Series Editor: William W. Huang, MD, MPH

Eye Findings in Dermatologic Conditions

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The author reports no conflict of interest.

Eye Finding	Definition/Pathogenesis	Dermatologic Condition(s)	Miscellaneous
Angioid streaks	Rupture of Bruch membrane (innermost layer of choroid); broad red-brown lines radiating from optic nerve	PXE, EDS (kyphoscoliosis and vascular types most commonly associated), Cowden disease	Associated with sickle cell anemia, β thalassemia, Paget disease of bone, and phosphatemia; often idiopathic
Ankyloblepharon filiforme adnatum	Fusion of all or part of eyelid margins	Hay-Wells syndrome (also known as AEC syndrome, a form of ectodermal dysplasia)	AD mutation in p63
Blue sclerae	Blue hue is due to underlying choroidal veins, which show through thin collagen fibers of sclera	Alkaptonuria, EDS, Fanconi anemia, Marfan syndrome, nevus of Ota, osteogenesis imperfecta types I–III, PXE	EDS type 6 (kyphoscoliosis; AR mutation in *PLOD*, which encodes lysyl hydroxylase): blue sclerae, retinal detachment, globe rupture, keratoconus; also found in alkaptonuria and nevus of Ota due to pigment deposition in sclera
Brushfield spot	White to gray spots at periphery of iris due to stromal hyperplasia	Down syndrome	Normal in children (Kunkmann-Wolffian bodies)
Cherry red spot	Bright red-orange color of fovea is contrasted against pale color of retina due to accumulation of lipid in retinal ganglion cells	Hurler disease, Niemann-Pick disease, Tay-Sachs disease, sialidosis, Sandhoff disease	Also seen in central retinal artery occlusion (fovea receives circulation from choroid rather than retinal artery); dapsone toxicity and carbon monoxide poisoning
Coloboma	Defect in one of the structures of the eye, such as the iris, retina, choroid, or optic disc	Epidermal nevus syndrome, Goltz syndrome (focal dermal hypoplasia), incontinentia pigmenti, Gorlin syndrome	Most commonly affects the iris; may be unilateral or bilateral
Comma-shaped corneal opacities		X-linked ichthyosis (occur in up to 50%)	Do not affect visual acuity but may cause corneal erosions

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Congenital hypertrophy of retinal pigment epithelium	Flat pigmented spots on outermost layer of retina	Gardner syndrome (familial adenomatous polyposis)	No impact on vision unless located over macula
Dendritic keratitis	Linear branching pattern of corneal ulcers	Herpes infection, Richner-Hanhart syndrome (tyrosinemia type II)	Herpes simplex keratitis is most common cause of infectious blindness in Western world; pseudoherpetic keratitis caused by accumulation of tyrosine
Ectopia lentis	Displacement of lens from its normal position	Upward: Marfan syndrome; downward: homocystinuria	
Juvenile posterior subcapsular lenticular opacity	Also known as juvenile cortical cataract	NF type II	90% of NF type II patients have ocular symptoms
Kayser-Fleischer rings	Dark rings encircling outermost portion of iris	Wilson disease	Caused by copper deposition in portion of cornea (Descemet membrane); early lesions best viewed via slit-lamp examination
Lester iris	Hyperpigmentation of papillary margin of iris	Nail-patella syndrome	AD mutation in *LMX1B* gene
Lisch nodules	Pigmented hamartomas of melanocytes, which appear as yellow-brown papules on the surface of the iris	NF type I	Present in >94% of NF patients >6 years of age; detected by slit-lamp examination; also optic gliomas
Phakomas	Astrocytic hamartomas of optic nerve, appear as grey-white lesions on retina	Tuberous sclerosis complex	May calcify

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Pinguecula	Yellow-white nodule on conjunctiva adjacent to limbus	Gaucher disease	Generally asymptomatic; differentiated from pterygium, which grows into cornea and may affect vision; most common cause is UV exposure
Retinitis pigmentosa	Salt-and-pepper appearance of retina with clumps of black pigment	Refsum syndrome, Cockayne syndrome, Sjögren-Larsson syndrome (atypical retinitis pigmentosa: intraretinal glistening white dots)	Progressive loss of vision due to degeneration of retinal pigment epithelial cells; first symptom is night blindness followed by loss of peripheral vision

Abbreviations: PXE, pseudoxanthoma elasticum; EDS, Ehlers-Danlos syndrome; AEC, ankyloblepharon-ectodermal defects-cleft lip/palate; AD, autosomal dominant; AR, autosomal recessive; NF, neurofibromatosis.

Practice Questions

1. Which type of EDS is most characteristically associated with blue sclerae and globe rupture?
a. arthrochalasia
b. classical
c. dermatosparaxis
d. hypermobility
e. kyphoscoliosis

2. Ankyloblepharon may be associated with mutation of which gene?
a. fibrillin 1
b. *LMX1B*
c. *NF1*
d. p53
e. p63

3. Which is a characteristic ocular tumor in patients with tuberous sclerosis complex?
a. congenital hypertrophy of retinal pigment epithelium
b. phakoma
c. pigmented iris hamartoma
d. pinguecula
e. pterygium

4. Which syndrome is *not* associated with blue sclerae?
a. EDS type 6
b. lipoid proteinosis
c. Marfan syndrome
d. osteogenesis imperfecta type II
e. pseudoxanthoma elasticum

5. Which term describes white spots at the periphery of the iris?
a. Brushfield spots
b. coloboma
c. Kayser-Fleischer rings
d. Lester iris
e. Lisch nodules

Fact sheets and practice questions will be posted monthly.