



# CdLS Foundation

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

## A Closer Look at Orthopaedic Issues

*Article in Reaching Out Spring '09*

It's not uncommon for children with CdLS to have orthopaedic issues. In many cases, therapy and/or surgery can help minimize, if not correct, the problem. Common orthopaedic issues in children with CdLS may include curving of the fifth finger, small hands, missing digits or limbs, or limitation of elbow motion, as was the case for Anna from Colorado. Five to ten percent of those with CdLS, like Nicole from California, have hip abnormalities. Orthopedic concerns may be addressed once diagnosed by a doctor. As both Nicole and Anna's mothers explain, children with CdLS can live happy, productive lives despite varying degrees of orthopaedic difficulties.

### **Nicole, age 16**

Karen noticed her daughter Nicole's right hip was popping out of its socket whenever she was upset. X-rays revealed that Nicole had hip dysplasia, which is the abnormal formation of the hip joint. The ball at the top of the thighbone was unstable within the socket because the socket was worn down. Nicole was experiencing great discomfort and pain as a result. Additionally, Nicole was continually crying and didn't sleep well.

Nicole had hip replacement surgery at Cedar Sinai Hospital in Los Angeles when she was 14 years old. She was then placed in a spica cast – a special body cast designed to immobilize the hip joints – for six weeks. "Upon removal of the body cast I was told that she would be able to put weight on [her hip] in a week or two," Karen explains. "She was up and running within the hour. The pain threshold for Nicole is unbelievable." Although Nicole still has some hip popping, her pain is eliminated.

### **Anna, age 12**

Years ago, after a "CdLS Lunch and Learn" in Colorado with Dr. Ian Krantz, member of the CdLS Foundation Clinical Advisory Board, Anna's mom Candee learned that all parents should have their child's arms x-rayed to look for bone fusion. If the ulna bones are fused, a child's range of motion in his or her arms may be limited and therapy to increase this range of motion may actually hurt the child. Candee noticed that Anna's elbow had an odd, large bump, which the pediatrician x-rayed.

Anna was diagnosed with radial head dislocation, in which ulna bone is shorter than normal, causing the radius bone to extend past the elbow joint. The extended bone would "lock up" and prevent Anna from fully rotating her arms. "In other words, she couldn't make the palms of her hands flat facing the ceiling without contorting her whole arm and shoulder," Candee explains. Anna's parents opted not to have corrective surgery, since Anna's quality of life didn't seem to be affected and she wasn't in pain. "We were told not to perform any



# CdLS Foundation

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

stretching exercises and let all physical therapists, physical education teachers, etc. know of her condition so they didn't try to force her arms where they wouldn't go," Candee says.

While she will have this physical limitation throughout her life, Anna doesn't allow this condition to slow her down. Although she has some difficulty doing such tasks as combing the back of her hair, brushing her teeth, and washing her back, she does not complain of any pain in her arms and can do everything she wants to do, especially playing ball and riding her bike.

As Anna moves into adolescence, her parents concerns now turn to developing hip issues. "When running track, we noticed Anna would run slightly hunched over and slap her feet onto the ground," Candee explains. "This unusual running position has gotten worse, and during Special Olympics basketball season I began to pay attention to how she walked and have noticed some awkwardness or limping in her gait." Anna's parents are currently waiting to receive a referral for an orthopaedic evaluation.