# Ear, Nose and Throat Health in CdLS

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The Cornelia de Lange Syndrome Foundation receives many inquiries about ear, nose and throat (ENT) health for children with CdLS. ENT health is not just a physical concern; it affects communication and development. Therefore, it is critical to address these issues when children are young and most able to acquire new skills. Since many otolaryngologists have no knowledge of the specific otologic problems of children with CdLS and their management and care, it is important for families to have as much information as possible.

CdLS occurs in approximately one of every 10,000 live births, affecting boys and girls equally. Infants with this syndrome often have a similar facial appearance including a small nose; anteverted (turned up) nostrils; down-turned mouth; prominent philtrum (space between nose and mouth); convergent eyebrows; and long, curly eyelashes. Other manifestations of the syndrome include low-set ears, heart defects, abnormalities of the limbs and developmental delays. Most people with CdLS will also have some degree of hearing loss and language delay.

## RESEARCH

Because of the rarity of this syndrome, the apparent developmental issues in many cases, and examination difficulties created by behavioral problems, ear, nose and throat abnormalities in patients with CdLS have not been studied extensively. Much of what we know comes from clinical experience and from one study of 45 patients published in 1990.

## EAR

Children with CdLS typically do not have normal eustachian tube (canal connecting the ear and the throat) and middle ear function. They often need tubes for many years to treat conductive (mechanical) hearing loss caused by middle ear fluid, not uncommonly until puberty. Some may require them even longer. The need for four or five sets of tubes is not unusual. It is important to keep the ear free of fluid in order to optimize hearing and prevent the development of additional middle ear problems. Up to 60% of individuals with CdLS have a high arched palate, and up to 20% may have either a cleft palate or a submucous cleft palate. Virtually everyone with a cleft palate has Eustachian tube dysfunction, and middle ear fluid that requires placement of tympanostomy tubes to correct the conductive (mechanical) hearing loss. Thirty percent of children have narrowed external ear canals, most of which can only partially be examined by the smallest pediatric ear speculum. At least fifty percent of children we studied had serous otitis media (middle ear fluid). Seven

additional children (15.5%) had tympanostomy tubes in place because of previously diagnosed ear fluid. All 22 of these children had narrowed external ear canals. Despite these findings, only three of the children (6%) had been evaluated previously for hearing loss and for hearing aids.

## COMMUNICATION

Audiograms and voice recordings were attempted on all 45 children in the original evaluations.
Twenty-seven children (60%) had language limited to just a few words, but no child was found to be mute. Correlating the degree of hearing loss with language development revealed an important trend. Nine children (24%) had mild hearing loss (the volume at which they could understand speech was 25 to 45 dB). Eight of these children had vocabularies ranging from 50 words to greater than 1000 words. One child younger than twelve months had no speech, but used expressive jargon appropriate for his age. Nine children (24%) had moderate hearing loss (45 to 70 dB). All of these children had vocabularies of fewer than 30 words. Twenty children (52%) had severe hearing loss (the volume at which they could understand speech was 70 dB). None of these children used more than three words, and even these were not used consistently or appropriately. Seven of 45 patients could not cooperate with audiometric testing. Their parents reported that they responded inconsistently to sounds, and all seven of them had no developed language. We suspect these seven children also had severe hearing loss. One set of twins was evaluated in which one child had serous otitis media (chronic fluid) bilaterally and his brother had no middle ear effusions. The child with the chronic fluid had less than half the language skills of his brother. Sensorineural (inner ear or hearing nerve) hearing loss also emerged as a component of CdLS. Because of the large number of physical issues and testing difficulties, this finding had not been emphasized in the past. However, it is clearly an important factor in children with CdLS who have moderate, severe or profound deafness.

## DEVELOPMENTAL ISSUES

It is particularly important for all physicians to recognize correctable causes of communication problems as early as possible. This is especially true for issues of hearing loss and language development. Children with hearing loss are often incorrectly thought to be delayed developmentally because of poor language skills. However, accurate, early differentiation is important for several reasons. First, children with mild or moderate mental retardation cannot afford the additional deficit caused by potentially correctable hearing loss and delayed language development. Second, there exists a “critical period” before the age of three years for the development of language in all children. During this time, a child’s central nervous system is most ready to receive, process and store auditory and linguistic information for the development and use of language. When language skills have not been acquired during this period, even intensive language therapy usually will not permit the child

to obtain optimal, age-appropriate language skills. Consequently, early evaluation is essential in children at risk for hearing loss. Hearing aids can be fitted to children of any age. There is a major advantage to fitting aids as early as possible.

## NOSE

Anecdotally, sinus disorders in individuals with CdLS are common. To the best of my knowledge, there has been no comprehensive study of this issue. We assume that the problems are related to the structural abnormalities common to CdLS: cleft and high arched palates, small nasal size, and perhaps mucosal abnormalities that are also associated with serous otitis media (ear infection). As with middle ear effusion (fluid in the ear), there really are no good preventative treatments that should be instituted prior to the first incident. If a child develops recurrent sinusitis, then treatment with decongestants, demulcents, antihistamines as appropriate, steroids, and antibiotics for active infections is appropriate. In severe cases, sinus surgery (usually functional endoscopic sinus surgery) may be indicated. Polyps are often present with chronic sinusitis (as well as allergy). Nasal polyps are benign growths that can partially or completely block the nose, obstructing nasal breathing. They are also accompanied often by mucopurulent (combination of mucus and pus) nasal discharge. They are diagnosed on physical examination by an ENT doctor. Surgical removal of polyps is necessary if there is nasal obstruction.

Children with CdLS may have other ear, nose and throat problems as well, including allergies, recurrent hoarseness, and other conditions. Most of these can and should be managed as they are for children without CdLS. It is important to note that tonsillectomy and especially adenoidectomy should be avoided in anyone with a true cleft or submucous cleft palate. However, ear fluid, infection and hearing loss clearly constitute the most common and significant otolaryngologic disorders in CdLS. If hearing loss goes unrecognized, untreated or undertreated, other problems are likely to be aggravated. Hence, diligent, expert otolaryngologic care is essential for all patients with CdLS.