CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

Management and Treatment Guidelines for Cornelia de Lange Syndrome

These cards highlight 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.

Please contact
the CdLS Foundation
at 800-753-2357
with any questions
or for more information.

OUR MISSION: The Cornelia de Lange Syndrome Foundation is a family support organization that exists to ensure early and accurate diagnosis of CdLS, promote research into the causes and manifestations of the syndrome, and help people with a diagnosis of CdLS, and others with similar characteristics, make informed decisions throughout their lifetimes.

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CORNELIA dE LANGE SYNDROME MANAGEMENT AND TREATMENT GUIDELINES

Based on 2007 Management Recommendations for Individuals with CdLS.*

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:

- Echocardiogram
- Renal ultrasound
- Pediatric ophthalmologic evaluation with cycloplegic refraction
- Hearing evaluation (otoacoustic emissions, or brainstem auditory evoked response if audiology is abnormal)
- An upper GI series to rule out malrotation and reflux – if malrotation is detected, early repair may be indicated.
- 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).
- Developmental assessment in infancy and continuing every one to three years.
- Early intervention services initiated and continued as long as needed.
- Growth assessment using appropriate CdLS growth charts [Kline et al., 1993a]. Treatment with high calorie formulas is often suggested, and may help with weight gain, however, typically, individuals with CdLS appear to grow at their own pace with a high metabolic rate.
- Support organization information should be given to the family whenever a diagnosis is made: the CdLS Foundation, 1-800-753-2357, www.CdLSusa.org.
- 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).
- Consider available molecular testing if parents are interested in further pregnancies and prenatal diagnosis options. Refer for genetic counseling if contemplating these prospects.

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

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Early Childhood (one to eight years old)

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

- In males, cryptorchidism (undescended testicles) should be repaired by 18 months.
- 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.
- Continue to monitor growth via CdLS-specific growth charts.
- Pediatric dentistry, or dentist familiar with patients with special needs, every six months.
- Pediatric ophthalmology evaluation once or annually, as indicated by findings on first examination.
- Audiology testing every two to three years.
- 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.
- 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.
- Follow-up with appropriate subspecialists as needed.
- 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.
- Support organization information should be given to the family whenever a diagnosis is made: the CdLS Foundation, 1-800-753-2357, www.CdLSusa.org.
- 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).
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Late Childhood (eight years – puberty)

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

- Orthopedic involvement may be needed for joint contractures, hip complications, bunions, development of scoliosis, or orthotic use.
- Behavioral assessment if issues arise, including ADHD, self-injurious behavior.
- 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.
- Continue to monitor growth via CdLS-specific growth charts.
- Pediatric dentistry, or dentist familiar with patients with special needs, every six months.
- Pediatric ophthalmology evaluation once or annually, as indicated by findings on first examination.
- Audiology testing every two to three years.
- 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.
- 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.
- Follow-up with appropriate subspecialists as needed.
- 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.
- 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).
- Consider available molecular testing if parents are interested in further pregnancies and prenatal diagnosis options. Refer for genetic counseling if contemplating these prospects.

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Adolescence (puberty – 20 years)

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

• 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.

• 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).

• Discuss recurrence risks if developmentally appropriate.

• Orthopedic involvement may be needed for joint contractures, hip complications, bunions, development of scoliosis, or orthotic use.

• Behavioral assessment if issues arise, including ADHD, self-injurious behavior.

• 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.

• Continue to monitor growth via CdLS-specific growth charts.

• Pediatric dentistry, or dentist familiar with patients with special needs, every six months.

• Pediatric ophthalmology evaluation once or annually, as indicated by findings on first examination.

• Audiology testing every two to three years.

• 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.

• 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.

Continued on back

- Follow-up with appropriate subspecialists as needed.

- 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.

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Adulthood

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

- Follow blood pressure, consider baseline EKG, routine breast, or testicular and prostate examination as per usual medical guidelines.
- Discuss job training or work issues, higher education.
- Behavioral or psychiatric assessment if issues arise, including ADHD, obsessive-compulsive symptoms, self-injurious behavior, depression.
- Consider DEXA scan to rule out osteoporosis.
- Dental evaluation should be every four to six months, depending on compliance, ideally with pediatric dentistry, or dentist familiar with patients with special needs.
- 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).
- Discuss recurrence risks if developmentally appropriate.
- Orthopedic involvement may be needed for joint contractures, hip complications, bunions, development of scoliosis, or orthotic use.
- 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.
- Continue to monitor growth via CdLS-specific growth charts.
- Pediatric dentistry, or dentist familiar with patients with special needs, every six months.
- Pediatric ophthalmology evaluation once or annually, as indicated by findings on first examination.
- Audiology testing every two to three years.

Continued on back

• 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.

• 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.

• Follow-up with appropriate subspecialists as needed.

• 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.

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