The Optician's Guide to Understanding GENETIC Eye DISEASES

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What Are Genetic Eye Diseases?

- Many of you are already aware that genes play a role in determining the hue of your eyes, but the fact is that genetics play a role far beyond than merely the color of your eyes. They are also responsible for inflicting various kinds of diseases, some of them very rare.
- *"Over the past one a half decade, NEI supported researchers have been able to* ***identify around 500 genes capable of inducing genetic eye diseases.***

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Genetic Disorders

- Genetic disorders can happen for many reasons. Genetic disorders often are described in terms of the chromosome that contains the gene that is changed in people who have the disorder. If the gene is on one of the first 22 pairs of chromosomes, called the autosomes, the genetic disorder is called an autosomal condition.
- If the gene is on the X chromosome, the disorder is called X-linked.

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Dominant VS. Recessive gENES

- Dominant diseases can be caused by only one copy of a gene having a DNA mutation.
- If one parent has the disease, each child has a 50% chance of inheriting the mutated gene.
- For recessive diseases, both copies of a gene must have a DNA mutation in order to get one of these diseases. If both parents have one copy of the mutated gene, each child has a 25% chance of having the disease, even though neither parent has it

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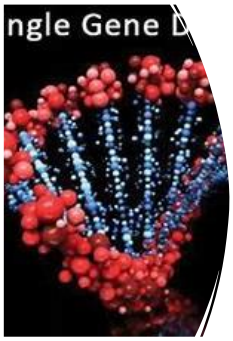

General eye exams are essential to screen for conditions that threaten 20/20 vision.

- Usually, the more common the eye condition, the less likely it's caused by a single gene. That factor helps determine whether genetic screening is worth your while.
- The frequency of eye disease among your ancestors and the age of onset suggest whether you should be concerned about suffering the same fate.

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Genetic Eye diseases/disorders?

1. Glaucoma?
2. Congenital Cataracts?
3: Retinitis Pigmentosa?
4: Juvenile Macular Degeneration?
5. Stargardt Macular Degeneration?
6. Best's Disease (Vitelliform macular dystrophy)?
7. X-linked Juvenile Retinoschisis?

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Prism eyeglasses for Star

Special prism based lenses are used in glasses to allow the refraction of light in such a way that the blind spot created by the macular degeneration is reduced.

Although the spot can't be removed completely, its size can be reduced a lot resulting in better vision. They're helpful in reading, writing, playing cards, etc. and other indoor activities.

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5 LEADING CAUSES OF VISUAL IMPAIRMENT IN INFANTS/CHILDREN

- Optic Nerve Atrophy:
- Albinism:
- Optic Nerve Hypoplasia:
- Retinopathy of Prematurity:
- Cortical Visual Impairment(CVI)

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CVI

- When students with these visual/behavioral characteristics are shown to have loss of acuity or judged by their performance to be visually impaired, they are considered to have CVI."

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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.

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Population with colorblindness?

- **According to the Howard Hughes Medical Institute, about 3.7% or 12 million Americans are colorblind (7 percent of the males, and 0.4 percent of the females).**
- **Achromatopsia is a relatively uncommon disorder, with a prevalence of 1 in 30,000 people.**
- **Less than 1%**

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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).

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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.**

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Acquired types of color blindness/ SELF TEST.

- Final Reason:
- **Alcohol consumption** – Reduced color discrimination can be symptom of alcoholism, with blue-yellow being particularly affected.

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Treatment for color blindness

Gene Therapy

Eyeglasses and Contact Lenses.

EnChroma glasses

An app called Color Blind Pal

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Causes of Dry Eye SyndroME

- Decreased tear production:
  - Aging
  - Certain Medical Conditions
  - Corneal nerve desensitivity caused by contact lens use
- Increased tear evaporation:
  - Allergies
  - Wind, Smoke, Dry Air
  - Meibomian gland Disfunction

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Moisture chamber eyeglasses

- How Do Moisture Chamber Glasses Work?
- Dry eyeglasses have a gasket (also called eyecup) that comfortably encompasses your eyes, creating a seal around them. As a result, it shields your eyes from wind, dust, pollen, and other drying factors in the air, including air conditioning.

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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

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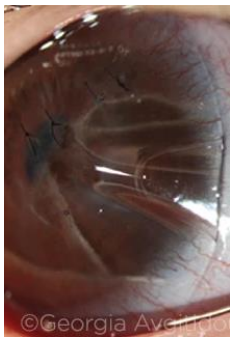

Less Well known corneal diseases/dysrophyies

- **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.

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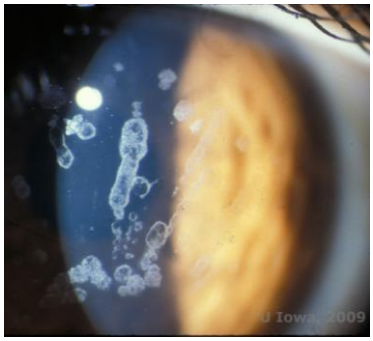

Granular corneal dystrophy

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What is Retinitis pigmentosa?

Retinitis pigmentosa (RP) is a group of rare eye diseases that affect the retina (the light-sensitive layer of tissue in the back of the eye). RP makes cells in the retina break down slowly over time, causing vision loss.

RP is a genetic disease that people are born with. Symptoms usually start in childhood, and most people eventually lose most of their sight.

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What causes RP?

- Most of the time, RP is caused by changes in genes that control cells in the retina. These changed genes are passed down from parents to children.
- Sometimes RP happens as part of other genetic conditions, like Usher syndrome.
- What is Usher syndrome?

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Treatments For RP

- Currently there is no cure for retinitis pigmentosa.
- Several companies are developing retinal implants (sometimes called bionic eyes) and other innovative treatments that are showing promise in providing or preserving some degree of usable sight for people affected by RP.

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What is Keratoconus?

KC is a degenerative disorder of the eye in which structural changes within the cornea cause it to thin and change to a more conical shape than its normal gradual curve.

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RECENT STUDY

- The annual incidence of keratoconus was 1:7500 in the relevant age category (13.3 cases per 100 000, 95% confidence interval [CI]: 11.6-15.2) and the estimated prevalence of keratoconus in the general population was 1:375 (265 cases per 100 000, 95%.
- These values are 5-fold to 10-fold higher than previously reported values in population studies. The mean age at diagnosis was 28.3 years and 60.6% of diagnosed patients were male.

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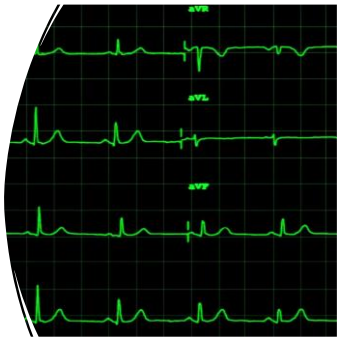
What is considered "normal IOP"?

- Currently normal range of intraocular pressure is considered to be between 10 and 20 mmHg (millimeter of Mercury) by some while **10-21 mmHg**



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Pigmentary Glaucoma

- **Pigmentary glaucoma. This rare form of glaucoma is caused by clogging of the drainage angle of the eye by pigment that has broken loose from the ????, reducing the rate of aqueous outflow from the eye**

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What is a Filtering Bleb?

- Glaucoma filtration surgery is indicated when other available treatment modalities such as medications and laser therapies have failed to control the intraocular pressure level or to prevent against progression of the disease (e.g. visual field loss or optic disc cupping).

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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.

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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

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3 Types of Cataracts

- Can You Name Them?
- Nuclear Cataract
- Cortical Cataract

Posterior Subcapsular Cataract

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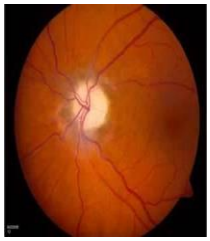
Optic Nerve Atrophy

- Optic nerve atrophy is damage to the optic nerve. The optic nerve carries images of what we see from the eye to the brain.
- Most common cause is poor blood flow, called ischemic optic neuropathy, which most often affects elderly people. The optic nerve can also be damaged by shock, various toxic substances, radiation, and trauma.



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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.

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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.

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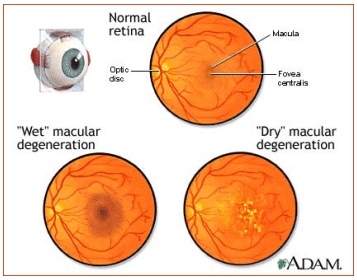
AMD



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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

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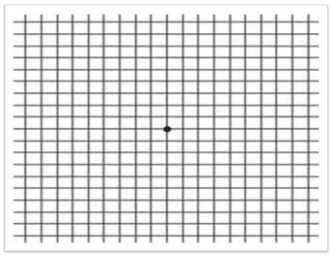
Amsler Grid



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