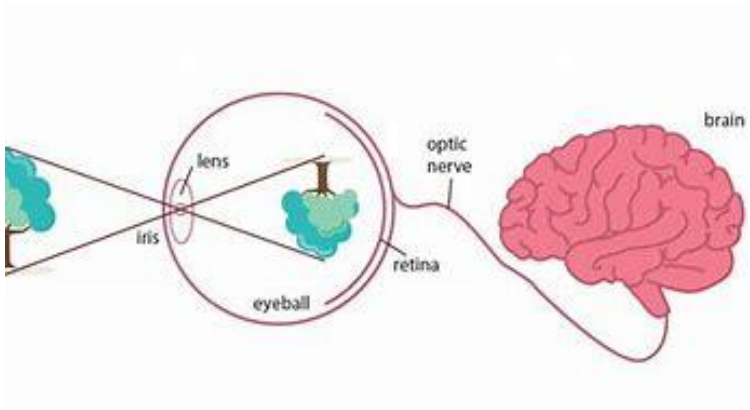
The Optician's Guide to Understanding GENETIC Eye DISEASES

B.W. Phiillips, NCLE, FCLSA

What is Genetics?

- Genetics is the scientific study of genes and heredity—of how certain qualities or traits are passed from parents to offspring as a result of changes in DNA sequence. A gene is a segment of DNA that contains instructions for building one or more molecules that help the body work. DNA is shaped like a corkscrew-twisted ladder, called a double helix.

The eye and the brain-are they on the same page?



- You look with your eyes, but you see with your brain. The brain does not receive an image the way a camera or videorecorder does. The brain is not passive, the light that falls on your eyes does not enter the brain as it is. It is processed in the eyes and in the brain. Light falls on the retina of your eyes

What Is Charles Bonnet Syndrome?

- Charles Bonnet Syndrome (CBS) is a condition that some people get when they lose some or all their vision. It causes them to have visual hallucinations (seeing things that aren't really there).

- A new study suggests this condition is surprisingly common among people with certain types of vision loss.

Juvenile Macular Degeneration?

- Stargardt's generally refers to a group of inherited diseases causing light-sensitive cells in the inner back of the eye (retina) to deteriorate, particularly in the area of the macula where fine focusing occurs. Central vision loss also occurs, while peripheral vision usually is retained.

What is the difference between color blindness and achromotopsia

COLOR BLINDNESS:

If you have color blindness, it means **you see colors differently than most people.** Most of the time, color blindness makes it hard to tell the difference between certain colors. Usually, color blindness runs in families. There’s no cure, but special glasses and contact lenses can help.

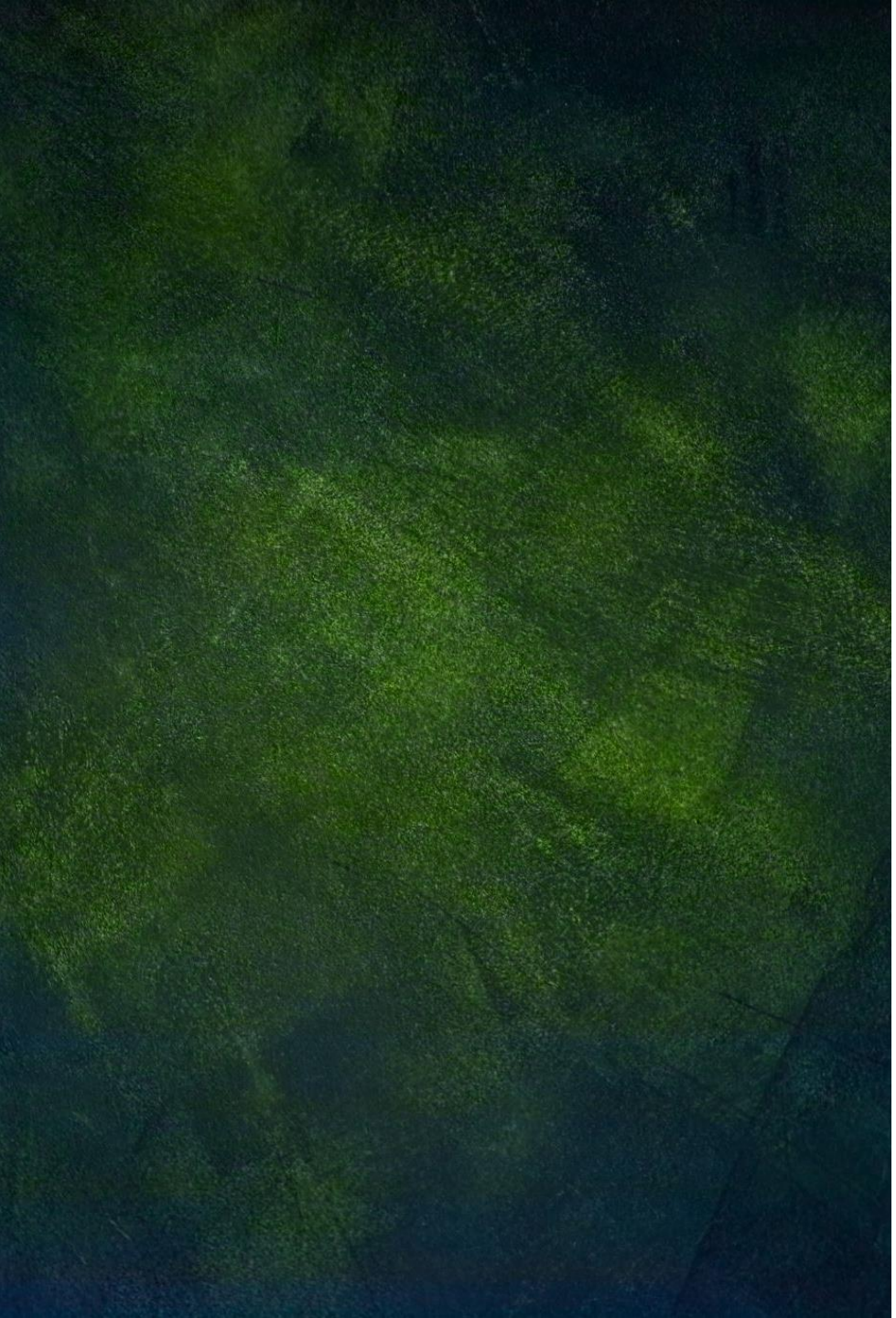
What is Achromatopsia?

- Achromatopsia is a condition characterized by a partial or total absence of color vision. People with complete achromatopsia cannot perceive any colors; they see only black, white, and shades of gray. Incomplete achromatopsia is a milder form of the condition that allows some color discrimination

- Achromatopsia also involves other problems with vision, including an increased sensitivity to light and glare (photophobia), involuntary back-and-forth eye movements (nystagmus), and significantly reduced sharpness of vision (low visual acuity).

Types of Color Blindness .

- **Red-Green:**
- Protanopia (aka red-blind) – Individuals have no red cones.
- Protanomaly (aka red-weak) – Individuals have red cones and can usually see some shades of red.
- Deuteranopia (aka green-blind) – Individuals have no green cones.
- **Deuteranomaly (aka green-weak) – Individuals have green cones and can usually see some shades of green.**



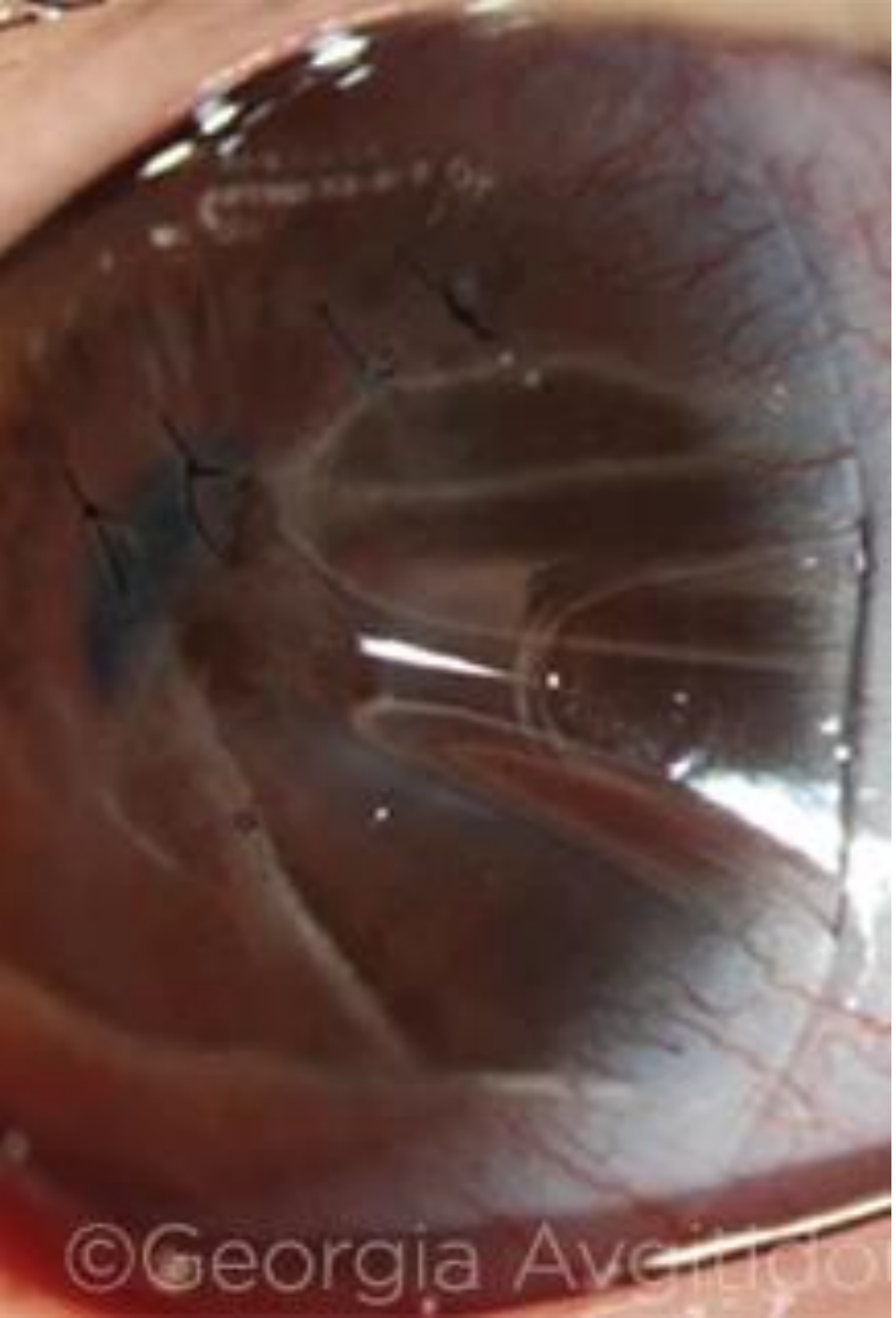
Scleral Lenses For Dry Eyes?

Dry Eye Statistics- At a Glance:

Currently between 16 million and 49 million Americans have dry eyes. This is between 5-15% of the population.

Dry Eye Disease in the USA costs over $55.4 billion to the economy each year.

Google trends for the topic “dry eye syndrome” have doubled between December 2010 and July 2020



Less Well known corneal diseases/dysrophyies

- **Brittle Cornea Syndrome:**

- Brittle cornea syndrome (BCS) is an autosomal recessive connective tissue disorder.
- Severe corneal thinning and carries an increased risk for spontaneous perforation or rupture from minimal trauma.

Diabetic Retinopathy

- Diabetic retinopathy is “the eye condition that results from diabetes, Type I insulin dependent, and Type II, typically non-insulin dependent. It occurs when blood vessels stop feeding the retina properly. In its early stages, the blood vessels may leak fluid in the retina, which can affect the macula, the entire retina, or the vitreous gel.

- In the later stages of the condition, new vessels may grow and send blood into the center of the eye, causing serious vision loss that can lead to blindness.

Diabetic Retinopathy- Symptoms?

- Blurry or double vision
- Halos or flashing lights
- Double vision
- Dark spots or floaters
- Pain or sensation of pressure in one or both eyes
- Diminished peripheral (side) vision
- Poor night vision

AMD

- **Age-related macular degeneration** (AMD) is a medical condition which usually affects older adults and results in a loss of vision in the center of the visual field (the macula) because of damage to the retina. It occurs in "dry" and "wet" forms. It is a major cause of blindness and visual impairment in older adults (>50 years).

- Starting from the inside of the eye and going towards the back, the three main layers at the back of the eye are the retina, which contains the nerves; the choroid, which contains the blood supply; and the sclera.

Wet vs. Dry – the difference?

- In the dry (nonexudative) form, cellular debris called drusen accumulates between the retina and the choroid, and the retina can become detached

- In the wet (exudative) form, which is more severe, blood vessels grow up from the choroid behind the retina, and the retina can also become detached. It can be treated with laser coagulation, and with medication that stops and sometimes reverses the growth of blood vessels.

Causes

- **Aging:** Approximately 10% of patients 66 to 74 years of age will have findings of macular degeneration. The prevalence increases to 30% in patients 75 to 85 years of age

- **Family history:** The lifetime risk of developing late-stage macular degeneration is 50% for people who have a relative with macular degeneration, versus 12% for people who do not have relatives with macular degeneration

Amsler Grid