

18th Annual International Scientific Meeting
The Velo-Cardio-Facial Syndrome Educational Foundation, Inc.
Hyatt Regency Hotel, New Brunswick, NJ, July 14-17, 2011

PROGRAM:

9:00 – 9:15: **Opening Remarks**

Welcome from the VCFSEF – *Karen J. Golding-Kushner, Ph.D., Executive Director, Christina Bush and Jennifer Stevens, NJ 2011 Local Arrangement Co-chairs*

9:15 – 9:30: The VCFSEF: Present, past and future; *Karen J. Golding-Kushner*

9:30 – 9:50: Keynote Address: How to determine if a therapy is based on science; *Gregory L. Lof*

- *Very funny talk about what to believe and whether information is based on science of quackery*

9:50 – 10:00: Questions and Comments

The Genome and VCFS

10:00 – 10:15: When does a 22q11.2 deletion not cause VCFS? *William D, Graf, Ryan Miller, Robert J.Shprintzen, Robert R. Lebel*

- *A presentation on the possibility that there are other disorders associated with 22q11 Deletion, caused uproar*

10:15 – 10:35: Heterozygosity of Tbx1 causes phenotypes in social behavior and spatial and working memory in mice; *Noboru Hiroi, Takeshi Hiramoto*

- *Very medical and I am unable to offer information*

10:35 – 10:55: Combined human and mouse genetic approaches to find genes for VCFS; *Bernice Morrow*

- *One again medical research*

11:20 – 11:35: Tissue interactions during inner ear development are regulated by the VCFS gene *TBX1*; *Dennis C. Monks, Evan Braunstein, Bernice E. Morrow.*

- *VCFS Patients with Hearing loss – 30% conductive hearing loss secondary to recurrent middle ear infections and 10% Sensorineural due to the mal development of the inner ear*
- *Tissue interactions regulated by Tbx1 are important for the development of the inner ear*
- *Loss of Tbx1 in the POM (tissue surrounding the Otic Vessel/ inner ear which helps form the bone to protect the inner ear) leads to changes in the retinoic Acid, WNT and SMP signaling to the Otic Vessel*
- *Understanding how Tbx1 regulates signaling during ear development can be applied to understanding the development of more severely affected systems in VCFS patients*

11:35 – 11:50: New directions in genome research; *Vandana Shashi, David Goldstein, Wendy R. Kates, Robert J.Shprintzen.*

- *Medical/scientific*

11:50 – 12:05: The prevalence of chromosome 22q11.2 deletions in 2,478 children with congenital cardiac malformations; *Peter Agergaard, Charlotte Olesen, John R. Ostergaard, Michael Christiansen, Karina M. Sorensen*

- *This study determined that the prevalence of 22q11.2 DS is approx. 1.9% in children with cardiac malformations*
- *The prevalence of 22q11.2ds is approx. 7% among children with conotruncal malformations*
- *¼ were not diagnosed in the usual clinical setting prior to the testing*
- *VSD or TOF are the most common malformations among children with 22q11.2 DS*

12:05 – 12:20: Genetic Modifiers of Cardiac Outflow Tract Anomalies in Humans with the 22q11.2 Deletion Syndrome; *Tingwei Guo on behalf of the 22q11.2 International Consortium*

- *Mecial/scientific*

12:20 – 12:35: Phenotype to phenotype correlations: the relationship of IQ to physical phenotypes in VCFS; *Nathaniel Miletta, Marcia Friedman, Cheryl Roe, Dongliang Wang, Bernice E Morrow, Wendy R. Kates, Kevin Antshel, Anne Marie Higgins, Robert J. Shprintzen*

- *A cooperative of a Irish Scientist (Dr Allan Holland) and The VCFS Centre in Syracuse to apply machine learning techniques to a large database of patients with VCFS to better understand complex Phenotypic relationships (Phenotype -observable features or characteristics in contrast to Genotype which represent genetic characteristics)*
- *Machine Learning-*
 - *Development of Algorithms allowing computers to alter behaviour based on data*
 - *Recognition of complex patterns not readily apparent with manual observation*
 - *Based on the data set the data mining program they use builds a decision tree composed of clinical features that are most likely to lead to the phenotype of interest*
 - *In VCFS it works if there is a large cohort of patients, large number of characteristics (VCFS has 180) and use of Data Mining to predict the likelihood of future phenotypic development based on characteristics that may observable at the time of diagnosis*
- *Having a CHD is associated with lower IQ*
- *Having a Cyanotic Heart Disease is also associated with lower IQ*
- *Other causes of decreased IQ could be due to Cyanosis, hospitalisation, ICU stays etc..*
- *ADD/ADHD and Chronic Otitis Media(COM)*
 - *There was a positive association between ADD/ADHD and COM.*
- *There are associations between COM and Craniofacial findings such as Prominent nasal bridge, attached ear lobules, VPU and palate anomalies, bulbous nasal tip and small ears.*

Speech and Language Disorders and Treatment

1:45 – 2:15: Logic, theory and evidence against the use of non speech oral motor exercises; *Gregory L. Lof*

- *Do not use non speech oral motor exercises; Gregory L. Lof*

2:15 – 2:35: Early intervention speech services: effective or not? Panel session (*Dennis Ruscello, Eileen Marrinan, Sue Carneol, moderated by Karen J. Golding-Kushner*)

2:35 – 2:55: Normal speech in VCFS: achieving the goal; *Karen J. Golding-Kushner*

- *Normal Speech is attainable through Speech Therapy, Surgery if needed, Dental treatment, Home Management, Coordinated treatment as long as there is a common goal.*
- *Difficult to relay people should consult via distance or find a therapist who understand VCFS*
- *Do not use Non Speech Oral Motor Speech Exercises" they should work on articulation, as speech = speech*

Velopharyngeal Dysfunction and Surgery

3:20 – 3:35: Velopharyngeal valving during speech in patients with velocardiofacial syndrome and patients with non-syndromic palatal clefts; *Antonio Ysunza, Ma. Carmen Pamplona*

3:35 – 3:50: Surgical Management of Hypernasal Speech in VCFS: results and complications from a series of over 100 consecutive cases; *Sherard A. Tatum III*

3:50 – 4:10: Panel: What to do if VPI is detected early: surgery now or later? *Antonio Ysunza, Sherard Tatum III, Robert J. Shprintzen*

4:10 – 4:20: Questions and Comments

Personal Success and Independence: Adults with VCFS

4:20 – 4:40: Marriage and VCFS: a panel of successful adults with VCFS and their spouses; *Quinn and Pari Bradlee, Karen and Douglas Lindsay.*

- *Quinn and Pari Bradlee plus Karen and Doug Lindsay answered questions from the audience about issues growing up, family and the struggles with drink and drugs.*
- *A very inspiring panel.*
- *It was clear though that these two VCFS adults grew up in very affluent families and I ponder the reality of how other young adults without this type of family may stand in the future.*

4:40 – 4:55: The Joys and Challenges of being a Parent, when the Parent has VCFS; *Karen Lindsay*

- *Karen talks about the decision to have a baby naturally over that of using VCFS. They have a healthy non VCFS baby.*
- *Karen talks about the struggles of being a parent with VCFS and coping with the day to day life and issues of raising a child*

4:55 – 5:15: VCFS Around the World; *Altuna, Campbell, Glaser, Gothelf, Natsume, Ysunza,*

- *Updates from around the world on what they have done and achieved. The VCSFEF brochure is now in several languages and the newsletter is also translated each quarter.*
- *Europe is expanding and the main group Max Appeal is moving forward quickly. The Irish group will host the conference in 2013 in Dublin*
- *Australia is by far the leader when it comes to awareness campaigns.*

5:15 – 5:25: Questions and Comments

5:25 – 6:15: Annual Business Meeting and Elections (Everyone welcome, only Members vote, be sure to join)

- New committee –
 - Executive Director – Dianne Altuna Speech Therapist from Texas
 - President – Phillipe ??? France
- A new direction

Saturday, July 16, 2011

☑ Peer relations and dealing with bullies; *Tracy Vaillancourt, Merav Burg-Malki*

- *Tracy Vaillancourt from Canada hosted this breakfast. It was fabulous and the main component was that advocacy and knowledge stops bullies*
- *Bullies usually stop within 10 seconds if an adult or another person intervenes on behalf of the victim.*
- *Bullies who continue without remorse are usually the minority and are likely to be adult psychopaths*
- *A great way to stop bullying is to educate the children about the specific differences of the child, usually don't have the child present. Don't make it a massive issue but explain that these are the reasons eg; little jonny has hearing aids, etc....*
I have asked for information from Tracy

Dental and Orthodontic Issues

9:00 – 9:15: Longitudinal soft tissue and skeletal facial development in VCFS; *Michael Arvystas*

9:15 – 9:30: Dental and orthodontic disorders and treatment in VCFS; *George Cisneros*

9:35 – 10:05: Platybasia in VCFS; *Ariela Nachmany, Yehuda Finkelstein, Doron Gothelf*

9:45 – 9:55: Questions and Comment

Behavior, Psychiatry, Cognition, and The Brain

9:55 – 10:20: Integrating cognition, emotion, and psychiatry for treatment in VCFS; *Tony J. Simon* *See DVD Talk*

10:20 – 10:35: The development of cognitive control in VCFS; *Heather Shapiro, Tony J. Simon*

- *Impairments in cognitive control are generally predictive of Schizophrenia*
- *VCFS Children are impaired on tasks that measure cognitive control*
- *It is not clear how these cognitive control impairments relate to the risk of Schizophrenia*
- *A study of VCFS and Typically developing (TD) children on Task Inhibition (one component of cognitive control) in order to characterize development*
- *The study that was conducted showed that TD improve with age and suggests that some children with VCFS do not improve with age on a task of response inhibition while other improve similarly to typically developing children*
- *Increased performance variance in older children with VCFS suggests a subgroup with atypical development of response inhibition*
- *Individual performance patterns might identify those with more or less typical cognitive control abilities*

11:00 – 11:20: What happens to ADHD across time in children with VCFS? *Kevin Antshel, Kaitlyn Hendricks.*

No Notes

11:20 – 11:35: Hippocampal volume and hormonal correlates of stress in children with VCFS; *Elliott A. Beaton, Kathleen Angkustsiri, Margie H. Cabaral, Yingratana A. McLennan, Ingrid Leckliter, Janice Enriquez, Tony J. Simon.* *See DVD presentation*

11:35 – 11:50: Spatial cognition impairments and possible atypical hemispheric connectivity in VCFS; *Frederick C. Bassal, Margarita H. Cabaral, Monika Farhangi Oskuei, Ling M. Wong, Tony J. Simon.*

- *This talks about a study using a computer based task measuring the way that a person accurately judges the location of objects in space.*
- *A target image appears on a horizontal line, either in the centre, offset to the left or right (offset is by 2mm increments from 2-16mm)*
- *The Landmark Task - Participants are asked to push a button indicating whether they believe the target is directly centred or offset to the left or right.*
- *The Line Dissection Task- Pencil and Paper task involving perception and motor skills. This measures the accuracy to which one measures the centre point of a line. Participants are asked to hand draw a vertical line as close as possible through the centre of a pre marked horizontal line (repeated 10 times)*
- *The results show that VCFS children perform poorer than typical Developing (TD) on both tasks.*
- *It also showed with age that the VCFS children did not improve with age on the Line Dissection task.*
- *The results of both tasks lend support to prior findings that children with VCFS have compromised spatiotemporal/visiospacial cognition compared to TD peers*

11:50 – 12:05: How coping skills affect symptoms of anxiety and depression in young adults with VCFS; *Elise Parson, Ulrich Schall, Linda Campbell.* *Research not yet published.*

- *60% of the VCFS population have anxiety*
- *16% of the VCFS population have depression*
- *25-30% of the VCFS population have psychosis*
- *Risk factors for anxiety and depression in VCFS*
 - Individual Factors*
 - *Social Functioning*
 - *Dependency vs Independence*
 - *Self –Perceived Efficacy*
- *The research focused of the coping mechanisms and the relationship to anxiety/ depression in VCFS*
 - *Is Maladaptive coping more common in VCFS than the control group*
 - *Is there a relationship between coping strategies and anxiety/depression*
 - *Is the relationship between maladaptive coping and anxiety/depression mediated by individual factors*

- *The results show that the VCFS group had higher levels of Anxiety and Depression and the VCFS group had lower social functioning , lower rates of independence and less perceived efficacy – No groups differed in coping styles*
- *The results showed there was a significant correlation between maladaptive coping styles and anxiety and Depression in the VCFS group- individual factors did not mediate relationship between Maladaptive coping and anxiety or depression*

☑ Lunch for Parents: Raising awareness and supporting each other: Starting and growing a support group; *Maria Kamper*

- *Discussions on setting up a support group, parents taking control and advocating*
- *Joining together to form alliances and raise awareness*
- *Made some contacts internationally*

Behavior, Psychiatry, Cognition, and The Brain: Interventions

1:30 – 1:45: Computerized cognitive remediation in adolescents with VCFS: a preliminary report; *Wendy R.Kates, Kerri Tang, Lauren K. Sanderson, Matthew Kurtz*

- *Reports on the Preliminary results of a study of the effectiveness of a computerized cognitive remediation program that is being administered to adolescence with VCFS*
- *The participants were administered the Challenging our minds (COM) program*
- *www.challenging-our-minds.com/*
- *The findings of the study show that remediation program MAY lead to overall improvements in overall mental ability.*
- *There is a cross over study phase 3 of the project. This will go for 24 months*

1:45 - 2:00: The effect of methylphenidate on prefrontal cognitive functioning, inattention, and hyperactivity in VCFS; *Doron Gothelf, Tamar Green*

- *Risk factors for ADHD in VCFS*
 - *Rate of ADHD in first degree relatives*
 - *VCFS with ADHD 12%*
 - *VCFS without ADHS 2.2%*
- *Methylphenidate in VCFS – Why Not*
 - *Side effects*
 - *– Depression , Psychotic*
 - *Increased risk of sudden death in individuals with Cardiac anomalies*
 - *Putatively high brain levels of catecholamine due to reduced dosage of COMT*
- *Methylphenidate in VCFS – Why Yes*
 - *ADHD is common and debilitating problem in half the children with VCFS*
 - *It has a robust clinical effect on symptoms of inattention and impulsivity which are common in VCFS*
 - *No good alternatives for ADHD*
- *A study was conducted to evaluate the efficacy of Methylphenidate on prefrontal cognitive functioning in VCFS, to evaluate the long term safety and effectiveness in VCFS*

and evaluate the effect of COMT genotype on clinical response to Methylphenidate and adverse events.

- The conclusions showed Methylphenidate to be effective and well tolerated by children with vcfs and co morbid ADHD treated for 6 months*
- It had a positive effect both on ADHD clinical symptoms and some prefrontal cognitive functions*
- It may increase the heart rate which is important in some cases*
- A full cardiac evaluation is recommended for all children with VCFS prior to initiation of Methylphenidate treatment*

2:00 – 2:15: Intervention for social skills deficits in children with VCFS; *Carrie Bearden, Jennifer Ho, Carolyn Chow, Chelsea Anderson.*

- This presentation outlines the characterization of social deficits in children and adolescents with VCFS 22q11 DS, the treatment approaches for social deficits, exercise-induced brain plasticity, neuro plasticity as a pre-requisite for treatment response and development of possible interventions.*
- Characteristic social deficits*
 - Autism spectrum*
 - Schizotypal Spectrum*
 - Cycle of bullying/teasing*
- The study showed*
 - Those children with VCFS/22q11DS are at increased risk for a variety of psychiatric symptoms and social deficits are components of many disorders.*
 - Poor social function very common*
 - Early intervention programs can help*
 - Exercise may provide the optimal platform in which remediation effects can occur*
 - New treatment will integrate social activities with Physical exercise*
 - While physical exercise can boost brain development, social element may differentially improve social function and self esteem*

2:15 – 2:30: Mentoring program as an effective psychosocial intervention to encourage social engagement in children with VCFS; *Merav Burg-Malki, Doron Gothelf.*

- This is a presentation on the mentoring program in Israel.*
 - The program has been operating for over 10 years with 100 psychology students and 50 children and adolescents with VCFS.*
 - The program provides a caring, sensitive, supportive adult friend to the child*
 - Building therapeutic intervention that is designed specifically for the needs of the child*
 - Helps the child to develop new abilities and skills*
 - It is based on weekly meetings – sessions are 2 hours*
 - Over one academic year*
 - At the child's house or surrounding areas*
 - The psych student writes a weekly report*
 - There is group supervision every 2 weeks*

- *The mentor encourages social engagement*
 - *Through playdates*
 - *Uses social autopsy to jointly analyse social errors and design alternate strategies*
 - *Provides a child with a positive model of appropriate skills*
 - *Observes the child in a variety of social situations to gain a deeper understanding of their strengths & weaknesses*
 - *Encourages and reinforces the child's social initiative*
 - *Talks with the child about friendship*
 - *Teaches the child to judge the quality of their relationships with peers*
 - *This program in Israel is free of charge*
 - *It strengthens the relationship between the family and the clinic*
 - *Increases awareness of VCFS*
 - *The other part of the presentation was presenting a study of the Mother Child interactions to obtain if it is specific to VCFS*
 - *The results conclude that children with VCFS display lower level of engagement during mother child interaction and that mothers to vcfs children tend to be more active during the interaction and acted in a manner that encouraged the child towards more intense social engagement*

2:55 – 3:15: Self-Hypnosis to Reduce Anxiety in Patients with VCFS; Ran Anbar

- *Very interesting presentation*
 - *Hypnosis does not involve an unusual condition, sleep, placing the subject under the control of the hypnotist.*
 - *The goal in VCFS is*
 - *to reduce anxiety from psychosocial stressors*
 - *reduce discomfort from medical procedures*
 - *help control reactions to stress*
 - *provide a coping skill*
 - *help increase self esteem*
- *I have the slides should anyone be interested*
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3:15 – 3:35: Web-based socio-emotional remediation for persons with VCFS; Bronwyn Glaser, Danny Dukes, Sonia Martinez, Amélie Dubuc, Catherine Pasca, Stephan Eliez.

- *A new program will be out in August to assist with remediation of socio emotional issues of people with VCFS*
- *Social deficits in VCFS can be enriched using Social Enrichment programs*
- *The Vis-à-vis program specifically aids attention reasoning abilities, facial emotion recognition and behaviour*
- *The program appears to help two groups of developmentally delayed children and adolescents in developing their attention and emotion recognition skills*
- *Due out in August*

3:35 – 3:50: Should psychiatric treatment be aimed at symptoms or at the genome? *William D. Graf, Robert J. Shprintzen.*

More for the Doctors

Implications for School and Peer Relations

4:00 – 4:30: Dealing with bullying and victimization; *Guest Faculty Tracy Vaillancourt*

- *Bullying can take many forms*
 - *Physical, verbal, social, cyber, racial, sexual*
- *Long term consequences*
 - *Academic difficulties, truancy, increase in absences, somatic complaints, stress related illness, physical health problems, low self-esteem, depression, social withdrawal, social anxiety, loneliness, suicide, aggressive behaviour*
- *During an average week teens spend less than 8% of their time talking with adults and 1/3 talking to peers and it is reported they feel most unhappy when talking to peers*
- *The need to belong is inbuilt in all of us*
- *Social ties are essential for optimal development*
- *Preventive efforts to end bullying in schools and communities needs to be accelerated*
- *Teachers and principals must take all acts of bullying serious and not disregard any child who makes a complaint. They need to be vigilant and watch for changes in the most susceptible children*

4:30 – 4:50: Focus on Math Interventions for Students with VCFS; *Donna Cutler-Landsman*

- *Most children with VCFS will have a maths difficulty of some kind*
- *We need to focus on number sense*
- *Emphasis on math language*
- *Formal teaching of math symbols and relationships to number concepts*
- *Use a visual approach*
- *Instruction in mental visualization to aid understanding*

4:50 – 5:05: Addressing Bullying and Social/Written Language through the IEP; *Yolanda Ortiz*

5:05 – 5:25: Education Panel, Questions and Comments; *Donna CutlerLandsman, Yolanda Ortiz, Tony Simon, Tracy Vaillancourt*

Sunday, July 17, 2011

Medical Issues and Management

9:00 – 9:15: Increased Medical Interventions in Children with VCFS; *Emily King, Betsy Leech, Kevin Stanford, Robert Hopkin, Howard Saal*

- *Mostly for doctors*

9:15 – 9:30: Is Oral feeding in Children with Severe Medical Issues a Possibility? *Jill Merrow*

- *This talk was aimed at explaining feeding issues in young babies with severe medical issues. Referral to Jill / Dr Shprinzen would be best as this was a very touchy subject*

9:30 – 9:45: VCFS Plan of Care; *LuAnn Weik, Stephen F. Conley, Adriane L. Baylis, Sue O. Carneol*

- *A copy will be posted to our website*

9:45 – 9:55: Questions and Comments

9:55 – 10:10: Family disclosure of a VCFS diagnosis; *Dana Faux, Sonja Eubanks, Vandana Shashi, Kelly Schoch*

- *A very small study on a research study about family disclosure.*
- *The study showed that disclosing the diagnosis need not be that serious, it could be as simple as telling the child on the way to the doctors to explain why they are going*
- *Parents lacked the confidence to tell people*
- *Concern over what the child may think – self concept*

10:10 – 10:35: How do I tell my child? When do I tell my child? *Betsy Leech*

- *This was the best presentation of the conference in my view. It asked us to go back and think about how we felt when we received the diagnosis. Then Betsy hypothesized that a child does not feel the same emotion or understanding that we as adults do. They do not feel the same remorse and once told will probably just brush it off and want to go back to playing ball or whatever they were doing.*
- *If we make the name a part of our everyday life, it should not bring too many mental implications. Children are resilient. In teens it's a great way to explain a life of difficulties and proof that they are not stupid, ugly etc....*

10:35 – 10:55: Panel discussion and Refreshments: Disclosure of Diagnosis; *Anne Marie Higgins, Betsy Leech, Vandana Shashi, LuAnn Weik*

10:55 – 11:25: Scoliosis and its treatment; *Guest Faculty Steven Reich*

- *This was mainly for medics to help determine scoliosis and treatment*

11:25 – 11:55 VCFS and Pediatric Care- There's No Place Like a Medical Home; *Guest Faculty Jack Levine*

- *This is mostly related to USA*

11:55 – 12:05: Questions and comments

International Advocacy and Support

12:00 – 12:10: Texas Senate Bill 1612: inception to outcome; *Dianne Altuna*

- *Relates to USA*

12:10 – 12:20: The Importance of VCFS Support Groups; *Maria Kamper*

12:20 – 12:25: Being a VCFS Advocate: How one person can make a difference; *Raymond Tanner.*

- *Read by Karen Kushner;- talks about all that Raymond has done to advocate for VCFS and encourages us all to advocate*

12:25 – 12:30: Concluding remarks; *Karen J. Golding-Kushner*

12:30 Adjourn.