

KEEPING  
IN  
TOUCH



APRIL  
2005

**Cornelia de Lange Syndrome Association (Australasia) Inc.**

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## From the Editor

Welcome to the first issue of KIT for 2005. As was mentioned last time, I am including one or two Family Directory entries in each issue of KIT. Some of the ones I have are maybe a bit old now (my fault, not yours). So if you sent in an entry a while ago, please consider an update – just a few lines – of any new activities, height, weight, photo, etc, so I can add this to your original entry. Thanks very much. Any other news from families is also very welcome.

**Welcome to New Members** from the Philippines & Queensland.

## ASSOCIATION NEWS:

### Genetic Research.

Dr. Ian Krantz and the CdLS research team from The Philadelphia Children's Hospital in the USA are requesting contact information from families who gave blood samples at the Sydney conferences in 1999 and 2003 - just an e-mail address or post address will do. ***Parents who have a child with CdLS and would like to have another child but have been reluctant to take the plunge are also invited to contact Dr. Krantz.***

Please contact Marie Jackson by email [jacksonmari@email.chop.edu](mailto:jacksonmari@email.chop.edu) or by phone to Dinah Yaeger, genetic counsellor, at the hospital. From Australia the direct number is 0011 1 609 3973286. Philadelphia is 15 hours behind Sydney time.

## **NEW ZEALAND NEWS:**

Hi from New Zealand.

What was going to be a quiet year for CdLS has turned into a busy one. We are looking forward to a gathering in July with Peter and Phyl who will call in on us in NZ on their way back from the CdLS Conference in Italy. They will be in NZ for about a week seeing as much as possible.

We have managed to arrange to have a geneticist, Komudi Siriwardena, come and talk to us at the same time that Peter and Phyl are here.

Therefore on 2 July 2005 we will have a lunch at Wilson Home in Auckland, followed by a talk from Peter and/or Phyl with the latest news from the Italian Conference. Following their talk, Komudi will explain the latest developments of the genetic discoveries of CdLS.

We will send a programme out to all the NZ CdLS families shortly. All families and friends welcome.

Peter and Phyl will be travelling to Wellington, so if anyone would like to catch up with them there I am sure we can arrange something.

Liz Molloy

## **QUEENSLAND NEWS:**

### **QUEENSLAND PICNIC *TAKES TO THE ROAD***

Due to popular demand from families living outside Brisbane, this year's picnic will be held in Bundaberg. Details are:

Date: Sunday, 1 May 2005  
Place: Alexandra Park, Quay Street, Bundaberg  
Time: 11 am onwards  
Contact: Catz @ [kleche@bigpond.net.au](mailto:kleche@bigpond.net.au) or  
Rose @ [dave.humphrey@optusnet.com.au](mailto:dave.humphrey@optusnet.com.au)

The riverfront Alexandra Park has recently undergone a major refurbishment and now includes a free zoo. The park is 1 min walk from the train station and approx a 4 hour drive from Brisbane. Monday, 2 May is Labour Day in Queensland so if you want to enjoy all that Bundaberg has to offer, why not make a weekend of it? For information on motels contact Catz or check out <http://www.bundabergcity.qld.gov.au/>

## NSW NEWS:

### CdLS Clinic Day

The second CdLS Clinic Day was held in March at the Ryde Developmental Disability Clinic. Dr Jane gave two patients thorough medical examinations, and arranged follow-up appointments with other specialists.

The third clinic day is set for Wednesday 21<sup>st</sup> September 2005. Two appointments are available. If you would like your child/client to use one of these appointments please contact Jenny. Contact details on front page of KIT. This offer is for NSW residents aged 16 years and over.



Nicky, Clinic Nurse Co-ordinator for CDDS



Brett and Stephanie with Jessica



Dr Jane with Frederick and Anne and Wilfred, who is recovering well after knee surgery.

## **Collis Curve Toothbrushes**

We still have some collis curve toothbrushes available in both adult and youth sizes. These are free to Australian resident financial members of the Association. Non-financial members a charge of \$5 to help cover postage and packaging applies. For our members outside Australia, the postage and packaging costs are \$7. Please contact Jenny, noting the size brush required, and if there is a colour preference. Youth size is suitable for baby teeth.

## **OZNZ Online Group**

I've just gone through the email addresses on OZ-NZ and found quite a number have a "bouncing" status which means that the address is invalid. Could you please go in and update your details.

Log in to <http://groups.yahoo.com/group/oznz-cdls/>

Click on Edit Membership

Delete all invalid addresses

Thanks very much

Your Moderator - Rose

## **International Conference**

The countdown is nearing its end, with only 2 months to go before the opening of the International CdLS Conference in Italy.

International CdLS conferences are an opportunity for the National Leaders of CdLS support organisations around the world to meet, offering a forum to discuss and meet the needs of our member families. Australia will be represented in Federation Council by Peter and Jenny. Peter is the past President of the Federation, and present executive member. The Scientific Advisory Council consists of medical and other experts representing each member country. Australia's representative, Dr Meredith (Genetics, Children's Hospital, Westmead) will be attending the SAC meetings as well as the professional and family conferences.

In total we have 20 delegates from Australia expected to attend, and while the early-bird deadline for registrations has now passed, it's not too late to register.

The Italian Association has planned an exciting agenda with speakers from many countries, informative and useful workshops, as well as the usual doctor appointments, girl scout childcare and social opportunities. Subjects concerning the needs of babies to adults are covered in this program.

For further information and registration details go to the Italian support group's web site [www.corneliadelange.org](http://www.corneliadelange.org) or contact Jenny.

# COMMUNICATION CHARACTERISTICS IN THE CORNELIA de LANGE SYNDROME WITH PROGNOSIS AND RECOMMENDATIONS FOR TREATMENT

CdLS International Conference: Penrith, Australia

July 3-6, 2003

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## Background

- I began providing therapy and assessment for individuals with Cornelia de Lange Syndrome (CdLS) in 1982
  - First to document successful speech therapy with a child with CdLS, delineate prognostic factors, and indicate the presence of oral-motor apraxia in almost all individuals with CdLS
  - I am indebted to the children with CdLS and to their families for making my research possible. Also want to thank Jenny Rollo and many others for helping to make these conferences possible.

## Early Observations and Goals

- Discovered at my first CdLS conference in the U.S.A. (1984) that
  - Almost none of the children had communication goals on the Individualized Education Plans (IEPs) in their schools
  - Few were receiving speech therapy
  - Almost no specific information was available about aspects of communication in this syndrome
  - Unusual results of hearing tests
  - Uncertainty about why some children spoke and others did not
- Initial Goals and Results
  - IEPs that included communication goals
    - Now, these goals exist for almost all of the children in the U.S.
    - Problems accessing services in some parts of Canada, Ireland, Columbia and other countries
  - Determine why some children with CdLS spoke while others did not
    - Now, there is prognostic information for varying levels of communication
  - Determine what treatment approaches are available
    - Now there is information available, which I have included in this paper

## General Development of Communication

- The answers to many of the above questions were published in 1993,<sup>2</sup> based on a database of 116 individuals with the syndrome
- That information will be presented today, along with additional information I have learned about communication in the syndrome

## Early Sounds, such as Cooing and Babbling

- Cooing and babbling, when they do occur, appear to be very limited
- Parents typically report the periods of babbling are unusually brief, perhaps for only a day
- When babbling is present, the repertoire of consonants is typically reduced, for example, perhaps only “m,d” rather than the “m,b,p,d,t,n,g,k” expected in the normally developing child
- Re-duplicated babbling (ba-ba-ba) and variegated babbling (ah-ba-ga-mo) are not usually present
- Jargoning,
  - Described as the production of the *melody of sentences without words*
  - Occurs between about 14-18 months in the normally developing child
  - Some children with CdLS do seem to use jargoning after they have acquired about 5 words

## Oral-Motor Structure

- Retro-micrognathia (the lower jaw is retruded and small)
- Retrusion of the lower jaw may result in more difficulty making the necessary tongue movements for speech
- Also can create misalignment of the teeth, which may affect pronunciation

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<sup>2</sup> Goodban, M.T. (1993) Survey of speech and language skills with prognostic indicators in 116 patients with CdLS. *Am J Med Genet* 47:1059-2063.

## Oral-Motor Abilities

- Oral-Motor apraxia is evidenced by the majority of the individuals with CdLS<sup>2</sup>
  - Defined as difficulty imitating or producing voluntary movements of the mouth, in the absence of paralysis or weakness
  - Child can execute vegetative movements such as eating or retrieving food like peanut butter from the upper lip
  - Great difficulty with voluntary imitation of tongue, lip or jaw movements
  - May see groping movements of the mouth and a puzzled look

## Development of Speech Sounds

- First oral attempts at communication usually consist of a single schwa vowel: /ɜ/ as in “sofa”
- Next productions are typically consonant-vowel combinations: “ma, ba, pa”
- Errors in articulation (pronunciation) are common
- Some of the misarticulations persist into adulthood

## Typical Speech Sound Errors

- Many consonants are omitted, distorted, or other sounds are substituted for them, for example, “\_ao/ball”
- *Front* sounds (speech sounds made in the front part of the mouth, such as “t”) are often substituted for *back* sounds (sounds made in the back part of the mouth, such as “k”), for example, “tat/cat”
- Final consonants, or consonants at the ends of words, are frequently dropped, for example, “do\_/dog≡ or “\_o\_/dog”

## Overall Speech Intelligibility

- Combination of missing and distorted consonants, with the soft, muffled voice characteristic of the syndrome, results in low overall intelligibility
- Higher functioning child with CLS usually 75% intelligible, in context, by age 6-7 years
- In contrast, normal child is typically 90-100% intelligible, in context, by age 3-4 years

## Factors Related to Articulation Errors

- Oral motor apraxia
- Overall developmental delay
- Hearing loss, if present
- Retro-micrognathia (the lower jaw is retruded and small)
  - Retrusion of the lower jaw may make the tongue movements necessary for pronunciation more difficult
  - Can create misalignment of the teeth, which may affect pronunciation

## Unusual Vocal Quality (and Vocalics: volume or loudness, pitch, intonation, phrasing)

- Most of the children exhibit an unusual vocal quality that is guttural and low in pitch; occasional dips into glottal fry
- Overall range of vocal inflection tends to be limited
- Soft voice, difficulty speaking loudly
- Although some children have a vocal quality that is more guttural and hoarse than other children, there does not seem to be a relationship between vocal quality and the development of expressive language abilities
- GERD (gastroesophageal reflux disease) and hearing loss may contribute to some of the vocal quality differences

## Resonance (the vibration of sound)

- Voice may sound muffled and not very loud as a result of the retro-micrognathia
- The smaller oral cavity results in a lessening of oral resonance

## Hypernasality

- Excessive resonance or sound vibration in the nasal cavity, usually associated with cleft palate or submucous cleft
- Glottal stops, pharyngeal fricatives, facial grimacing
- Voice will sound even more muffled
- Nasal emission of air may be heard, especially on consonants like “s, d, p”

## Normal and Delayed Expressive Vocabulary Development

- Normally developing children have about 300 words at age 2 years and 4000 words by age 4 years
- A small percentage (3 – 4%) of the children with CdLS in my 1993 study had normal to low-normal speech and language abilities
- Approximately 35-40% of children with CdLS acquire their first words between 12 – 48 months
- Approximately 20-25% do not acquire their first word until 7 – 10 years of age
- My 1993 database indicated 25-30% of the children with CdLS acquire no spoken words at all – or just one “word”

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<sup>2</sup>Nine doctors present research update at Arizona convention, U.S.A. (1994) *Reaching Out: The Newsletter of the CdLS Foundation*. XIII: 4, p. 11.

### **Words Are Not Language**

- The ability to use words does not guarantee the child with CdLS will be able to use normal grammatical structures
- Most children with CdLS who acquire a minimum of 30-50 words will be able to use simple, short sentences
- Many who use 3-5 word sentences on a regular basis usually do not develop full grammatical abilities, such as the ability to transform words into the standard question form, but nevertheless communicate their ideas well

### **Asynchronous Development of Language Development**

- Many aspects of the language development in this syndrome are marked by discrepancies between skills
- Expressive (spoken) language abilities are far below cognitive (IQ or mental age) abilities and receptive (understanding what is said) language abilities
- Syntactic skills
  - Syntax or grammar, ability to combine words into sentences, transform words into the standard question form
  - Below that expected given total number of vocabulary words and utterance length
- Recent research on children with specific genetic syndromes reveals performance not accounted for by their general cognitive abilities. For example, similar to my 1993 findings in CdLS
  - In the Down syndrome population, children evidence deficits in language production in comparison to language comprehension and nonverbal cognitive skills, as well as better development in vocabulary skills than in syntactic skills. A similar comparison is seen in the fragile X syndrome
- In contrast, children with Turner syndrome appear to have better language communication skills than would be suggested by their general cognitive skills

### **Unusual Findings**

- Unexpected Utterances
  - Clinical observations and reports from parents indicate that some children with CdLS clearly utter a word or phrase once or twice and then never use it again
  - This utterance is often many levels above the current level of performance, e.g., a child who is still struggling with simple, single-syllable words may unexpectedly produce a multisyllabic utterance with perfect articulation
  - This behavior is also observed in adults with apraxia and in children with autism

### **Receptive Language Skills Much Higher than Expressive**

- Child's ability to understand is much higher than ability to speak
- Nonverbal cognitive abilities (what the child is able to understand by watching) appear to be higher than expressive language abilities, but more study needed

### **Hearing**

- Many children have conductive hearing losses (reduction in hearing sensitivity, often caused by fluid in the middle ear cavity), often correctable with time, antibiotics and other treatment
- Hearing aids may be recommended, although parents often report their children seem to hear as well without the aids
- A number of children have sensorineural hearing losses (nerve damage), which create more problems for language development; hearing aids are often recommended

### **Unusual Results of Hearing Tests**

- Numerous reports from parents indicate that children are often diagnosed as deaf or profoundly hearing impaired immediately after birth, moderately hearing impaired at about 12 months, and mildly hearing impaired or with normal hearing at 18 - 24 months
- These are highly unusual findings because hearing typically gets worse instead of better. We do not know why they happen.

### **Auditory Neuropathy?**

- Is the condition of auditory neuropathy/auditory dys-synchrony (AN/AD) the cause of the unusual hearing tests?
- If so, relevant to note that normal populations report that although they can hear speech, they can not always understand it, especially in the presence of background noise
- More information is needed about why the unusual hearing tests occur

### **Parents Need to be Informed about these Unusual Hearing Test Results**

### **Facial Expression/Gesture/Nonverbal Behavior**

- Faces tend to lack the range of expression typical of normally developing children
- Gestures and nonverbal behaviors appear to be limited and are probably commensurate with verbal expressive language
- More information is needed in these areas

### **Pragmatics** (appropriate use of language in social settings; includes turn taking, interactions in conversation)

- Most of the children talk very little, even when they have highly developed vocabularies.

- Some children exhibit difficulty in their relationships with peers
- The social behavior of some of the children with CdLS is similar to that of autism
- A small number appear to exhibit selective mutism

#### **Deficits in Social Relatedness**

- Autistic-like behaviors in approximately 25% of my data base
- Impairment in social interaction: eye gaze, facial expression, body posture, gestures, lack of seeking to share attention and interests with others, failure to develop peer relationships, withdrawal from people
- Impairment in verbal and nonverbal behavior
- Insistence on sameness
- Child may benefit from a therapy program for children with autism

#### **Prognostic Factors Associated with NonTalkers in CdLS**

- Children who do not talk at all tend to have at least one of the following characteristics:
  - Moderate-to-severe hearing impairment
  - Upper-limb malformations
  - Severe motor delay: sitting up later than 25 months or walking later than 30 months
  - Deficits in social relatedness: autistic-like behaviors, eye contact, overall ability to relate to people
  - Birth weight under 5 lbs. (probably the least important factor)

#### **Oral Communication Behavior**

- 53% of children 4 years or older combine 2 or more words into sentences
- 33% of children use no words, or only 1-2 questionable words
- 3-4% exhibit normal or low-normal speech and language development

#### **Groupings Based on Observed Characteristics. *Please note: There will be exceptions in the following categories.***

- **Group I: ATalkers**
  - Approximately 3 - 4% of the population in my database
  - Developmental milestones such as sitting and walking are close to normal; may walk as late as 18 months; abilities in all areas are at low-normal to normal levels; no upper limb malformations; normal ability to relate socially; minor hearing problems, if any
  - Communication development is close to normal or maybe a little late; these children usually begin talking on their own but will probably benefit from speech therapy and other interventions
  - Educational placement may be in normal classroom with resource help for speech, reading, math, etc.
- **Group II: ALate Talkers**
  - Approximately 35-40% of the children I have seen
  - The children in this group typically sit at or by 18 months and walk at or by 30 months; no upper limb malformations; normal hearing, except for a possible mild conductive loss in one or both ears or a moderate conductive loss in one ear; moderate-to-good skills in social relatedness; birth weight at or greater than 5 pounds (although birth weight does not seem as important as the other factors)
  - The children in this group will communicate verbally but these skills will develop later than for the Group I children and may not develop to the same extent; children in this group usually exhibit a relatively good ability to imitate words they hear, although pronunciation may not be precise
  - First Areal words" such as mama/dada may emerge as early as 12-18 months but often not until 24-30 months, and some not until 4-8 years of age, although these later developing children may have 6 – 15 signs; additional words will slowly emerge; earlier talkers will have two-word utterances by 36 - 40 months and a number of short sentences by 6 years; sentence length may never exceed 5-7 words and will probably average 3-5 words per utterance; may be persistent problems with the formation of the standard question form; consistent omissions of substitutions of some consonants may persist; tendency to be very shy except around family members
  - Until age 5-7, attention to activities is typically very brief
- **Group III: ALimited Talkers**
  - Approximately 20-25% of the children I have seen
  - Developmental milestones, such as sitting and walking, are achieved at a later age than described for Group II children; typically no upper limb malformations; skills in social relatedness may not be as good as for the Group II kiddies
  - The children in this group will communicate verbally but these skills usually do not develop as early or to the same extent as found in the Group II children
  - First real words may not emerge until 8-10 years of age; additional words may develop as late as 11-12 years of



age

- **Group IV: ANonTalkers**
  - Approximately 25-30% of the children I have seen
  - One or more of the following is almost always present: Upper limb malformations; moderate to severe hearing loss; autism or autistic-like behaviors; very late attainment of developmental milestones, e.g., may not walk until after four-five years of age
  - The children in this group may not develop verbal communication skills, or may never have more than 1-3 words

## TREATMENT

### Typical Therapy Procedures for Groups I – IV

- **Group I**
  - Speech-language therapy may be necessary for some speech sounds, standard grammatical forms, increase in loudness, and optimal social interactions, such as social greetings, offering snacks, etc.
- **Groups II and III**
  - Assumes the necessary prognostic factors for success with verbal language: physical ability to produce speech sounds; sufficient level of cognition; adequate vision; sufficient attention span; ability to imitate sounds and words; adequate hearing; prepubescent; lack of autistic-like behaviors; and absence of upper-limb malformations.
  - Therapy moves from gross motor imitation, to vocal imitation, to sound and word imitation.
  - The earlier sessions are marked by
    - Playful activities involving oral-motor play and use of noise-making objects
    - Maximum visual and auditory stimulation
    - Activities designed to approximate those of normal daily routines, such as pretend grocery shopping, meal time, playing with doll houses, using a vacuum cleaner
    - Also see my *Prespeech and Speech Intervention Procedures sheet*
  - A very successful stimulation approach for eliciting vocal imitation consists of saying "ba" or some other sound, word or phrase into a plastic ring or similar object and encouraging the child to imitate this utterance, while holding a ring near his/her mouth. My videotape provides many examples of these and other therapy activities.
  - For the most part, therapy should follow the lead of the child. Be prepared with many interesting toys/items so that the child maintains interest and is frequently surprised. These items also serve as motivators
  - Try to find something the child will work hard to obtain. Food, toys, or fascinating items often work.
  - Tactile (touch) stimulation is often avoided because of the disturbing effect this has on many children with CdLS.
  - The use of sign language or gestures frequently facilitates the development of oral language. We all gestured before we talked, and many of us still rely on gestures to communicate or facilitate our recall of words.
  - Use the procedures of
    - **Self-talk** (describing to the child what you are doing)
    - **Parallel talk** (describing to the child what the child is doing)
    - **Expectant waiting** (using an expectant manner while waiting for the child to respond)
  - For the child who has developed some expressive vocabulary, drill and repetition are useful in maintaining and developing longer utterances. The "Wh-" question form often requires much practice.

### Therapy for Oral Motor Apraxia

- Although this therapy approach seems easy to learn, I know it is not because of the difficulty I have encountered teaching it to student clinicians.
- Features of therapy for oral-motor apraxia
  - highly inflected simple, single word or utterance (eventually may be extended to longer utterances)
  - syllables must be prolonged a minimum of 2 seconds per syllable, e.g., b-a-a-a-b-y-y-y
  - presentation should be a little louder than normal
  - similar to motherese
  - numerous repetitions of the same word or phrase, accompanied by holding the stimulus item or a plastic ring near the mouth, or placing ones fingers near the parent=s/therapist=s mouth
  - expectant waiting: look at the child as though you expect a response (usually includes raised eyebrows)
- The features of prolongation, slightly louder volume, and exaggerated inflection are more successful at eliciting a response
  - They intensify auditory and visual stimulation
  - Allow for greater auditory processing time
  - Are closer in form to the motherese appropriate for the child's cognitive age

- Are more “musical”
- Use gesture, movement, tapping with a block, or clapping to mark the rhythm of each syllable or word is important because gesture facilitates oral communication
- Reinforcement of successive approximations by the child, i.e., praise all attempts, particularly the ones that come closer to the desired behavior
- **Groups III and IV**
  - **Communication Board:** If your child is completely nonverbal, this procedure is useful. At the same time, you will be teaching your child the basic nature of communication.
    - Provide two-to-four actual objects, imitation objects, or large, very clear pictures or photographs of objects, per context. By “context” I mean, mealtimes, playtime, selection of toys, etc.
    - For example, at meal or snack time, you may have a communication board with four different pictures of food, or these pictures may be on the refrigerator door. When your child indicates a desire to eat, pair the touching of the picture (first by the parent as an example; later by the child) with the presentation of that particular food or drink. This provides a method of communication for the nonverbal child who has no other method of communication.
    - There are a number of variations on the above approach. One is to have laminated pictures of objects on a large, circular key ring. This method is very portable and can be used by individuals other than the parents.
  - **Picture Exchange Communication System (PECS):** This system is often used in schools for children in Group IV. PECS is usually established by a speech-language pathologist and used by the therapist as well as by the teacher and/or an aide. The basic idea of PECS is that the child learns to exchange a picture, and gradually a more sophisticated form of communication, for the desired outcome.
  - **Object Communication:** Similar to PECS. Dr. Mary Morse does an excellent presentation on the use of Object Communication in a school setting.
  - **Oral-Facial Stimulation:** Increases tolerance of stimulation of the structures in and around the mouth
  - **Therapy for Children with Autism Spectrum Disorders.** Some children exhibit behaviors that are similar to those seen in children who are diagnosed with autism. According to reports from parents, such children have qualified for treatment programs for children with autism.
  - **Therapy for Children who are Deaf/Blind.** A few parents have reported success with their children in this type of setting.
  - **Therapy for Oral-Motor Apraxia** may be appropriate for many children in Group III.
  - **Signing while Singing:** One report was made of a group of children being taught signing while singing. Similar to approaches used in nursery schools.

### Other Treatment Procedures

- Selective Mutism
  - Defined as a disorder of childhood that is characterized by the total lack of speech in at least one situation, despite the ability to speak in other settings
  - Duration is at least one month
  - This persistent lack of speech in some places is not accounted for by a communication disorder
  - Assessment of the child will probably be conducted by a multidisciplinary team consisting of the pediatrician, a psychologist or psychiatrist, the teacher(s), and a speech-language pathologist who will interview the family
  - Underlying anxiety often involved
  - A behavior therapy program is developed to increase verbalizations, usually coordinated by a psychologist or a speech-language pathologist
- Important Note
  - Many children with disabilities are likely to become passive and give up on all communication attempts because the few attempts they do make are not noticed or are misunderstood
  - These impairments disrupt the familiar communication routines found in most interactions between young children and their parents. Children with CdLS are less likely to initiate and to imitate behaviors
  - It is critical that children learn that their behaviors are meaningful to at least one other person
  - Because we do not want to lose time until traditional procedures are established, I am recommending that caregivers react to the behaviors of their children as though they are meaningful communication.
    - Pay attention to posture and body shifts or vocal behaviors.
    - Example: With a very young child, watch what behaviors occur when the child wants to be picked up. If his/her shoulders are raised, touch the shoulders and say, “I see you are raising your shoulders. Do you want

up?” Then pick up the child. Communication has occurred and it was successful.

- Also see Buddy and Sammy under the Case Studies section of this handout

### General Intervention Recommendations

- All children should receive a communication assessment as early as possible. Preverbal and verbal assessment can be obtained from interviews of caregivers, formal test administration, informal observations, and medical and educational reports.
- Communication intervention should be initiated as early as possible. Above all, the parents should talk to their child as though they expect a response and continue to expect a verbal response for as long as appropriate.
- Hearing ability is a critical factor in the development of speech and language.
  - Early and frequent tests are necessary, particularly with the child who has a suspected hearing loss.
  - Babies with CdLS have very tiny structures and testing may be difficult. It is advisable to consult an audiologist and/or otolaryngologist who is familiar with CdLS or who is experienced in working with infants.
  - Tubes may be useful for middle-ear drainage but again it is necessary to consult with a physician experienced in working with small infants.
- If a hearing loss is suspected, headsets and hearing aids may be prescribed for infants and children. Even a mild hearing loss can result in a speech and language delay.
  - Smaller aids are available so it is not necessary or advisable to use an adult-sized aid.
  - If the child will not leave on the aid, an audiologist or behavioral therapist may be helpful.
  - Appropriate audiological management should include selection and fitting of suitable amplification for all listening environments.
  - While the child=s personal hearing aid may be sufficient some of the time, the use of FM amplification may be necessary in the school setting.
- Cleft lip and palate should be closed as early as possible. This improves the ability to eat, enhances the normal speaking process, and reduces the likelihood of ear infections leading to hearing loss. Evaluation and treatment for cleft lip and/or palate, insufficient velopharyngeal closure, and submucous cleft requires a team approach, usually comprised of a speech-language pathologist, surgeon and dentist.
- For children with esophageal reflux, the parents are advised to seek early treatment or surgery to help reduce pain and discomfort, thus improving behavior. This treatment may also avoid irritation of the oral-pharyngeal-laryngeal areas as well as the eustachian tube and help promote better vocal quality and hearing.
- Gestures and sign language are encouraged as methods to facilitate and motivate oral communication.
- Children with CdLS from bilingual families seem to do equally well in both languages (or not do equally well in both languages as the case may be). I do not recommend that intervention be limited to only one language.
- If the child has a gastric tube, feeding therapy may be indicated so the oral mechanism functions as normally as possible. However, it is not necessary for the child to have eaten normally in order for talking to occur.
- Work with the child and other therapists as necessary to reduce deficits in social relatedness. For the older child who has autistic-like behaviors and constantly plays with or holds the same toy, strive to have that object be an age appropriate object
- Also see my Prespeech and Speech Intervention Procedures.

### CASE STUDIES

**Buddy** The following case study illustrates a child who benefited from non-vocal strategies.

The description of Buddy by Buzolich (1987)<sup>1</sup> demonstrates the successful use of blissymbols with a 12-year-old ambulatory nonverbal boy with Cornelia de Lange Syndrome. Buddy received his first non-oral communication evaluation at the age of 12 years upon admittance to a private school for severely behaviorally and educationally handicapped children. At the same time he was placed in a group home due to the death of his foster parent. He was described as having no symbolic communication system on admittance to this private school and it was determined he had a severe oral-motor, verbal, and motor apraxia. In addition he had an attention deficit disorder, and he exhibited both impulsive and compulsive behaviors.

Buddy's eye gaze, facial expressions, gestures, and vocalizations were his primary means of communication. He reportedly had mastered 60 signs although they were idiosyncratic versions of standard signs. Expressive verbal language was below the one year level and receptive language functioning was at approximately the four year level. His strength was in his visual modality; he could readily recognize letters of the alphabet and some common words. After 8 months of training he

was easily trained to express himself with 100 Blissymbols and was able to combine these symbols into more complex expressions. After outgrowing his communication book of symbols, Buddy was trained to use the Wolf Voice-Output Communication Device (cited in Buzolich, 1987) and achieved an improved level of functional communication. Through an appropriate assessment, educational and therapeutic program, Buddy was able to learn and compensate for his handicapping condition.

**Sammy** The following case study illustrates the use of facilitative communication.

According to the parent, Sammy,<sup>2</sup> a six year old girl with CdLS, has benefited greatly from the use of facilitative communication (Ask the Doctor, 1993). Both the therapist and the parent believe this approach has enhanced this child's ability to communicate.

Facilitative communication is a procedure involving a therapist or a Afacilitator≡ who assists an individual with physical and communication disabilities to point to desired objects, pictures, printed letters and words, or to a keyboard. **The scientific validity and reliability of this treatment technique have not been established** (American Speech-Language-Hearing Association, 1995). As such, this procedure should not be used without the informed consent of the individual and family. The critics of this technique claim the facilitator and not the child is the one communicating. Proponents claim there are instances in which they had no prior knowledge of the information that ultimately unfolded.

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Buzolich, M.J. (1987). Children in transition: Implementing augmentative communication systems with severely speech-handicapped children. *Seminars in Speech and Language*. 8 (2), 199-213.

Ask the doctor. (1993 March/April). *Reaching Out: The Newsletter of the Cornelia de Lange Syndrome Foundation*, pp. 3-5.

## DISCLAIMER

This newsletter is not intended for diagnostic purposes or self treatment. The Cornelia de Lange Syndrome Association and its committee do not necessarily endorse or recommend any products, services, methods or literature mentioned within. Any questions about treatments should be discussed with your child's doctor.

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