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Communication Intervention in CdLS

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Abstract

This article provides comprehensive information about speech and language abilities in the Cornelia de Lange syndrome (CdLS), also known as the Brachmann-de Lange syndrome, and describes successful therapy approaches with this population. It presents a review of the literature and summarizes information gained from a database of 116 children diagnosed with CdLS along with general information resulting from more than 275 evaluations of children with this syndrome. Also included are prognostic indicators for the acquisition of speech and language as well as intervention recommendations for speech-language pathologists. The asynchronous development of receptive versus expressive language abilities and the presence of oral-motor apraxia are documented. Also suggested is the likelihood of higher cognitive functioning than expressive language skills would indicate.

In 1984 when I began working with a child with Cornelia de Lange Syndrome (CdLS) I became aware of the lack of information available in the speech-language journals. Except for one article (Moore, 1970) there was little information about the range of expressive language abilities or the documentation of successful therapy procedures. Both the article by Moore (1970) and the literature available in the medical journals indicated speech skills were often significantly delayed, even in the more mildly affected. I was uncertain of the current validity of this literature because much of it was at least 10 years old. I also questioned whether the data actually reflected the potential for speech and language in this syndrome, particularly since considerable data were collected on institutionalized children. In addition, indications were that many of the individuals in these studies had not been provided with speech-language therapy either because it was not expected they could talk or because early intervention was not available.

I became involved with the Cornelia de Lange Syndrome Foundation in 1986 as a result of documenting successful therapy for expressive language in a child with this syndrome (Goodban, 1985). This involvement included attending annual conferences of the CdLS Foundation where I presented papers, led workshops, presented videotapes of my therapy with two children with CdLS, and provided evaluations and consultations for many of the families who brought their children to these conferences. I serve as the speech and language advisor on the Scientific Advisory Council for the CdLS Foundation.

My attendance at the earlier conferences of the CdLS Foundation confirmed many of my concerns regarding speech and language intervention for these children. Numerous parents told me that speech and language goals were not included on the Individualized Education Plans (IEPS) for their children at school. In addition, many of the parents reported that before they were aware of the CdLS Foundation they had been advised that their children would not survive or they should be institutionalized; they would never walk, they were deaf and they would never talk. Other parents reported they were told their children were deaf at birth and as a consequence the parents did not talk to their babies while they bathed, fed



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or changed them, only to discover later their children were not deaf. Furthermore, there were other problems frequently present, such as feeding difficulties or serious medical problems which made demands on parental time and energy. All too often the expectation was that these children would not talk. It seemed in part these expectations had become self-fulfilling. Consequently, I believed that much opportunity for language stimulation had been lost and a true picture of potential expressive verbal skills in this syndrome was not available.

In contrast, there was a second but smaller group of parents who had made every attempt possible to provide stimulating language experiences for their children, and in spite of the best attempts, their children had acquired little or no speech. There was also a third but still smaller group of parents who had children who were speaking in sentences and achieving success at school. What was not clear was why some children talked and others did not, particularly among the children without obvious differences in physical appearance, such as severe upper-limb malformations, profoundly noticeable facial features, or very obvious difficulties in social relatedness. One of my goals was to determine why some children acquired expressive language and others did not. Another goal was to discover intervention strategies for the severely non-verbal children.

In an effort to increase the knowledge about communication skills in this syndrome, I began to collect systematic information about each child I evaluated. The 116 individuals included in my database (Goodban, 1991; 1993) were evaluated at CDLS Foundation conferences in Atlanta, Boston, New Orleans, and San Francisco, USA; and in Blackpool, England. For a more complete description of the procedures see Goodban (1993).

Before describing this syndrome it is important to present some information about the Correlia de Lange Syndrome Foundation. This Foundation was formally established in 1981 as a result of an informal meeting of parents in the mid-70's. In 1977 the first newsletter was mailed out to 9 families. This newsletter is now mailed out to over 4,000 parents and another 2,500 professionals and friends. The CdLS Foundation has an active membership of more than 1000 families in the United States and a considerable international membership. Professional advice is provided by the Scientific Advisory Council, a group of individuals who specialize in medical genetics, neuropsychiatry and behavior, pediatric dentistry, molecular genetics, ophthalmology, pediatric genetics, radiographic imaging, pediatrics, orthopedics, and genetic counseling. The CdLS Foundation is an extraordinarily successful organization, essentially started and organized by parents of children with CdLS, with professional advice volunteered by the Scientific Advisory Council. Families travel from all over the United States and the world to its meetings.

Characteristics of Cornelia de Lange Syndrome

The de Lange Syndrome (Brachmann, 1916; de Lange, 1933) is most commonly referred to as the Cornelia de Lange syndrome (CdLS) but is also known as the Brachmann-de Lange syndrome. Dr. Cornelia de Lange first described this syndrome in 1933 although Dr. Brachmann wrote about a similar child in 1916. CdLS is a syndrome with an estimated birth prevalence between 1:10,000 (Opitz, 1985) and 1:30,000 (Facing the Challenges, 1993).

There are no definitive biochemical or chromosome markers (Hawley, Jackson, & Kurnit, 1985) for the diagnosis of this syndrome although severe biochemical markers have been

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suggested (Rappaport, Falik-Borenstein, Clark, & Rimoin, 1989). At the present time diagnosis is made on the basis of clinical observations. The most frequently observed facial characteristics include thin, downturned lips; elongated philtrum; broad nasal bridge with arched nostrils; and the chin may be smaller than expected. The eyebrows are often confluent and thin with a characteristic arch and the eyelashes may be very long (Facing the Challenges, 1993; Hawley et. al., 1985).

Other characteristics often associated with this syndrome include delayed growth and small stature; language delay, even in the more mildly affected; microcephaly (small head size); hirsutism (excessive body hair); and simian creases. Low set ears may also be present. Low birth weight is typically a feature of this syndrome with the average birth weight being five pounds and one ounce (Facing the Challenges, 1993).

Mental retardation is present in almost cases, ranging from mild to severe. According to literature available from the CdLS Foundation (1993) the average IQ is 53. Although mental retardation is typically present in this syndrome, a total of 9 cases have been reported in the literature as having normal or slightly below normal intelligence (Borghi, Giusti, & Bigozzi, 1954; Brylewski, 1978; Cameron & Kelly, 1988; Gadoth, Lerman, Garty, & Shmuelewitz, 1982; McIntire & Eisen, 1965).

Also reported are hearing losses, visual problems, dental abnormalities, feeding difficulties, upper limb malformations, and recurrent respiratory tract infections (Hawley et. al., 1985). The palate is often narrow or high and arched (Ptacek, Opitz, Smith, Gerritsen, & Waisman, 1963) and in approximately 10% of the cases, it is cleft (Berg, McCreary, Ridler, & Smith, 1970). Laird Jackson, the CdLS Foundation Medical Advisor, has documented that approximately 15% of these children are born with heart problems which are usually diagnosed within the first three months and corrected by surgery (Facing the Challenges, 1993).

These children may have faces that are devoid of expression (Barr, Grabow, Matthews, Grosse, Moti, & Opitz, 1971) and their behavior has been described as autistic-like or lacking in relatedness (Nyhan & Sakati, 1976). They may also be tactilely defensive (Goodban, 1985) or show a lack of sensitivity to pain. There is often a characteristic vocal quality in the crying of these children which has been described as feeble, low-pitched, growling, guttural, deep and raucous (Moore, 1970; Ptacek et al., 1963).

Characteristics of Speech, Language, and Hearing

Speech and Language

The absence of speech (Barr et. al., 1971; Hawley et al., 1985; Nyhan & Sakati, 1976) or the development of only minimal speech (Joubin, Pettrone, & Pettrone, 1982) has been well documented, even in the more mildly affected (Moore, 1970). Fraser and Campbell (1978) found that only three of the six patients they studied had useful language.

Beck (1987) in a study of 36 de Lange patients aged 5-47 years found the area of verbal communication to be specifically retarded. It should be noted that of these 36 patients six children had been institutionalized by age 9 years and seven patients had been institutionalized by age 19 years. Twelve (30%) of the patients in this study were reported to have language equal to or better than children of 3 to 4 years of age which was defined by Beck as the ability to recount independently. It was also reported in this study that only the



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patients older than 10 years had useful language. Sixteen patients (44%) were not able to express themselves verbally, although they were vocal, and one patient was completely mute. An additional eight patients were said to have some useful language although it is not clear how useful language was defined in this study.

McArthur and Edwards (1967) in a report of 20 cases aged 4 months-18 years noted that the major retardation was that of speech although mental retardation was evident in all cases. These two authors also point out that speech was far more retarded than comprehension.

Cameron and Kelly (1988) report on a child with CdLS and normal intelligence. In this study it was reported that although this 2:7 (years:months) old girl had initially exhibited delays in gross motor skills and in receptive and expressive language, she responded well to both physical therapy and speech and language intervention. As a consequence she achieved age-appropriate language skills.

I reported on 116 evaluations that took place at three CdLS Foundation conferences in the United States and one CdLS Foundation conference in England (Goodban, 1991; 1993). The purpose of these evaluations was to survey the speech and language skills of individuals with CdLS and to determine the prognostic value of related variables.

Each of the 116 individuals I evaluated had had an extensive genetic evaluation and had a confirmed diagnosis of CdLS. The age range of the patient group was from 2 months to 29 years, with a mean age of 5.6 years and a median age of 4.6 years. Fifty-five of the patients were male and 61 were female. Whenever possible the data were controlled for by age in order to prevent chronological age from being a factor in the acquisition of speech and language skills. Although a total of 116 patients was evaluated it was not possible to use this figure for most calculations because some of the patients were infants and thus were not yet talking or because some information about motor development, hearing loss or other factors was not available. All of the individuals were living with their parents except for the 29 year old, another child who stayed in an institution during the week, and two children who were in foster care. The data from these evaluations were analyzed using the database program of Paradox, 3.5.

As indicated in Table 1, a total of 56 individuals (61%) 2 years or older ($n = 92$) and 42 individuals (67%) 4 years and older ($n = 63$) had an expressive vocabulary of at least 3-10 words. Thirty-nine individuals (44%) aged 2 years or older ($n = 89$) and 32 individuals (53%) aged 4 years and older ($n = 60$) had the ability to combine two or more words into sentences. Twenty-one individuals (33%) 4 years and over had no words at all or had only 1 to 2 words in their expressive vocabulary.

Four individuals (4%) ($n = 100$) were judged to be within normal to low normal limits for language development. Other developmental skills were reported to be and appeared to be within normal limits. From a sample of 73 children aged 2 to 9 years, one child each at the age of two-years, three-years, five-years, and nine-years exhibited expressive language skills well within normal limits. The results from this database (Goodban, 1991; 1993) indicate a higher percentage of the population with CdLS has slightly better speech and language skills than has been previously reported (Beck, 1987; Barr et al., 1971; Hawley et al., 1985).

Most of the children with CdLS that I have evaluated have exhibited errors in articulation. Consonants were typically distorted or missing. Some of these children appeared to exhibit

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severe oral-motor and verbal apraxia which is the loss of the voluntary aspect of speech and motor movement. This diagnosis is based on the observation that although these particular children could spontaneously chew, swallow, cough and smile, there was difficulty in imitating oral-motor movements, phonation or speech. In the children with more advanced language skills, articulatory apraxia appeared to be present because of observed difficulties as articulatory complexity increased, with frequent sequencing and substitution errors.

My observations also confirmed previous reports of the tendency of children with CdLS to unexpectedly utter a meaningful word or phrase only once, using completely clear articulation and often performing at a level higher than previously observed. But then this performance is rarely if ever repeated. It is not known why this behavior occurs in these children but it is not an uncommon occurrence among apraxic adults with aphasia. This raises the question of a similar phenomenon occurring in CdLS.

The history in infancy and early childhood of abnormal vocal quality and feeding difficulties, particularly poor chewing and swallowing, raises the possibility of dysarthria as a cause for later speech difficulties. However, this does not seem to be the prevailing etiology for speech difficulties because these children rarely drool past the appropriate age and there is usually not consistent or persistent difficulties with vegetative movements. The low-pitched cry frequently present at birth and early infancy often disappears by 12 months. The abnormal vocal quality that is present in approximately half of the populations does not seem to be associated with speech or language abilities.

In addition to the above measures of expressive language other characteristics were noted. In almost all cases the ability to produce language was remarkably inferior to the ability to comprehend language. There was also considerable discrepancy between vocabulary measures and syntactic skills in that children who had highly developed vocabulary usually did not exhibit the expected syntactic skills. Similarly, children who were using an average utterance length of 4-5 words per utterance typically were not using question transformations.

Overall there did not appear to be a synchronous relationship between cognitive abilities and expressive language abilities. Although there is an assumption that a synchronous relationship exists between IQ and speech and language development, research on children with specific genetic syndromes reveal distinct profiles of performance not accounted for by their general cognitive abilities (Miller, 1991). One example of this is in the Down syndrome population where children evidence deficits in language productions in comparison to language comprehension and non-verbal cognitive skills. In addition, within language production, children with Down syndrome evidence better developed vocabulary skills than syntactic skills. Similarly, in the fragile X syndrome, language deviances can not be attributable to level of mental retardation (Jung, 1989; Miller, 1991).

My most predominant impression of these children as a group is they are among the most nonvocal children I have encountered. There was no indication of elective mutism; the majority of these children are just very quiet. They often make eye contact and there is often a sense they understand what is being said, but they are not usually vocal. Even among the children who have good language skills few of them can be described as talkative.



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I briefly talked to two adults who were mildly affected with CdLS, one female and one male, who attended the conferences because they had children with CdLS. Until recently (Robinson, Wolfsberg, & Jones, 1985), it was not known that individuals with CdLS could parent children. These adults reportedly had not been diagnosed with CdLS as children and it is not known whether they experienced difficulty with academic subjects as students. Their own syndrome did not become apparent until they parented children who were severely affected by the syndrome. It was not possible to conduct a formal evaluation on these adults but on an informal basis they appeared to have adequate expressive language abilities. At another conference I observed a third adult with CdLS. He was reported to be a bright individual who worked as a computer technician.

Hearing:

Almost all children with CdLS are diagnosed with a mild to moderate and sometimes severe hearing loss (Facing the Challenges, 1993). An extensive study of 45 patients with CdLS (Sataloff, Spiegel, Hawkshaw, Epstein, & Jackson, 1990) showed that virtually all of these patients had a hearing loss. Other studies have documented two children with verified sensorineural hearing loss (Egelund, 1987) and three out of seven patients with hearing losses (Marres, Cremers, & Jongbloet, 1989).

My evaluations of children with CdLS revealed unexpected reports from their parents regarding the history of audiological results. Many of these parents reported their children were diagnosed as severely hearing impaired at birth, moderately impaired at 12 months, and mildly impaired or without impairment at age 2 years. Since it is unusual for hearing to improve rather than worsen, it seems correct audiological assessment is difficult (Goodban, 1987). Infants with CdLS are often very small with narrow ear canals and difficult behaviors, making examinations a challenge.

Parents often report their children seem to hear much better than their test results would indicate. Considerable confusion exists regarding hearing ability for some children. Many children fitted with hearing aids will not tolerate the use of them or do so only sporadically.

Common Misconceptions

It was apparent after conducting numerous evaluations and attending several conferences that a number of misconceptions were being conveyed to parents by individuals unfamiliar with CdLS. For example, a few of the parents whose children were diagnosed with cleft lip or palate reported they were told it was not necessary to close these clefts since their children would not talk anyway. In truth, these clefts should be repaired as early as possible. Not only does this repair improve the ability to eat and reduce the likelihood of ear infections leading to hearing loss, it also enhances the speaking process.

Other parents have reported they have been told speech therapy could not begin until their children were talking! Additional parents reported they were told their children could not receive speech therapy or learn to talk until the gastric tube was removed and their children were eating normally. This of course is not true. Furthermore, I have evaluated children who were talking who had never received nourishment except through their gastric tube. However, when appropriate it would be beneficial to work with a speech-language pathologist on feeding therapy so the oral mechanism functions as normally as possible.



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A substantial number of parents have reported they have had difficulty finding an otolaryngologist who can insert pressure equalization tubes into the auditory structures of their babies or toddlers because they were so tiny. These parents need to consult with physicians who are experienced in working with small infants. There have also been reports of young children fitted with adult-sized hearing aids when child-sized aids are available and more appropriate.

A Summary of Prognostic Factors for the Acquisition of Speech and Language

As indicated in Table 3, children who weighed at least 5 pounds at birth, who had no or mild hearing loss, who had no severe upper-limb malformations, who sat by the age of 18 months or walked by the age of 30 months and who were judged to have good social relatedness were much more likely to acquire expressive language skills than those who did not meet these criteria. Social relatedness included factors such as eye contact, the appearance of comfortableness, alertness, and the child's overall ability to relate to people. Analysis of the database did not suggest a significant difference in language skills according to gender.

The analysis of this database (Goodban, 1991; 1993) confirmed the relationship of a lower birth weight with more severe involvement or delay of skills (Hawley et al., 1985); difficulties in relatedness (Nyhan & Sakati, 1976); the delay or absence of speech and language (Beck, 1987; Hawley et al., 1985); the presence of normal to low-normal speech and language skills (Cameron & Kelly, 1988); and the association of speech and language delays with hearing losses (Sataloff et al., 1990).

Many of the children had a voice that was guttural and low in pitch and loudness while some of the children had voices that were normal. Although the association between vocal quality and the ability to combine words in a sentence was not analyzed, in the database there did not seem to be a relationship between these two factors. Based on my evaluations of these children I do not believe vocal quality is a prognostic indicator of success in expressive language.

Concomitant Factors

Feeding Difficulties:

It is not unusual for a child with CdLS to have difficulty eating, swallowing, digesting food and gaining weight. Moreover, gastroesophageal reflux is the most common medical condition associated with CdLS (Facing the Challenges, 1993). When this reflux is not treated the child will more than likely be in pain and yet not be able to communicate verbally. Instead the child may act out, resort to self-injurious behavior or exhibit the Sandifer Syndrome (Sommer, 1991). Sandifer Syndrome is characterized by severe gastroesophageal reflux and unusual body movements such as wiggling and moving constantly, turning the head to one side or throwing the head back. The treatment for this reflux usually consists of special diets, medications, and elevating a child after eating. When these treatments are not successful surgical procedures such as a Nissen fundoplication and a gastrostomy may be necessary. The Nissen results in a narrowing of the lower esophagus and the gastrostomy provides a hole in the stomach, thus allowing for feeding by a G-tube and providing an outlet for stomach gases.

Behavior:

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Many children with CdLS have no behavioral problems although there may be conditions of pain, discomfort, frustration or dismay which makes self-injurious behavior more likely to occur. Gualtieri (1990) describes the typical young child with CdLS as hypersensitive and dysrhythmic. They are hypersensitive in that they may have strong reactions to ordinary stimuli and these reactions may continue long after the stimulus is gone. They may also be prone to behavioral problems such as hyperactivity, short attention span, and oppositional or repetitive behavior. They are dysrhythmic in that they are irregular in vegetative behaviors such as eating and sleeping and in emotional response. The lack of sensitivity to pain and/or heightened sensitivity to touch suggests some children may have neurological impairment.

Vision:

It is not unusual for children with this syndrome to be nearsighted, have recurrent redden, discharge or tearing or have ptosis of the eyelids. If ptosis is severe the children may lift their chins or arch their eyebrows in order to improve their vision. Many parents opt for surgery to correct the ptosis.

Many children with CdLS may not engage in normal gaze behaviors. Gaze averting may happen for a number of reasons. It may give the child time to process visual information, it may mean the child perceives the task as too difficult, or it may mean the child is feeling uncertain or stressed. Children with CdLS may also use peripheral vision more frequently than direct gazing because they have greater difficulty choosing which of the varied stimuli should receive their attention. It also tends to be true that children who are lower functioning show greater sensory rejection and sensitivity to stimulation in their environment (Morse, 1992).

Orthopedics:

In the more mildly affected children curving of the fifth finger (clinodactyly), small hands, a short thumb placed closer than usual to the wrist and some limitation of elbow motions are often present, with webbing of one or more fingers (syndactyly) less common (Facing the Challenges, 1993). Abnormalities of the hip occur in five to ten percent of the children with CdLS and may interfere with the ability to walk. Surgery may be used to correct this condition.

Of greater relevance to communication because of the interference in the use of sign language or other augmentative strategies for communication are the more severe upper-limb malformations. In some cases fingers, metacarpals and the long bones of the arm are absent.

Dental:

Dental problems can include small jaw development, poor oral hygiene, crowded teeth, small teeth, periodontal disease, and the erosion of teeth caused by stomach acids from reflux (Facing the Challenges, 1993).

Communication Intervention Procedures

In this section I will briefly discuss the therapy I provided for Becky and Michelle, two children with CdLS. Becky was the subject of the first documentation (Goodban, 1985) of successful speech-language therapy for a child with CdLS. Cameron and Kelly (1988) provided a second documentation of successful speech-language therapy for a child with



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CdLS and normal intelligence, although specifics about the therapy procedures were not provided.

Summary of My Therapy for Two Children with CdLS

Becky:

Becky is an ambulatory child who has received individual speech-language therapy from the age of 8 months. I began providing treatment for her at the age of 27 months and have continued with her up to her present age of 11:4.

My first impression of Becky was very similar to my later first impressions of almost all children with CdLS: she was a remarkably quiet child. Not only was she nonverbal she was also completely nonvocal. Becky, like most children with CdLS, had a face that was devoid of expression and she had the characteristic low-pitched vocal quality that approximated vocal fry. She had been diagnosed as tactilely defensive and she was reported to have a high threshold to pain.

During my first 30-minute session with Becky I was only able to elicit the CV utterance 'ba' and shape it into the word 'baby'. I left the session with a feeling of concern because Becky had vocalized 'ba' and 'baby' only a few times and only in response to unwavering stimulation. In contrast to my feeling, her mother, who was watching the session, was elated because she had never seen Becky be so vocal.

I was discouraged when I read the journal article findings that the area of speech and communication could be expected to be significantly delayed, even in the more mildly affected. Nevertheless, I believed Becky had the necessary prognostic factors for success with verbal language. She had the physical ability to produce speech sounds; a necessary level of cognitive development (an estimated IQ of approximately 50); adequate vision; the ability to attend to me and to imitate sounds and words (although this was not consistent); and she seemed able to hear even though she had been diagnosed with a mild hearing loss.

Becky was also well under the age of 5 years when therapy began, which I believed to be a critical factor. Other prognostic factors in her favor were the lack of severe upper-limb malformation and difficulties in social relatedness. She also had a mother who was confident Becky would talk.

Frequent throughout the treatment were concerns about the ultimate success of therapy. There were times when we wondered whether the goal of a vocal system should be abandoned and instead primary emphasis placed on sign language or some other form of nonverbal communication. Even when Becky began using two-word utterances her verbalizations were infrequent and not always intelligible. However, I continued to believe that although progress was slow, language would continue to emerge. For a more thorough description of this therapy see Goodban (1985).

By the age of six years Becky exhibited sufficient verbal skills to easily communicate in the verbal world, although intelligibility was still occasionally a factor and Becky did not talk readily to all people. Now at the age of 11 years Becky communicates with much greater facility. She telephones friends; she easily makes her needs known; and she has a sense of humor which she is able to express verbally. Her typical length of utterance is four-to-five words per utterance although she occasionally produces eight-word utterances. Intelligibility is approximately 90% with the subject unknown because of difficulty with articulation in

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longer utterances. Small function words are still occasionally omitted from longer utterances and higher level question-transformations are correct approximately 50% of the time. Vocal quality remains low-pitched and guttural, as with many children with CdLS.

Michelle

Michelle was 4:7 when I first began providing therapy for her. Although she reportedly had been enrolled in a total communication program since the age of 2 years my initial evaluation of her revealed an absence of meaningful words. This observation was confirmed by her mother. Michelle communicated her needs by vocalizing /ma/ or by using approximately 1-2 signs, according to her mother. She continued to receive sign language instruction in her school program. Michelle was ambulatory and wore bilateral hearing aids for a mild-to-moderate (20-40 dB) hearing loss in both ears. She missed numerous sessions because of frequent illnesses and was particularly susceptible to respiratory and ear infections.

Michelle was also very nonvocal as well as being nonverbal although vocal quality was more normal than it is with most children with CdLS. Like many children with CdLS her face was frequently devoid of expression and she was somewhat tactilely defensive.

Progress was slow but within the first year Michelle was able to verbalize approximately 5 words on a consistent basis. Both consonants and vowels were difficult for Michelle to initiate and imitate but her utterances became true meaningful words.

Michelle exhibited the same puzzling behavior as has been seen in some of the other children with this syndrome. This behavior consists of the child unexpectedly uttering a completely clear word or phrase that typically can not again be elicited, although the lower-level behaviors remain intact.

When Michelle was 7 years old it was discovered by an audiologist that unfortunately Michelle had been incorrectly fitted with hearing aids that amplified the low rather than the high frequencies where hearing acuity was insufficient. Apparently this incorrect amplification had occurred for at least one year. An additional factor relevant to Michelle's hearing acuity and subsequent verbalizations was the mother's observation that it was critical that Michelle's ear canals be irrigated at least every six months.

By the age of 8 years, Michelle was using approximately 85 words occasionally and infrequently combining two and three word utterances. Her mother reported she also had 68 signs. Michelle was able to make simple verbal requests and her mother stated she was very happy that Michelle had another channel of communication opened for her, even though it was still a limited one. Michelle continues at a treatment program closer to her home.

Therapy Procedures for Oral Communication for Becky and Michelle

Initial procedures included insistent stimulation and reinforcement for any imitations, both nonverbal and verbal. These earlier sessions primarily consisted of playful activities involving oral-motor play and use of noise-making objects. Sessions were designed to provide maximum visual and auditory stimulation and to increase attending behavior. Tactile stimulation and reinforcement was avoided because neither Becky nor Michelle liked to be touched. Activities were designed to approximate those of normal adult-child relationships, such as pretend grocery shopping, eating, and playing with dolls.

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With both children the most successful stimulation for eliciting vocal imitation consisted of saying "ba" or some other sound, word or phrase into a plastic ring and encouraging imitation of this utterance, likewise holding a ring near their mouths. Another successful technique was to cue the children by holding my fingers near my mouth as I stimulated for a response. These behaviors were more successful in eliciting responses than any other procedures. These and other visual cues clearly facilitated vocal responses and provided an indication oral-motor apraxia was a component of their disorder. Similarly, in order to develop consistent syllable or word inclusion, the rhythm of the utterance was tapped out with blocks, clapping, table tapping or gestures in the air.

Another successful stimulation technique consisted of using highly inflected verbal models. If I said 'baby' I rarely obtained a response. But if I said 'b-a-a-a-by-y-y-y' a response was much more likely to occur. This exaggerated inflection was probably more successful because it intensified auditory and visual stimulation, allowed for greater auditory processing, and was closer in form to the motherese appropriate for children at a younger language age.

Numerous standard approaches were used for stimulating for receptive and expressive vocabulary. Drill and repetition appeared to be useful in establishing the correct syntactical form for Becky when she began to combine three and four words into utterances. 'Wh-' question forms still require practice.

One or both parents were involved in most of the sessions and were taught the procedures of expectant waiting, self-talk and parallel talk. It was particularly important to teach the procedures of re-stimulating and waiting for a response. In addition, both children had been enrolled in early intervention programs and continued to receive special services through their school district.

Augmentative and Alternative Communication

For some children with CdLS, non-vocal or non-verbal strategies will need to be considered. For these children it may not be possible to achieve normal or even adequate speech for communicative purposes. Instead the ultimate goal should be to communicate at a level adequate to meet communication needs. Examples of available strategies include a communication board; American Sign Language; American Indian Hand Talk or Amer-Ind gestural code; Blissymbolics; Total Communication; Pantomime; a manual alphabet; eye-blinking encoding; or electronic communication aids. For a thorough description of all of the available non-vocal strategies see Silverman (1989), and for an overview of vocal and non-vocal strategies see Musselwhite and St. Louis (1982).

For children with severe upper-limb malformations there are gestural-assisted and neuro-assisted strategies available (Silverman, 1989). Unfortunately almost all augmentative communication strategies are difficult to learn for children who have difficulty understanding nonverbal or gestural communication. Regardless of the handicapping condition, it is important that all children be taught some means of indicating 'yes' and 'no.' Before considering the use of augmentative or alternative communication, a number of factors need to be considered. These include the level of cognition, motor abilities, receptive language abilities, and the motivation to communicate. It will often be most beneficial when speech-language pathologists function as communication therapists rather than as speech therapists (Silverman, 1989) and consult as appropriate with professionals such as physical



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therapists, occupational therapist, physicians, psychologists, engineers, social workers, vocational counselors, wheel-chair seating specialists, nurses, and teachers.

The following case study illustrates a child who benefited from non-vocal strategies:

Buddy

The description of Buddy by Buzolich (1987) demonstrates the successful use of blissymbols with a 12-year-old ambulatory nonverbal boy with CdLS. 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.

Facilitative Communication

Facilitative communication training (FCT) is described by Crossley (1992) as a useful teaching strategy for children who have severe speech impairments as well as neuromotor impairments affecting hand use. The hand function impairments are severe enough so that these children are not able to use sign language systems or handwriting nor are they able to use scanning systems. Candidates for FCT include those who have been diagnosed as intellectually impaired and/or autistic.

This method typically involves a helper or facilitator who holds and steadies the hand and wrist of the child who cannot speak, thus allowing the child to point to letters on a keyboard or alphabet board. There has been controversy about this technique. Its critics 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.

Sammy

According to the parent of Sammy, a six year old girl with CdLS, her daughter 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.

Intervention Recommendations

I have found the following recommendations to be useful for both professionals and caregivers.



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Become members of the CdLS Foundation as quickly as possible. The benefits are substantial. The Foundation provides expert assessment and advice on all aspects of the syndrome and immediately disseminates up-dated information to its members, long before it appears in professional journals. The Foundation also functions as a support group for its members, providing speakers and counseling sessions dealing with loss, sibling issues and stress within the family.

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 when necessary 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. Pharyngeal-esophageal tubes may be useful for middle-ear drainage as needed but a physician experienced in working with small infants is usually necessary.

If a hearing loss is suspected, headsets and hearing aids should 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 other situations.

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 sub mucous cleft requires a team approach of 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.

The therapy procedures of self-talk, parallel talk, and expectant waiting are recommended for the parents and the therapists. Gestures and sign language are also encouraged as methods to facilitate and motivate oral communication. All babies gesture before they use words and everyone uses gestures to facilitate 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.



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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. Augmentative and alternative strategies are available for the child who is severely impaired and/or has oral-motor apraxia. All children should be provided with a means to indicate 'yes' and 'no.'

Swallowing therapy may be beneficial for children who have swallowing disorders. A speech-language pathologist provides this treatment.

Conclusions

The results from the most recent survey of speech and abilities in the CdLS population (Goodban, 1991; 1993) indicates a higher percentage of this population has slightly better speech and language skills than was previously reported (Beck, 1987; Barr et al., 1971; Hawley et al., 1985). There are a number of reasons why speech and language skills appear to be improving in this population. Members of the Scientific Advisory Council for the CdLS Foundation acknowledge they are seeing an increase in the more mildly affected children, possibly as a result of earlier recognition of the more mildly affected. There also is a definite tendency toward higher expectations for these children, earlier and more effective speech and language intervention, as well as a tendency toward a lower incidence of institutionalizing these children. Another ameliorating effect is the one provided by the CdLS Foundation in alerting parents to the need for earlier intervention in other areas such as feeding difficulties and visual and hearing problems.

Documented in this article is the probable presence of severe oral-motor apraxia among many children with CdLS. Also noted but documented in a previous article is the asynchronous development of receptive versus expressive language. More thorough investigation is needed in the area of oral-motor apraxia to determine prevalence rate. There is considerable need to develop intervention strategies for the children who are most seriously impaired because of severe problems with social relatedness, upper-limb malformations, and hearing impairment.

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