

Eye Problems & Ectodermal Dysplasias

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Introduction

The ectodermal dysplasias are a broad group of disorders that show abnormal development of the ectodermal layers of the developing embryo. The ectoderm of the embryo forms the hair, teeth, nails, sweat glands, and some parts of the eyes.

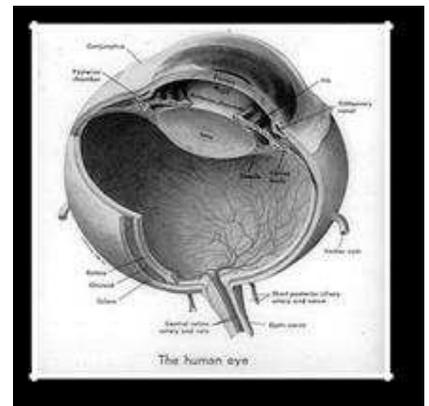
To understand the eye problems in ectodermal dysplasias, you must understand the basic structures and function of the eye. First, let's briefly describe the anatomy of the eye and how the eye develops in the embryo, in particular the role of the ectoderm. How different structures and functions of the eye are affected by ectodermal dysplasias will be covered, leading into the treatments available for different ocular problems, and to finish, a look to the future.

Basic Eye Structure

The eye consists not only of the globe but also the surrounding structures - the lids, the tear drainage passages, and the tear or lacrimal glands. The globe of the eye consists of an outer coat, middle layer and inner layer. The outer coat is made up of a tough fibrous, white layer - the sclera, and a clear transparent front window - called the cornea. The middle layer contains pigment and forms the iris and some deeper structures in the wall of the eye. The inner layer is the light-seeing layer or the retina. The lens is an oval disc which sits behind the pupil and iris.

The cornea does two-thirds of focusing of the light entering the eye and the lens does one-third. The cornea must be healthy and transparent for the eye to function. The cornea depends on the lids, the conjunctiva (the transparent membrane inside the lids and covering the white of the eye), and tear or lacrimal glands to maintain its function. These structures and tears keep the eye healthy, moist, and comfortable.

Tears form a film over the surface of the eye - cornea/ conjunctiva/sclera. The tear film has three layers forming a sandwich over the eye. The outer layer is made up of an oil layer. The oil is produced by glands found in the lids called meibomian glands. The inner layer is a mucous layer which is made by glands in the conjunctiva. Sandwiched between these layers is a thick filling of water from the tear or lacrimal gland. This water layer contains chemicals and substances which nourish the eye surface. The tears eventually either evaporate or drain into the nose through a drainage system called the nasolacrimal duct.



Eye Development

The formation of the eye is a complex process which starts as early as the fourth week after conception. The eye forms from three main tissues - the brain/nervous system tissue (neuro-ectoderm), a middle layer (mesoderm), and an outer layer (the ectoderm). The ectoderm forms the outer parts of the embryo - skin, hair, sweat glands, teeth, and several parts of the eye and ear.

A cystic outgrowth from the developing brain will form the eye. The surface layer (skin) overlying this cystic area is the ectoderm. The lens buds off this outer ectodermal layer to lie inside the developing eye. The ectoderm closes the gap forming the outer part of the cornea, conjunctiva, lids, lacrimal gland, and nasolacrimal duct.

Eye Involvement in Ectodermal Dysplasias

Eye problems occur in ectodermal dysplasias but how common and the specific ways the eyes are involved are not fully appreciated. Eye problems seem to differ among the different types of ectodermal dysplasias and can vary in severity. The structures of the eye formed from the ectoderm may be affected by ectodermal dysplasias. The function and comfort of the eye may therefore be disturbed.

Tears

- The health of the surface of the eye - conjunctiva and cornea - depends on the tears. The tears bathe the ocular surface with nutrients and moisture like the water and fertilizer in a garden. The lacrimal gland produces the 'water' component of the tears which contain growth factors and substances to ensure ocular health and comfort. The function of this gland can be affected by ectodermal dysplasias. The oil layer which floats on the 'water' layer seals the tears against the eye and is secreted from the meibomian glands in the edge of the eyelids. These glands may be absent in some forms of ectodermal dysplasias - failing to form in the embryo. Alternatively, abnormalities in the production of this oil layer may allow the tears to evaporate more readily than normal, thus causing tear deficiency. The 'water' layer sits on a thin layer of mucus produced from the conjunctiva which allows the tears to flow and spread across and wet the eye surface.
- Thus, the oil, water and mucus layers can each be affected by the ectodermal dysplasias. Defects in the tear film causes changes in the conjunctiva and cornea resulting in a diseased 'ocular surface'. This can predispose to infection, lead to corneal ulceration and delayed healing.

Cornea

The outer surface of the cornea, the corneal epithelium, forms from the ectoderm and requires the tears to function normally. The cornea is the window of the eye focusing and allowing light to enter the eye; its function is essential for vision. The surface of the cornea is renewed every 7-14 days. Tissue at the edge of the cornea continually forms the corneal epithelium from stem cells. In some types of ectodermal dysplasia, these stem cells are affected. This leads to an unhealthy cornea which can result in defects in the corneal epithelium, ulcers, and scarring.

Lens

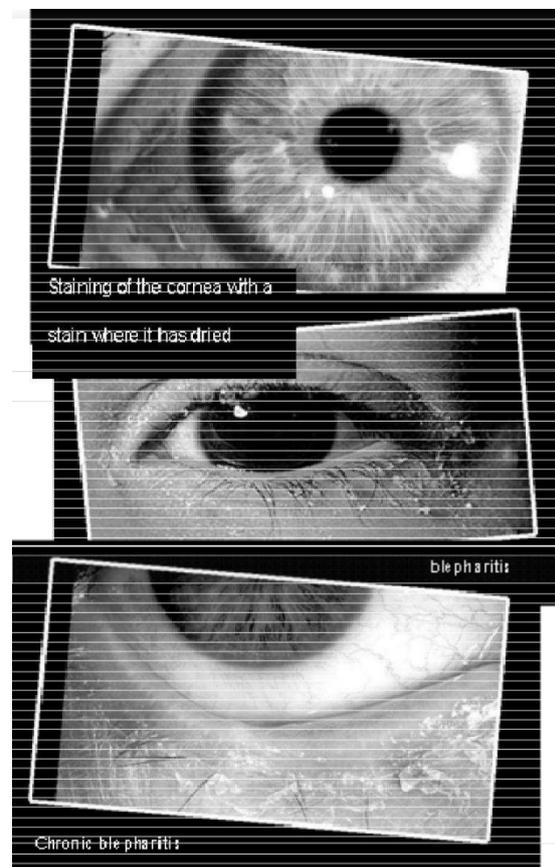
The lens forms from the surface skin or ectoderm and can have opacities within it in rare forms of ectodermal dysplasia. Opacity of the lens (loss of its transparency) results in defects in focus and clarity of vision and is termed a cataract.

Tear drainage

The tear drainage system drains tears from the eye into the nose. Obstruction in this system can occur leading to watery eyes, especially from birth, and increases the risk of infection within this drainage system. Infection of the tear drainage sac (inside the nose) is called dacryocystitis. Several forms of ectodermal dysplasia have abnormal embryological development of the lacrimal drainage system as an expected component, such as the EEC Syndrome.

Treatment

The underlying causes of eye disease in ectodermal dysplasia are not fully understood, and so treatment is aimed at symptomatic relief, rather than cure or prevention. Tear supplements increase both lubrication and comfort, but natural tears contain many substances other than just 'water', so the surface of the eye can still remain unhealthy. Antibiotics and topical steroids may help to control infection and reduce inflammation. Photophobia (painful discomfort in bright light) may result from a diseased ocular surface and in my experience seems particularly severe and common in the EEC syndrome. Corneal ulcers are treated by a variety of measures - antibiotics, tear supplements, patching, and occasionally special contact lenses termed bandage contact lenses.



Surgery may be required for blockage or non-functioning of the nasolacrimal duct; this includes syringing and probing the system (usually done in childhood) and an operation to 're-plumb' (actually completely reconstruct) the duct into the nose - a dacryocystorhinostomy or DCR for short.

Cataract surgery can be performed if cataracts reduce the vision. Rarely corneal transplants have been performed to replace a scarred cornea with healthy tissue. The outcomes from corneal transplantation are uncertain due to the other changes in the ocular surface - like planting a seed in dry, barren soil. One should be wary of a recommendation for corneal transplantation in the ectodermal dysplasias because of the associated risks from poor healing in the face of poor tear production. Recent advances in reconstructing the ocular surface and corneal transplantation may be more successful in the future.



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