



## Orthopaedic Issues in CdLS

*Carmen P. Pichard--Encina, M.D., Assistant Professor, Johns Hopkins Orthopaedic and Spine Surgery, Good Samaritan Hospital Baltimore, MD, and member of the Foundation's Clinical Advisory Board*

Musculoskeletal problems are common in people with CdLS. Some problems are obvious at birth such as missing hands or fingers. The upper extremities are usually more involved than the lower extremities. Upper extremities are involved in almost one third of cases. Other problems develop with age. One study looked at twenty individuals with CdLS ages 13-50 and found that 90 percent had some degree of decreased bone density, which is commonly associated with older aged individuals. Many individuals also appeared older than their actual age. Premature aging is a topic that is being studied at this time.

The most common orthopaedic findings in CdLS are as follows:

**Hand abnormalities:** The differences include small hands (in 90 percent of cases), incurved and short fifth fingers (in 74 percent of cases), and short thumb placed low in the hand. More severe findings include missing fingers and hands. Surgery is rarely helpful when digits or hands are missing. Prostheses are also rarely useful. In general individuals with CdLS will find ways to use their residual limbs efficiently. Occupational therapy might be useful to help kids find tools or adaptations to make daily activities easier.

**Forearms:** Some individuals with CdLS have radio-ulna synostosis which is the fusion of the radius and ulna bones. The use of physical therapy on somebody with a synostosis could cause injury. Therefore it is recommended that a person diagnosed with CdLS have an x-ray of his or her forearms.

**Stiff elbows:** A common finding in CdLS is congenitally dislocated radial heads, simply said, the two bones that make the elbow hinge do not fit perfectly. This is found in 64 percent of patients. This can cause the elbow to be stiff and in most cases it will not extend all the way. This does not require surgery and stretching by a physical therapist will not help. Recent studies of activities of daily living such as using a fork, knife and cell phone have shown that you do not need the entire arc of elbow motion; you can lack 30 degrees of extension and still do all of your functional activities. In other words, a stiff elbow should not stop individuals with CdLS from using their elbows fully. It is possible to detect a dislocated radial head on a physical exam but a radiograph is the gold standard for a definitive diagnosis.

**Hip problems:** Congenital dysplasia of the hip, or an abnormal hip ball and socket relationship at birth has been documented as well in up to 10 percent of individuals with



### CdLS. Treatment

will depend on whether one or two hips are involved, whether the hip is subluxed (slightly out of socket) or dislocated (completely out of socket) and whether the hip can be manipulated back into the socket and maintained there with a cast or not. Depending on the various presentations, different surgical reconstructions are available.

**Feet abnormalities:** Individuals with CdLS might have small feet (90 percent of cases), webbing between the 2nd and 3rd toes (80 percent of cases) or club feet at birth. Small feet do not interfere with walking and need no intervention. Children born with club feet should be initially treated with the Ponseti method. This is a method of casting where children get casted weekly and the cast rearranges the position of the bones in their feet to realign their feet. As in individuals without CdLS, this is the gold standard, followed by surgery only if casting fails. With aging, patients may develop bunions and tight Achilles tendons.

At the CdLS Multidisciplinary Clinic, I've seen a very high proportion of bunions. Just as with bunions in the general population, they should be initially treated with wide toe shoes for comfort. Surgery should only be considered if they become painful or interfere with walking. We have also seen tight Achilles tendons, or tight heel cords, very frequently. The initial treatment once again should be stretching exercises which parents can ideally do daily. Surgery for release of the tendon is again reserved for failure of stretching exercises. You can tell a heel cord is tight if you cannot bend the foot to get it to a flat position, sometimes children with tight heel cords will walk on their toes and their feet are "stuck" pointing down. Despite all these possible findings, the majority of individuals with CdLS will walk independently. Often later than their peers but they will be ambulatory by adulthood.

Treatment for these orthopaedic manifestations of CdLS focuses mainly on maximizing function and preventing deterioration of such function. Physical therapy and occupational therapy can help patients achieve good functional abilities whether they have short, missing or malformed limbs. Stretching and ambulation can help maintain good muscle tone, good flexibility and range of motion. Weight bearing exercises also prevent premature bone loss in the general population and should be recommended for patients with CdLS until we get a better understanding of the pathology of their early bone loss.

Published Fourth Quarter 2013

