

CdLS AND CONGENITAL HEART ISSUES

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People with Cornelia de Lange Syndrome are known for their unique outward appearance. Indeed, the characteristic facial features, small stature and limb differences often signal the diagnosis of CdLS. However, what we can't see (namely, the heart) is of utmost importance to the individual's well being. This article details our current knowledge of congenital heart conditions and CdLS.

Occurrence

While the occurrence of heart issues for children with CdLS is higher than their typically developing peers, there have been many advances in the medical field that give us a greater prognosis for treating them than ever before. The likelihood that any baby is born with a heart defect is 8 in 1,000 (or just under 1%). At the 2008 CdLS Foundation National Conference, Dr. Bonnie Lynch presented data showing that 29% of U.S. respondents indicated that their child with CdLS had heart problems/defects.¹ A recent European study found that 45% of patients with CdLS had congenital heart defects.² The exact reason why children with CdLS have a higher incidence of congenital heart disease is not known at this time. Other chromosomal abnormalities (such as Down syndrome) are also known to have a higher incidence of congenital heart defects. Further research will help us determine if specific gene mutations correlate to congenital heart disease within CdLS.

Types of Heart Issues

The kinds of heart issues that children with CdLS may be born with run from mild to severe. One of the most common heart defects seen in CdLS is a ventricular septal defect (VSD). A VSD is a hole in a particular wall of the heart. If the hole is relatively small at the time of birth, it may close up spontaneously without the need for surgery or medications. Larger VSDs require surgery, usually before the child's first birthday. There are other more complex heart defects that have been seen in people with CdLS. Tetralogy of Fallot is one example. This defect requires early surgical intervention. Even then, the surgery does not cure the heart – it only makes it work better than it had. Most patients with Tetralogy of Fallot will have to undergo more open-heart surgeries in the future (perhaps once every 10 years). While the distribution of the types of heart defects that people with CdLS often have is similar to the general population, there is some evidence to suggest that individuals with CdLS are at increased risk of heart valve disease in particular.³ Thirty years ago, there were many heart defects that include a devastating prognosis. In this day and age, surgery affords a much better outcome for patients with congenital heart defects.

What to Look For

Many heart defects will produce signs and symptoms that parents can identify. Babies with heart defects may demonstrate "cyanosis." This is a bluish or pale coloration of the lips, tongue or fingernail beds. It indicates that there is a low level of oxygen in the baby's blood. Low oxygen can be caused by lung problems or congenital heart disease. Most babies with cyanosis are identified before they are discharged from the hospital, but some are missed. A simple test called "pulse oximetry" can verify if the baby's oxygen levels are appropriate or not. Babies with



congenital heart defects may also have fast breathing, tiring or sweating with feedings, and poor weight gain. You should notify your doctor if you notice any of these features in your child. Older children may have a decrease in energy, dizziness, fainting spells, or chest pain. If your child is incapable of performing physical activities that they once enjoyed, this could be a sign of heart disease. Unfortunately, it is sometimes the case that these symptoms (poor weight gain, decreased exercise tolerance) are written off as merely a part of the overall CdLS manifestation, when in reality they are due to an underlying heart defect. It is important to trust your instincts as a parent in this respect. It is also important to note that sometimes even significant heart defects may show no outward sign and routine check-ups are very important.

Who Should Be Evaluated?

Because their risk of congenital heart defects is greater than the general population and because some heart defects have no symptoms, all people identified with CdLS should have an evaluation by a pediatric cardiologist and an echocardiogram performed at the time of their diagnosis. An echocardiogram (ultrasound study) is the best way to determine if a person has a congenital heart defect. It is a painless procedure that takes about 15 minutes to perform. It does not require sedation as long as the patient can lie relatively still. A baby born with suspicions of CdLS should have an echocardiogram performed prior to discharge from the hospital. If you have an older child/adult with CdLS but an echocardiogram was never performed, it is recommended that the individual obtain an echocardiogram now to insure a “silent” heart defect does not exist. Clearly, the sooner a problem is identified, the better the outcome will be.

How Is It Treated?

Treatment of congenital heart defects is dependant on the particular one that is identified. For some mild heart defects (such as a mild valve narrowing), treatment may not be required. These types of defects may require follow-up by a pediatric cardiologist to insure that the defect does not worsen with time. Other types of heart defects may require medication. Medication can sometimes serve as a “bridge” until the body can heal itself. A moderatesized ventricular septal defect is a good example of this. Medications can be used to help the patient along until the hole becomes sufficiently small as to not require further medication or surgery. Some heart defects are amenable to non-invasive procedures. This usually involves placing a catheter (long skinny tube) into a blood vessel in the leg and advancing the catheter to the part of the heart that needs to be fixed. A balloon can be inflated to fix a valve problem, or a “plug” can be inserted to close a hole in the heart. These non-invasive procedures reduce complications and shorten hospital stays. The most serious types of heart defects often require surgery. Surgery can sometimes truly fix the heart defect so that the resulting heart works just as well as if the defect had never existed in the first place. Sometimes, however, surgery can make the heart work a lot better but it cannot restore the heart to complete normality. Further surgeries may be needed as a child ages.

CdLS Characteristics Play a Role

Some of the characteristics of CdLS may play a role in treating congenital heart disease.

- **Slow Growth:** Ventricular septal defects (hole in the heart) make up the majority of heart defects found in people with CdLS.

Many VSDs will spontaneously close in typically-developing people as long as the individual has



normal growth. Because individuals with CdLS don't grow as quickly, a similar sized VSD may require surgery.

- **Nutritional Status:** Because of the myriad of feeding issues encountered in CdLS, nutritional status can sometimes be compromised.

The ability to successfully undergo open-heart surgery can be influenced by the nutritional health of the patient. Other factors of recovery, such as wound healing, may also be affected.

- **Respiratory Illnesses:** After heart surgery, the lungs play an important role in the recovery process. Many people with CdLS have a history of lung illnesses that puts them at increased risk for heart surgery complications. The development of pneumonia after surgery has the potential to severely affect the recovery process.

- **Anesthesia:** An early critical part of an operation is placing a breathing tube for the mechanical ventilator. The facial structure of children with CdLS makes it more difficult to safely place the breathing tube. Recently, evidence has been collected that patients with CdLS may have adverse reactions to certain anesthesia medications.⁴ More research is on-going to help anesthesiologists choose the safest medications for these individuals. Should you have questions/concerns about specific anesthesia for a procedure, please contact the Family Service Coordinators at the Foundation office.

- **Behavior Issues:** Because of developmental delays, some people with CdLS may not fully understand the necessity of medical interventions. Post-surgical management can sometimes be difficult with patients pulling out intravenous tubing or indwelling drains. Surgical wounds need a few weeks to heal and can be compromised by infection if picked at constantly.

Who Should Be Treated?

Most children with CdLS can live well into adulthood. In the past, some have suggested that babies with CdLS should not have heart operations. With improvements in heart surgery and a better understanding of CdLS, it is recommended that all people with CdLS be treated equally to their typical peers. There arises, rarely, a situation where intervention is not recommended. Clearly, these issues are dealt with on a case-by-case basis between the parents and the doctor. However, identifying and fixing congenital heart defects as early as possible may show improvement in other aspects of a child's life.

Our understanding of congenital heart defects continues to improve. Many of the heart operation techniques that surgeons perform today did not even exist ten years ago. There is great hope that more and more non-invasive procedures will replace current open-heart operations. So while it is with heavy heart that a parent hears that their child has a congenital heart defect, the outcome for most of these children is quite good.

Contact the CdLS Foundation at 1-800-753-2357 or familysupport@CdLSusa.org if you have questions or concerns about your child's cardiac health.

Endnotes

¹ Lynch, B Parental Report on Development and Coordination of Care with the Education and Medical System of Children with CdLS. Presentation given on June 26, 2008 at the 2008 CdLS National Conference in Lincolnshire, Chicago.

² Barisic I, Tokic V, Bianchi F, Calzolari E, Garne E, Wellesley D, Dolk H; EUROCAT Working Group. Descriptive epidemiology of Cornelia de Lange syndrome in Europe. *Am J*





CdLS Foundation

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³Oudit GY, Chow CM, Cantor WJ. Calcific bicuspid aortic valve disease in a patient with Cornelia de Lange syndrome: linking altered Notch signaling to aortic valve disease. *Cardiovasc Pathol.* 2006 May-Jun;15(3):165-7.

⁴Lumb AB, Carli F. Respiratory arrest after a caudal injection of bupivacaine. *Anaesthesia.* 1989 Apr;44(4):324-5.

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