



CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.
Reaching Out, Providing Help, Giving Hope

Puberty and Adolescence in CdLS: a Survey of the Changes

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Although CdLS was originally described in the early 1900's, the syndrome has been well characterized only during the last quarter of the century. Many individuals with CdLS, diagnosed when they were infants, are now entering into adulthood. A large number of families and caregivers, therefore, have multiple questions about both the physical and mental changes that will occur during puberty, adolescence and adulthood. Previously, our answers as healthcare providers had been based on personal experiences with individual adult patients of different ages. There are few publications addressing adolescent and young adulthood issues for any genetic syndrome, although these are of great interest to both primary care providers and geneticists.

In the past several years we have collected specific information about puberty and adolescence from families, as part of an ongoing global project to evaluate aging in CdLS. A puberty questionnaire was originally distributed to member families of the CdLS Foundation through the USA office eight years ago. Seventy-four have been evaluated for this part of the aging project, and the collated results follow.

There were almost twice as many surveys received on females than on males. Both males and females experienced worsening gastroesophageal reflux during adolescence, often requiring treatment. Sixty percent reported circulation problems with their hands and/or feet. Other medically related issues include scoliosis (curvature of the spine), nasal polyps (growth in the nose), seizures, and psychiatric disorders.

Of the responses received, the average age of puberty onset was 13 years in females and 14 years in males, both slightly later than in unaffected children. There was a growth spurt reported during puberty in both height (in 40% of the females and half of the males) and weight (in 60% of the females, and in 40% of the males). Pubic hair developed in most of the individuals (in 85% of the females and 90% of the males); however axillary hair (under the arms) was infrequent (in 20% of the females and 35% of the males). Of the survey answers received on the females with CdLS, 80% reported breast development, and only 75% ever "got" their period. Premenstrual syndrome was common; irregularity of the periods was present but less common. Various treatments for menstruation included, with varying degrees of success: hysterectomy, hormonal replacement (e.g. taking hormones via a pill), and gynecology procedures. Of the surveys received on the males with CdLS, two-thirds reported facial hair, and half had increased size of genitalia (penis, testicles).

Three-quarters of the surveys discussed that behavioral issues were very common and worsened with the onset of puberty. Thirty-eight percent reported moodiness, 25% had obsessive-compulsive disorder. Other behavioral concerns included change in sleep patterns



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and masturbation. Management of these issues provides the most difficult challenge to the caregivers.

This information will enable us to address your questions about physical changes, mental development, and specific concerns as your children enter puberty and adolescence. In addition, we will be able to provide specific answers to queries from your primary care provider. As we pursue our “aging” project, we hope to continue to be able to supply information that can be useful for each of you. As always, please do not hesitate to contact us with specific questions.