

# How Ectodermal Dysplasias Affects Eyes and How to Treat

Syndrome	Eye Problem
Anykyloblepharon-ectodermal dysplasia-cleft lip and/or palate (AEC) syndrome	Fused eyelids, chronic epiphora, corneal opacities, photophobia, ectropion of eyelids or the lacrimal papillae
Clouston Syndrome	Absent 2/3 of eyebrows, underdeveloped or missing eyelashes
Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome	Deficient tears, recurring infections, corneal scarring, absent lacrimal punca or drainage apparatus in the nose.
Hypohidrotic ectodermal dysplasia (HED)	Absent or underdeveloped eyebrows and eyelashes, deficient tears, photophobia
Hypohidrotic ectodermal dysplasia with corneal dysplasias	Corneal opacities
Hypomelanosis of Ito	Myopia, microphthalmia, strabismus,iris heterchromia, retinal pigmentary abnormalities, optic atrophy
Incontinentia Pigmenti	Strabismus, nystagmus, retinal changes similar to retinopathy of prematurity, leading to retinal detachment, secondary cataract, and microphthalmia (small eyes)
Marshall Syndrome	Juvenile onset cataracts, myopia, fluid vitreous, and increased risk for retinal detachment
Oculodentodigital syndrome (ODD)	Microphthalmia, narrow lid fisures, epicanthal folds, microcornea, strabismus, optic atrophy, congenital cataract, glaucoma, persistent pupillary membrane, coloboma of the optic nerve head

## Eye Problems in Ectodermal Dysplasias

In most individuals affected by the ectodermal dysplasias, eye and vision problems are no different than those in the general population. Many eye problems relate to the altered tear function or decreased tear supply or to blepharitis (smoldering infections in the oil glands of the eye lashes).

However, an ophthalmologist (a physician with specialty experience in the breadth of disorders of the eye and vision) should perform a complete medical eye examination if vision appears to be abnormal. Cataracts, corneal scarring, infections, and retinal changes may cause visual problems in some of the ectodermal dysplasias. An ophthalmologist must evaluate, diagnose, and treat these problems.

## Causes of Increased Sensitivity To Light

Photophobia or sensitivity to light or painful aversion of bright sunlight has several causes: dry corneal surfaces caused by insufficient tears, corneal abrasions,



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and blepharitis. Abrasion of the cornea may be caused by distichiasis (eyelashes that grow in an abnormal direction and then touch and scratch the cornea). These abnormal eyelashes may be removed to provide comfort and prevent recurrence. Removal may have to be repeated as new, abnormal eyelashes may grow in quickly. After repeated removal of aberrant lashes in the same area, the ophthalmologist may use electrolysis or a laser or freezing probe to “kill” the lash follicle to prevent those lashes from growing again.

Tears that normally lubricate the eyes consist of a watery component from the lacrimal gland, a mucous protein from cells in the conjunctiva, and oil from the glands in the eyelids. Any one or all three parts may be altered in the ectodermal dysplasias. Inadequate tears, which do not lubricate the surfaces of the eye and the linings of the lids, will cause drying and result in small abrasions, irritation, pain, and aversion to light.

Drops of artificial tears placed in the eyes several times a day usually provide relief. A non-prescription eye ointment, such as DuraTears or Lacrilube, may be appropriate at bedtime because these products provide a sterile grease that, when melted, lubricates the eye but does not evaporate.

Another treatment is to plug the tiny ducts in the corners of the eyes that normally drain tears; this allows salvage of the sparse tears. It is recommended that

dissolvable collagen plugs be used first as a trial. If they are successful, “permanent” silicone plugs may be put in. These plugs may improve poor tear volume substantially by “salvaging” what tears are made. Generally, however, “permanent” closure of the tear drain (called the punctum) with a laser or freezing is not recommended.

Infection of the eye causes red eyelids, red conjunctiva, pus, and crusting of the eyelashes, and may be accompanied by aversion to bright light (“photo aversion”). An infection should be evaluated and treated promptly by a physician (ophthalmologist).

Proper hygiene may include daily hot, moist compresses to soften the crusting and then scrubs with baby shampoo to remove debris at the base of the eyelids. Occasionally, antibiotic eye drops may be used, but the chronic and continuing use of antibiotics is NOT appropriate, since the bacteria often become resistant to continuing use of the same antibiotic for an extended time.

### Discolored Eyelids

Mild abnormalities of skin pigmentation in ectodermal dysplasias are seen more easily in the thin skin of the eyelids. This “pigmentation” is really seeing the blood within the blood vessels through the thin lid skin and underlying tissues and is only an esthetic problem that may be covered with makeup.

*The content of this document is for informational purposes only. Questions regarding specific patient issues should be directed to the appropriate professionals for resolution.*