



CdLS Foundation

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

CdLS and the Eye

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CdLS is a multisystem, multifocal genetic anomaly. Major characteristics of CdLS include delayed growth and development, as well as skeletal and facial abnormalities. In this article we review a number of the facial and ocular (eye) findings in CdLS.

External Characteristics

The typical CdLS face demonstrates a number of characteristics involving the eye – in fact, the findings involving the eye are the most consistent clinical features of CdLS.

In CdLS, the eyebrows are fused centrally to create a single continuous brow. Often the central fused area has a v-shaped dip. The fused brow is medically called a *synophrys brow* and is seen in 99% of people with CdLS. The eyebrows, as well as the supporting bone beneath the brow, function to protect the eye. It is similar to the way in which the roof of a house extends beyond the walls in order to protect the house from falling debris (branches, etc.). The central fused area does not interfere with the eyebrow's function (to protect the eye itself) so we are left with only a cosmetic issue. Some families choose to shave or epilate this area and this should be considered as an optional grooming issue.

Children with CdLS not only have extra brow hair but generally have more hair on their bodies. *Hirsutism* is the term for excess body hair. The eyelash hairs are unusually long with a smooth curved shape. The elongated lashes are a highly consistent clinical finding seen in 99% of patients with CdLS. This is merely a cosmetic issue if the long lashes are in a proper orientation from the eyelid. However, they sometimes grow sideways or even backwards into the eye. This can produce irritation, abrasions (scratches) on the cornea, and may lead to scarring and damaged vision. The irritation will manifest itself as a red eye with discharge on the lid and lashes. Its significance could be just cosmetic (with messy lids and lashes), to mildly medically important (due to pain and irritation), to urgent (due to possible corneal ulceration). Depending on the specifics of each case, treatment might require antibiotic drops, mechanical removal of the misdirected eyelash, or surgical rotation of the lid margin.

Blepharitis

Commonly, the lids are inflamed and/or matted due to inflammation of the 40 or so oil glands of each lid. This is not terribly different from teenage acne except that it is in a localized area. Warm compresses are soothing for this type of irritation. Diluted baby shampoo can be used as a scrub to remove the debris and excess oil and to make the area less hospitable for bacteria. Sometimes a topical antibiotic is required. Control of the problem, rather than a cure, is the primary goal. Blepharitis often improves with age.



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Crossed Eyes

Normally both eyes are precisely aligned in the same direction to simultaneously target the same object. Failure to do so will send different sets of signals to the brain and result in double vision. This is obviously unpleasant and sometimes the brain will ignore one image to avoid double vision (diplopia). The unused eye becomes lazy (amblyopic) and over time may be unable to work even though it is a physically normal eye.

The eyes could be malpositioned inward (esotropia) or outward (exotropia). Treatment goals are to reestablish bilateral, simultaneous vision to enhance depth perception and to improve cosmetic appearance. The cosmetic benefit in these cases may be more important to development than the technical goal of stereoscopic vision. Treatment methods include glasses, surgery, and occasionally eye exercises. Typically, children with CdLS reject eye glasses.

Nystagmus

Nystagmus is a condition in which the eyes may be aligned adequately but the guidance system for smooth tracking motions is defective. The eyes oscillate (wiggle) back and forth in a horizontal or vertical pattern. This is more noticeable and distressing to the observer than to the patient. Think of your own head and eyes as you drive down a very bumpy road; you bounce but your image of objects outside the car remains stable. Unless there is a specific, identifiable cause, this may be secondary to brain development and no therapy is suggested.

Ptosis

Ptosis (pronounced toe-sis) is a malposition of the upper or (rarely) the lower lid resulting in droopy lids. It is found to some degree in almost half of all patients with CdLS. It is often mild and limited to a cosmetic effect. A severe case of ptosis, which allows the upper lid to fully block the pupil of the eye, can prevent the image from getting to the back of the eye and then to the brain. This could result in failure of the brain to develop its visual pathways – just like the amblyopic (lazy) eye mentioned in the section on crossed eyes. If the ptosis produces partial or intermittent interference, the child may develop a backward head tilt or chin lift in order to see from under the drooped lid. A slight head tilt is a cosmetic issue. A pronounced head tilt can alter posture/balance and interfere with walking. Treatment for ptosis would be surgical and may be advised for both eyes even if only one is drooped.

Cornea

The cornea is the anterior layer of the actual eye. It is normally clear and covers the blue or brown iris and black pupil. Its function is similar to the crystal in your wristwatch. It protects the tissues behind it and focuses light. Recent reports suggest the corneal diameter is smaller than average in children with CdLS. Issues involving the cornea include scarring from misdirected lashes. Treatment of the lash problem should prevent further scarring.



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Lens and Vitreous

The lens and vitreous comprise the bulk of the middle of the eye. The vitreous is the jelly-like substance that fills the inside of the eye and helps to keep the shape of the eye. The light path to the retina is through both of them. They usually remain healthy and clear in patients with CdLS and don't typically cause problems.

Retina

The retina is the light sensitive tissue that transmits a signal to the brain. Usually, the retina of patients with CdLS remains structurally healthy. However, because some patients with CdLS are very nearsighted (myopic) they may be at risk for retinal detachment. This condition may occur rarely in patients with very high myopia, but may be a bit more common in patients who have trauma to the eye. Some children with CdLS exhibit self-injurious behavior, either head banging or hitting themselves in the head and face. That behavior increases risk for retinal detachment. When a detachment occurs, the eye does not see well. It would be difficult, however, for a caregiver to detect a detachment because most patients with a detachment in one eye will continue to use the other eye to navigate. Surgery may be required to repair a detachment if it is found, however some retinal detachments cannot be repaired. Regularly scheduled eye exams can help to detect this problem.

In summary

Each person with CdLS potentially has a unique mix of ocular issues. Some are curable, many are controllable. Periodic eye exams by an empathetic ophthalmologist (a pediatric ophthalmologist for anyone under 18) and a prepared