

VCFSEF News

VCFSEF, Inc. Newsletter

Spring, May 2001

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Stay tuned to the Foundation Web Site for updated conference information, including registrations & clinic sign-ups.

<http://www.vcfsef.org>

*2001 Annual Meeting,
Boca Raton / Deerfield
Beach, FL
Agenda on page 6.*

Straight Forward VCFS Parent Advocacy

by Paula Long

Any parent reading this newsletter is most likely aware that they must be informed advocates to optimize their child's care. However, the path of advocacy can be diverse. The following parent written articles offer two different perspectives. One, written by an anonymous parent reflects largely personal experience and the other, written by Donna Landsman, reveals the perspective of a parent and educator.

I am a parent in the throes of advocating for my child in the U.S. public school system. I am writing under an assumed name in order to protect my child and family from retaliation. I struggled with writing this article because parents don't dare risk jeopardizing their relationships with the schools, which have so much power over their children's lives. I would have so much appreciated an article like this when I first found out that I needed to work with the special education department at my child's school. It found out that I needed to work with the special education department would have saved my family and me time, money, and a lot of worry, heartache and bewilderment.

*Thus began
the process of
awakening. . .*

I have spoken with many parents who have told me these same stories over and over. Someone and break through the phytes on this journey, I hope I can be the first to help them through the initial stages faster to get their voice. Someone and break through the phytes on this journey, I hope I can be the first to help them through the initial stages faster to get their voice. For those already well along in their advocacy, I hope this is an affirmation that they are not crazy. For professionals and lawmakers, I hope this is an eye opener of what it is like to deal with the special education maze in this country. There may be those who take offense and disagree with some things in this article, but I made sure that I am not alone in having experienced these things.

When my pediatrician notified me that the genetic test for our youngest child was positive for velo-cardio-facial syndrome (VCFS). I went through shock and grief. When our neuropsychological specialist notified me that our child was a bright child not a dull child, as the school system had labeled him, I began experiencing significant anger. Thus began the process of awakening, so that I could begin advocating for my child. Next, I moved into acceptance and action.

Dumbing Down The Child

This phenomenon frequently occurs in the American school system. If a child is labeled dull, expectations can be set low. Services and resources required are less. Then if the child does not do well, the school is not held accountable. Furthermore, if the child falls behind, the school is not accountable to remediate the child to grade level.

Beware of averaged sub-scores on standardized tests. The schools know they should not average scores for children with learning disabilities, but they do it anyway. They count on the parents not knowing any better.

The Child is Doing Just Great!

"He is performing at grade level. How do we know? Because we think so!" Beware of reluctance to do frequent standardized testing. This is the only way to know if your child is working at grade level. Make sure that the school repeats the same tests so that comparisons can be made from test to test.

You Must Get Your Own Neuropsychological and Educational Testing Done

If the school system does or pays for the neuropsychological and educational testing, it will be biased in the school's favor. If you want to really know what is going on with your child, take out a loan and get it done independently. Be sure to research and find someone who is familiar with VCFS.

Children as Defective

I will be forever grateful to Peter Wright, Esq., special education lawyer of www.wrightslaw.com. In his Tactics Manual, he has a chapter on how the educational system views the child and parent. Children with special needs are defective parts in the system and the school is not responsible for defective parts. Children with VCFS are especially defective because they have a genetic cause of their learning disabilities. If you know this going in, you know how to engage in advocacy for your

child and save a lot of time and bewilderment.

Ignore VCFS and It Will Go Away

School systems are wary of newfangled diagnoses. If the school does not mention it, maybe it is a figment of the imagination. Maybe it is not really such a problem after all. Then it does not have to be addressed.

Bad/Pushy Parents

Schools view parents who advocate for their children as pushy. Period. Save yourself a lot of time and anguish and just accept it. Remain polite, businesslike and move on.

The School Will Not Tell You What the Child Needs

Parents must become experts in both their child's disabilities and in educational techniques for those particular disabilities. This is an enormous undertaking. Most parents do not have the education or training in these areas and do not know where to begin to find out these things. Schools will not share this information because if the parents find out, they will ask for these services, which under the law, the schools are obligated to provide if the child needs them. Schools cannot afford this. One thing that would be absolutely priceless to parents is if the VCFS professionals could develop a comprehensive educational reference list that would match interventions to problems so that parents would know

what to ask schools to provide for their children.

The System is Set Up to Be Adversarial

Funding for special education (SPED) is set up so that it seems as though it is taking away from regular education. Non-SPED parents view SPED families as pariahs. Schools do not get adequate funding for SPED in many systems. Therefore families must fight for services for their children. Schools cannot cope

with the volume of services needed. They do not have the manpower, talent, resources, training or classroom space to provide adequately for children with special needs.

Given this scenario, schools systems are usually very entrenched,

bureaucratic and unimaginative. As a consequence, huge amounts of resources are dedicated to fending off the demand for services that special needs children require instead of spending those precious resources on needed services. Children with very obvious disabilities e.g. cerebral palsy cannot be ignored and often get needed services. However, if a child has "invisible disabilities" i.e. learning disabilities, their chance of receiving adequate services are far less. Unfortunately, many VCFS children fit in the latter category. One caveat – if a child can play well at football or basketball, there is a better chance of receiving services.

The Stalling Tactic

School systems count on parents getting exhausted and giving up. They try to wear the parents down. It is all too often a very effective tactic. The victim is the child

School systems are wary of newfangled diagnoses. If the school does not mention it, maybe it is a figment of the imagination.

who needs services and does not get them. Insist on having high expectations for the child and do not back down. Every child deserves a chance to live up to his/her potential. Comedian Phyllis Diller once said "Aim high so you don't shoot your own foot off." The top goal may not be met, but at least something gets accomplished.

Schools Cannot and Will Not Provide All Needed Services

In the process of stalling, your child is getting older. It is better to get the services or remediation the child needs now on your own and keep fighting to get the school system on board in the meantime. If not, the child will be 18 and nothing will have been done to help him. Live with it and move on – as fast as you can. It is best to try at all times to work with the schools, but go in with eyes wide open. If your child needs services that the schools cannot or will not provide, you must provide them. Then fight to get the schools to fund the services if you want to go that route. If you wait to get the school to provide the services, the child will fall further and further behind.

School Professionals Did Not Go Into the Profession to Do A Bad Job

Special education professionals across the U.S. feel bad about the system too. They want to do a good job and feel good about their jobs.

So What is a Mother To Do?

Use anger to get energized to help the child, not to get even. Get counseling to get going. Read laws, medical literature, go to seminars, network, volunteer for the local special education parent advisory council, talk to the Department of Education for the State, contact the Federation for Children with Special Needs, join the VCFS Educational Foundation, search the Internet, look up Wrightslaw.com and advocate at the legislative level. And get an advocate. Now.

Advocates are for all VCFS children, not just for hearings. Even if an advocate does not go to team meetings, their expertise and knowledge are indispensable. Free or low cost advocacy is available through the Federation for Children with Special Needs and your state Department of Education. Do something. There are so many parents who think they cannot make a difference. Every little thing makes a difference.

Most importantly, maintain a cordial business relationship with the school system – it really is easy and easier than fighting. In the process, get organized and document everything. I have included some resources for parents at the end of this article for getting started on the advocacy journey. There are many links in the websites. Never give up. Care for each other in your family and may the power of love be with you.

Resources

www.fcsn.org Federation for Children with Special Needs Website

The Complete IEP Guide: How to Advocate for Your Special Ed Child by Lawrence M. Siegel, Nolo.com Inc. Berkeley, CA, 19992.

www.wrightslaw.com Peter and Pamela Wright's website

Tactics and Strategy Manual by Peter D. Wright and Pamela Darr Wright, Harbor House Law Press, Hartford, VA, 1999

Wrightslaw: Special Education Law by Peter D. Wright and Pamela Darr Wright, Harbor House Law Press, Hartford, VA, 1999

Audio Tapes will again be available from 2001 conference. If you want a full set, let us know and we'll provide your name to the vendor.

RESERVE NOW -- pay at the conference.

Details on the Foundation web site at: www.vcfsef.org/vcf_ref.html

So, you moved!

Please notify the Foundation when you have a change of address and or email, so we can keep our database current. It is costly to mail newsletters to absolute addresses.

Email is our fastest and most frequent means of communication, and we get back about 30% of our email messages, which means that many of you are not getting the information we're sending, as quickly as you might..

So, if you have an email address, and would like to receive information or updates more quickly, please provide us with a current email address.

Presently, we mail about 1300 newsletters worldwide and we'd like to reduce that number, if possible.

If your system can read and print a .pdf file (Adobe Acrobat), we can decrease our postage costs by emailing you the newsletter, or simply notifying you when it is available on the web site for download or viewing.

If the "E-news" option is acceptable for you, and if you haven't already done so, please email us at vcfsef@mail.upstate.edu.

In the subject of the email, just write: "E-Newsletter Okay".

Thank you! and have a great day!

7th Annual Meeting

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

Deerfield Beach/Boca Raton, Florida, July 20-22, 2001

This CME event is co-sponsored by the Office of Continuing Medical Education, SUNY Upstate Medical University at Syracuse. The State University of New York Upstate Medical University is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians. The State University of New York Upstate Medical University at Syracuse takes responsibility for the content, quality, and scientific integrity of this CME activity.

The State University of New York Upstate Medical University at Syracuse designates this continuing medical education activity for 13 credit hours in Category 1 of the Physician's Recognition Award of the American Medical Association.

CEUs for Speech Pathologists are offered by the Floride Speech-Language Hearing Association

Friday, July 20, 2001

- 12:00 noon: Registration
- 12:30 P.M.: Introductory remarks
Robert J. Shprintzen, Ph.D., Executive Director
Barbara Dandron, President
- 1:00 P.M. Velo-Cardio-Facial Syndrome, Congenital Heart Disease, and Asthma: A Prospective Study
Michael J. Ackerman, M.D., Ph.D.; Arlene VanGalder, R.N.; Mark E. Wylam, M.D.; Robert H. Feldt, M.D.; Coburn J. Porter, M.D.; Gordon Dewald, Ph.D.; Paul D. Scanlon, M.D.; David J. Driscoll, M.D.
- 1:30 P.M. Vasomotor Instability in VCFS: Implications for Surgical Correction of Congenital Heart Disease.
Vandana Shashi, Wake Forest University Baptist Medical Center, Winston-Salem, NC
- 1:50 P.M. Questions and Discussion
- 2:00 P.M. Speech Therapy in VCFS: A Model for Professionals to Follow
Karen J. Golding-Kushner, Ph.D., Kean University, Union, NJ.
- 2:45 P.M. Speech Therapy at Home: A Mother's Personal Experience
Amy McDonald, Lake Orion, MI
- 3:05 P.M. Questions and Discussion
- 3:15 P.M. Coffee
- 4:05 P.M. A Three-Year Prospective Study of Leg Pains in VCFS
Ahmad Al-Khattat, University College, Northampton, United Kingdom
- 4:35 P.M. Immunizations in the Child with VCFS
Anne Marie Higgins, N.P., R.N., Upstate Medical University, Syracuse, NY
- 4:50 P.M. Questions and Discussion
- 5:00 P.M. Business Meeting
- 6:30 P.M. Annual Dinner

Saturday Morning, July 21, 2001

- 8:30 A.M. Registration and Continental Breakfast
- 9:00 A.M. Modeling the complex and variable VCFS phenotype in the mouse.
Elizabeth A. Lindsay, Ph.D., Baylor College of Medicine, Houston, TX
- 9:35 A.M. Tbx1 function(s) in congenital heart and vascular disease
Antonio Baldini, M.D., Baylor College of Medicine, Houston, TX
- 10:10 A.M. Gene dosage and genomic disorders on chromosome 22q11
Bernice Morrow, Ph.D., Albert Einstein College of Medicine, Bronx, NY
- 10:45 A.M. Coffee
- 11:00 A.M. Can Tbx1 Account for the Spectrum of Anomalies in VCFS?
Robert J. Shprintzen, Ph.D., Upstate Medical University, Syracuse, NY
- 11:35 A.M. Questions and Panel Discussion: The Interface Between Molecular Genetics and Clinical Findings In VCFS
Antonio Baldini, Elizabeth Lindsay, Bernice Morrow, Robert Shprintzen, Vandana Shashi
- 12:00 P.M. Lunch

Saturday Afternoon, July 21, 2001

- 2:00 P.M. Brain Changes and Intellectual Function in Velo-Cardio-Facial Syndrome
Wendy R. Kates, Ph.D.; Courtney Burnette; Leslie Abbott; Walter E. Kaufmann; Godfrey Pearlson; Johns Hopkins University School of Medicine, Baltimore, MD.
- 2:30 P.M. Educational Goals in VCFS
Donna Landsman, M.S., Madison, WI
- 3:15 P.M. Advocacy for the Child with VCFS
Nadine O. Vogel, MBA, Founder and Director, S.N.A.P. (Special Needs Advocate for Parents)
- 4:00 P.M. Questions and Discussion
- 4:15 P.M. Coffee
- 4:30 P.M. A Concert from the Heart
Karen Ruckman

Sunday, July 22, 2001

- 9:00 A.M. Establishing and Maintaining Support Groups for VCFS
Maureen Anderson, Salem, NH, NE Support Grp
- 9:30 A.M. The List: Using the Internet Wisely
Panel Session
- 10:00 A.M. Surgery for Speech: Goals and Outcomes
Sherard A. Tatum, III, Upstate Medical University, Syracuse, NY
- 10:30 A.M. Questions and Discussion
- 10:45 A.M. Coffee
- 11:00 A.M. Pediatric Cardiology and VCFS: A Panel from Miami Children's Hospital
1. Common Cardiac Abnormalities in VCFS; *Elizabeth Welch, M.D.*
 2. Noninvasive Techniques in the Management of Common Heart Abnormalities in VCFS; *Evan Zahn, M.D.*
 3. Surgical Management and a Team Approach to Complicated Cardiac Defects in VCFS; *Redmond P. Burke, M.D.*
- 12:00 Noon Adjourn

Concurrent Clinics

Speech Clinics and Leg Pain Clinics will be held throughout the course of the meeting. The speech clinics will be staffed by Karen J. Golding-Kushner, Robert J. Shprintzen, Eileen Marrinan, Susan Marks, and Robert Beecher. The Leg Pain Clinic will be staffed by Ahmad Al-Khattat and Stewart Goldman.

The preceding program is a tentative schedule and subject to change to accommodate last minute additions in an attempt to keep the program as current as possible.

PREREGISTRATION FORM

(Preregistration will be accepted until June 30, 2001)

Seventh Annual Meeting, July 20 - 22, 2001, Deerfield Beach, FL

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

Name: _____

first

middle initial

last

Address: _____

Home Phone: _____

Work Phone: _____

please include area code and extensions; if not in U.S./Canada, include country & city code

Fax: _____

E-Mail: _____

I am a member of the Foundation: yes _____ no _____ Lay _____ Professional _____

If you are not a member, contact us for a membership form, send it with the preregistration, you will receive the member's registration fee

Accommodations: The entire meeting will be held at the Hilton Deerfield Beach/Boca Raton Hotel. The hotel is close to both the Fort Lauderdale and Palm Beach airports and is also close to Boca Raton. All rooms are \$75 (rate per room not per person). Reservations should be made directly with the hotel or with Hilton International. Call 1-800-624-3661 or 1-800-HILTONS, you are attending the Velo-Cardio-Facial Syndrome Educational Foundation meeting. The hotel has a beautiful pool area, exercise facility, and restaurant. The beach is two miles away. Golf is readily available with courses accessible to the hotel. Shopping at the Mizner Park is close to the hotel.

Registration Fees: *Registration includes attendance at the meeting, continental breakfast, and refreshment breaks. Annual dinner is a separate fee. Enclose a check or money order made out to **The Velo-Cardio-Facial Syndrome Educational Foundation.** For registrants from outside of the U. S., please send a bank draft payable in U.S. dollars*

Single preregistration, member	\$50.00 U.S. _____
Family preregistration, 2 members (please list names below)	\$70.00 U.S. _____
Single preregistration, nonmember:	\$60.00 U.S. _____
Family preregistration, 2 nonmembers (please list names below)	\$90.00 U.S. _____
Family preregistration, 3 or more members (please list names below)	\$80.00 U.S. _____
Family preregistration, 3 or more nonmembers (please list names below)	\$100.00 U.S. _____
Student member preregistration:	\$40.00 U.S. _____
Student nonmember preregistration:	\$50.00 U.S. _____
Optional Friday Night Welcome Dinner:	\$30.00 per person _____

Total Enclosed _____

Please list all family members to be registered (family registrations only):

I would like my child seen at the speech clinic _____ I would like my child seen at the leg pain clinic _____

I am interested in a sibling workshop _____ I am interested in a teen/young adult get-together _____

I am interested in child care during the meeting _____ I am interested in sharing a room _____

Registration fee at the door will be an additional \$10.00 per person, if space is available.

Send checks with completed form made payable to: Velo-Cardio-Facial Syndrome Educational Foundation

address: c/o Dr. Robert J. Shprintzen, Jacobsen Hall 714

Upstate Medical University

750 East Adams St., Syracuse, NY, 13210

Receipts will be available at the door.

Understanding the Educational System, Advocating and Surviving Raising a VCFS Child

Donna Landsman, MA

As a growing organization whose mission is to educate, it is extremely important that we work to benefit our children's lives by helping others to understand their challenges and hopefully to assist them. Even though there is increasing knowledge of this syndrome, the vast majority of doctors, teachers, lawmakers, etc. have never heard of it. Consequently, little is in place to assure that our children receive the services that many of the so desperately need. It is therefore not surprising that parents are frustrated in their attempts to find the best education for their VCFS child. There are no easy answers or magic methods that erase the problems associated with VCFS and learning. Very little research has taken place to determine which educational approaches work best and little is documented assessing the long-term outcome that can be expected. We are on a pioneering journey where we need to keep an open mind, listen to each other, and learn together how to best proceed.

While the public school system is traveling with us, they also will need time to try new approaches and techniques. It is inappropriate, however, to assume that the school system is in the practice of undermining our children or purposely working to withhold services. Most educators do not alter test scores or assume that children are too dull to learn. Like all bureaucratic agencies, they are working under a set of guidelines that spell out particular criteria that, by law, need to be used to place children in special educational programs. The laws were set up to provide services to children who show severe deficits in learning, speech and language or have behavioral problems that impede learning. Certainly many VCFS children do meet the criteria and in most cases will be allowed to receive services. Many parents have had success using the Other Health Impaired category to access the special education programs. This category covers chronic health issues that

diminish a child's strength and alertness; therefore affecting the ability to learn without special interventions. Attention issues, hypotonia, frequent surgeries, thyroid problems, immune system dysfunction, etc. are all health matters that would affect a child's ability to be alert and ready to learn. Parents can, and should inform the school personnel of the medical difficulties their children are having so teachers can better understand the syndrome and make necessary accommodations.

Parents must also understand that they will need to be more actively involved in their child's education than they would be with a normally developing child. They will either need to take the time to work 1-1 at home, or hire a tutor to help with assignments and studying for tests. Without this more individualized help, many VCFS children will struggle to keep up in class and will likely not master the concepts presented in a large group format at school. Unless the child is enrolled in a private, specialized school, it is unlikely that the intense tutoring will take place. Public schools are not set up to offer this kind of support for a child. While children may be given small group instruction in some areas, they will be mainstreamed for many classes with 20+ students.

Teachers in these large classes can not take the time it takes to slow down, re-explain concepts, and give the specialized help VCFS children need. Parents can ask for modification in these large classes, (such as study guides before tests, modified grading, notes, teacher aides, etc.) to help their child be successful. Eventually, as the child reaches high school age, a program that focuses on job, social, and functional living skills may be appropriate.

Parents *should advocate for their child and it is wise to understand the IDEA law and the rights they have to request services. Pulling together to educate legislators about this syndrome may help to get services provided without so much difficulty. Setting up an adversarial relationship with the school system, however, does not always work in the child's best interest. Parents certainly should not go into the schools expecting to have difficulties accessing services.*

I do feel that our Educational Foundation can (and does) give valuable help by assisting with a better understanding in the following areas:

1) Educate professionals that VCFS offers unique learning challenges—children with this syndrome will need specialized educational help. Even though they may decode words at an early age, and appear to be progressing normally they likely will be deficient in math concepts, problem solving, comprehension, abstract reasoning, spatial relationships, and social skills. Insist that these areas be tested and addressed by school districts.

2) Collect data on successful interventions—we need better documentation of programs that work. At this point, case studies and anecdotal information needs to be assembled. I would be happy to start keeping track of some of these if parents wish to e-mail me. Try to be as specific as possible.

3) Collect data on successful high school and adult transition programs—we need more direction on what we can expect from our children as they mature. Will they need assistance as adults? What kinds of jobs have VCFS adults found? What classes in school will better prepare them for these jobs? Are there colleges/schools/camps that can help? Perhaps a better understanding of a possible future for our children will alleviate some of the frustration associated in dealing with the school system.

4) Continue to provide a forum for the wonderful professionals who are researching VCFS to report results of their studies so that we can all improve our understanding. What we learn from this research will help us design appropriate programs, tailor our expectations, and seek appropriate help.

5) Assist with lobbying efforts to get this syndrome recognized by the legislators who fund programs.

Raising a child with this syndrome is challenging to say the least. We need better treatments, school settings, educational interventions, and flexible teachers. This will take time, research, advocacy and education. We can all do our part by staying positive, focused and supporting each other in our efforts.

The Challenge of Appropriately. . .

Defining and Addressing Speech and Language Problems in Children with VCFS by Eileen Marrinan, MS, CCC, MPH

This is the first in a series of three articles covering this topic. The next issue will focus on children three to six years of age and the subsequent newsletter will address the school-aged child.

PART I. BIRTH TO THREE YEARS:

As parents interact with each other in support groups or at conferences, they become increasingly aware of the great diversity in communication issues in their children. Many parents have relayed to me their nomadic journey in quest of understanding the nature of their child's communication problem. Most are appropriately leery of "one-size fits all" answers to their questions. Recently published evidence (Scherer, et. al) suggests that there may be one main speech and language profile in young children with VCFS and potentially several distinct sub-profiles. Of course, while there are trends, it is important to remember that every child is unique.

How can parents become strong advocates for their children in obtaining appropriate care? Given the broad range of possible problems over time, perhaps it is best to uncover all stones along the path, beginning in infancy. The first step is to find a speech-language pathologist who has expertise in children with VCFS and cleft palate. Information about Cleft Palate Teams can be obtained from cleftline.org (1-800-24CLEFT) and the VCFS Educational Foundation (vcfsef.org). A FULL speech and language evaluation should be conducted before the first birthday. Evaluations should be conducted yearly and more frequently should new concerns arise. This allows for appropriate goals and a reference point to measure the child's progress at each assessment. Parents should expect a detailed picture of their child's communication. This explanation must be to the parents' satisfaction so they may become effective advocates. General information such as "your child's understanding of language is 'mildly delayed'" is less helpful in planning therapeutic and educational guidelines.

POSSIBLE PROFILE:

Scherer, D'Antonio and colleagues have shown that children with VCFS have an asynchrony in their speech and language development that manifests itself before one year of age. Children from 6 to 30 months were found to have a relative weakness in their speech sound and language output as compared to their non-verbal cognitive abilities and understanding of language. Specifically, the quantity and quality of speech sounds (in babbling and later in words) and vocabulary use were all relatively delayed.

ASSESSMENT:

The following outlines the speech and language areas that should be investigated and addressed in every child with VCFS during the first three years of life.

Play Development

- These skills are a window of a child's early non-verbal cognitive development. A standardized tool such as the Symbolic Play Test (Lowe and Costello) is preferable to observation alone.

Language Comprehension

- Vocabulary knowledge
- Understanding of relationship between words
- Morphological markers (the present progressive -ing, plural markers, etc.)
- Linguistic concepts (size adjectives, prepositions, etc.)
- Abstract language

Language Production

- Number of words produced
- Classes of words used (verbs, nouns, descriptive words, etc.)
- Early sentence structure (mean length of utterance and flexibility of early grammatical structures)
- Early conversational skills (turn-taking, considering a listener's perspective, etc.)
- Early oral narrative skills (ability to tell stories)

Speech Sound Development

- Mean Babbling Length (classification of babbling complexity calculated by dividing the level of babbling complexity by the number of vocalizations (Stoel-Gammon))
- Speech sound repertoire- full inventory of English consonants and vowels produced by child.
- Presence of deviant speech sounds (e.g. glottal stops, pharyngeal fricatives, etc.)
- Ability to control temporal and spatial properties of speech (speech motor skills)
- Are there assimilatory error patterns such as "pup" for "cup" even though she can produce a "cuh" sound?)
- Does she neutralize vowels in multi-syllabic words ("puhpuh" vs. "puppy")?
- Does she delete consonants known to be in her repertoire ("ha" vs. "hat")?
- Velopharyngeal Function. Although a child this age is too young for surgical intervention, speech characteristics (e.g. hypernasality, reduced oral pressure, etc.) associated with velopharyngeal incompetency should be documented, if present.

INTERVENTION:

Speech

- Normal speech production can and should be expected. Odd as it may seem, speech therapy can begin before first words appear. Therapy can be viewed as preventive (avoiding the onset of glottal stops) as well as facilitative of normal speaking patterns. Since language is a social phenomenon, group therapy can be most effective. However, speech articulation therapy is not conducive to a group setting. Individual attention is necessary to address the specific motor skills and complexity level for each child.

Language

- As many of you may know, signing as a means to promote communication while a child is actively working on improving her speech articulation skills is not an agreed upon treatment approach (VCFS newsletter, 1998). Nor is it necessary for all children. It is my strong personal bias based on extensive clinical experience that if a child has the underlying capacity to speak but does not yet have the speech sounds to do so, it is essential for her to express herself symbolically. This is not just to reduce frustration (it does) but to optimize the child's language learning potential during this critical phase of development. There is old adage - "We learn to talk and we talk to learn". The longer a child communicates by pointing and grunting (EH!) the more likely glottal stops will develop.

Traditional techniques to promote the understanding and production of language in therapeutic or educational settings are appropriate. It is most important for parents to be aware of the weekly goals set forth by the speech-language pathologist.

GLOSSARY:

Language is an understanding of what's said and an expression of one's communicative intent.

Speech represents the actual sounds made.

Velopharyngeal Incompetency refers to a lack of closure between the mouth and nose during speech resulting in air-flow through the nose.

Glottal Stop is a deviant articulation pattern where the child stops the air in the vocal folds (voice box) rather than in the mouth.

A **Pharyngeal Fricative** is another example of a deviant articulation pattern where the tongue is pulled to the back of the throat to make "long" sounds (e.g. Sssss).

REFERENCES:

Scherer NJ, D'Antonio LL, Kalbfleisch JH. Early speech and language development in children with velocardiofacial syndrome. *Amer J Med Genet.* 1999; 88:714-723.

Scherer NJ, D'Antonio LL, Rogers J. Profiles of communication disorders in children with velocardiofacial syndrome: comparison to children with Down syndrome. *Genet Med* 2001;3:72-78

Scherer NJ, D'Antonio LL. To sign or not to sign: pro. *VCFSEF Newsletter, VCFS Educational Foundation; Syracuse, NY: 1998.*

Stoel-Gammon C. Prespeech and early speech development of two late talkers. *First Lang.* 1989;9:207-224.

GENERAL GUIDELINES FOR A SUCCESSFUL SPEECH ARTICULATION THERAPY PROGRAM

The goal of speech therapy is termination of the program. Normal speech production must be the expectation of all. A specific plan of action to achieve this goal should be fully understood and agreed upon by all those helping the child improve their communication skills.

Speech Pathologist as the Coach

Parents and other caregivers such as grandparents and older siblings should be enlisted as an integral aspect of the therapy process. Unlike other motor skills such as playing the piano, we are speaking all the time. Thus, without a strong home therapy component, children have infinitely more opportunities to practice the wrong speech patterns than the target ones that are addressed in the therapy room. There are endless ways to practice speech and language tasks in a non-punitive manner (story time, in the bath, traveling in the car, etc.).

Child as active participant in the therapy process

Therapy cannot be done to a child. It must be done with a child who is fully aware of the goals. Even a very young child should be able to tell another what he does in speech therapy ("play games" is not the right answer!).

Four stages of learning:

Each therapy session should be appropriately challenging to the child but must allow many opportunities for success. The hierarchy of difficulty should consider these four stages at each task level.

- Unconscious incompetence (e.g. child does not realize she is making sound in throat (glottal stop) versus her mouth)
- Conscious incompetence. In a way a young child can understand, they become aware of the problem (e.g. identifying a "cough" vs. "mouth" sound as modeled by the speech-language pathologist).
- Conscious competence. The key step to learning. Therapy sessions and home activities should be replete with opportunities here (e.g. very purposely lifting the tongue tip to behind the upper teeth to produce a T sound). This step must follow a hierarchy of success (tongue movement alone, to sound to words to sentences etc.).
- Unconscious competence. The muscle memory for the correct production at each level of difficulty is mastered in and out of the therapy room.

Give child a road map to his mouth.

Speech is a complicated affair. Most adults do not realize how individual speech sounds are made. Children need to be taught in a way that is comprehensible and interesting. I often use a train (tongue), tunnel (mouth that is opened and closed with lips) and train-gates (teeth) metaphor. Thomas train is the tongue tip, the mid tongue is the passenger car and the caboose is the back of the tongue. For example, to introduce a T sound I would use the above named toy trains, a mouth puppet and a mirror to teach a child to elevate their tongue tip for the T speech sound.

THERAPY RESOURCES

Step by Step: Speech Therapy Techniques for Cleft Palate Speech. Teaching video produced by The Hospital for Sick Children. Cleft Lip/Palate Program, 555 University Ave, Toronto, ON Canada, M5G K8 (416-813-7490) \$100 US.

Therapy Techniques for Cleft Palate Speech & Related Disorders by Karen J. Golding-Kushner. Singular Press. Available through Amazon.com. \$49.95.

Our membership is growing steadily, and there are many areas with no support structure. If you are willing to be -- or participate in -- support activity in your area for other VCFS parents, drop us a line or an email at [The Foundation](#), and let us know. . .

Thank you!

The VCFS Educational Foundation gratefully acknowledges the contributions from the following individuals:

Ms. Carol Kelleher
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Lakewood, CA
\$450.00

“TBX1” is Responsible for Cardiovascular Defects in Velo-Cardio-Facial / DiGeorge Syndrome

The following is a summary of an article just published in the journal **Cell** (*CELL 104:619-929, FEB 23, 2001*):

SUMMARY

Velo-cardio-facial syndrome (VCFS)/DiGeorge syndrome (DGS) is a human disorder characterized by a large number of clinical features including cardiovascular defects. Most VCFS/DGS patients have a deletion of a region on one of their two copies of chromosome 22, termed 22q11.

To investigate the etiology of this disorder, we generated mice that have a deletion corresponding to that on 22q11. The mice had one normal copy of its chromosome and one copy harboring a small deletion. Approximately half of these mice died soon after birth, or around birth, and had cardiovascular defects. The mice also had a missing parathyroid gland, important for maintaining normal calcium levels. A gene therapy approach was used to return some of the genes that were deleted in the mice.

The cardiovascular defects in mice were partially rescued by adding back four human genes, including TBX1. These data allowed us to localize the gene(s) responsible for the cardiovascular defects. Mice overexpressing the four genes also had defects in structures affected in VCFS/DGS patients including the thymus gland. This suggested that the function of one of the four genes is very sensitive to altered gene copy number. Based upon its expression in the developing structures affected in patients with VCFS/DGS, we specifically inactivated one copy of Tbx1 in the mice. The resulting mice developed cardiovascular defects, and they were identical to those in the mice carrying the deletion and similar to those in VCFS/DGS patients.

These results, together with the expression patterns of Tbx1, suggest a major role for this gene in the molecular etiology of VCFS/DGS.

Sandra Merscher^{1,*}, Birgit Funke^{1,*}, Jonathan A. Epstein^{2,*}, Joerg Heyer¹, Anne Puech³, Min Min Lu², Rannik J. Xavier⁴, Marie B. Demay⁵, Robert G. Russell⁶, Stephen Factor⁶, Kazuhito Tokooya⁷, Bruno St. Jore³, Melissa Lopez¹, Raj K. Pandita¹, Marie Lia¹, Danaise Carrion¹, Hubert Schorle⁸, James B. Kobler⁹, Peter Scambler¹⁰, Anthony Wynshaw-Boris⁷, Arthur I. Skoultschi³, Bernice E. Morrow¹ and Raju Kucherlapati¹

Notes:

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In addition, several more publications have cited the role of TBX1 in the development of VCFS, including a recent paper by our guest speakers at this year's meeting in Florida, Elizabeth Lindsay and Antonio Baldini. Their paper is listed below:

Lindsay EA, Vitelli F, Su H, Morishima M, Huynh T, Pramparo T, Jurecic V, Ogunrinu G, Sutherland HF, Scambler PJ, Bradley A, Baldini A. Tbx1 haploinsufficiency in the DiGeorge syndrome region causes aortic arch defects in mice. *Nature*. 2001 Mar 1;410(6824):97-101

ABSTRACT

DiGeorge syndrome is characterized by cardiovascular, thymus and parathyroid defects and craniofacial anomalies, and is usually caused by a heterozygous deletion of chromosomal region 22q11.2 (del22q11) (ref. 1). A targeted, heterozygous deletion, named Df(16)1, encompassing around 1 megabase of the homologous region in mouse causes cardiovascular abnormalities characteristic of the human disease. Here we have used a combination of chromosome engineering and P1 artificial chromosome transgenesis to localize the haploinsufficient gene in the region, Tbx1. We show that Tbx1, a member of the T-box transcription factor family, is required for normal development of the pharyngeal arch arteries in a gene dosage-dependent manner. Deletion of one copy of Tbx1 affects the development of the fourth pharyngeal arch arteries, whereas homozygous mutation severely disrupts the pharyngeal arch artery system. Our data show that haploinsufficiency of Tbx1 is sufficient to generate at least one important component of the DiGeorge syndrome phenotype in mice, and demonstrate the suitability of the mouse for the genetic dissection of microdeletion syndromes.

Upcoming Regional Meetings

California

Date: May 20, 2001, 1 to 3 pm
Place: Stanford University, Psychiatry Building, Room 2209
Speaker: Stephan Eliez, M.D., and colleagues
Topics: What we know about genes, brain development and behavior in children with VCFS

Colorado

Date: June 17, 2001, afternoon
Place: TBA
Event: Family picnic
Contact: Shelly Czopek 303-494-8071

Florida: Miami 22q Support Group for VCFS and DiGeorge

Date: May 10, 2001, 10:30 to 1:30
Place: Romano's Macaroni Grill, Boca
Event: Mother's Day Luncheon
Contact: Barbara Dandren 561-394-9791

Mid-Atlantic Support Group

Date: May 12, 2001, 7 pm to midnight
Place: Tall Cedars
Event: Fundraiser for support group
Contact: [Diane Holland 410-335-2887](tel:410-335-2887) or ladydi722@home.com

Date: September 9, 2001, 2 to 4 pm
Place: TBA
Speaker: Mary Heppel, educational consultant and advocate
Contact: [Diane Holland 410-335-2887](tel:410-335-2887) or ladydi722@home.com

Nebraska 22q Family Support Group

Date: Meets the second Monday of every month, 7 pm
Place: Horizons Community Church Office
5930 South 58th Street, Suite Q, Lincoln
Contact: Anne Stalker 402-435-0221

North East VCFS Support Group

Date: June 2, 2001, 1 to 4 pm
Place: National Birth Defects Center, PARCenter conference room, floor 1
Speaker: Ann Helmus, PhD and Rafael Castro, PhD
Topic: Neuropsychological Testing
Contact: Maureen Anderson 603-898-6322 or mladja@aol.com

We Apologize...

This is a new item for us, and we fully expected to have the newsletter out in time to disseminate the information regarding regional meetings. We'll do better next time. *IN THE MEANTIME...* please furnish us this information as far ahead as possible, and we'll get it on the web site *and* put it in the newsletter(s) so as many as possible may benefit.

***JULY 1st !
is the cut-off for
Conference Pre-
Registration.
Registrations not
postmarked by July
1st MUST be paid
on-site at the
conference.***

VCFSEF Newsletter

The VCFSEF newsletter is the official publication of the Velo-Cardio-Facial Syndrome Educational Foundation, an international not-for-profit, self-help organization dedicated to providing support and information to individuals who are affected by Velo-Cardio-Facial Syndrome, their families, physicians, and other practitioners.

This document is published for informational purposes only, and should not be used as a substitute for competent, medical advice.

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