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 or the brows are arched in a way that they almost resemble a pencilled in arched brow. Often the central fused area has a v-shaped dip. The fused brow is medically called a synophrys brow and is commonly seen in individuals 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 over 90% 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 mattered 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.

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Sometimes a topical antibiotic is required. Control of the problem, rather than a cure, is the primary goal. Blepharitis often improves with age.

Crossed Eyes

Typically 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.

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.

In the case of strabismus in CdLS, strategies for the general population should be followed.

Some individuals with CdLS have been reported to have abnormal optic nerves (Nallasamy, S. et al. Ophthalmologic findings in Cornelia de Lange syndrome: a genotype-phenotype correlation study. Arch. Ophthalmol. 124, 552–557 (2006). Another finding is a ring of pigment found around the optic nerve, seen on an eye exam in over 80% of children with CdLS, although this does not cause any harm. There is risk for retinal detachment in CdLS, either due to very severe near-sightedness or self-injurious behaviour relating to poking the eye.

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.

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

Cerebral/Cortical Vision Impairment (CVI)-What You Should Know

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This article expands on the March/April 2005 article (on "CdLS and The Eye" written by Drs. Berger, Schloff and Schoedel, describing the implications of many structures of the eyes and various conditions common to persons who have CdLS) in regard to the educational, communication, and social aspects related to visual functioning. "CVI" or "cortical/cerebral visual impairment" is an often overlooked condition that affects many children with CdLS, impinging on not only their vision, but also their ability to learn and interact with their environment.

Vision is complex. It requires:

(1) the ability of the eyes to receive visual information,
(2) the brain's ability to process the visual information in combination with and at the same time as other sensory information and
(3) motivation to use the visual system.

All people with CdLS should have examinations by an ophthalmologist or optometrist to determine, as much as possible, the health of the eyes, how clearly they are able to see visual information and/or if they have conditions that may be relieved by surgery, glasses and/or other means. It is the eye that takes in the visual information.

However, it is the brain that processes the information and allows us to understand the visual messages the eyes receive. Understanding all the information the eyes receive may be difficult for some persons because it takes many areas of the brain, working as a team, to process such information. Thus, a major challenge for many people is in understanding the visual information.

Finally, effective use of the visual system requires motivation. It is a lot of work to:

(1) determine what to look at from the vast array of visual images that bombard us almost every waking hour and
(2) coordinate the movements of the ocular muscles in the right direction, track and scan. The most critical aspect, as related to motivation, is the desire to learn from and socially interact with the environment.

Understanding Visual Information

There are some persons with CdLS who have visual behaviours similar to those individuals who have been diagnosed with cortical/cerebral visual impairment – otherwise referred to as CVI. CVI is a disability resulting from either an insult to the brain or how the brain was configured during prenatal development. CVI affects how the brain processes the visual information received by the eyes. Many persons with CVI display wide variations in the functional use of their vision, not just day-to-day but also minute-to-minute.

People with CVI also tend to display some/many of the following behaviours (very few persons have all these behaviours):

- Great variability in how they use their eyes from being visually attentive at one moment and visually inattentive at another moment. May appear to stare into space.
- A tendency to use peripheral vision more than central vision. The midline is rarely the preferred gazing orientation.
- Limited visual attention and a seeming lack of visual curiosity about their environment/ surroundings.
- Great difficulty in looking at crowded or complex visual displays for informational purposes.
- A tendency to want to get close to the object(s) they are viewing either to magnify the image or to reduce the complexity of what they are looking at.
- A tendency to be light sensitive and/or a tendency to gaze at lights. Some individuals gaze at light even though they are light sensitive.
- A tendency to look away when reaching.
- A tendency to avoid looking at the human face OR a tendency to stare at the human face OR a tendency to stare at just one part of the face.
- Difficulty in managing multi-sensory demands and planning/implementing motor responses; relying mostly on other senses for help/information.
- Possible depth perception difficulties which may affect accurate reaching for objects and/or mobility.
- Many people with CVI have difficulty in making the quantum leap of learning from three-dimensional objects to two-dimensional line drawings or photographs, except when trained picture-by-picture. Even then, it is difficult for many to generalize the understanding of these pictures to different environments and situations.
- For many who are print readers, there may be inconsistent difficulties interpreting print, a need for larger and bolder size print even though visual acuity is near-normal, and a short attention span when using print materials. The latter frequently is due to visual fatigue as a result of the effort to see and interpret the crowded visual symbols. The major difference between readers with CVI and readers with learning disabilities is that the former is inconsistent. At one moment the person may be able to read well and in the next moment, not be able to read at all. With learning disabilities, there is more consistency in the type of problems related to reading.
- Difficulty in using vision when moving OR difficulty when the object is moving. Some people may have difficulty when there is no movement. This may make moving around in the environment difficult.
- Difficulty in discriminating between and identifying people based solely on vision, relying on the other senses for information. This is called prosopagnosia or, in its more extreme form, facial agnosia.

Numerous other possible behaviours may be associated with CVI including difficulty in understanding the full range of language used by others as well as the language they, themselves, use. Such communication difficulties may range from people who understand only the emotional and melodic aspect of language to other people who may talk a lot but have significant difficulty in understanding the social aspect of the verbal exchange.

<u>The Implications of Cortical/Cerebral Visual Impairment (CVI) for Students Who Have CdLS:</u>

A primary implication of CVI is the confusion that is created for families and professionals unfamiliar with this condition when visual interest is not obvious or is demonstrated only occasionally. Such confusion is even greater when the medical eye examination is relatively normal. Such variability in visual responsiveness may be highly dependent on the person's level of fatigue, stress, medication, motivation for the task, competing sensory demands, position, and motor requirements of the task. Visual behaviours also may be highly influenced by physical challenges that do not allow the person easy visual access to the environment and by the nature of a seizure condition, should that exist.

Another primary implication of CVI, especially in relating with students who have CdLS and do not talk, is the monumental task in determining what type(s) of communication systems would be the most appropriate. It must be understood that the use of line drawings (e.g., Mayer-Johnson), photographs and/or print assumes the ability to interpret abstract visual symbols. As part of this consideration, it is critically important to remember that when a person looks at something, the visual gaze does not automatically translate into understanding (although it does show interest). The difficulty in understanding may be due to a lack of appropriate visual experiences, a cognitive status that does not support understanding two-dimensional abstract representation or a form of CVI. Thus, when planning what type of communication system might be introduced to a student who does not talk, the following must be considered:

- Does the student demonstrate many of the behaviours described previously that might suggest there is a possibility of some degree of cortical/cerebral visual impairment?
- Does the student consistently demonstrate knowledge of object function and anticipation of the subsequent activity based solely on looking at an object that is an integral component of the activity? If the student cannot visually interpret the three- dimensional world (e.g., objects), how can the student be expected to interpret the two-dimensional world (e.g., pictures, line drawings)?
- Does the student prefer to use objects to communicate while rejecting the use of pictures, line drawings and/or communication devices (for more than cause & effect purposes)?

Finally, the diagnosis of CVI is not restricted to persons who have severe and obvious physical and/or cognitive challenges. Nor is CVI restricted to persons who do not talk or those who have acuity measurements in the visual impairment range. Recent research has demonstrated that CVI also may be a "hidden disability" for many walking, talking academic students who also have near-normal visual acuity. Diagnosing CVI is difficult when there is no focal brain insult. The best diagnostic methods to determine CVI include the medical eye care examination and systematic observations of functional visual behaviours by families and involved professionals. It is especially helpful to have a certified teacher of the visually impaired, who has training and experience with cortical/cerebral visual impairment (CVI), as part of the diagnostic team. Many students, with and without CdLS, have been given visual communication systems without appropriate and thorough observations of their understanding of visual information. Such decisions may have a life-long impact on the educational, communication, and social aspect of these students' lives.

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