

## EYE CONCERNS IN THE ECTODERMAL DYSPLASIAS

The ectoderm (or outer layer) of the embryo develops into many parts in and near the eye: eyebrows, skin of the eyelids, eyelashes, tear glands, conjunctiva, cornea, and lens, as well as most structures of the face. All these structures may be normal in individuals with Ectodermal Dysplasia, or one or more tissues may develop or function abnormally. Scanty, short, or fragile eyebrows and eyelashes may present a cosmetic concern, but may also cause irritating debris in the tear film. In some forms of ED, because the skin may be unusually thin and because the skin of the eyelids is already among the thinnest skin of the body, a darkness caused by the blood vessel patterns (usually veins) in the deep tissues may cause some people to use cosmetics.

In some ED's the eyelid margin may be loose, while in other forms, eyelids may be tight and scarred. If the eyelids are tight either because of excessive retention of the outer layers of the skin's surface as seen in some forms of ichthyosis, or if there is scarring and vertical shortening of the eyelid skin, the eyelids do not close on casual reflex blinking or when the eyes are at rest (while asleep, for example). Careful attention must be directed to lubricate the cornea and the conjunctiva. Exposure caused by failure of the eyelids to close quickly permits drying of the outer surface of these tissues and encourages infection or scarring. Lubricating sterile ointments (e.g., Lacrilube® or Duratears®) or artificial tear supplements may be necessary as much as several times an hour to protect the ocular surfaces and at bedtime. Rarely, if the eyelids are lax or floppy, or if the lower eyelid curls outward away from the surface of the eye, then a surgical procedure ("tarsorrhaphy") may be done to support the eyelid and close the eyelid.

Sometimes eyelashes become misdirected and curl inward and scratch against the conjunctiva and cornea. These aberrant lashes can be removed, but relief is usually temporary. As soon as the lashes regrow, they may almost always be misdirected again. Misdirection of the lashes of this type can be repaired permanently by freezing the abnormal lashes, by burns with a laser, or by electrolysis.

Dry Eye Syndrome (caused either by insufficient tear volume or by inadequate tear components, such as underproduction of oil or mucus in the tear film) leaves the surface of the eye irregular and irritated. The frequent use of artificial tears (these may be purchased without prescription) or holding a warm compress to the closed eyelids often will provide comfort. Sometimes, obliteration of the puncta, the "drains" near the inside corner of the eyelids that conduct tears into the nose, must be closed. The simplest way to do this semi permanently is with silicone plugs; alternatives include cautery or laser to scar the puncta closed. Whenever possible, use the plugs, as they can always be removed if circumstances change.

If a thick, purulent discharge or crusting of the eyelashes occurs, then this should be treated with an appropriate antibiotic. The occurrence of corneal opacities sufficient to affect vision is uncommon, but if it should develop, then visual improvement would be expected following corneal transplant. However, if the scarring has resulted from corneal drying or from exposure and poor blinking, the success of corneal transplant surgery is very limited. If a cataract develops prematurely, then cataract surgery may return vision to pre-cataract levels. However, restorative procedures including artificial lens implants are substantially different and more complex in children and adolescents than in older adults; therefore, anyone contemplating cataract surgery should discuss carefully and in great detail the risks and complications of the specific procedure(s) proposed with the surgeon well in advance of any decision to undergo the surgery.

Many of the eye findings seen in young individuals affected by ED occur frequently in non-ED individuals as they get older. Dry eyes, thin eyelid skin, misdirected eyelashes, diminished tears, and cataracts are common events in an older adult population. These same treatment options are used by many people after age 50-60 years.

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