of Genetics and Pediatrics at the University of Alabama in Birmingham. His talks on genetics at our Atlanta and Syracuse meetings were of outstanding quality and his round-table discussions are among the most well attended. We welcome him to the Board as Professional Council member. Also elected to the Board as Lay Council Member was Dominique Pfeiffer, an active member of the European Federation and President of Generation 22, the VCFS Support Group in Strasbourg, France.

Outgoing Board members Wendy Kates, Ph.D., and Fred Berg were recognized and presented with plaques that expressed the gratitude of the Foundation for their years of service, as was Nancy Robbins, who assumed the coveted role of Past President. Nancy agreed to accept the position of Chair of Public Relations

and remains the contact person for the VCFS Awareness Wristbands.

The Board and Membership voted on venues for some future meetings. I am happy to share with you that we will meet in July 2007 in the Dallas area of Texas. We thank Keri and Steve Alexander for proposing Dallas and agreeing to work on the local arrangements along with Dianne Altuna, a speech-language pathologist there. Fred and Joanna Berg proposed Detroit, Michigan, and we will meet there in July 2008. They and Jennifer Lewandowski have already been busy securing local arrangements in that area.

Several conference attendees submitted index cards with questions that were not answered at the meeting. Look for those answers elsewhere in this newsletter. If you missed any of the presentations or want to hear them again, please remember that audio recordings are available from Conference Copy. An order form is included in this issue of the newsletter.

As soon as we adjourned at noon on Sunday, I met with committees to plan next year's meetings in Strasbourg (July 7-9, 2006) and Brisbane (Nov 2-4, 2006). Both of these meetings promise to be extraordinary and unique. An abstract form for the Strasbourg meeting is already posted on our website ([www.vcfsef.org](http://www.vcfsef.org)) and on the website of our hosts for the meeting, Generation 22 ([www.generation22.asso.fr](http://www.generation22.asso.fr)). As always, we invite the participation of professional and lay persons with experience to share. We look forward to strong participation by our European colleagues and families and hope many of our American members will take advantage of this opportunity to travel to one of the most beautiful regions of France. Stephan Eliez, VCFSEF Regional Director of Europe, and Dominique Pfeiffer, President of Generation 22 and a recently elected lay Council Representative to the VCFSEF Board, have been working tirelessly to be sure our first bilingual meeting will be a success. Yes, presentations at the meeting will be in French and English, with simultaneous translation!

Please remember that we look forward to receiving your comments. We want to be sure the Foundation continues to meet the needs of the professional and lay community. You can reach me by phone (1-866-VCFS5) or email ([kgkushner@vcfsef.org](mailto:kgkushner@vcfsef.org)).

### 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).



## FREQUENTLY ASKED QUESTIONS....

Answered by Dr. Robert J. Shprintzen

The following are among the questions submitted in writing by registrants at the 11<sup>th</sup> Annual Meeting of the Foundation. We will continue to publish the answers in upcoming newsletters to all of the questions submitted at the meeting, so keep an eye out for your question.

### Can you have VCFS even if your FISH test is negative?

This is actually a commonly asked question, and one that was recently the subject of an article written by Drs. Robin and Shprintzen and published in the Journal Of Pediatrics this past summer. According to Robin and Shprintzen, the answer is "probably not." FISH is a highly accurate test, and it is specific for VCFS. Although it is possible for the testing itself to have technical difficulties, these errors are extremely rare. Because the DNA probes used for the test are specific to distinct locations within the 22q11.2 region, it is also possible for someone to have a deletion within 22q11.2 but that deletion may not include the the specific location of the probe. However, such instances are also

extremely rare, and it not really known if this type of deletion results in a typical case of VCFS. It is also possible that one of the important genes involved in the development of VCFS, such as *TBX1*, has something known as a point mutation altering the function of the gene, but the gene is not deleted. There have been a few such examples reported. However, it is not clear that such point mutations result in a disorder consistent with the entire expression of VCFS. For all practical purposes, FISH for the 22q11 deletion is as close to accurate as a test can be, and if the deletion is not detected, it most likely means that the diagnosis is not correct. The suggestion is that for cases where the deletion is not detected, review by other experts who know a lot about VCFS is warranted to see if the diagnosis is consistent with VCFS to see if other types of molecular genetics testing are indicated. Stated another way, the laboratory tests are more accurate than clinical diagnosis which relies on human judgment more than DNA probes.

*(Continued on page 17)*

## NEW PRESIDENT & BOARD MEMBERS

### Steve Russell President, VCFSEF

Steve Russell is the founding President of the VCFS Foundation of Queensland, Australia, and also chairman of the VCFS Foundation of Australia Ltd, the national body for the VCFS movement in Australia.

Steve and his wife Kathy set up and launched the Queensland Foundation in 1996. Their daughter, Amy, aged 13, has the deletion. Amy is one of six children. Like many families, Steve and Kathy wanted not only to know all there is to know about VCFS, but they also wanted to get active in supporting other VCFS kids and families. So, they began.

The Queensland Foundation has gone from small beginnings to an organisation which now has two part-time paid staff and an annual budget of over AU\$100,000.00.

Steve is 45, and a lawyer in private practice in Brisbane. He is managing partner of the firm he established in Brisbane in 1992. He enjoys reading, golf, music, wine (and will argue untiringly over the merits of Australian shiraz) rugby, travel, running and is a (lapsed) marathon runner.

### Nathaniel H. Robin, MD Professional Board Member

I am a medical geneticist, with an interest in craniofacial anomalies in general, and velocardiofacial syndrome in particular. My interest in VCFS is long standing, as I was exposed to the disorder throughout my training. After completing medical school at Albert Einstein College of Medicine I did my residency in pediatrics at Montifiore Medical Center. It was then that I first met Dr. Shprintzen and had the opportunity to learn first hand about this syndrome that was named after him. I went on to do my genetics training at the Children's Hospital of Philadelphia with Dr. Elaine Zackai at the time FISH testing for the 22q11 deletion was just beginning. Dur-



Steve Russell

*(Continued on page 17)*

## FAQ....

*(Continued from page 16)*

**Can someone explain the issues with teeth in the syndrome? Why do problems occur (decay) and what to do about it. Local dentists aren't educated and accuse parents of not brushing their child's teeth. Also, what should we not do?**

Dental decay problems in the primary (baby) dentition are fairly common in VCFS, although much less common in the permanent (secondary) dentition. The exact mechanism for this is not yet understood well. One can surmise that because of calcium metabolism problems in some kids with VCFS, the enamel may be thinner and more prone to decay. Also, the teeth come from some of the same embryonic tissues affected by the deletion. We do know that the primary teeth in VCFS are slightly smaller than normal. This may be an indication of thinner enamel. It also means that the spaces between the teeth are therefore bigger and this can make it more difficult to clean the teeth so that bacteria have more chance to grow. Also, if the immune system is depressed, the bacteria may also be more

likely to gain a better foothold in the mouth of a child with VCFS. The secondary dentition comes in at a time when most children with VCFS have outgrown most of their immune problems. In addition, the hypocalcemia issues that may be present during infancy are often not present later in childhood when the secondary tooth buds are forming. In infancy, the body may need to use all of its calcium reserves for metabolic issues that deprive the teeth of the calcium. In terms of what to do, the answer is probably already what you are doing. Bring your child to a pediatric dentist by 2 years of age. Brush well, but be careful in children with heart anomalies that you avoid brushing so hard that you make the gums bleed. A soft bristle brush is probably best. If your water is not fluoridated, speak to your dentist and pediatrician about a fluoride supplement. Don't let your child go to bed with a bottle because the sugars and acid in milk can cause significant decay. If your child drinks milk before bed, clean the teeth and rinse with warm water. If decay occurs anyway because of congenital abnormalities of the teeth, don't worry too much. Have your dentist treat the teeth appropriately and remember that in most cases the secondary teeth will be fine.

## BOARD...

*(Continued from page 16)*

ing this period we came to appreciate the wide clinical variability of VCFS, and the how common it was. I moved on to Cleveland for my first faculty appointment as a medical geneticist, and in a short period of time had identified dozens of VCFS patients at all ages. I also realized that few physicians knew about VCFS, and began to make it my mission to educate healthcare professionals, educators and parents about the condition. I continued to follow these children regularly, supervising their medical care and providing assistance to parents in educational and medical matters. I relocated to the University of Alabama at Birmingham in 2003, but have continued my involvement with individuals with VCFS. While we do not have the facilities to compare with Dr. Shrpintzen's in Syracuse, we aim to offer all the necessary components to our patients here. In an effort to improve this care we have recently begun a multidisciplinary clinic for children with VCFS (and other genetic syndromes with behavioral issues). While staying busy with my clinical responsibilities, I have tried to maintain some research efforts, and am currently involved in studies that are looking at why some children with VCFS have a cleft palate, while others do not, and in another we are trying to find the cause for the severely abnormal brain development that is seen in a very small percentage of VCFS children. Joining the VCFS Educational Foundation was a natural extension of my commitment to helping those affected with VCFS. I am very excited about the opportunity to be a member of the board, and look forward to my involvement in the coming years.

### **Dominique Pfeiffer** Lay Board Member

I am Dominique Pfeiffer. I have been married for 36 years and we have 4 girls (28, 25, 23, and 22 years old). Our fourth daughter, Elise has the micro délétion and is now in a French university. What else? I am, as we say in France, a mother at home; I founded the French association Generation 22 in 1997 and I am its président for this time.



Dominique Pfeiffer

# SYRACUSE CONFERENCE—SUMMER 2005

Snapshots from this summer's VCFS Educational Foundation's annual conference held in Syracuse, New York USA.



Bob Shprintzen and Anne Marie Higgins



Edna & Harry Keleshian  
former treasurer and past president



Fred Berg & Wendy Kates



Karen Golding-Kushner & Steve Russell

*Photography by  
Kelvin Ringold*



The sessions were well attended

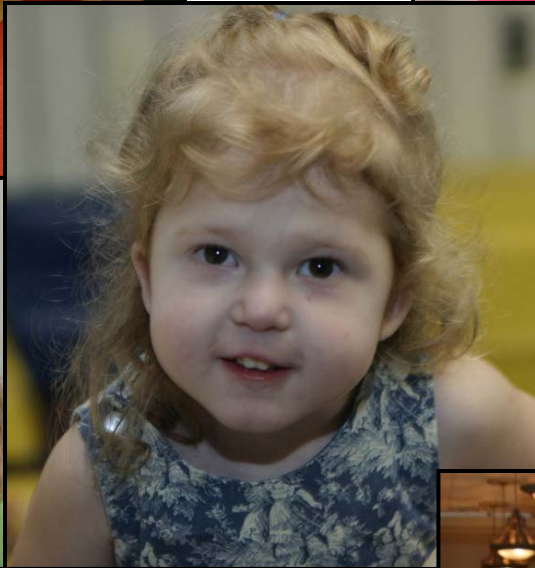


The Pfeiffers promote the Strasbourg Meeting

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<p>01 (2 Tapes)</p>	<p><b>Moderator: Susan Marks</b> <b>Speech Disorders and Their Treatment:</b></p> <p><b>Velopharyngeal Function in VCFS: What Advanced Diagnostic Techniques Have Taught Us;</b> Robert J. Shprintzen</p> <p><b>Asymmetry of the Pharynx, Larynx, and Palate;</b> Burke Chegar</p> <p><b>Muscle Volume of the Palate in VCFS;</b> Robert Brown, Wendy Kates, Robert J. Shprintzen</p> <p><b>Facial Nerve Function in Individuals with VCFS and Facial Asymmetry;</b> Antonio Ysunza</p> <p><b>Surgical Management of Hypernasal Speech in VCFS;</b> Sherard A. Tatum III</p> <p>Questions and Comments</p> <p><b>The Computer in the Speech Clinic for: Therapy, Productivity, and Resource;</b> John E. Riski, Ph.D.</p> <p><b>Speech Therapy to Correct Articulation Impairment in VCFS;</b> Karen J. Golding-Kushner, Ph.D.</p> <p>Questions and Comments</p>
<p>02</p>	<p><b>Moderator: Robert J. Shprintzen</b> <b>Molecular Genetics and Developmental Biology Presentations:</b></p> <p><b>Understanding the basis for the 22q11 deletion;</b> Melanie Babcock, Bernice Morrow</p>
<p>03</p>	<p><b>Pediatric Issues</b></p> <p><b>A Primer on the Immune System: When and How to Look for Problems;</b> Joseph Domachowske</p> <p><b>Self-Hypnosis to Reduce Anxiety in Patients with VCFS;</b> Ran Anbar, M.D., Kim Hummell, C.S.W.</p> <p>Questions and Comments</p> <p><b>VCFS Around the World</b></p> <p><b>Developments in Australia;</b> Stephen Russell</p> <p><b>Developments in Argentina;</b> Antonio Ysunza</p>
<p>04</p>	<p><b>Brain Imaging in VCFS: where will the studies lead us?</b> <b>Moderator: Doron Gothelf, Petah Tiqwa</b></p> <p><b>Brain Structure and Function in VCFS: Does COMT Make a Difference?</b> Wendy Kates</p> <p><b>Multiple Attention system Impairments in Children with VCFS;</b> Tony J. Simon, S. Ferrante, V. Nguyen, H. Ferrante, Donna McDonald-McGinn, Elaine Zackai, J. Bish</p> <p><b>Brain Structure, connectivity and function changes and their relation to cognitive impairments in VCFS;</b> Tony J. Simon, J. Bish, L. Ding, Machado, Nguyen, Donna McDonald-McGinn, Elaine Zackai</p> <p><b>Polymicrogyria in VCFS;</b> Nat Robin</p> <p>Questions and Comments</p>

05	<p>COMT and VCFS and Treatment Effects Moderator: Tony Simon</p> <p><b>The Effect of Genetic Variation in COMT on Risk for Psychiatric Disease Neural Function;</b> Andreas Meyer-Lindenberg</p> <p><b>COMT Polymorphisms and Language Impairment in VCFS;</b> Eileen Marrinan</p> <p><b>Inconsistent Evidence for an Effect of COMT Polymorphisms on Brain Morphometry and Cognition in VCFS;</b> Bronwyn Glaser, Martin Debbané, Christine Hinard, Michael Morris, Sophie Dahoun, Stylianos Antonarakis, Stephan Eliez</p> <p><b>Drugs in the Management of Behavior in VCFS,</b> Wanda Fremont, M.D.</p> <p>Questions and Comments</p>
06	<p><b>Saturday P.M., Moderator: Anne Marie Higgins</b> <b>Psychiatric and Behavioral Treatment of VCFS</b></p> <p><b>Mentoring Program as an Effective Psychosocial Intervention in VCFS;</b> Merav Burg, Tamar Steinberg, Petah Tiqwa, Doron Gothelf</p> <p><b>Longitudinal Study of Behavior in VCFS,</b> Doron Gothelf, Petah Tiqwa, Allan L. Reiss</p> <p><b>Social Skills Training &amp; VCFS: The Moderating Influence of ADHD;</b> Kevin Antshel, Ph.D.</p> <p>Questions</p>
07	<p><b>Neuropsychological and Psychiatric Findings in VCFS</b></p> <p><b>Relative Strengths and Weaknesses in Visuo-Spatial Abilities in Individuals with VCFS;</b> Bronwyn Glaser, Martin Debbané, Stephan Eliez</p> <p><b>Cognitive and Psychosocial Aspects of Children with VCFS;</b> Neil Nicoll, Belinda Barton, Meredith Wilson</p> <p>Questions</p>
08	<p><b>Chronic Problems in VCFS</b> <b>Moderator: Nancy Robbins</b></p> <p><b>Autoimmune Thyroiditis as a Previously Unrecognized Cause of Hypothyroidism in VCFS;</b> JI Mendez-Inocencio, MB Bueso, JA Bellanti</p> <p><b>Use of Intravenous Immunoglobulin (IVIG) Therapy in Patients with VCFS: Replacement or Immunomodularity Effects?</b> JI Mendez-Inocencio, JA Bellanti</p> <p><b>A Perspective on Leg Pain;</b> Marc Heatherington</p> <p><b>Annual Meetings in 2006: An International Affair</b> Stephen Russell, Dominique Pfeiffer</p>
09	<p><b>Cardiology, Molecular Genetics, Gene Interactions: Moderator – Nat Robin</b></p> <p><b>VCFS: The Heart of the Matter;</b> Roger Ruckman</p> <p><b>Alteration in neural crest cell function by inactivation of Tbx1 in mouse models;</b> Vimla Aggarwal</p> <p><b>Identification of genes in the genetic pathway downstream of Tbx1 by microarray analysis;</b> Jun Liao</p> <p><b>Interactive Partners for Tbx1 in Skeletal Muscle Development;</b> Evan Braunstein</p> <p>Questions and Comments</p>

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**Developmental and Educational Issues**

**Moderator: Maureen Anderson**

**Developmental Milestones in VCFS;** Nuria Abdul Sabur, Nancy Roizen, M.D.

**Education, Advocacy, and Wrights Law: helping your child achieve;** Donna Landsman, M.S., Judy Gaughran, Yolanda Ortiz, Debbie Lightfoot

Questions and Comments

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