

Older Individuals with CdLS

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Aging is a process that no one can reverse, impacting everyone at some point in his or her life. Individuals with CdLS are no exception. Certainly, for their caregivers, aging can be a cause for a great deal of concern. The life-expectancy for children with CdLS has improved in the last thirty years as knowledge of CdLS and treatment options have improved. Because individual with CdLS can live well into adulthood, it is important for caregivers to recognize and monitor the changes their children encounter as they age.

Aging is complex, involving all organ systems and body functions. The Baltimore Longitudinal Study of Aging, America's longest-running scientific study of human aging, addresses what happens as people age by using 1,400 men and women from their 20's into their 90's as study volunteers. Sponsored by the National Institute of Aging, the goal is to sort out changes due to aging from those due to disease or other causes. Interestingly, there is very little evaluation of individuals with syndromes, and, in fact, this is an underrepresented area even in genetics research. For CdLS, the little that we know about the aging process has come from the members and families' reports, as well as an ongoing project in which I, and a number of other professionals in Baltimore, have been involved. (See page 14 for description of new CdLS research projects).

Puberty

Regarding adolescence (likely ages 12 to 19), many changes are similar to those of children without CdLS. Hormones do increase in CdLS and lead to body changes as well as behavioral effects. Physically, most individuals undergo puberty, however up to 10% of males and 15% of females have very few changes. In addition, only one quarter to one third of adolescents with CdLS develop axillary hair (hair under the arms). 75% of females achieve menstruation, which frequently can be irregular. Hormones (female sex hormones) can be used to help regulate cycles. DepoProvera (progesterone) is a hormone shot given every three months. It will eventually decrease the blood flow of the menstrual periods, or stop them altogether. This is not thought to be dangerous and has the advantage of no more bleeding. Initially, though, there can be increased blood flow that could last up to 9-12 months. Oral contraceptives, "the pill," may also be used to control menstrual bleeding, and there is no general contraindication for this use in persons with CdLS. There are medical conditions that make the pill a poor choice, such as seizure disorder, family history of clotting disorders and so on. There is now a patch form, which makes a good choice for girls with significant gastrointestinal problems. Premenstrual syndrome may occur, but manifests itself in many ways. Motrin, if tolerated, or similar anti-inflammatory agents could be helpful. Some parents have reported difficulty with their daughters understanding their menstrual periods. I would recommend for parents of young, adolescent daughters with



CdLS to discuss several issues with their health care provider preferably prior to the onset of menses, including control of bleeding, prevention of pregnancy, and sexuality. Some females with CdLS do have a hard time with the idea of the bleeding.

Behavior

The behavioral manifestations of adolescence include: emotional swings, anger without apparent cause, frustration, sexual tension (which may be alleviated by masturbation or acting out), sleep disruption, and worsening of current behavioral manifestations of CdLS such as self-injurious behavior, aggression, obsessive-compulsive tendencies, anxiety, and depression. Behavior may be cyclical, potentially more obvious in females. Often, “difficult” behaviors seem to resolve or be curtailed by late adolescence. Management techniques for behavior problems include: counseling (ideally, family counseling), behavioral modification (positive reinforcement for “good” behaviors and consistency in the approach), and medications if unable to be controlled in other ways. Calm music and quiet time alone in the bedroom may be calming if needed. Issues of sexuality occur in individuals with CdLS as in all adolescents. Masturbation is a frequent area of concern. There are several general suggestions that can be given, and the developmental age of the individual plays a role: allow personal touching only in the bedroom and/or bathroom, consider having the individual wear overalls or other clothes that make it difficult to reach the genital area, and discuss privacy and that touching is not wrong. Do not allow your child to be aggressive sexually with other individuals. Do the same with inappropriate use of words.

Medical Issues

As individuals with CdLS age, they have variable medical issues, which may be dependent upon physical changes that were present from birth. Those with congenital heart disease, cleft palate, scoliosis and other major findings should continue to be checked by the various subspecialists. Gastroesophageal reflux can worsen during puberty and/or in the adult years, and needs to be monitored. Anyone who has had a diagnosis of Barrett’s esophagus should have an annual esophagoscopy with biopsy for two or three years until deemed stable. Constipation should be treated with diet and medications if needed. If there are still gastrointestinal complaints, food allergies are possible and lactose intolerance, milk protein intolerance, or gluten sensitivity could be investigated. All individuals with CdLS, regardless of age, should have an upper GI series to rule out a bowel malrotation. In addition, seizures may occur for the first time in older individuals, or may recur if they had been present in childhood and seemingly resolved. Ongoing studies currently include an evaluation for osteoporosis and hormonal imbalance as individuals age; the results will be available by Conference in June 2006. Prostate enlargement has been noted. Tumors and cancers have been reported only very rarely and are not likely an increased risk in CdLS. Unlike Down Syndrome, Alzheimer’s Disease (early senility) does not appear to be an issue with age. Many families even report a calming of behavior in the 30’s.



Development

An important aspect of aging in CdLS is that the learning process continues. Many parents of older individuals with CdLS have reported that there appears to be major developmental learning occurring. For example, one male began walking in late adolescence and another learned to use the telephone at age 32 years. I would urge continuation of therapy as long as possible. Most states will discontinue their services at age 21 years, but the insurance may pay for additional service.

Causes of Death

Causes of death with aging may be related to the congenital malformations (e.g. congenital heart disease), but also include GI issues (e.g. volvulus, or twisting of the bowel), aspiration pneumonia, or unknown infection. Some of these are potentially preventable, with appropriate screening studies and immediate treatment for complications. The oldest individuals that I have seen and confirmed as having CdLS have been in their 40's, however I recently received a report of a man in his 60's. According to Dr. Laird Jackson, the former Foundation Medical Director, the major causes of death are still an area that needs to be studied.

Continued Health

Maintaining appropriate medical care is crucial for all individuals with CdLS as they age. Many pediatricians will continue extended care past 18 years, but it may be difficult to obtain procedures in a children's hospital. There are internal medicine and family service physicians who have large practices with some experience in individuals with special needs. Often, the clinical geneticist or pediatric gastroenterologist may be able to recommend these physicians. The subspecialists are harder to find, but are likely to be affiliated with children's hospitals. Parents should always inquire, at the first meeting with these individuals, whether or not he or she is comfortable taking care of a patient with CdLS. Any of the Clinical Advisory Board (CAB) members, particularly myself, would be happy to field questions from your physicians.

If you have questions or concerns about any issue related to aging, more information can be found through the Foundation. Parents can submit questions to the CAB through the www.CdLSusa.org "Ask the Expert" section or browse the numerous answers in the "Ask the Expert Answers" section. Family Service Coordinators are also happy to address your questions at 1-800-753-2357.

