
VCFSEF Newsletter

VOICE OF THE VCFS EDUCATIONAL FOUNDATION

FALL - NOVEMBER 20, 1998

University Hospital, 750 E. Adams St, Jacobsen Hall Rm 707, Syracuse, NY 13210 Telephone: (315) 464-6590

1998 Meeting in Boston Another Great Success.

The 4th Annual Meeting of the Velo-Cardio-Facial Syndrome Educational Foundation was held in Boston, MA, June 26 - 28 at Children's Hospital. Over 225 people attended from all parts of the United States, including California, Florida, Texas, Washington, and Wisconsin. Registrants from England, Israel, Japan, and Australia were also on hand to give the meeting a strong international flavor.

Highlights included a wide variety of professional presentations, the first "great debate" (see this issue of the Newsletter), presentations from parents, and a stirring and memorable talk by Alexandra Oppenheimer who was one of the first patients identified in 1978 and who recently graduated with a Master's Degree in Education. Her eloquent and awe inspiring talk was followed by a presentation by her mother, Suzi Oppenheimer, a New York State Senator who described her own problems as a child with reading disorders. Nick Tawa, an eminent surgeon in Boston and the father of a child with VCFS, provided his personal perspective as someone who knows both the professional and personal side of the syndrome.

Plans were cemented for future meetings through the year 2002. This coming July 16 - 18, 1999, the Foundation will meet in Milwaukee. The Inner Harbor of Baltimore will greet us in 2000. The beaches of Miami will beckon in 2001, and our first meeting out of the U.S. will be held in England in 2002.

Though there was a core of familiar faces at this meeting, including Past Presidents Joe Abruzzi and Harry Keleshian, the majority of the registrants were new to the meeting. Announcement of the 5th Annual Meeting is in this edition of the Newsletter. Be a part of the process of

guiding and shaping the meeting. Send any suggestions to the VCFSEF office by mail, fax, or email so we can continue to make the meeting as meaningful as possible to everyone. (Photos, page 12) ❖

The Great Debate

The following pages summarize a feature of this past summer's annual meeting of the Velo-Cardio-Facial Syndrome Educational Foundation. Under the format of a debate, the issue of using sign language for children with velo-cardio-facial syndrome was presented. On the "pro-signing" side were Drs. Linda D'Antonio (of Loma Linda University Medical Center) and Nancy Scherer (of Eastern Tennessee State University). On the other side of the issue were Drs. Shprintzen (Center for the Diagnosis, Treatment, and Study of Velo-Cardio-Facial Syndrome in Syracuse, New York), and Karen J. Golding-Kushner (East Brunswick, New Jersey). The debate's major points are summarized below. Each passage was written independently and neither side had the advantage of seeing their opponents' material. ❖

To Sign or Not to Sign?

Nancy J. Scherer, Ph.D.
Linda L. D'Antonio, Ph.D.

We know that many children with VCFS have severe early communication impairment. Communication is a complex process that involves many components including; speech, receptive language, and expressive language. In older children and adults these various components of communication can be isolated. But in young children who are attempting to learn to communicate, speech and language are highly interrelated. This leads to a controversy regarding the most efficient and appropriate therapy approaches for children with severe speech production disorders. Specifically, the question arises, what do you do to help them communicate?

The approach one takes to intervention is influenced by what one believes the origin of the problem is. Some individuals believe that the origin of many of the speech production problems demonstrated by children with VCFS is related to abnormalities of the pharynx (throat) and velopharynx. This model is very linear and suggests that if you treat the errors related to velopharyngeal valving problems, this will improve articulation, and improvement in language deficits will follow. While it is true that children with VCFS who have severe early speech problems

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often show great improvement in these problems as they grow and develop, language deficits appear to persist and may become even more problematic over time.

Speech and language are highly interrelated in the early stages of development. If a child is limited to using only words which contain the sounds he is capable of producing, they will be able to say few words. Our colleagues would argue that increasing the number of sounds a child can make correctly would permit language to emerge. However, evidence from normally developing children and children with language delays indicate that improving the child's language use facilitates the production of more sounds as the child needs them to express greater language complexity. This phenomenon is called "boot-strapping".

In this case, language supports or bootstraps sound development. Language has been shown to "boot-strap" sound development in young children with cleft lip and/or palate who show early speech and language delays. Therefore, most practicing speech-language pathologists advocate a holistic model of intervention that targets the velopharynx, articulation, language and pragmatics. The ability to communicate is essential to the ability to develop cognitively. Put another way, children learn to talk, and they talk to learn. Recent evidence suggests that a number of young children with VCFS emerge from the prime language learning years with no functional oral communication system. These children are already restricted in their communicative partners. They become increasingly frustrated in communicative situations and in turn this can add a behavioral component to their disabilities. Therefore, many children and parents naturally devise a system of gestures that they both understand. Children naturally use gestures for communicating, e.g. pointing for something they want or putting hands up to be lifted. Sign is simply a conventional gesture system. However, it has greater capability to express a variety of language content and in a form paralleling spoken language.

We are suggesting that this early gesturing be augmented with formal signs in addition to (not to the exclusion of) talking. The holistic model recognizes the very important interaction between the development of sound production and language development and seeks to give the child an effective means of communication early on so that all areas can grow.

In this holistic model where sign is used, it is a bridge to improved communication and learning, is not an end point goal. Used in this manner, literature suggests that sign facilitates spoken language. This finding is very robust. It has been shown for children with many types of learning and developmental disorders including hearing children with language impairments, children with developmental delays and pragmatic disorders and

children with multiple handicaps. We are not suggesting that sign replace spoken language but that the two should be presented simultaneously. In this application, sign serves as a bridge to spoken language.

What constitutes a bridge sign program? First, the child must have prerequisites that underlie the language concepts. That is, he or she must understand the words or symbols and, be interacting. The child must know how to make wants and needs known by pointing or gesturing, i.e. *he must be trying to communicate*. Secondly, *sign and spoken words and sentences are always presented together*. These children are not "deaf", they live in a hearing world and they are exposed naturally to oral input. In a bridge sign program, this oral input does not change. We simply give the child sign as an additional form of input and therefore a means to interact with his or her world.

In addition to giving the child a means of communication, bridge signing delivers multi-modality cues that are; auditory, visual, and tactile. Our goal is to use sign only to give the child an effective, successful way to communicate and to learn while the oral motor mechanism is developing.

The scientific literature clearly supports this approach. There is a strong tradition of bridge sign programs which have been used successfully in many other populations including; normal hearing children with severe speech/language impairment, multiply handicapped, autistic, down's syndrome. Drs. Golding-Kushner and Shprintzen will tell you that they are concerned if you use sign with your preschool child, oral communication will not develop. However, in all of the studies in the literature, with all of the populations studied, signing falls away as the child learns to use oral communication. In fact, the literature shows advantages of signing even for normal children. For example, in one study of hearing children of deaf parents who signed, the children signed before they could speak and showed more rapid language acquisition than matched children who had hearing parents without sign. And importantly, these children had no difficulty switching between oral language and sign language.

Given the strong scientific support for this common sense approach we advocate the use of bridge signing programs for young children with VCFS with severe expressive communication impairment. ❖

To Sign or Not to Sign?

Karen J. Golding-Kushner, Ph.D.

Robert J. Shprintzen, Ph.D.

The two of us have been fortunate to have a long history in studying the speech and language of children with VCFS. In fact, we were both there at the beginning and it

is our data which is cited when scientists report the disorders of speech and language associated with the syndrome. The notion that sign language could be useful in helping children with VCFS communicate came along after we had already been finding ways to help children communicate verbally for more than a decade. We have had many years to learn from our patients and we have incorporated that experience in our therapeutic approaches which has spanned hundreds of patients. In our opinion, resorting to signing is giving in to one mistaken presumption, and adding a second mistake on top of it.

Our esteemed colleagues on the “pro” signing side may point out that the early use of signing avoids frustration and allows language development. However, we believe that speaking, even abnormally, accomplishes more than signing and does it faster and better if the attempts at speech are recognized and reinforced by those to whom the child is talking. We have learned that children with VCFS have a number of problems with their early communication, including language delay that often is mild, velopharyngeal insufficiency with hypernasality, and a severe “compensatory” articulation impairment. It is a very complicated communicative disorder. What we have found is that in the overwhelming majority of children with VCFS, the component which interferes most with the children being understood and communicating is the articulation impairment, not the language delay or the hypernasality.

Hypernasal speech can be fully intelligible if articulation is accurate. However, children with VCFS almost always take an alternative route to articulation, substituting sounds like glottal stops which are so abnormal as to make them unintelligible to even those who are closest to them. However, nearly all children with VCFS do talk, want to talk, and attempt to talk. If the child were severely language impaired, signing would be as difficult as speech. It is our experience that the most “frustrated” children are the ones who are most motivated to speak and progress the most rapidly in speech therapy.

Therefore, clinicians can take one of two paths....the path of least resistance which is teaching them to sign that enables them to express a small number of specific needs, or teaching them to articulate more normally so that they can express their own needs *and* their feelings and humor with inflections which can not be expressed with signs which are not acquired in a natural way like spoken language. In other words, signing limits the child to a few specific symbols selected by the speech pathologist or the parent, not the child. The first option, teaching them to sign, eliminates some of the necessary reinforcement required for the development of normal speech.

Unfortunately, in too many cases we have seen, the child more consistently follows the easier path and speech becomes the secondary mode of communication when it can and should be primary. The second option, teaching them to speak, is solving the presenting problem and maximizing the child’s ability to express himself or herself while increasing the number of people the child can communicate with. The opportunity to develop language is, in large part, enhanced by the opportunities to use language. Signing severely restricts opportunities to communicate with both children and adults. The only reason to pursue the first option (signing) is if children with VCFS can not speak and require sign language. This is simply not the case.

Many years of experience has taught us the following. Using proper techniques for stimulating and shaping early speech and articulation development, it is possible for children with VCFS to have intelligible speech from the outset. If a clinician does not come in contact with a child until 2 or 3 years of age, it is still possible to redirect speech development to the point of intelligibility. The techniques involved are much like many other medical treatments which need to be applied for VCFS. The speech pattern is nearly syndrome specific, but the technique for resolving the disorder is very straightforward. When a child fails to develop intelligible speech, the problem is not with the child...it is with the therapist. If the child does not improve, the techniques being applied are incorrect.

Many speech pathologists today are fascinated with newfangled treatments like “phonological” approaches, or sometimes they have been taught older approaches such as palatal massage or blowing exercises. Neither these newer approaches nor the old ones have any chance of success in the child with VCFS who has the typical severely disordered speech pattern. “Speech therapy” is simply a generic term. It does not imply that the therapy is the right approach. We can name many procedures which will not work, and only a few which will. Because most speech pathologists have little or no experience with VCFS, and because many have had no educational exposure to techniques designed to eliminate glottal stops (or prevent them), they will simply fail in treatment. It is also important for the therapist to know how children with learning and behavioral problems must be reinforced differently from other children. Therefore, should we compromise therapy to do something which is less than optimal? Every minute, every second which is spent teaching a child signing is time spent not teaching the child speech.

Our esteemed colleagues on the “pro” side of signing will argue that giving the child the means to communicate will cause their language skills to grow. We wholeheartedly agree. The difference in our position is that their language skills will grow exponentially faster if the communication skill they are taught is speech. Speech is

preferable to signing because it presents the child with a far more rich and complex language system than signing. In addition, the time spent teaching signing must be spread to family members. Valuable resources must be applied *away from* speech. The child's ability to communicate with signing is limited to those who understand it. Not so for speech.

It is true that many parents will give strong testimonials to their experience with signing. They will point to the fact that early in childhood, their children could not speak or could not be understood and after signing was taught, the children began to communicate. These kinds of testimonials have always been problematic for scientists. They speak to valid observations by caring parents who are working arduously for their children's benefit. Could their observations have another explanation? Possibly.

For example, while the teaching of signing is going on, the children are also getting older. The improvement seen may be as much a function of age as of the signing. While signing does not hinder language development, there is no evidence that it enhances speech development in children who would develop it anyway. Also, specifically related to VCFS, there is a different pattern of speech and language development than in other children, but the ability to develop verbal language is almost always present. There are many parents who can provide equally compelling testimonials to the effectiveness of early speech therapy. Scientists do not typically accept anecdotal evidence because the data provided by them can not be systematically analyzed. Regardless of what our opponents in this debate may say, there have been no studies whatsoever which have looked at the effectiveness of signing in children with VCFS when compared to speech therapy. None. It may even be true that children who are taught to sign and those who are taught speech from the outset end up in exactly the same place at the same time. If this is so, then the outcome may be more dependent on the syndrome and less on the signing therapy.

One of the great incentives to learning speech is the reinforcement of provided by the necessity of verbal output. Young toddlers always gesture. We encourage this. While gesturing is a simplistic form of signing, there are limits to how much can be understood. This drives the child to increase the variety of expressive language by speaking. Signing takes away this necessity which is the primary reinforcer of verbal output. Our observation of children who have been in total communication programs is that they initially rely exclusively on signing, and it is not until much later that they speak. We prefer for them to speak earlier.

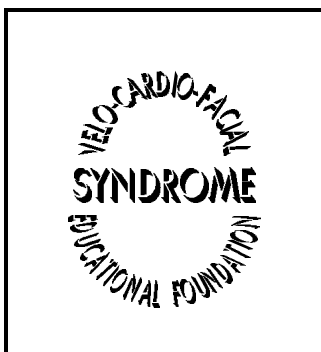
In summary, to those of us who understand the communication disorder associated with VCFS, we have

no hesitation to teach these children to speak while avoiding the use of sign language. The outcome is directly related to the speech pathologist's ability, not the child's. The frequency and intensity of therapy are also important. If a child with VCFS could not learn speech, then we would be the first to agree with our colleagues that signing should be taught. It is not that we *believe* that children with VCFS can be taught intelligible speech...we *know* they can because we have done it and seen it literally hundreds of times. To withhold a normal means of communication from a child is inappropriate. To foist alternative communication on the child because of our own failure as clinicians is admitting defeat. ❖

You need to know:

In the rear of this newsletter, we have included the following forms:

- Order form for the tape sets from the 4th annual conference. If you'd like to order tapes, please do so directly from the vendor, using the form and/or number(s) provided.
- Membership application / renewal / dues form. If you'd like to renew your membership, please fill out the form and mail it us at the address indicated. Feel free to copy the form as needed. *Note: Please complete both sides of the form; it contains an authorization to release information, which will determine whether or not we may release your name, etc. to other VCFS individuals.*
- Announcement for 5th annual meeting in Milwaukee, WI, as well as a *Call for Abstracts*. *Please observe the instructions on the forms.*



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DUES NOTICE

As approved unanimously by the full membership of The Foundation, voluntary dues have been assessed according to the schedule of suggested minimums listed below:

Single Membership	\$25.00
Family Membership which includes immediate family (spouses, children)	\$30.00
Student Membership	\$10.00

We also urge pledges of \$5 for the Caitlin Lynch Memorial Fund, to provide scholarships for our annual meeting, and \$5 to the Tony Lipson Memorial Fund, to allow a member from Australia to attend our annual meeting. Please make your checks payable to The Velo-Cardio-Facial Syndrome Educational Foundation. Your payment is tax deductible, as are any additional donations you might wish to make. In cases of hardship, The Foundation will grant membership without the collection of dues. In such cases, The Foundation will waive dues, if the appropriate box is checked below and returned to us. Dues, or requests for waivers, must be received no later than January 31, 1999 in order to maintain membership, remain on the Foundation's mailing list, and to assure delivery of the next periodic newsletter. Your canceled check will be your receipt unless otherwise requested. Thank you for helping to fulfill the mission of The Foundation.

Robert J. Shprintzen, Ph.D.
Executive Director

Donna L. Landsman
President

Lisa Jennings
Secretary-Treasurer

My membership category is: Individual Family Student Professional

For family dues, please list additional family members: _____

Enclosed is my check or money order for \$ _____ for my 1999 dues, plus:

\$5 for the Caitlin Lynch Memorial Fund (optional)

\$5 for the Tony Lipson Memorial Fund (optional)

Total enclosed: \$ _____

Waiver:

I request a waiver of my dues. Please retain me as a member of the Foundation.

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The Velo-Cardio-Facial Syndrome Educational Foundation

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first middle initial last

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Date of Birth: _____ / _____ / _____ Work Hours: _____
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What is your interest in velo-cardio-facial syndrome? I have velo-cardio-facial syndrome

I am the parent of someone with velo-cardio-facial syndrome (if so, please provide name, date of birth, and sex of your child) _____

I am a student or resident interested in learning more about velo-cardio-facial syndrome

Other (please explain):

I am a professional/health care provider working with individuals with velo-cardio-facial syndrome

If you are a professional or health care provider interested in joining, what is your professional field? _____ Degree(s): _____ Are you certified or licensed? yes ___ no ___

How did you find out about the Foundation? _____

Send this completed form to: **Dr. Robert J. Shprintzen**
Communication Disorder Unit, Jacobsen 707
Center for the Diagnosis, Treatment, and Study of Velo-Cardio-Facial Syndrome
SUNY Health Science Center at Syracuse
750 East Adams Street
Syracuse, NY 13210

Or Fax to: 315-464-5321 or email data to vcfsef@hscsyr.edu
Telephone: 315-464-6590

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New Membership

Renewal Membership

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FOURTH ANNUAL MEETING

The Velo-Cardio-Facial Syndrome Educational Foundation
CHILDREN'S HOSPITAL, HARVARD MEDICAL SCHOOL, BOSTON, MA

June 26-28, 1998

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**** MEETING NUMBER VC9801 ****

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Minutes from the 1998 Business Meeting (4th Annual Meeting)

The meeting was called to order at 5:00 P.M. by the President, Maureen Anderson. The President discussed the arrangements for the 1998 meeting and thanked all of those who had contributed to its success. She acknowledged the financial and logistic support of Children's Hospital and the Harvard Medical School. Grants were also received from the Boston Children's Heart Foundation, Inc., The Sullivan Associates (facilitated by Nick Tawa), and the Mahoney Family in memory of David Mahoney.

The Treasurer's Report was delivered by the Executive Director because the current Treasurer, Edna Keleshian, could not be present. Dr. Shprintzen indicated that the treasury was solvent with a current balance of approximately \$3,000 not including the receipts from the 1997 Annual Meeting or the expenses to be paid for the meeting. Dr. Shprintzen expressed some disappointment at the percentage of members who had chosen to pay voluntary dues which was well under 50% of the membership. However, even at this level of support, he anticipated that the Foundation would remain solvent into the next year. He indicated that current membership was in excess of 1,200, approximately 60% lay members and 40% professional.

Under Old Business, the issue of forming a separate 501(c)3 corporation was discussed. A vote indicated that this should be done over the next year. Jeff Landsman, a member of the Board of Directors and an attorney, indicated he would explore the matter. At present, the Foundation maintains tax exempt status through the SUNY Health Science Center.

Under New Business, sites for future meetings were discussed. The Foundation had committed to Milwaukee in 1999 and Baltimore in 2000. Two future meeting sites were suggested: Miami, by Barbara Dandron and Northampton, England by Ahmad Al-Khattat. There was discussion of these two sites, including discussion of the advisability of having a meeting in Europe. A vote was taken and Miami was chosen for 2001 and England for 2002.

The last order of New Business was elections. The Board of Directors put forward a slate of nominees as suggested by the nominating committee. Donna Landsman (Madison, Wisconsin) was nominated as President; Lisa Jennings (Weymouth, Massachusetts) was nominated as Secretary-Treasurer; Ahmad Al-Khattat was nominated to fill the vacancy for the Professional Member of the Board of Directors and Barbara Dandron was nominated to fill the Lay Member slot on the Board. Linda Walters was nominated for the Chair of the Liaison Committee. The Committee on Committees was elected. Dr. Shprintzen thanked Maureen Anderson for all of her hard work over the past year. The meeting was then handed over to Donna Landsman. There was no other New Business and the meeting was adjourned.



Recent VCFS-related Publications

- Papoulos, DF, et al. Ultra-ultra rapid cycling bipolar disorder is associated with the low activity catecholamine-O-methyltransferase allele. *Mol. Psychiatry*. 1998 July; 3(4): 346-349.
- Kirov G, et al. Low activity allele of catechol-O-methyltransferase gene associated with rapid cycling bipolar disorder. *Mol Psychiatry*. 1998 July;3(4): 342-245.
- Devriendt, K, et al. Deletion in chromosome region 22q11 in a child with CHARGE association. *Clinical Genetics*. 1998 May; 53(5): 408-410.
- Chow, LY, et al. Velo-cardio-facial syndrome, schizophrenia and deletion at Chromosome 22q11. *J. Intellect. Disabil. Research* 1998 April; 42(Pt2): 184-188.
- Smith CA, et al. Increased prevalence of immunoglobulin A deficiency in patients with the chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Clin Diagn Lab Immunol*. 1998 May.; 5(3): 415-417.
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SAVE THE DATE:
July 16 through July 18, 1999
Children's Hospital of Wisconsin

The Velo-Cardio-Facial Syndrome Educational Foundation
Fifth Annual Meeting

A Meeting for Parents and Professionals Interested in Velo-Cardio-Facial Syndrome/The 22q11.2 Deletion

To be Held At:
Children's Hospital of Wisconsin
9000 West Wisconsin Avenue
Milwaukee, Wisconsin

Additional Details and a Call for Abstracts are in this issue of the Newsletter

General Information: Contact the Local Arrangements Chair:

Susan Marks

Phone: (414) 266-2922; Fax: (414) 266-6189;

E-Mail: smarks@chw.org

For Specifics Regarding the **Scientific Program:** Please Contact:

Robert J. Shprintzen, PhD

Phone: (315)-464-6596

Fax: (315)-464-5321

E-Mail:

shprintr@mailbox.hscsyr.edu

For a registration form, complete the information below and return to:

Velo-Cardio-Facial Syndrome Educational Foundation
c/o Dr. Robert J. Shprintzen
C.D.U., 707 Jacobsen Hall
SUNY Health Science Center at Syracuse
750 East Adams Street, Syracuse, NY 13210

NAME: _____

ADDRESS: _____

CITY: _____ STATE: _____

ZIP CODE: _____ PHONE: _____

FAX: _____ E-MAIL: _____

- FUTURE MEETINGS:** **2000 - Baltimore**
 2001 - Miami
 2002 - Northampton, England

Big plans are underway for these meetings, from Baltimore's Inner Harbor to Miami's beaches, to our first international venue in England. Dates will be announced shortly. Be a part of the process by telling us what features you would like to see at these meetings. ❖

FORM FOR ALL SUBMISSIONS

**Fourth Annual Meeting, July 16-18, 1999, Children's Hospital of Wisconsin, Milwaukee
The Velo-Cardio-Facial Syndrome Educational Foundation**

TITLE:

SUBMITTED BY (underline person presenting):

TYPE OF PRESENTATION (check appropriate category):

Professional paper Professional Seminar Clinical case presentation

Lay Presentation: Sharing the Good Times Lay Presentation: Helpful Hints

Other (describe) _____

ADDRESS:

TELEPHONE: FAX: E-mail:

TIME REQUESTED FOR PRESENTATION:

AUDIOVISUAL EQUIPMENT REQUIRED:

**Type or print the abstract (without title or authors) in the space provided below. Submit 3 copies to be received by May 15, 1998. Mail to Dr. Robert J. Shprintzen at:
Center for the Diagnosis, Treatment, and Study of Velo-Cardio-Facial Syndrome, Jacobsen Hall 707, State University of New York Health Science Center, 750 East Adams St, Syracuse, NY 13210 USA
Abstracts may also be faxed (single copy) 315-464-5321 or emailed to shprintr@mailbox.hscsyr.edu**