CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

Dear Primary Care Provider:

This letter is written from the Cornelia de Lange Syndrome (CdLS) Foundation in conjunction with the pediatric gastroenterologists on the CdLS Clinical Advisory Board. I am writing to you with our gastrointestinal evaluation recommendations. This is an area affected in CdLS with potentially very serious complications and sequelae related to other body systems.

It is important that children with CdLS be evaluated and followed at some regular interval for gastroesophageal reflux (GER), preferably by a pediatric gastroenterologist. At the minimum, any child with CdLS and GER needs an upper-gastrointestinal series to evaluate for malrotation, a congenital abnormality of bowel rotation that can be "silent" until an acute presentation where the bowel twists on its vascular pedicle, resulting in necrosis of the intestine, a life-threatening emergency, and a not infrequent cause of mortality in CdLS. There are a variety of different gastrointestinal tests, which may be used to evaluate for GER (and even other intra-abdominal problems) including: contrast studies (upper GI, small bowel follow-through, barium enema), endoscopy, pH probe (both on and off medications) (often poorly tolerated in CdLS), and various nuclear medicine studies.

It is best that the use of these tests be tailored to the individual child, depending on the present symptoms and past findings. These decisions are best made jointly by the doctor and parents or guardians. Likewise, it is sometimes the best decision to first consider a trial on a reflux medication, and then to reassess for any improvement in symptoms; however, this can be more difficult and subjective in the setting of silent reflux, which occurs not infrequently in CdLS.

As far as the interval between appointments, I would recommend an appointment with a pediatric gastroenterologist at the time of the initial CdLS diagnosis. Although none of the pediatric gastroenterologists on the Clinical Advisory Board for the Foundation have recommended routine annual gastrointestinal exams for all children, even if asymptomatic, we probably should be seeing even the mildest individuals every few years to evaluate for symptoms of silent reflux. Pediatric gastroenterologists are familiar with silent reflux, and they will evaluate for the presence of subtle symptoms (such as arching, behavioral problems, hoarseness, respiratory problems, poor growth relative to the CdLS growth curve, etc.) while speaking with the family and evaluating the child.

Of course, follow-up will need to be more frequent if the child has symptoms, significant findings on any of the studies (such as an ulcer), or if the child is prescribed any gastro-intestinal medications. Follow-up could be more infrequent if prior evaluations were negative for reflux and the child remains asymptomatic off of all gastrointestinal medications, including the over-the-counter gastrointestinal medications.

Older teenagers and adults with CdLS who have either not been evaluated or were managed less-aggressively for their reflux; should have an assessment for reflux, such as an upper endoscopy. This is particularly important since there are a few cases of Barrett's esophagus (a pre-malignant change) in CdLS reported in the literature, and at least 2 cases of esophageal cancer in CdLS adults in their 20s. Once an endoscopy has found evidence for severe changes (e.g. ulceration or stricture) or Barrett's esophagus, a follow-up endoscopy should be determined clinically until stable. If improved or no change, then this can be spaced out to every three years or so.

Please do not hesitate to contact me with any questions or concerns. Thank you for your attention to this matter.

Sincerely,
Antonie D. Kline, M.D., CdLS Foundation Medical Director

Antonie D. Kline, M.D.
Director of Pediatric Genetics
Greater Baltimore Medical Center
6701 North Charles Street, Ste. 2326,
Baltimore,MD 21204