CANADIAN CDLS FOUNDATION

# Management and Treatment Guidelines for Cornelia de Lange Syndrome (CdLS)

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It is always best to have information before it is needed. In medicine, we call this preventative care. Having a child with CdLS increases the need for advance information, although the routine preventative care in paediatrics still applies, such as immunizations. For caregivers of children who have CdLS, whether mildly or more severely involved, there are certain medical and behavioural issues about which you need to be informed. It is recommended that diagnostic studies be done at the time of diagnosis or specific ages on all individuals with CdLS. Regular follow-ups with several sub-specialists should occur annually throughout life.

This excerpt from our recent publication in the *American Journal of Medical Genetics* [June, 2007] highlights routine care for people with CdLS needed by specific age groups, including infancy, early and late childhood, adolescence and adulthood. At each age, individuals with CdLS will have specific health care needs, as listed below. We hope that this will be useful, both for your doctors and for you. We are available for you and your doctors for questions or support.

## MANAGEMENT RECOMMENDATIONS FOR INDIVIDUALS WITH CdLS

[American Journal of Medical Genetics, AD Kline, et al., 143A:1287-1296, 2007]

### 1. Infancy and at the time of diagnosis

When the diagnosis of CdLS is considered, a karyotype should be obtained (blood chromosomes should be sent and evaluated), although it will typically be normal. Once the clinical diagnosis has been made, a number of studies and services are recommended:

A. Echocardiogram;
B. Renal ultrasound;
C. Pediatric ophthalmologic evaluation with cycloplegic refraction;
D. **Hearing evaluation** (otoacoustic emissions, or brainstem auditory evoked response if audiology is abnormal);
E. An **upper GI series** to rule out malrotation and reflux – if malrotation is detected, early repair may be indicated;
F. **Evaluation for gastroesophageal reflux disease (GERD)** including pH probe and/or endoscopy, and, if found, treated medically (e.g. prokinetics) or, if that fails, surgically (e.g. Nissen fundoplication, gastrostomy tube);
G. **Developmental assessment** in infancy and continuing every one to three years;
H. **Early intervention services initiated** and continued as long as needs are being addressed or until adulthood;

I. **Growth assessment using appropriate CdLS growth charts** [Kline et al., 1993a]. Treatment with high calorie formulas is often suggested, and may help with weight
J. gain; however, typically, individuals with CdLS appear to grow at their own pace with a high metabolic rate;
K. **Support organization information should be given to the family** whenever a diagnosis is made: in Canada the CdLS Foundation, support@canadiancdlsfoundation.com, web site **www.canadiancdlsfoundation.org.**
L. **Ensure that family has the CdLS Medical Care Card**, available from the CdLS Foundation Web site, which would be helpful in an emergency situation (e.g. risk for volvulus);
M. Consider available **molecular testing** if parents are interested in further pregnancies and prenatal diagnosis options. Refer for genetic counselling if contemplating these prospects.

## 2. Early Childhood (one to eight years old)

An individual with CdLS should have regular evaluations and immunizations with the primary care provider and:

A. In males, **cryptorchidism (undescended testicles) should be repaired** by 18 months;
B. Ongoing developmental services, with school placement and therapy issues individualized. It is likely that most individuals will benefit from **physical, occupational and speech therapy**. The use of sign language is encouraged since this will help facilitate oral communication;
C. Continue to **monitor growth via CdLS-specific growth charts**;
D. **Pediatric dentistry**, or dentist familiar with patients with special needs, every six months;
E. **Pediatric ophthalmology evaluation** once or annually, as indicated by findings on first examination;
F. **Audiology testing** every two to three years;
G. With any clinical suspicion of worsening or initial signs of **GERD, a repeat evaluation** should be performed. Endoscopy will often have the greatest yield, but pH probe could be considered;
H. **Any sign of potential volvulus** (e.g. bilious emesis [vomiting bile] or bilious withdrawal from gastrostomy tube, sudden acute abdominal pain) should merit an **immediate visit to the emergency room**, work-up and potential surgery;
I. **Follow-up** with appropriate sub-specialists as needed;
J. Whenever any surgery is performed, all involved specialists should be consulted in order to **maximize the use of anesthesia** and so that the individual can undergo diagnostic or management studies as needed at the same time.
K. See 1.j-l. from “Infancy” recommendations.

### 3. Late Childhood (eight years – puberty)

An individual with CdLS should have regular care through the primary care provider and:

A. **Orthopaedic involvement** may be needed for joint contractures, hip complications bunions, development of scoliosis, or orthotic use;
B. **Behavioural assessment** if issues arise, including ADHD, self-injurious behaviour;
C. See 2.b-k. above.

### 4. Adolescence (puberty – 20 years)

An individual with CdLS should have regular care through the primary care provider and:

A. **Ongoing developmental services.** School placement and therapy issues should be individualized. Plans should be initiated early for school or workplace placement after high school, job training and/or higher education;
B. For females, consider **pelvic examination with Pap smear** regularly, at least every three years, depending on sexual activity, from late adolescence throughout adulthood. Discuss hormonal therapy with patient and family, both from the pregnancy prevention point of view, and management of menstruation (individualized to specific patient and family);
C. **Discuss recurrence risks** if developmentally appropriate;
D. See 3.a-c. above

### 5. Adulthood

An adult individual with CdLS should have regular evaluations with primary care provider and:

**A. Follow blood pressure, consider baseline EKG, routine breast, or testicular and prostate examination** as per usual medical guidelines;
**B. Discuss job training or work issues, higher education**
**C. Behavioural or psychiatric assessment** if issues arise, including ADHD, obsessive compulsive symptoms, self-injurious behaviour, depression;
**D.** Consider **DEXA scan** to rule out osteoporosis;
**E.** See 4.b-d. left. **Dental evaluation** should be every four to six months, depending on compliance, ideally with paediatric dentistry, or dentist familiar with patients with special needs.

**Contact the Canadian CdLS Foundation at support@canadiancdlsfoundation.com with any questions or concerns.**