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***Review article*** ***Pathophysiology***

# Pathophysiology and pharmacological study of colour blindness

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## ABSTRACT

Color blindness occurs when the person is unable to see colors in a normal way. It is also known as colour deficiency. In Trichromacy, three types of cones are present and working properly. Patient can see all colors on the visible spectrum of light in the traditional way. This is full colour vision. In Anomalous trichromacy. There are three types of cones, but one type isn't as sensitive to light in its wavelength as it should be. As a result, person doesn't see colors in the traditional way, with variations from normal ranging from mild to severe. In Dichromacy, one type of cone is missing. So, only two types of cones (usually S cones along with either L cones or M cones) are present. In Monochromacy: Person has only one type of cone or no cone function at all, so very limited or no ability to see color. There are four main subtypes: a.Protanopia: In this condition, in the person L cones are missing. So, person can't perceive red light. Mostly see colors as shades of blue or gold. b.Deuteranopia: In this condition, in the person M cones are missing. So, person can't perceive green light. Mostly see blues and golds. c.Protanomaly: In this condition, the person has all three cone types, but L cones are less sensitive to red light than they should be. Red may appear as dark gray, and every color that contains red may be less bright. d.Deuteranomaly: In this condition, the person has all three cone types, but M cones are less sensitive to green light than they should be. Mostly see blues, yellows and generally muted colours. The colour blindness can be diagnosed by the Ishihara test, Colour vision test. Colour vision tests and Genetic testing.

**Keywords:** Colour Vision, Trichromacy, Monochromacy, Ishihara Test, Genetic Testing, Cones.

## INTRODUCTION

Color blindness occurs when the person is unable to see colors in a normal way. It is also known as color deficiency. Color blindness often happens when someone cannot distinguish between certain colors. This usually happens between greens and reds, and occasionally blues.(1) Colour blindness (colour vision deficiency) affects approximately 1 in 12 men (8%) and 1 in 200 women. In the UK there are approximately 3 million colour blind people (about 4.5% of the entire population), most of whom are male. Worldwide, there are estimated to be about 300 million people with colour blindness, almost the same number of people as the entire populations of the USA.There are different causes of colour blindness. For most colour blind people their condition is genetic, usually inherited from their mother, although some people become colour blind as a result of other diseases such as diabetes and multiple sclerosis or it can be acquired due to ageing or from taking drugs and medications.(2)

## *Types of color blindness*

There are several types of color blindness, defined according to which types of cones aren't working well. To understand the types of color blindness, it helps to know a bit about cones. Cones are nerve cells in your eye that detect colors in the visible spectrum of light. This spectrum includes all the wavelengths that humans can see. These range in length from 380 nanometers (short), or nm, to 700 nanometers (long). Normally, you're born with three types of cones:

- **Red-sensing cones (L cones)**: These cones perceive long wavelengths (around 560 nanometers).
- **Green-sensing cones (M cones)**: These cones perceive middle wavelengths (around 530 nanometers).
- **Blue-sensing cones (S cones)**: These cones perceive short wavelengths (around 420 nanometers)[1].

## *Pathophysiology*

Most people have all three types of cones, and these work as they should. However, if the person having color vision deficiency, at least one type of cone isn't working properly. Problems with cones affect the ability to see colors in the traditional way.

General categories of cones and their working process:

***1. Trichromacy**:*

All three types of cones are present and working properly. Patient can see all colours on the visible spectrum of light in the traditional way. This is full-color vision.(3)

***2. Anomalous trichromacy**:*

There are three types of cones, but one type isn't as sensitive to light in its wavelength as it should be. As a result, person doesn't see colors in the traditional way, with variations from normal ranging from mild to severe. In mild cases, may just confuse pale or muted colors. In more severe cases, may also confuse vivid and pure (fully saturated) colors. These types of color blindness have names that end in "anomaly" (which indicates partial vision of a specific color).

***3. Dichromacy**:*

One type of cone is missing. So, only have two types of cones (usually S cones along with either L cones or M cones) are present. Patient can see the world through the wavelengths that those two types of cones can perceive. It's hard to tell the difference between fully saturated colors. These types of color blindness have names that end in "anopia" (which indicates absence of vision of a specific color).

***4. Monochromacy**:*

Person has only one type of cone or no cone function at all,so very limited or no ability to see color. Instead, see the world in varying shades of gray.

Within these general categories, there are many specific types of color blindness.

## *Red-green color deficiency*

Red-green color deficiency is the most common type of color blindness. It affects how you see any colors or shades that have some red or green in them. There are four main subtypes:

**a.Protanopia**: In this condition, in the person L cones are missing. So, person can't perceive red light. Mostly see colors as shades of blue or gold. Person may easily confuse different shades of red with black. He or She may also confuse dark brown with dark shades of other colors, including green, red or orange.

**b.Deuteranopia**: In this condition, in the person M cones are missing. So, person can't perceive green light. Mostly see blues and golds. . Person may confuse some shades of red with some shades of green.may also confuse yellows with bright shades of green.(4)

**c.Protanomaly**: In this condition, the person has all three cone types, but L cones are less sensitive to red light than they should be. Red may appear as dark gray, and every color that contains red may be less bright.

**d.Deuteranomaly**: In this condition, the person has all three cone types,but M cones are less sensitive to green light than they should be. Mostly see blues, yellows and generally muted colours.(5)

Protanopia and deuteranopia are examples of dichromacy. Protanomaly and deuteranomaly are examples of anomalous trichromacy.Other terms you might hear are "protan" and "deutan." Protan and deutan are shorthand ways to talk about red-green colorblindness. Deutan refers to green (impaired or missing green-sensing cones, or M cones). Protan refers to red ( impaired or missing red-sensing cones, or L cones).(6)

Red-green color blindness is much more common among men and people assigned male at birth (AMAB) compared to women and people assigned female at birth (AFAB). This is because the genes for the color vision cone light-sensitive proteins are on the X chromosome, of which males have one and females have two. So if the one X in a male contains abnormal genes, the color blindness will reveal itself, while females can compensate with the other normal gene on the second X chromosome**2-5**.(7)

## *Genetic Causes*

The color blindness is typically an inherited genetic disorder. The most common forms of colorblindness are associated with the Photopsin genes, but the mapping of the human genome has shown there are many causative mutations that don't directly affect the opsins. Mutations capable of causing color blindness originate from at least 19 different chromosomes and 56 different genes.

## *Non-genetic causes*

Physical trauma can cause color blindness, either neurologically – brain trauma which produces swelling of the brain in the occipital lobe – or retinally, either acute (e.g. from laser exposure) or chronic (e.g. from ultraviolet light exposure).Color blindness may also present itself as a symptom of degenerative diseases of the eye, such as cataract and age-related macular degeneration, and as part of the retinal damage caused by diabetes. Vitamin A deficiency may also cause color blindness.

Color blindness may be a side effect of prescription drug use. For example, red–green color blindness can be caused by ethambutol, a drug used in the treatment of tuberculosis. Blue–yellow color blindness can be caused by sildenafil, an active component of Viagra. Hydroxychloroquine can also lead to hydroxychloroquine retinopathy, which includes various color defects. Exposure to chemicals such as styrene or organic solvents can also lead to color vision defects. Simple colored filters can also create mild color vision deficiencies. John Dalton's original hypothesis for his

deuteranopia was actually that the vitreous humor of his eye was discolored.(8)

### *Blue-yellow color deficiency*

Blue-yellow color vision defects (tritan defects) are much less common and include:

- **Tritanopia**: In this condition lack of S cones. So, the person can't perceive blue light. Patient may be seeing mostly reds, light blues, pinks and lavender.
- **Tritanomaly**: In this condition patient have all three cone types, but your S cones are less sensitive to blue light than they should be. Blues look green, and see little or no yellow. Blue-yellow color blindness equally affects people AMAB and people AFAB (AFAB/AMAB meant for "assigned female/male at birth").

### *Blue cone monochromacy*

This is the rarest form of color blindness. In this type, don't have working L cones or M cones.patient have only S cones. It's hard to tell the difference between colors, and see mostly grays. They may also have other eye problems, including sensitivity to light (photophobia), nystagmus and near sightedness.

### *Rod monochromacy (achromatopsia)*

Achromatopsia is when all or most of the cones are missing or don't work properly. Patient may see everything in shades of gray and also have other vision issues that may greatly impact quality of life.

### *Causes of color blindness*

Color blindness can be either inherited (you're born with it) or acquired (you develop it later in life). The causes are different in each case.

### *Causes of inherited color blindness*

A change (mutation) in the genes causes inherited color blindness. The most common form, red-green color blindness, follows an X-linked recessive inheritance pattern. Conditions inherited in this way usually affect babies AMAB and are rare among babies AFAB.(9) Here's a breakdown of the genetics for red-green color blindness.

A male baby:

- Will inherit red-green color blindness if the mother has the condition.
- Has a 50% chance of inheriting red-green color blindness if the mother is a carrier (this means the mother carries one copy of the genetic mutation but doesn't have the condition). The other copy is normal, hence the 50:50 chance.
- Won't inherit the condition if only the father has it because the father contributes the Y chromosome to male babies and the X chromosome to female babies.

A female baby:

- Will inherit red-green color blindness if both parents have the condition.
- Will be a carrier if the father has the condition but the mother doesn't (and isn't a carrier).
- Will either inherit red-green color blindness (50% chance) or be a carrier (50% chance) if the father has the condition and the mother is a carrier.

### *Causes of acquired color blindness*

Acquired color blindness, which usually develops as blue-yellow color deficiency, has many possible causes. These include:

- Exposure to chemicals that harm your nervous system, such as organic solvents, solvent mixtures and heavy metals.
- Long-term exposure to welding lights.
- Medications, including hydroxychloroquine .
- Eye conditions, including age-related macular degeneration, glaucoma and cataracts.
- Medical conditions that affect your brain or nervous system, including diabetes, Alzheimer's disease and multiple sclerosis (MS).

Acquired color blindness is less common than inherited forms.

### *The symptoms of color blindness*

- Telling the difference between certain colors or shades.
- Seeing the brightness of certain colors.

### *Diagnosis and tests*

***1. The Ishihara test***

This is the most common test eye care providers use to diagnose red-green color blindness. For this test, a provider shows you a series of color plates. Each plate contains a pattern of small dots. Among those dots, there's a number (or shape for young children). You identify what you can see on each plate. Some plates include numbers that you can only see with full-color vision. Others include numbers that you can only see with color vision deficiency. Based on the results of the Ishihara test, provider may recommend further testing to confirm a diagnosis and learn more.

***2. Colour vision test***

The main method for diagnosing a color vision deficiency is in testing the color vision directly. The Ishihara color test is the test most often used to detect red–green deficiencies and most often recognized by the public. Some tests are clinical in nature, designed to be fast, simple, and effective at identifying broad categories of color blindness. Others focus on precision and are generally available only in academic settings[9].

- **pseudoisochromatic plates**, a classification which includes the Ishihara color test and HRR test, embed a figure in the plate as a number of spots surrounded by spots of a slightly different color. These colors must appear identical (metameric) to the colorblind but distinguishable to color normals. Pseudoisochromatic plates are used as screening tools because they are cheap, fast, and simple, but they do not provide precise diagnosis of CVD.
- **Lanterns**, such as the Farnsworth Lantern Test, project small colored lights to a subject, who is required to identify the color of the lights. The colors are those of typical signal lights, i.e. red, green, and yellow, which also happen to be colors of confusion of red–green CVD. Lanterns do not diagnose colorblindness, but they are occupational screening tests to ensure an applicant has sufficient color discrimination to be able to perform a job.

- **Arrangement tests** can be used as screening or diagnostic tools. The Farnsworth–Munsell 100 hue test is very sensitive, but the Farnsworth D-15 is a simplified version used specifically for screening for CVD. In either case, the subject is asked to arrange a set of colored caps or chips to form a gradual transition of color between two anchor caps.
- **Anomaloscopes** are typically designed to detect red–green deficiencies and are based on the Rayleigh match, which compares a mixture of red and green light in variable proportions to a fixed spectral yellow of variable luminosity. The subject must change the two variables until the colors appear to match. They are expensive and require expertise to administer, so they are generally only used in academic settings[10].

***3. Genetic testing***

While genetic testing cannot directly evaluate a subject's color vision (phenotype), most congenital color vision deficiencies are well-correlated with genotype. Therefore, the genotype can be directly evaluated and used to predict the phenotype. This is especially useful for progressive forms that do not have a strongly color deficient phenotype at a young age. However, it can also be used to sequence the L- and M-Opsins on the X-Chromosome, since the most common alleles of these two genes are known and have even been related to exact spectral sensitivities and peak wavelengths. A subject's color vision can therefore be classified through genetic testing, but this is just a prediction of the phenotype, since color vision can be affected by countless non-genetic factors such as your cone mosaic.(10)

## *Management and treatment*

Currently, there's no medical treatment or cure for people with inherited color blindness. If you have acquired color blindness, your healthcare provider will treat the underlying condition or adjust your medications as needed. This may help improve your color vision.

Color-blindness glasses may provide a richer color experience for people with mild forms of anomalous trichromacy. The glasses enhance the contrast between colors so people with color vision deficiency can see the differences more clearly. But they don't allow you to see any new colors, and the results vary based on the individual. Plus, it's important to know that these glasses aren't a cure and won't correct any issues with your cones.

## *Prevention*

Prevention of inherited color blindness is not possible. However, able to lower your risk of acquired color blindness. Visit a healthcare provider for yearly checkups and ask about risk for developing color vision deficiency. Some questions to ask include:

- Do any of my medical conditions put me at risk for color blindness?
- Can any of my medications cause color blindness?
- Should I be concerned about any chemical or environmental exposures at my job?
- What can I do to lower my risk?

## *Regarding children's care*

Certain careers may be too challenging or unsafe to pursue with color deficiency. These include careers as an electrician, pilot, fashion designer or graphic artist. But you can encourage your child to pursue other careers where color vision won't play a major role. Talk to counselors or mentors at your child's school to access resources on career options.

Connect with others who have color vision deficiency or parents of children with the condition. They can share advice and resources for living with color vision deficiency from day to day. Some tips include:

- Find a color buddy who can help with shopping for items like clothes or paint.
- Memorize the correct order of colors on things like traffic lights.
- Download apps that help you identify colors in the world around you.

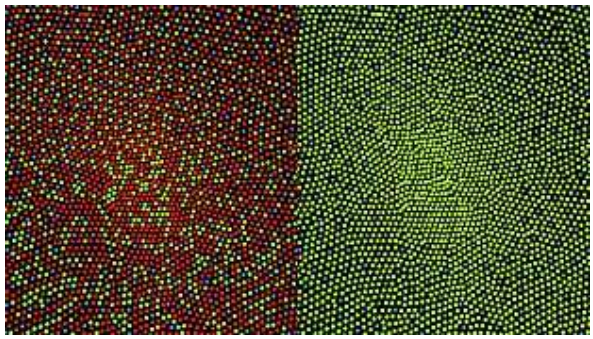

***Illustration of the distribution of cone cells in the fovea of an individual with normal color vision (left), and a color blind (protanopic) retina. The center of the fovea holds very few blue-sensitive cones.***

## CONCLUSION

In this condition, the person has all three cone types, but M cones are less sensitive to green light than they should be. Mostly see blues, yellows and generally muted colours. The colour blindness can be diagnosed by the Ishihara test, Colour vision test. Colour vision tests and Genetic testing.

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