

Speech-Language and Audiological Profile of Cornelia De Lange Syndrome: A Case Report

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ABSTRACT

The present study aims towards delineating the speech, language and hearing difficulties in a case of ‘Cornelia de Lange Syndrome (CdLS)’. Pediatrician diagnosed the child by observing the phenotypic characteristics, like syndactyly in left leg, tongue tie (repaired), kyphoscoliosis, bushy eyebrows, long philtrum, small stature, small head size, history of delayed eruption of teeth and delayed language and motor developmental milestones. Pedodontist surgically corrected the ankyloglossia and reported that the lower incisors were missing congenitally with only the canines and molars being present at this point of time. Psychological evaluation revealed borderline mental retardation, along with presence of autistic-like behavior, by administration of tests like ABC, CARS and VSMS. Speech and Language Analysis conducted via formal (REELS, PAT, Dr. Speech) and informal (PCC & GRBAS) protocols depicted delay in language development (with expression being inferior to comprehension), misarticulation (substitution and distortion errors), severely affected speech intelligibility, and severe hoarse vocal quality. Lastly, a comprehensive audiological evaluation (PTA, Immitance Audiometry & BERA), showed presence of mild conductive hearing loss. These myriad findings are the first of its kind to be documented from Central India, and such informations will aid us in gaining a better understanding about multidisciplinary diagnosis and management options for CdLS.

Key Words: Cornelia de Lange Syndrome (CdLS), Syndactyly, Speech and Language Delay, Autistic-like Behaviors, Conductive Hearing Loss, Multidisciplinary Team.

INTRODUCTION

Cornelia de Lange syndrome (CdLS) is a syndrome of multiple congenital anomalies characterized by a distinctive facial appearance, prenatal and postnatal growth deficiency; feeding difficulties, psychomotor delay, behavioral problems, and associated malformations that mainly involve the upper extremities. This is a developmental disability first reported by Brachman in 1916 and further investigated by De Lange in 1933 (Goodman & Gorlin, 1977). It may also be referred to as De Lange’s syndrome, Brachmann- de Lange syndrome, Amsterdam

dwarfism syndrome, and *typus degenerativus amsteiodamensis*. The syndrome tends to be relatively uncommon, with reported incidence rates varying from 1:30,000 to 1: 50,000 live births and appears to affect males and females equally (Corlett, 2003). Currently, no definitive tests or genetic analysis to confirm the diagnosis exists. However, it is suspected of being caused by a deletion on the short arm of chromosome 5 (5p12) (Goodart et al., 1994; Overhauser et al., 1994). Diagnosis rests on the presence or absence of a number of physical, cognitive and behavioral characteristics. Diagnosing classic cases of de Lange syndrome is usually straightforward, but diagnosing mild cases may be challenging, even for an experienced clinician.

Cornelia de Lange infants show a lower than normal birth weight and length, and can be described as failing to thrive. The majority are found to be functioning in the lower reaches of the moderately retarded range. A few reported cases have shown functioning levels approaching the low average range. Motor problems are pronounced (Porcella, 2007).

Principal clinical characteristics include: Delay in growth and development, hirsutism (excessive hair growth), bluish mottled skin, structural anomalies in limbs, missing or joining phalanges, dental problems, vermian hypoplasia, feeding difficulties, psychomotor delay, behavioral problems, sensorineural hearing loss (90%), self-injurious behaviors, and gastroesophageal reflux (Shprintzen, 1997; Scacheri, 2002; Percy et al., 2007 Robb & Reber, 2007; Lefler, 2008).

Distinctive facial characteristics include: Short, upturned nose with anteverted nares (88%), depressed nasal bridge (83%), thin & downturned lips (94%), long philtrum , low set ears, possible cleft palate, dentition problems such as eruption difficulties, confluent eyebrows that meet at midline (synophrys), long curly eyelashes (99%), low anterior and posterior hairline (92%), underdeveloped orbital arch (100%), high arched palate (sometime cleft) 86%, late eruption of widely spaced teeth (86%) , and micrognathia (84%). (Shprintzen, 1997; Scacheri ,2002; Acs, Ng , Helpin , Rosenberg , and Canion ,2007; Percy et al.,2007 ;Lelfer , 2008).

Other dental problems include misalignment, delayed teething, microdontia (small teeth), dental erosion due to gastric reflux, and periodontal disease. About 50 percent have ophthalmologic manifestations including the following: Myopia (Nearsightedness), ptosis (drooping of eyelids), blepharitis (inflammation of eyelids), epiphora (excessive secretion of the lacrimal glands producing an overflow of tears), microcornea (small cornea), strabismus, nystagmus, astigmatism (unequal curvature of the eyes refractive surface, i.e. cornea, creating a visual problem), optic atrophy, coloboma of the optic nerve (defect of optic nerve, usually congenital), and congenital glaucoma (Vinson, 2012).

Developmental and cognitive delays predominate in individuals with Cornelia de Lange syndrome. A severe speech delay is typical. Only about 50% of the children of 4 years of age or older combine words into sentences, and 33% have no word, or possibly one or two words. Only

4 percent have normal or lower than normal language skills. Severe speech delays are likely to co-occur with intrauterine growth retardation, hearing impairment, upper limb malformation, severe motor delays, and poor social interactions. Typical IQ range from 30 to 85, with an average IQ of 53. Those with higher IQs tend to be those who have normal cephalometric measures and high birth weight. The degree of occurrence of behavioral characteristics tends to vary with the degree of mental retardation and the presence of autistic-like behaviors. In terms of prevalence, hyperactivity occurs in around 44 percent, and daily aggression is noted in approximately 49 percent.

The absence of speech or the development of only minimal speech has been well documented, even in the more mildly affected. There is often a characteristic vocal quality in the crying of babies, which has been described as feeble or low-pitched. The low-pitched cry frequently present at birth and early infancy often disappears by 12 months. Most people with CdLS exhibit errors in articulation. Consonants are typically distorted or missing. In addition, there have been some reported observations of severe oral-motor and verbal apraxia, which is the loss of the voluntary aspect of speech and motor movement. There have also been unconfirmed reports that individuals have a tendency 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 rare if ever repeated. In almost all individuals 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. Those people 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. The majority of individuals are 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 individuals who have good language skills, there are few who can be described as talkative.

In spite of vision problems, visuospatial memory, along with perceptual organization and the fine motor skills are relative strengths. Activities that provide stimulation for the vestibular system are typically pleasant to individuals who have de-Lange syndrome. Speech-language therapy should focus on feeding problems, behavioral issues, articulation errors, apraxia, language delays, cognitive impairment, and AAC for those who are nonverbal (Percy et al., 2007).

NEED OF THE STUDY

CdLS is not typically diagnosed in children, since within the syndrome there is a wide spectrum. Diagnosing classic cases of CdLS is usually straightforward; diagnosing mild cases may, however, be challenging even for an experienced clinician. Early detection thus becomes important because early intervention can have a significant impact on the long-term prognosis of

many children. The need of the present study was to increase the professionals' knowledge about children affected by CdLS, along with contributing towards evidence based research.

AIM

This investigation was aimed towards highlighting clinical features related to speech, language and hearing in a case with "Cornelia de Lange Syndrome" reported at the department of Speech and Hearing in Sri Aurobindo Institute of Medical Sciences (SAIMS), Indore.

METHODOLOGY

I. SUBJECT

An eight years old male case has been selected as the subject of the study (with informed consent of the family), who basically reported the Speech and Hearing Department of Sri Aurobindo Institute of Medical Sciences (SAIMS), Indore one year back with the presenting complaint of unclear speech.

II. RESEARCH DESIGN

A descriptive (explanatory case study) research design was selected for the study.

III. PROCEDURE

Taking into consideration the phenotypic characteristics of the subject, we decided to do a detailed multidisciplinary investigation by a team of Pediatrician, Pedodontics, Psychologist, Speech Language Pathologist and Audiologist, so as to get an overview of such rarely reported case of CdLS.

IV. TOOLS

All the professionals used a varied range of tools in order to deduce the characteristics of the selected subject. The tools being used are as follows:

S.NO	PROFESSIONAL	TOOLS USED
01.	Psychologist	Wechsler Intelligence Scale for Children-3 rd Ed. (WISC-III; Wechsler, 1992). Autism Behavior Checklist (ABC; Krug et al, 1980b).

		The Childhood Autism Rating Scale (CARS; Schopler et al, 1980) .
		Indian adaptation of Vineland Social Maturity Scale (VSMS; Malin, 1992).
02.	Speech Language Pathologist	Receptive Expressive Emergent Language Scale, 3 rd Ed. (REELS-3, Bzoch, League & Brown, 1991).
		Hindi version of Picture Articulation Test: PAT (Ali Yavar Jung National Institute for the Hearing Handicapped and Regional Rehabilitation Training Centre Madras, India, under UNICEF sponsored project, 2004).
		Dr. Speech Version 4 (Vocal Assessment for Windows, Version-4.30; 1999, Tiger DRS, Inc.).
		GRBAS Scale (Hirano, 1981).
		Percentage of Consonants Correct (PCC; Shriberg& Kwiatkowski, 1982).
03.	Audiologist	Otoscope.
		Pure tone Audiometer (MAICO MA 53).
		Immittance Audiometer (Madson Zodiac 729).
		Distortion Product Otoacoustic Emissions (DPOAEs) & Brainstem Evoked Response Audiometry (BERA) through NEURO-SOFT.

RESULTS AND DISCUSSION

Initial case history revealed that the child was the first baby of non consanguineous marriage and had vaginal delivery at 37 weeks gestational age with low birth weight (< 2500gms). Post-natal history depicted presence of pathological jaundice and pneumonia after 3 days of birth. At present scenario, the child is under medication for the treatment of gastro esophageal reflux. He was able to communicate verbally through simple sentences and comprehends few familiar complex sentences. The scholastic performance was good, though had difficulties with reading, and there was parental suspicion of probable hearing loss. CdLS is characterized by low birth weight (Porcella, 2007) and 85% experience some type of gastro esophageal reflux (Cornelia de Lange Syndrome Foundation, 2010b), leading to a variety of behavioral problems.

The findings of the present study have been divided into five sections, based on the reports of multidisciplinary professionals. However special emphasis has been given over the findings by ASLP.

Section I: Pediatrician's Findings

The child had syndactyly in left leg, tongue tie (repaired), kyphoscoliosis, bushy eyebrows, long philtrum, small stature, small head size, history of delayed eruption of teeth and delayed language and motor developmental milestones with no other features of ectodermal dysplasia. Based on these cardinal features, Pediatricians diagnosed the case as “Cornelia de Lange Syndrome”.

The recent literature also supports that at the present time diagnosis is made on the basis of clinical observations (Porcella, 2007). The most frequently observed facial characteristics include thin, downturned lips; elongated philtrum; broad nasal bridge with anteverted 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 (Hawley et. al., 1985). Clinodactyly on the toes and fingers is common (Toker et al., 2009).

Figure 1(A), 1(B), and 1(C) depicts, few of the cardinal features observed in an eight year old male child diagnosed with CdLS.



Figure 1(A): CdLS child, with depressed nasal bridge, anteverted nostrils, elongated philtrum, thin and downturned lips, bushy eyebrows, and low set ears.



Figure 1(B): Syndactyly in left leg of the CdLS child.



Figure 1(C): Short stature of eight year old male child, diagnosed with CdLS.

Section II: Pedodontic's Findings

The child's lower incisors were missing congenitally with only the canines and molars being present at this point of time along with a delayed eruption of maxillary incisors. Many of the teeth had carries. Ankyloglossia was also present which was surgically corrected at the age of 7.11 years. Both speech and non speech functions of lips and tongue (even after lingual frenectomy) were restricted. All the vegetative functions were normal except difficulty in chewing harder substances.

Kline et al (2007), reported that there is poor oral hygiene and dental carries may be problematic, increasing the risk of periodontal diseases with aging. Dental problems may include misalignment, crowded teeth, delayed teething, microdontia (small teeth), dental erosion due to gastric reflux, and periodontal disease (Vinson, 2012).

Section: III: Psychologist's Findings

Psychological evaluation revealed borderline mental retardation, which is in accordance with the data of Cornelia de Lange Syndrome Foundation (2010b), which states that an average IQ score in individuals with CdLS is 53, which is within the mild to moderate range of mental retardation. Wierzba & Selicorni (2009) also reported that the psychomotor retardation varies from mild to profound and is combined with a lack of speech or speech impairment. Though a passed criterion had been attained on ABC, but administration of CARS revealed the child to be

non autistic. Due to the presence of autistic-like behaviors, the child's self help, self direction, communication and socialization skills were affected as depicted by VSMS.

Nyhan & Sakati (1976) described the behavior of CdLS child as autistic-like or lacking in relatedness. They may also be tactilely defensive or show a lack of sensitivity to pain. Oliver et al (2008) stated that individuals with CdLS may have an increased likelihood of showing autistic like behavior, while Richman et al (2009) also revealed that "individuals with CdLS may exhibit increased level of behaviors commonly associated with autism".

Section IV: Speech Language Pathologist's Findings

Speech and Language Analysis depicted delay in language development as receptive and expressive age ranged in between 33-36 months and 24-27 months respectively, on administration of REELS. PAT revealed that vowels were articulated correctly, however almost all the consonants at word level were predominantly substituted with other phonemes, followed by distortion. PCC of conversational speech sample was found to be 42% suggestive of severely affected speech intelligibility. These findings were in contrast to literature review which says that 53% of CdLS children who were four years or older combined two or more words into sentences, though the receptive language skills were typically much higher than the expressive language skills. Apart from that, sound substitution pattern is common along with an unusual vocal quality that is guttural, hoarse, low in pitch, muffled and not very loud (Goodban, 1993).

Objective voice evaluation through Dr. Speech suggested presence of severe hoarse vocal quality with predominance of breathiness (Mean Fo: 198.67 Hz, Jitter: 0.54%, Shimmer: 6.43%, NNE: -7.07dB, and SNR: 13.87dB), whereas perceptual evaluation through GRBAS, revealed that qualitatively the voice is moderately rough, aesthenic and strained and severely breathy, reflecting an overall gradation of extremely disordered voice. In Contrast, literature revealed that 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 (Cornelia de Lange Syndrome Foundation (2010b). Many exhibit a characteristic low-pitched gravelly voice early in infancy (Smith & Jones, 1982).

Although documented in the literature (Woliver, 2009) is the probable presence of severe oral-motor apraxia among many children with CdLS, however no such findings have been revealed in this child.

Section V: Audiologist's Findings

Audiological evaluation revealed PTA of right and left ear as 36.6.dBHL and 33.3 dBHL respectively and BC thresholds within normal limits, suggestive of bilateral mild

conductive hearing loss (Figure 2). Tympanogram was 'B' type bilaterally, suggested of presence of middle ear pathology.

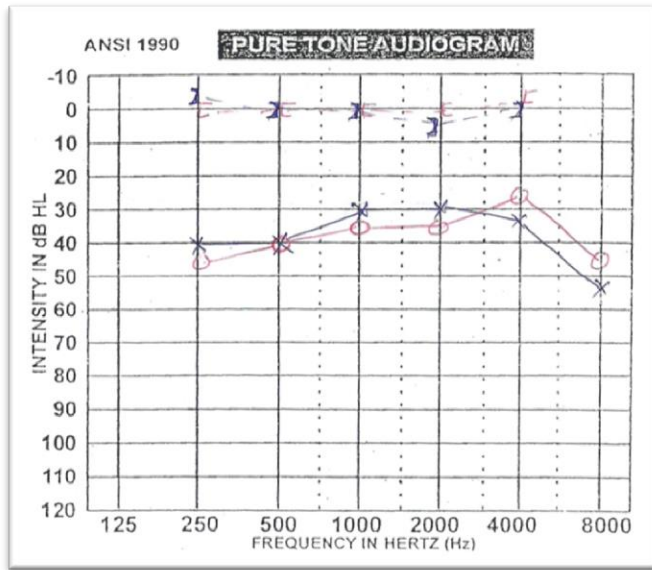


Figure 2: Pure tone Audiometry findings of CdLS child.

ABR testing showed well-formed responses to click stimuli down to 40dBnHL by air conduction and down to 20 dBnHL by bone conduction. This is consistent with a mild hearing loss in the 1000Hz to 4000 Hz frequency range.

Almost all children with CdLS are diagnosed with a hearing loss, which may be conductive, sensorineural or mixed (Marres et al., 1989; Ichiyama et al., 1994; & Marchisio et al., 2008). Even, Sataloff et al (1990) reported that some degree of hearing impairment is present in over 90% individuals with CdLS.

In Marchisio et al.'s (2008) investigation of 50 children, 1-18 years with CdLS, hearing loss was found in 40 (80%), with conductive hearing loss alone in 60% and in combination with a sensorineural hearing loss in 20%. Otitis media with effusion was found in 94% and prevalence was reported to be the same in all age groups. Sensorineural hearing loss was found in two boys examined by Ichiyama et al (1994). One had no responses on an auditory brainstem response test at 100 dBHL, the other had a wave V threshold of 40dBHL.

CONCLUSION

Children with CdLS 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.

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There exists a myth in Indian scenario that “speech therapy can not begin until their children are talking”. However, the decision to begin communication intervention should not be delayed, and 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. It is advisable to consult an audiologist and/or otolaryngologist who is familiar with CdLS or who is experienced in working with infants. Even a mild hearing loss can result in a speech and language delay.

Apart from this, children with CdLS usually have a wide array of health problems, making it important for all specialists to be aware of the child’s special needs. Multidisciplinary treatment approach is the key to success in managing children with syndromes. Ongoing developmental services such as physical, occupational, and speech therapy, should begin simultaneously. Apart from that, an annual audiological evaluation, evaluation of GERD, a dental visit by a dentist familiar with patient needs, and an ophthalmology evaluation should be conducted. An “Audiologist & Speech Language Pathologist”, may be the first healthcare personnel to identify such a child and may lead the multidisciplinary team in treating their problems.

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