

Common Eye Disorders in CdLS

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Eye disorders are a common manifestation in CdLS. Although external eye findings, such as the shape of the eyelids, long eyelashes and eyebrows that meet in the center (synophrys), are the most consistent facial signs of CdLS, none affect vision function. The most significant impacts to sight and behavior are ptosis (pronounced toe-sis) and severe nearsightedness.

Ptosis is mild to severe drooping of one or both eyelids, affecting approximately 44 percent of patients with CdLS. The most common cause is a weakness of the upper eyelid muscle that is present at birth (congenital ptosis). Normal functioning lids protect the eyeball. Similar to a windshield wiper, blinking lubricates and cleans the front surface of the eye. Significantly weak and drooping lids can block vision and lead to poor visual development amblyopia (lazy eye) in one or both eyes.

If vision is compromised by a droopy eyelid, the muscles in the scalp and brow are used, subconsciously, to help raise the lid. A child's head posture can change from lifting the chin, which clears a light pathway for the child to see from under the droopy lids. In some cases, children tilt their heads so far backwards that they fall over when trying to look upward. Although uncommon, ptosis can interfere with walking or can impair interaction with the environment.

There is no magic age for ptosis surgery, and while mild ptosis—defined as intermittent blockage—does not require surgery, it may be done to normalize the child's appearance. The decision to go forward with surgery should be based on whether or not vision is being blocked (especially when only one eye is being blocked and the child, six years old or less, is in the early years of vision development) and whether walking and interacting with the environment is affected. If there is severe blocking of vision in both eyes, surgery can be done in infancy.

In a study of patients with CdLS, only four of 117 had a ptosis repair. In a more recent study, none of 41 individuals required ptosis treatment.

Nearsightedness (myopia) is seen in almost two thirds of patients with CdLS. Being mildly or moderately “sighted for near” is actually an advantage for children at chronologic or developmental ages of under two as they live in a “near” world. As children get older and



mature, they become more interested in the “far” world and may benefit from glasses. Unfortunately, about half of the children with CdLS who might benefit from glasses won’t wear them. It may simply be an aversion to having something on or near their face or they may not recognize the difference in their visual world enough to give them incentive to wear the glasses.

High degrees of nearsightedness (more than -6.00) can result in an elongated (egg shaped) eyeball. This causes the inner lining of the eye—a tissue called the retina—to be stretched. The retina is like film in a camera, capturing pictures of the world and sending them to our brain to create vision. A stretched retina can develop tiny thin spots that may lead to holes that allow fluid to enter and separate the retina. This is called a retinal detachment.

Retinal detachment can damage sight and even cause blindness and requires immediate surgical repair. Fortunately, it is rare in patients with CdLS. Head or eye trauma from self-injurious behavior can also cause retinal detachment, even if the patient is not nearsighted. Early detection of retinal detachment is a challenge, particularly in young children or older patients with significant developmental delays. Since normal vision allows the image of the right and left eyes to almost completely overlap, even typical adults can fail to notice the loss, or partial loss, of vision in one eye. A child or individual with developmental delays may not recognize vision changes from one eye at all.

Changes in behavior, increased clumsiness and difficulty locating objects on one side may be the best clues. Periodic eye exams are critical. For patients with high myopia or self-injurious behavior, every six months is recommended.

We encourage you to discuss any vision concerns with your child’s pediatrician or ophthalmologist.

Blepharitis

Recurrent red eye, eye discharge, tearing, crusty eyelashes, styes, blinking, and/or eye rubbing are often caused by blepharitis, a condition in which the eyelid glands that lubricate the eye flow slowly. While blepharitis is common in the general population, it's even more common in individuals with CdLS.

The best treatment is washing the eyelash are with baby shampoo once or twice daily. Your doctor can show you how to do this. If this treatment fails, then other causes, such as an infection or blockage of the tear ducts (nasolacrimal duct obstruction) should be explored.

