



VCFSEF NEWS

Tony J. Simon, Ph.D.
Kelvin Ringold, Sr.

Editor
Design & Layout

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FROM THE EDITOR

It is with great pleasure and humility at being asked to assume such an important role that I begin my term as Editor of the Newsletter. During my tenure I plan to maintain the Newsletter as a vehicle for a lively exchange of information between families who are experiencing life with the disorder and all the professionals who are trying to understand its complexities and figure out ways to reduce or remove them. It is also my hope to use this time to add a slightly increased emphasis on brain and behavioral issues to the wide mix of topics that will be covered in these pages. This is partly because that is my own field of research. However, it is mostly because it is clear that these issues are becoming increasingly relevant as questions about the syndrome move beyond trying to gain a basic understanding of what it is like and towards understanding issues of how to live a long and productive life with it.

There are many questions about issues relating to school and work, dealing with mild and more serious psychiatric issues, questions about medications to help with learning, thinking, emotion and behavior and dealing with adult relationships that are among many of increasing interest to this community. While the regular columns, and personal accounts will remain a staple of the Newsletter, we will try to include information and discussions of some of these topics a few times a year. I am hoping to have a good deal of input from families and professionals in order to guide those discussions and so this is a call to anyone and everyone to respond to requests for input and have your voice heard. Our input needs to come from community members all around the world, and not just in the USA. It is clear that the Foundation is now a global entity and so I hope everyone will participate.

To help me do this job as well as possible I am honored to have the help of two enthusiastic colleagues. One is Cheryl Fox-Dultz. Cheryl is a parent of a beautiful 9-year old girl who was diagnosed with a chromosome 22q11.2 deletion in 2000. Cheryl is also a 3rd grade teacher at Kingswood Elementary School in Citrus Heights, California. Cheryl has an interest in educational interventions and accommodations for students with the deletion and she has taught such students, other than her daughter, in her class. The other is Joel Johnson M.D., who is a Psychiatry Resident at the University of California, Davis School of Medicine and who is building a strong research program

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SAVE THE DATE!

14th International Scientific Meeting

July 20–22, 2007

Marriott Dallas/Plano
at Legacy Town Center
Plano (Dallas), Texas

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EDITORIAL...

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within my own team. Joel's interest is in indicators that might predict later psychiatric problems and how to use them to guide treatment. Joel is also a practicing member of the Early Diagnosis and Preventive Treatment Clinic in the UC Davis Psychiatry Department. I look forward to a lively exchange of ideas with you all over the next two years and also to meeting as many of you in person as possible.

Tony J. Simon Ph.D

Frequently Asked Questions

Edited by Robert J. Shprintzen, Chair, FAQ Committee

Question: My 2 year old Child was diagnosed as having a mosaic 22q11 deletion. What is mosaic deletion? How is it tested? If my child presents with a mosaic deletion, are the symptoms milder? Does this include both physical and mental problems? Based on this can you predict that his chance of developing more severe developmental/psychological problem in the future would be lower?

Answer: A mosaic refers to a deletion that happens after the egg was fertilized so that not all of the cells in a person carries the deletion...it could be 50%, 25%, 12.5%, etc. depending at what stage the deletion occurred. It is also possible for only certain lines of cells to be affected if the chromosome rearrangement happens later in the developmental cycle. Mosaic chromosome rearrangements are seen in Down syndrome and other chromosomal disorders. One would expect a milder expression if the deletion were a mosaic. However, the expression of VCFS is so variable to begin with, that you cannot determine if a person has a mosaic by any way other than doing FISH on a large number of cells...usually 20 or more, to see if any have normal signals. Because reported mosaics in VCFS are rare, it is not known how it affects individual features, physical or mental, but in general the expression would be expected to be milder.
(Answered by Robert Shprintzen)


Question: My son is 16 and has recently developed trouble with his jaw and may require surgery. Is this common in people with VCFS?

Answer: Although not very common, there are some people with VCFS who have problems related to jaw alignment issues. Malocclusions can be caused by a retruded lower jaw, a chronic open-mouth posture from oral breathing, and sometimes a constricted upper jaw that is sometimes referred to as a "high palate" or "high arched palate." If the bite is abnormal, this can result in temporomandibular joint problems, usually called TMJ. This can cause headaches, clicking in the ears from the jaw joint, and limited oral opening. This can be evaluated by an orthodontist familiar with TMJ problems. Such problems are actually more common in people with relatively low muscle tone which is common in VCFS. However, it would also be wise to rule out juvenile rheumatoid arthritis (JRA) that does occur in VCFS. JRA will alter possible treatment plans and is a progressive disorder. Although JRA is very rare in VCFS, it is a possible clinical feature and should be ruled out. Mention these issues to your primary care doctor.

(Answered by Robert Shprintzen)

Sacramento Conference Focus on Behavior and Learning

On March 3-4, 2007, Pennsylvania-based Elwyn, Inc. and the M.I.N.D. Institute of the University of California, Davis (www.mindinstitute.org) will be teaming up to co-sponsor a two-day conference on education and behavior in children with the chromosome 22q11.2 deletion syndrome.

This meeting will be similar to previous Elwyn conferences in Philadelphia and Atlanta, offering families on the west coast a chance to access this important information. Save the date and plan to attend next March. Complete conference information and online registration will be posted to the conference website in late December at www.elwyngenetics.org. 

New Book on the Near Horizon!

Practical Handbook for Educating Children with Velo Cardio Facial Syndrome and Other Developmental Disorders. Author: Donna Landsman. Available for pre-order from Amazon.com for \$55.00. *Note: Donna Landsman has lectured more about VCFS and education than any other person in the world.*

Regional Events

(See Regional Events on the VCFSEF Web Site!)

March 11, 2007

North Carolina 22q11 Deletion Syndrome Support Group Mtg
Guest speakers, including Dr. Shprintzen [Click for Program](#)

Attention Florida 22q Families!

A Notice from Barb Southwick and Maria Kneipple:

The VCFSFlorida Group is creating a database of all Florida families who have a family member with a diagnosis 22q11 DS, for possible use at a Florida conference and hopefully future meetings. Your privacy is assured. Information shared only with another 22q/VCFS non-profit organization. If interested — or for more details — send an email to Barb Southwick at bsouth@swfla.rr.com, or miami22qinfo@yahoo.com. You can also visit our VCFSFlorida group and add your info at <http://health.groups.yahoo.com/group/vcfsflorida>.

We need parent support to make it happen! Thank you!

RAYS OF HOPE

Compiled by:

Maureen Anderson

mladja@aol.com

In the prior issue of the VCFS Educational Foundation Newsletter we premiered "Rays of Hope". This portion of our newsletter introduces us to those in our community who have overcome the challenges of VCFS and are willing to share their "Rays of Hope" with us all. If you would like to share a story of a photo with us, please contact Maureen Anderson at mladja@aol.com

Apryl

My name is Apryl Fox. I am 24-years-old. The doctors diagnosed me with VCFS when I was only seven, because I kept having so many health problems. They told my parents that I would not be able to read, write, do math, or finish high school. I have done all of those things. In fact, I like writing so much, that I decided to publish a book of poems.

My book, "This Ain't No Paradise" contains about 52 poems that were written between the ages of 21 and 24-years-old. I have been published in several ezines and magazines, including "Strange Horizons," "Verse Libre Quarterly," and "Dark Animus." I am an Assistant Poetry Editor at an online magazine called "The Rose and Thorn" E-zine and I am a freelance writer and editor. If you'd like to see my book online visit www.lulu.com/apryl_fox

The one thing I want everyone to know is this: we can ALL do great things-regardless of VCFS, or where we come from, or how much money we have. Greatness comes from inside of us. ***

Lindsay (Minnesota)

My life began as a struggle to survive. I weighed only 5 pounds 15 ounces when I was born. I quickly lost my ability to suck, which made me unable to eat and quickly I lost a lot of weight.

My first surgery occurred when I was a year and a half to repair a vascular ring. I also kept getting infections: pneumonia specifically. At five years old, I had my second surgery to remove the middle and lower lobes of my right lung.

My parents were young, only 19 and 20 years of age. My illnesses were a great struggle for them. They couldn't understand why these random illnesses were happening.

The first five years of my life were spent mostly in a hospital room at St. Mary's. I was also treated by doctors at Mayo Clinic in Rochester. By the time I was two, I was able to point out letters to my doctors. They were quite impressed! I learned to read and write during my hospital stays.

As I got older, I became healthier and was able to attend regular public school. I struggled with math and science especially. I specifically remember that I couldn't tell time in second grade. I remember looking at the massive paper clock that my teacher had made and I couldn't re-

member the difference between the big hand and the little hand. I couldn't remember that a quarter to three was at the two and nine mark on the clock. I also almost didn't pass sixth grade because I couldn't read measurements. But, with the help of friends and family, I made it through school and graduated high school with the rest of my friends, although I was second to last in my class.

I also struggled through the first part of college. I dropped out after two years of failed grades. With the encouragement of my boyfriend (now my husband), I began working. This gave me great confidence and I finally figured out that I should go back to college. When I went back, I began writing for our college newspaper, the Echo, and became Editor-In-Chief after a year or so of being a staff writer. I found my 'calling' in writing. I found that all of the reading I did as a child stuck in a hospital room gave me the knowledge of English that I needed to become a good writer. I was on the Dean's List several semesters after I figured this out.

I have now been accepted to MSU Mankato and will take two classes this fall while working full time at the only newspaper in our town, The Post-Bulletin. My ultimate goal is to eventually become a Technical Writer (my major) and write about genetics/genome programs. I will minor in Biology, which is a subject I once hated.

I still struggle with hypothyroidism, a symptom of 22q11 deletion, for which I have to take medicine every day for the rest of my life. I also have to be careful of infections, as I still get pneumonia, bronchitis, the flu, or pleurisy once in a while.

I have to worry about when my husband and I decide to have children, which will be in a few years. We have to get genetic testing done and we have to understand the consequences of what could happen by having our children naturally. However, I am confident that with the knowledge I now have, and having met others who are dealing with 22q11 deletion, my husband and I will be in better shape than my parents were twenty years ago. Having this defect has made me the person I am today. I am kind, understanding and care about my fellow human beings, even though they are not always kind to me or to others. I am empathetic toward people who are struggling or have struggled to survive because I've been there. ***

WORDS FROM THE EXECUTIVE DIRECTOR

Karen Golding-Kushner, Ph.D.

This year, for the first time, we held two international scientific meetings and each was a huge success. Planning two conferences in the same year is something we have not done before, but it was worth the effort because it enabled us to reach close to 800 people. We met in Strasbourg in July and, as I write this column, I am 31,000 feet above the Pacific Ocean, returning to the US from our 13th International Scientific Meeting in Brisbane, Australia. Like everyone who attended the meeting, I am tired but energized. Anyone who has attended one of our conferences knows what I mean. The response of professional and lay participants at both meetings was extraordinary. I heard comments about the high quality of the scientific and clinical presentations. Several researchers shared results of studies that are in press but not yet available in professional journals, so attendees were privileged to hear about the absolute latest research in VCFS. At the same time, appreciation was expressed that the information was presented in a way that made it understandable to professionals doing similar work, to professionals other fields, and to parents who were veterans at our meetings or attending for the first time. Scientists and clinicians planned new international collaborations. Parents made connections with each other and with international experts. Professionals listened to parents and children, gaining new insights about the impact of their research and clinical work and about priorities. This could not have happened without the presenters, who traveled from five continents, gave generously of their time and shared their research. They made no effort to hide the passion, compassion, and sensitivity with which they approach their work. Among the guests at the Brisbane meeting was Roopa Nagarajan, who is Chairperson of the Department of Speech Language and Hearing Sciences at Sri Ramachandra Medical College and Research Institute in Chennai, India. VCFS is not commonly diagnosed in India, but we know that will change with the information now available there.

Thank you to the Dominique and Jean-Louis Pfeiffer, who, with the support of Generation22, organized and hosted the Strasbourg meeting, and to Stephen and Kathy Russell and the VCFS Foundation of Queensland, who organized and hosted us in Australia. We plan to have articles in next issue of the newsletter about the meetings in Strasbourg and Brisbane. For those who were not able to attend the meeting in person, we can offer the next best thing! Presentations of speakers who gave permission are available in power point format on our website (from the Homepage, click on "About VCFS"

then "Power Points" and pick your subject of interest). Audio recordings of most speakers are also available.

Thanks to the efforts of Publications chair Nancy Robbins, our brochure about VCFS is in press. We will be sending copies of the brochure to geneticists, cardiologists, pediatricians, and speech-language pathologists. The mass mailing is possible because of generous donations to the Amanda McPherson fund. Members of the Foundation will also receive a copy. Watch the Foundation website for information on how to order copies of the brochure if you are not a current member or if you want additional copies to distribute. That reminds me please be sure to join or to renew your membership by downloading a membership form from the website, completing it and mailing it to Lisa Jennings, Treasurer. Our membership year begins January 1, so 2007 payments are now due. As always, dues are voluntary. If payment of dues would present a hardship, indicate on the form that you want to have them waived and we will do so. Your membership in the Foundation is more important than your dues. However, if you are able, your dues (and any additional donation) may be paid online with PayPal or with a check enclosed with your membership form. Please complete the form, even if you are renewing your membership. The form enables us to maintain a current mailing list and requests some new information this year.

Given that dues are voluntary, I am often asked why we collect dues at all. Dues and other donations support the Foundation by enabling us to maintain the website (server space is expensive!), the toll-free phone, and to hold the annual scientific meetings. All of the funds go directly to direct Foundation expenses. Not a penny goes to salaries- everyone involved with the Foundation volunteers their time.

Our blue "Knowledge is Hope" wristbands have been selling around the globe, spreading awareness of VCFS and helping inform by directing people to our website. Over 7000 wristbands have been distributed worldwide already! Why not purchase a few for yourself and your family, and a few extras to give out to teachers, doctors, even friends and neighbors? Each is individually packaged with a small explanatory card. They make great gift-tag attachments!

We are now excited to introduce tee-shirts that display our logo, which we hope will also increase awareness. Information on ordering wristbands and tee-shirts is on pages 13 of this newsletter.

FROM THE EXECUTIVE DIRECTOR...

So, to summarize, since the last issue of this newsletter, we have held two international scientific meetings, elected a new board, completed an informational brochure about VCFS and created logo tee-shirts, all of which further our mission to increase awareness and disseminate educational information. We also welcome India into the family of countries with ties to the Educational Foundation.

Warm wishes to all,

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

DONATIONS

The VCFS Educational Foundation relies on membership dues and donations to fulfill its mission to disseminate information regarding the syndrome. We gratefully acknowledge the following individuals and organizations for their donations that were made since our last newsletter.

General Gifts:

Maureen Alberts
Abraham & Hetty DeLeeuwe Family Fund
John Krol
Jeff & Donna Landsman
VCFSEF Benefit at Lilly Pulitzer store

"Benefit for Life" Sigma Alpha Iota, Eta Mu Chapter, Hartt

School Fundraiser:

Barbara Johnson
Lisbeth Steen, DMD
Lillie H Feierabend

In Honor of Rhett Alexander (Happy 40th):

Deborah and Stephen Jacobs
Harriet Martin
Lee Alexander

In Honor of Monika and Joel Levanson

Lenore & Robert Cohen

In Honor of Kim Coates:

Agonian Alumni Association, Alpha Kappa Phi

In Memory of Elizabeth Landsman

Karen Golding-Kushner

To date we have received donations in the amount of \$2080 since the last newsletter

Thank you to all of our generous donors and to those who solicited their support.

WORDS FROM THE PRESIDENT

Hi Ya'!!! We are excited about the 2007 Conference set for July 20-22, in the Lone Star State. I wanted to introduce myself and tell you a little bit about my family, our support group, the 2007 VCFS conference and me. My husband, Steve, and I have two wonderful young boys. Ty, our oldest, is a precious seven year old and a first grader. Rhett, our always loving and "monster truck fan", has VCFS and is 4. I knew something was "not quite right", with Rhett and I had diagnosed Rhett with many syndromes myself, after countless hours on the internet when he was just 3 months old. However, I was told that his craniosynostosis, spitting up out of his nose and frequent ear infections were all isolated problems. At 2.5 years old, Rhett's speech had not progressed like that of his peers and we began to seek speech therapy. Speech therapy led us to a speech pathologist named Diane Altuna, who happened to be a personal friend of Dr. Bob Sprintzen.

Within minutes Diane felt Rhett had VCFS. After the fish test was confirmed we asked ourselves, "What is VCFS and what does the future hold for our son.

It didn't take long, three weeks later, I was on a plane bound for Atlanta to the 2004 VCFS conference. Two months later, with the help from Dianne, we started a support group in the North Texas area and we currently have 22 families. One year later at the 2005 VCFS Conference in Syracuse, Diane Altuna and I offered to host the 2007 conference in Dallas. Our support group meets quarterly and at least twice a year will have guest speakers who help educate us on VCFS or other related issues. Our support group will arrange all the local plans for the conference and organize and staff "Kid's Zone". Parents who attend the conference can feel assured that their child is having fun and will be well supervised. Already, we have planned a fundraiser for April 21st at our home, in which we hope to raise \$7,500.00.

The foundation has created t-shirts with the VCFS logo and our VCFS slogan, "Knowledge is Hope" t-shirts will be on sale soon!! We ship them from our home in Dallas.

Dianne Altuna and I will co-chair the conference this summer. Dianne is a Speech Pathologist in the Dallas, Texas area. Currently she works in the Fogelson Plastic Surgery Center at Children's Medical Center, and is a Lecture/Supervisor at the University of Texas at Dallas, Department of Communication Disorders. She has specialized in the diagnosis and treatment of feeding, speech, language and resonance disorders in children with craniofacial differences for the past 20 years. In 2002, Dianne developed Zone C.A.R.E. through UT Dallas. This summer program offers intensive speech therapy to children with communication differences. The "clinicians" are graduate students through UT Dallas.

Children with Velo-Cardio-Facial Syndrome hold a special place in Dianne's heart, and helping with this conference brings the opportunity to learn more about these special children and their families. Dianne lives with her husband Patricio Altuna, and her daughter Gabriela in Dallas. The Altuna family have two adopted rescue dogs. Family favorites are to spend time together biking, traveling and reading.

The 2007 conference will be held at the Legacy Center in Plano, conveniently located just 20 miles from DFW airport. Legacy Center

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MEET THE BOARD

Elections were held at the annual membership meeting in Strasbourg in July. Stephen Russell completed his term as President, and Keri Alexander of Richardson, Texas was elected to succeed him. Eileen Marrinan completed her third term as Editor and we welcome Tony Simon, Ph.D., of the M.I.N.D. Institute at University of California, Davis who has assumed that position with this issue. Each year we elect two new Council members who will serve three-year terms. Elected this year were Bronwyn Glaser (Professional) and Amyel Loy (Lay). They move into the positions that were held for the last three years by Jay Riski and Raymond Tanner. The Foundation thanks Steve, Eileen, Jay and Raymond for their dedication, and we wish Keri, Tony, Bronwyn and Amyel well as they begin their service on the Board.



Keri Alexander, President

Keri Alexander is married to Steve Alexander and has 2 young boys, 7-year old Ty and 4-year-old Rhett. Rhett was diagnosed at 2-1/2 years of age with VCFS. He loves cars, trains and anything with wheels! He has a great role model- his older brother who watches over him and makes him "TOUGH!" The Alexanders live in Dallas, Texas and are excited about the opportunity to host next year's annual meeting of the VCFSEF. Keri said, "I consider it a blessing to help create awareness for Velo-Cardio-Facial-Syndrome in our community and our local medical world. We plan on everyone enjoying our southern hospitality and learning all we can about this syndrome!" Keri is a stay at home mom who volunteers at her children's school and many church activities. They have lots of family that live close to them and love their big family gatherings. Rhett is surrounded by those who love him and they feel fortunate to have such support. Keri founded the Dallas/Fort Worth VCFS Support group along with Dianne Altuna two years ago. Dianne is the

speech pathologist who suggested Rhett be tested for VCFS and will co-chair the 2007 VCFSEF International Scientific conference along with Keri. The Alexanders have a Bichon named Calli who loves her two boys! They hope to meet you all and look forward to a great time in Big "D" next summer!!



Tony Simon & son Benjamin

Tony Simon, Editor

Tony Simon is a pediatric cognitive neuroscientist, which means that he studies the relationship between the "hardware" of the brain and the "software" of the mind as they change during childhood. His research focuses on explaining and eventually remediating cognitive impairments seen in genetic conditions that can produce mental retardation, developmental disability or psychiatric disorders. His main interest is in children with deletions of chromosome 22q11.2 or VCFS. A native of London, England Tony moved to the US in 1988 and now lives very happily with his wife, Virginia (Ginger) and young son, Benjamin in Northern California. Tony's interest in children with VCFS came about almost by accident because of his earlier work on how the human brain comes to represent and process numbers. He is thrilled to be working on this exciting project with these wonderful children and their families and finds his current work to be the most fulfilling

thing he can imagine doing. Tony was surprised and honored to have been elected to the board of the VCFS Educational Foundation and to be serving as the Newsletter Editor. He recently attended the Australian conference and took time to get to know some of the locals. Tony is currently an Associate Professor in Psychiatry at the University of California, Davis where he works at the M.I.N.D. Institute. He previously worked at the Children's Hospital of Philadelphia.



Bronwyn and "friend" in Brisbane, 2006

Bronwyn Glaser, Professional Council Member

Since completing her B.A. at Stanford University in 1998, Bronwyn Glaser has been doing clinical research with children affected by neurogenetic syndromes, including velo-cardio-facial syndrome, fragile X syndrome, and Williams syndrome. In 2004, Bronwyn earned her M.A. at the University of Denver in Child Clinical Psychology, where she was studying the development of psychopathology. She is currently working on her Ph.D. in Psychology at the University of Geneva in Switzerland while doing research devoted to understanding the specific cognitive and behavioral phenotype in VCFS. Her work is now focused on VCFS, and she has worked with VCFS families at Stanford University in California and the University of Geneva, under the mentorship of Allan Reiss and Stephan Eliez. Bronwyn's current doctoral work is focused on the

contribution of visual perceptual impairments to abnormal social cognition in VCFS. After her doctorate, she would like to combine her research and educational interests to develop syndrome-specific cognitive remediation therapies and ways to cognitively monitor children with atypical developmental trajectories. Being a Californian transplanted in Geneva, Bronwyn takes great pleasure in being exposed to differences in treatment approaches and strategies, as well as the overall richness of language and cultural diversity in Europe.



Amyel in front of a painting she made of Giuseppe

Amyel Loy, Lay Council Member

Amyel was born in Rome the 7th of December 1969. She thinks that what makes life meaningful is children and art. That's why she has two children, Giuseppe and Orlando. She is an art historian and writes art books for children. She considers herself a very lucky woman because she discovered that beauty and love can raise only from complexity and effort. She wrote, "Thank you, VCFSEF, for the wonderful opportunity you are giving me to share and learn."

WORDS FROM THE PRESIDENT

(Continued from page 5)

has a 3-acre lake just outside the doors of the Marriott with a walking trail around it. Within a 2-minute walk, there are a dozen restaurants, dozens of retail stores, a movie theatre, and ESPN zone. We are currently planning the kid's activities, speakers and logistics. Many more details will be worked out over the coming months.

In closing I am so excited to be a part of this great foundation. All of the people I have met and talked with associated with the foundation have been truly a joy to work with. The enthusiasm felt for our kids show in the countless hours our board and staff members put in. I am committed to planning a memorable conference for parents, our children and all the medical leaders who are participating in the 2007 International VCFS Conference. We hope you will join us, July 20-22, 2007.

Thank you and I look forward to a great year!

Keri Alexander

New Kid's Zone Fund

The VCFS Educational Foundation, Inc. has established a Fund called "Kid's Zone." All donations to this fund will be used to offset the cost of children's supervision and programming at our annual international scientific meetings.

We hope this will encourage and enable more professional and lay members of the Foundation to come with their families to the meetings. We continue to make every effort to provide the **Kid's Zone** programs without charge to participants, so your donations will be greatly appreciated.

Please click here to make a donation, or go to <http://www.vcfsef.org/donations> and choose Kid's Zone from the list.

MAX APPEAL CONFERENCE UK 2006 REPORT

Four speakers presented during the morning session of the 2006 Max Appeal conference. Dr Frances Bu'Lock, cardiologist from Glenfield Hospital in Leicester, explained how clinical teams ensure care standards are maintained. Dr Jacob Vorstman explained that 22q11 patients exhibit behaviours similar to those within the autistic spectrum, and thus that similar interventions may be appropriate.

The conference attendees received Dr Vorstman's findings constructively and were very interested and greatly appreciated Dr Vorstman's candidness and humour. Dr Anna Zeffert, Clinical Psychologist at Addenbrooke's Hospital in Cambridge, spoke about cognitive assessments that would be beneficial during the transition to secondary school education. Sophie Brigstocke, a research fellow from York University, has been traveling the UK researching learning patterns in children with VCFS and presented her preliminary findings.

After lunch Dr Andrew Gennery, Consultant Immunologist at Freeman Hospital, Newcastle Upon Tyne. Andy's presentation was titled "All sorts of things about immunology that you didn't know you wanted to know". Andy always manages to explain this complex topic humorously, helped by copious analogies. The next speaker, Aine Lawlor, stole the show. She is 23 years old and was only diagnosed in Ireland when she was 15 and received

little help. However, Aine's prevailed and represented her country in the 2003 Special Olympics and wrote her own speech for the conference. Aine is proof that people can live fulfilled lives with VCFS and go on to make real achievements. Andrea Parker explained the workings of the Kumon programme to help with math and English skills. Karen Marie-Morris explained how the

Parent Partnership Service can give practical help to parents needing special educational needs advice. Finally, Anne Keatly-Clarke, the Chief Executive of the Children's Heart Federation, kindly stepped in to explain how to improve your chances of receiving disability living allowance (DLA).

As the conference was a month earlier this year than normal, Father Christmas was still taking his holiday

and so Astronaut Buzz McHale (aka Granddad) kindly stepped in to deliver presents to all the children. This was Max Appeal's biggest conference, and its location at the British National Space Center helped both the children and parents to really enjoyed it. Our next annual conference will be at the "At-Bristol" centre on the 1st December 2007 (www.at-bristol.org.uk).

(See Photos, page 13.)

The next speaker stole the show.

Aine Lawlor is 23 years old ...

*from Ireland to speak
about her life with VCFS.*

ABSTRACT FORM FOR ALL SUBMISSIONS
14th International Scientific Meeting, July 20-22, 2006, Plano, Texas, USA
The Velo-Cardio-Facial Syndrome Educational Foundation, Inc.
Marriott Dallas/Plano at Legacy Town Center

THIS FORM (INCLUDING AUTHORIZATION AT BOTTOM) MUST BE COMPLETED BY ALL PRESENTERS

TITLE:

SUBMITTED BY (underline person presenting):

TYPE OF PRESENTATION (check appropriate category):

- Professional paper (up to 15 minutes*)
- Professional Seminar (up to 30 minutes*)
- Poster
- Lay Presentations: Sharing the Good Times (5-10 minutes*)
- Other (describe _____)

TIME REQUESTED FOR PRESENTATION* (if different): _____

(*Actual time allocated will be determined by the Program Committee)

PLEASE PRINT OR TYPE

ADDRESS:

TELEPHONE:

FAX:

E-mail:

This form **MUST** be completed and included with all submissions. Abstracts must be received by May 1, 2007. Submit to Dr. Karen J. Golding-Kushner by email at kgkushner@vcfsef.org or by post to: Dr. Karen J. Golding-Kushner, VCFSEF Educational Foundation, Inc., P.O. Box 874, Milltown, NJ 08850. Go to www.vcfsef.org for additional contact and meeting information.

Abstract:

Authors please check boxes as appropriate:

- I GIVE permission for this presentation to be audiotaped. (I do not give permission for taping)
- I give permission for this presentation to be uploaded to the website of the VCFSEF, Inc following the conference. (I do not give permission for upload to website)
- If my talk(s) includes data, photographs, or video that cannot be posted, I will provide an alternate version of my talk.

Signature

Print name

Date

MEDICAL RESEARCH ETHICS:

What You Can Do to Protect Yourself
by: Joel Johnson, M.D.

Raymond Tanner, who has lived with VCFS for over fifty years, suggested we post a link to an article about his experience with unethical medical research practices. You can read the article online by visiting the internet address printed below. In 1986, his newborn son succumbed to his cardiac malformations and died. After autopsy, his heart and brain were removed and studied without the Tanner family's permission.

Unethical research hurts us all. Clearly the participants and their families are harmed. Also, knowledge from such research is often tainted and not accepted by the scientific community. The majority of researchers who are conscientious face a community that becomes rightfully suspicious. In turn, all are disadvantaged because it is more difficult to learn about disorders.

Unfortunately, there will always be a few bad actors like those who took advantage of the Tanner family. Luckily, those who intentionally take advantage of their participants are few. As time has passed, both international guidelines and many countries' laws have become more and more stringent. In the United States, President Clinton's public apology for the infamous Tuskegee medical experiments helped galvanize the movement towards ethical medical research. Audits, review boards, criminal and civil suits have all helped to protect research participants.

Unethical research practices are usually unintentional. How are participants protected? In most cases, any research involving people or animals must go through a strict process of review by a government approved board of experts. These boards have both experts in science, community members, and participant advocates. They help the research catch ethical errors which are often very subtle! For example, a researcher may have to improve the wording on their consent forms to make them easier to read.

The VCFS community strongly supports and is active in research and with that support much has been learned about the syndrome in the past decade. We have come very far since the genetic cause of VCFS was discovered in 1992.

How do you protect yourself and your family from unethical research practices?

Find out if the study has been approved by an ethics board. Make sure that any research you are involved in is approved by one of these boards and that the board follows government guidelines. If the study you are involved in is not funded by the government, make sure that a third party government-approved board has approved the study.

Don't be shy! Ask the researcher exactly what the research is for and why you are being asked to take part. Don't be satisfied with not understanding a single part of the study. It is the responsibility of the researcher to explain everything to you in terms that you can understand.

Understand that studies may not be meant to benefit their participants directly. Direct benefits are limited on purpose. For example, to prevent the poor from being overly subjected to research, financial incentives are very limited. Even if you are part of a clinical trial, you should only expect limited clinical benefits. Some studies offer clinical evaluations, but researchers do not have the same legal responsibilities a health professional does to do a thorough assessment or follow up on diagnoses. Taking part in a study is meant to be good for society as a whole.

If you are part of a clinical trial, understand that a researcher has an unintended bias to keep you in the study. The success of the study depends on you staying in it. Remember this principle and maintain a healthy skepticism. Don't be afraid to ask which treatment group you are in. If anything goes wrong, don't be afraid to contact the investigator and get a second opinion.

None of us can be expected to reasonably protect ourselves from deception from an institution of public trust such as a hospital. In these cases, as in the Tanner family's case, it is up to the community to become active and notify the press and appropriate authorities.

Mr. Tanner's son died soon after he was born and he had no opportunity to plan his death. Most of us are fortunate to have this time. My experience as a physician has taught me that family meetings in an ICU during the last stages of a loved one's life is not the place to make these decisions. Please contact your local hospital for information on how to establish an advanced directive.

You can read about Mr. Tanner's experience at the following internet address or URL:

<http://womansday.ninemsn.com.au/article.aspx?id=154245>

Mr. Tanner reviewed and supported this article. He comments, "If I may add, the Adelaide Woman's and Children's Hospital are currently in the process of having a memorial sculpture made which will be unveiled in the next couple of weeks. In addition, in the next few years they will be creating/planning a memorial garden at the hospital where the sculpture will eventually be permanently based."

As mandated by our by-laws, the VCFS Educational foundation, Inc does not sponsor or fund research. However, as a service to professionals and families interested in studies about VCFS, we may, on request, act as a source of information about research projects by allowing researchers to provide information to prospective subjects about their studies on our website. The information is posted ONLY after the Foundation's research committee has conducted a thorough review of the research protocol and the approval of the IRB (Institutional Review Board) of the facility at which the study is being conducted.

GLOBAL COMMUNITY FORUM

ADHD: EXPERIENCES WITH DIAGNOSIS AND TREATMENT

Global Community Forum

ADHD: Experiences with Diagnosis and Treatment.

Moderator: Joel Johnson, M.D.

Welcome to our first printed global community forum for the Newsletter. The purpose is to collect and archive ideas from the community on important subjects in related to VCFS. Each forum will begin with a brief introduction to the topic. This will be followed by representative comments, selected by the Moderator, from members of the community around the globe who have written to us. We will strive to print as many responses as we can and represent as many views as possible. The views presented are not necessarily endorsed by the editorial staff or the Foundation but we hope they will foster communication. Please note that we cannot print solicitations or direct medical advice. However it is okay to discuss your own medical experiences. We advise that all personal information be removed, however. If medical information is shared, we reserve the right to remove any identifying information. We also reserve the right to edit responses for clarity and grammar, but will not change the content.

The Question

To start the first forum, we asked community members about their experiences with medications used to treat attention deficit/hyperactivity disorder (ADHD) in those with VCFS. As you are probably aware, up to 50% of children with VCFS have diagnosable ADHD symptoms that cause significant impairment in their daily lives. ADHD is not a simple diagnosis. In practice the diagnosis is often made in different ways and there are many problems that follow a diagnosis. These involve deciding on whether to treat it or not, what the positive and negative effects of either decision are, which drugs to use if treatment is chosen, what their potential side effects might be, and many other issues that we do not know about but which families have experienced.

For a review of ADHD and its treatment options in VCFS, see Dr. Gothelf's article "Attention Deficit Hyperactivity Disorder (ADHD) in VCFS." It appeared in the June 2005 issue of the VCFS EF Newsletter. It can be found at this web address:

<http://www.vcfsef.org/newspast.html>

Feel free to request a paper copy at the address below.

The Responses

from Maine

Speaking as a parent—

Our daughter, who is now 24, was diagnosed with VCFS at age 19. Looking back, it's clear that she had vivid symptoms of ADHD from early childhood, but this diagnosis was not formally made until she was in her middle teens. She was treated with Adderall and Tegretol at about age 15. I worried that her difficulty learning and remembering was related to the Tegretol and when she was 18 I persuaded the psychiatrist to drop it. She then became psychotic (oops). She's doing fine now.

One thing about VCFS is that the symptoms of ADHD you see can have a variety of origins. You can be staring into the face of bipolar disorder or schizophrenia and not realize it. What looks like ADHD can also turn out to be thyroid failure—this happened to our daughter at age 11. She had gradually developed Hashimoto's encephalopathy, but because she hadn't been diagnosed VCFS, none of the half-dozen doctors I took her to thought to do a TSH test. I suspect she lost some brain cells in the course of that fiasco. An ENT finally decided to rule out thyroid problems.

Speaking as a professional (I'm a neuropsychologist)—

I never cease to be amazed how many of my clients, both young and old, appear in my office with a working diagnosis of ADHD. The most memorable was a woman in her late fifties thought to have late-onset ADHD (?!), but turned out to have had a stroke. I also routinely see children who have attentional symptoms secondary to PTSD and early-onset bipolar disorder.

Thinking about medications... It's a delicate and difficult process to help family physicians (and some psychiatrists) realize that you really, really don't want to prescribe a psychostimulant and/or antidepressant (without mood stabilizer) to a kid with a major family history of bipolar disorder or schizophrenia. I wonder how many adults we see who have undiagnosed VCFS. Likely quite a few, though I haven't caught one myself, yet...

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GLOBAL COMMUNITY FORUM...

(Continued from page 10)

from Spain

My son is 11 years old and has been diagnosed with VCFS (1 year old) and ADHD (9 years old more or less). He has had a lot of surgery (heart, palate, hernia, and ear). The ADHD diagnosis was due to his bad behavior at the school. He always forgot his homework, and had a lot of problems with math and vocabulary.

We had an appointment with a psychiatrist and my son started with methylphenidate, commercial name in Spain is Rubifen. But he became very anxious, irritable, and his meals became like a torture due to decreased appetite. He slept poorly (sleeplessness, with nightmares). After some weeks he was very depressed and he took other pills for his depression (I don't remember the name), but the effects were the opposite. During this period my son was affected by bullying at school and probably the effect of the medications was hidden in some way because of the bullying. In spite of everything, we decided to not have more medications during the summer. When he started a new year at a new school, we tried to start with medications and we went to another different psychiatrist who suggested that we try long-acting methylphenidate (Concerta). He explained about the improved tolerance people have of Concerta due to its sustained delivery of the methylphenidate throughout the day. We started with a small quantity, half of the recommended starting dose for his age and weight - 18 mg. It worked quite well. The results were not spectacular, but his attention slightly improved. The psychiatrist tried to increase to the typical dose (36mg) and its effect was similar to that of Rubifen, including anxiety, irritability, poor appetite, poor sleep, nightmares, depression, and bad general behavior due to his inability to control anxiety. We decided to stop all the medications. Because the psychiatrist insisted, I take Concerta myself - a 36 mg pill each day for one week. I'm extremely absentminded, and have some characteristics of ADHD. But, I hope that I do not have ADHD. The pills worked really well and I had no side effects. I have spoken with other parents of children with ADHD and I have heard about both good and bad experiences with Concerta.

Today my son is not taking any medications. Sometimes it's too hard to work with him and his attention is very poor, but our experience is that the bad effects of methylphenidate in my son were more than the benefits in his attention. My son's behavior at school is not good and it requires extra work at home. We have known that there is another non-stimulant medication called Strattera, but now we are quite skeptical about any medications. At the moment we prefer to deal with ADHD symptoms without medications. I think that there is not extensive experience in Spain with ADHD. My personal opinion is that psychiatrists are too much confident with Concerta. In general they are very opposed to hearing anything bad about Concerta. Also, I think that the treatment can't only be a pill: parents need to have some education about how to deal with the children. I find this point particularly difficult in Spain.

from North Carolina

My son has VCFS with ADHD. He is 10 years old now but was diagnosed with ADHD at age 6. We have chosen to medicate him with Concerta, and he takes 72mg each morning. Without the medication, I feel my son's life as well as the lives of his family members would be severely impacted. When my son wakes up in the morning, he is clumsy, obstinate, distracted, difficult, and not enjoyable to be around. He has a hard time finishing his breakfast due to his distractibility. He makes annoying noises and bothers his brothers. After 45 minutes when the medication has had a chance to be absorbed in his system, he is a different child. He is much more coordinated, he stops bothering his brothers, he is cooperative, and he can stay on task. It is an amazing transition.

My son plays the piano quite well, but unmedicated his fingers are not able to play as well, and he inappropriately speeds through his pieces. He can read unmedicated, but he could never tell you what he has read. He can write unmedicated, but it looks like chicken scratch and is too large of a print for his age. With medication, he plays the piano beautifully, reads at grade level and is able to tell you what happened in the story, and writes quite well. At school he has kept As and Bs through the 4th grade, passed his fourth grade writing test, and made a 3 on the state end-of-grade testing, which is at grade level. He is generally chosen as one of the first "terrific kids" in his classroom for the school assembly due to his good behavior and diligence in his school work. Without the medication, I do not feel my son could maintain good grades or have good behavior. He would also be one of the children at school that the other students were frustrated with due to a lack of control of his behavior. ADHD kids are often the ones that lose privileges for the whole class because they are unable to behave at school and get their work done. My son is well liked and is never one of the ones that disrupts the class. He would only be frustrated at school and could not learn to the degree that he does if he were not medicated. In addition, our home life would be much less enjoyable if he acted all day like he does the first 45 minutes of his morning.

Now, I must add that the medication does not completely alleviate the ADHD. He is still more distractible than my other children, and the medication does not last until he goes to bed. We compensate by reading and doing homework earlier in the day or in the late afternoon. We cannot wait until after dinner to do homework, or he has lost his attention span. Also, he probably eats a little lighter lunch but makes up for it at dinner time. He does a lot of snacking after dinner instead of the afternoon, which is certainly fine. He sleeps well usually, but it takes him a few minutes longer to fall asleep.

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GLOBAL COMMUNITY FORUM...

(Continued from page 11)

All in all we find Concerta absolutely invaluable in treating my son's ADHD. I don't know what we'd do without it. It gives me back the son I know and love and he is able to achieve success in many areas of his life.

Professional Perspective –

Joel Johnson M.D. and Tony J. Simon Ph.D.

The letters above illustrate the many problems that parents face when trying to find treatment for ADHD in children with VCFS. First consider the children without VCFS. Increasing awareness of the diagnosis of ADHD and proof that treatment improves overall outcome has led to the situation that, for these children, the standard of care is to use medication. This standard is supported by hundreds of medicine trials with tens of thousands of participants and the large "Multimodal Treatment Study of Children with Attention Deficit Hyperactivity Disorder" study carried out in the USA. Despite this, treatment of ADHD with medication in typically developing children is still controversial. This is largely because of the side effects of the medication as well as the cost of purchasing the drugs. It is a difficult decision for any parent to make.

In VCFS, the decision is more difficult. There is only one published study on this topic (Gothelf, Gruber, Presburger, Dotan, Brand-Gothelf, Burg, Inbar, Steinberg, Frisch, Apter, & Weizman, 2003). The study involved the use of methylphenidate (trade name: Ritalin) and studied only 12 children with VCFS for one month. While the outcomes were positive it is hard to know if all children with VCFS would respond in the same way. Furthermore, there is good theoretical reason for parents to be concerned about how some of these drugs will affect their children. For example, as in the letter from Spain, children with VCFS may be more prone to the side effects of direct stimulants because they cannot process them very well. The natural variation in the now well-known COMT gene may have an effect on the occurrence and strength of those side effects but the role of that gene in this process is still not clearly understood. Children with VCFS are likely to be more susceptible to side effects such as psychosis and seizures due to some of these medications. On the other hand, children with VCFS would greatly benefit from treatment. With developmental delay and specific cognitive impairments, some early help in being able to focus one's cognitive abilities in order to learn better may be the edge that such children need to improve their developmental outcomes. Clearly this is the case in children without VCFS.

What is a parent supposed to do? Luckily there are many medication and therapy options for children with VCFS. The best strategy will be to work closely with a child psychiatrist, pediatric neurologist, or developmental pediatrician who is familiar with VCFS. As often is the case, parents will likely be in the role of educating their doctors. Specifically, parents should warn their doctors about the susceptibility of children with VCFS to side effects of medications for ADHD.

Gothelf, D., Gruber, R., Presburger, G., Dotan, I., Brand-Gothelf, A., Burg, M., Inbar, D., Steinberg, T., Frisch, A., Apter, A., & Weizman, A. (2003). Methylphenidate treatment for attention-deficit/hyperactivity disorder in children and adolescents with velocardiofacial syndrome: an open-label study. *J Clin Psychiatry*, 64(10), 1163-1169.

Next Forum Question

Parents of those with VCFS and those with VCFS are often in the awkward position of educating their health care providers. We need your assistance in gathering information about how you have tried to find knowledgeable providers, how you have educated the providers to whom you have access, or other strategies that have produced an improved outcome. We appreciate any type of response. It could be a personal narrative about a family's experience with the ups and down of finding informed care. For example, we are interested in how experiences vary between those living in large and small towns. As always, we are also interested in perspectives from different countries. We are also interested in hearing from professionals who had patients bring new information to their attention. What was the most effective way for patients to approach you with new or different information? What did you find helpful? What was not helpful? What do you think is the most effective way for patients to bring information to your attention?

Write to:

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PHOTOS FROM MAX APPEAL MEETING



Left... the main meeting hall at Max Appeal meeting with standing room only.



Harrison and Lindsey Stedman take in some sites while attending the Max Appeal meeting.

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 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 nicrobbins@aol.com.



Educational Foundation Logo Tee-Shirts

Promote VCFS awareness by purchasing "Knowledge is Hope" tee-shirts, available in youth sizes S, M, L and adult sizes S, M, L, XL, XXL, XXXL.

The tee-shirts are white and made of 100% cotton, and read KNOWLEDGE IS HOPE, above the VCFSEF logo.

You can purchase your tee-shirts directly from the foundation's web site at http://vcfsef.org/t_shirts.html. Each tee-shirt is \$15 .00 US, with free shipping in the U.S. and Canada. For orders of 4 or more, shipping outside the U.S. and Canada is also free. Online payments are processed through PayPal.

For questions regarding tee shirts, including shipping cost to other destinations, contact Keri Alexander directly at skalex1@tx.rr.com.



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