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Investigation into Congenital Color Vision Deficiency (CVD)

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Abstract. Most humans have trichromatic color vision due to adaptation for gathering food with more energy in rainforest habitats. However, about 8−10% of males and 0.4−0.5% of females have abnormal color vision, including dichromacy and monochromacy. With the trend of urbanization, the issue of difficulties distinguishing colors for individuals with congenital color vision deficiency is becoming increasingly severe. This essay aims to compare the explanations for phenomena related to color perception. The topics include the basic mechanism of trichromacy, the evolution of color vision in primates, and information about congenital color vision deficiency. After reviewing relevant literature, it is shown that the causes of anomalous trichromacy and dichromacy still lack consensus, and there are arguments about the basic mechanisms behind different classifications of colour vision deficiency. To support research on therapies for relieving abnormal color vision conditions, this essay summarizes the possible genetic causes of mutations.

Keywords: Color vision, Color blindness, Trichromacy

1. Introduction

Color vision deficiency (CVD) is also called color blindness, which is defined as the disability to distinguish colors. The prevalence of color vision deficiency exhibits sexual dimorphism, affecting approximately 8–10% of males and 0.4–0.5% of females [1]. This variation is due to red-green color blindness being X-chromosome linked recessive inheritance, the genes OPN1LW and OPN1MW, which code for L and M opsins, are located on the X-chromosome [2]. A female with two copies of X chromosomes has a greater chance for a functional copy to substitute the mutated chromosome.

Inborn or acquired factors can cause colour vision deficiency; the former is congenital and the latter is acquired. This disease bothers about 90% of people with color blindness and 67% of people with color weakness and half of the patients cannot distinguish traffic lights [3]. In many countries, people with colour blindness are excluded from obtaining a driving license, and their career opportunities are also restricted, especially in experimental subjects that involve observation in colours and jobs related to art [4]. To support research on therapies for relieving abnormal color vision conditions, this essay summarizes the possible genetic causes of mutations.

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2. Mechanism of color vision

2.1. Photoreceptors

Light is the foundation of vision, but the color that eyes detect are not the real color on the surface of materials. Eyes can detect different colours in three dimensions: hue, brightness and saturation. Hue is classified into two types, the chromatic hues contain different colors, which is detected by different wavelengths of light. Achromatic hues are only black and white; they only measure light intensity, instead of the wavelength of light. The last one, saturation, is measuring how pure the color is, describing the proportion of gray contained in a color. The color with high saturation is more pure, and the color with less saturation is dull [5]. Adjusting these three data allows any color on the color disc. After light reaches the environment's surface, it will be reflected and finally reach the eyes. Eyes detect light's brightness, hue and saturation and turn the light energy into electrical impulses the neuron can carry. The photoreceptor, which is located on the retina, achieves this function. Light that reaches the eyes is absorbed by the proteins, opsins, which are located on a membrane called the outer segment [6].

In vertebrates, the photoreceptors on the retina can be classified into two types: cone cells and rod cells. Rod cells are more sensitive in dim light environments, which function in dark environments. However, the disadvantage of a rod cell is that it only detects the light intensity; without detecting the wavelength of light, it provides an achromatic image with only white and black colours. Opposite to the rod cells, cone cells only function in normal light environments with higher light intensity, because higher light energy is needed to trigger the sensitivity of cone cells. The benefit of cone cells is that they can distinguish different wavelengths of light, providing a clear colour image. However, the disadvantage is that cone cells are less sensitive to light intensity and cannot function in dark environments [7].

2.2. Young–Helmholtz theory

In the 1900s, the study of colour vision started with an argument between two theories about the basic mechanisms of human color vision. The one that was introduced earlier, introduced by Thomas Young and Hermann von Helmholtz, was named “the trichromatic theory”. Thomas Young and Hermann von Helmholtz believed that three types of cone cells contain opsins to absorb light. The different structures of opsins classify these three types of cone cells they contain; each is sensitive to a specific wavelength of light. L cone (long wavelength sensitive), most sensitive to long wavelength, has the peak at 570 nm, located in red color in spectrum. M cone (middle wavelength sensitive), most sensitive to middle wavelength, has the peak at 540 nm, located in green color in spectrum. S cone (short wavelength sensitive), most sensitive to short wavelength, has the peak at 440 nm, located in the blue colour in the spectrum. The important fact is that the cone cells can detect a broad range of light with different wavelengths on the light spectrum, so there is also overlap between each type of cone cell. However, all these cone cells are most sensitive to the specific wavelength of light they specialise in [8].

The trichromatic theory posits red, green, and blue as primary colors. These are defined as three colors of equal luminance, maximum saturation, and distinct hues on the color wheel. Mixing these in varying ratios can generate any color within the visible spectrum. The three primary colors must be mixed in equal proportions to make the white light. The wavelength covered by these three types of cone cells is the visible light range on the light spectrum. The visible light has a wavelength of about 310 nm, near the ultraviolet for children, to about 1100 nm, next to the infrared red light [9].

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However, there is a significant limitation on the trichromatic theory. This hypothesis fails to explain why individuals with red-green color blindness, who lack either L or M cones, often perceive red and green as yellow. According to the trichromatic theory, these individuals can't sense the white light, due to the absence of one primary color.

2.3. Hering's theory

The limitations of the trichromatic theory failed to explain the phenomenon of color blind individuals having normal vision in distinguishing the brightness of light. Hering's opponent process theory explains color perception through four primary colors: red, green, blue, and yellow. These are arranged in opposing pairs (red-green, blue-yellow), with white-black handling brightness. Mixing opponent colors yields white light. An important feature of these opponent pairs is the absence of intermediate colors between the two extremes. For instance, there is no such thing as a "red-green" or "blue-yellow" color [5].

A pair of opponent colors will have an opposite effect on the same neuron. If one of the color will excite it, then its opponent color will inhibit it [5]. For example, color-opponent neurons compare red-green and yellow-blue signals: red/yellow excite, green/blue inhibit, white excites, and black inhibits. This ensures that the brain processes color contrasts efficiently, enhancing our ability to distinguish between complementary hues. In these three pairs of opponent colors, the white-black group is the most sensitive, needs less light energy to trigger a response. This group provides achromatic vision without color [10].

2.4. Compromise theory

Bowmaker, Dartnall, and Mollon's 1980 study used microspectrophotometry to identify three cone cell types in the human retina: S (short wavelength), M (middle wavelength), and L (long wavelength) cones. Their technique measured the precise light absorption spectra of each cone type. Their findings showed clear sensitivity peaks in human cone cells, S cone is most sensitive to 425nm light, M cone is most sensitive to 534nm light, and L cone is most sensitive to 564 nm [5]. This proves the Young and Helmholtz's theory, which suggests color perception is provided by coactivity of three types of photoreceptors, each sensitive to different wavelength lights. Yet, this discovery did not deny Hering's opponent-process theory. Instead, it clarified Hering's theory, which describes color perception in three pairs of opponent colors: red-green, blue-yellow and white-black. Unlike the color perception by photoreceptors on the retina, comparing pairs of opponent colors does not operate at the retina's or photoreceptors' level. These electrical impulses are modified in the neurons in the brain, then form images with color [5].

Valois and De Valois proposed the compromise theory in 2011, integrating trichromatic and opponent process theories for comprehensive color perception. Color processing occurs in two stages: cone cells convert light into electrical signals, which are then processed in the central nervous system to produce color perceptions [11]. In conclusion, the trichromatic theory is the colour collection in the retina, and the opponent theory is the processing in the brain. These two processes combined to form the foundation of human color vision.

3. Evolution of trichromacy in primates

Primates are a diverse order of mammals and the only group of trichromatic species. Old World and New World monkeys represent two major groups in this order. Continental drift initiated the

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divergence of these groups approximately 40-50 million years ago. Old World monkeys inhabit regions across Central and South America, Africa, Asia, and Europe, differing primarily in color vision: Old World species are trichromatic, whereas New World species exhibit variability between dichromacy and trichromacy. For example, in Saimiri sciureus, a New World monkey, two-thirds of females are trichromatic, while all males and the remaining one-third of females are dichromatic [12]. The difference in their gene sequence will be the key to discovering the evolution of trichromacy.

Humans (Homo sapiens) are a kind of Old World monkey, and most humans with normal color vision are trichromatic. The genes that code for L, M and S opsins are OPN1LW, OPN1MW, and OPN1SW. OPN1LW and OPN1MW genes are located on the X-chromosome at Xq28, arranged in a tandem array, and are very similar. And 98% of the gene sequences are the same, but they only share 40% identity to the OPN1SW gene located on chromosome 7 at 7q32. This suggests that OPN1LW and OPN1MW genes have a more recent relationship than OPN1SW [13]. The diversion of OPN1LW and OPN1MW genes may result from gene duplication, and the ability to distinguish fruit and young leaves that contain nutrients in the rainforest environment may be a driving force for the evolution [14].

Old World monkeys possess stable trichromatic vision due to fixed expression of OPN1MW (M cone) and OPN1LW (L cone) genes on the X chromosome, plus an autosomal OPN1SW gene. New World monkeys exhibit "allelic trichromacy" with a single, variable X-linked gene encoding opsins between L and M cones. Males are dichromatic. Heterozygous females can be trichromatic, while homozygous females remain dichromatic, similar to males [15].

4. Classification of congenital color vision deficiency

Congenital color vision deficiency, or color blindness, is a prevalent inherited visual disorder affecting color discrimination, with an incidence of approximately 8% in males and 0.5% in females. Typically, it arises from mutations in genes encoding photoreceptor opsins. Classification is based on the number of discernible primary colors, categorized as anomalous trichromacy, dichromatism, or monochromatism.

4.1. Anomalous trichromacy

Anomalous trichromacy has the most mild symptom among congenital color vision deficiency types. According to the type of color perception that is abnormal, this situation will be classified as protanomaly, deuteranomaly, or tritanomaly. Protanomaly, the most common, results from L cone pigment changes, causing aberrant red perception. Deuteranomaly, the second most frequent, stems from M cone pigment alterations, leading to abnormal green perception. Tritanomaly, the rarest, arises from S cone pigment changes, causing aberrant blue perception [16].

There are two different theories about the mechanism of this situation. Scientist MP Simunovic suggests the individuals with anomalous trichromacy have the same three types of cone cells L, M and S cone cells that can provide three primary colors: red, green and blue as normal trichromacy and the main problem that leading to anomalous color perception is the abnormal mixture of colors [17]. However, another scientist, Sung Eun Song Watanabe, and Scientist Paula Yuri Sacai suggest that it is due to a change in the structures of cone pigments, which leads to the abnormal functioning of cone cells [8]. Scientist MP Simunovic suggests the problem is at the color processing stage, and scientists Sung Eun Song Watanabe and Paula Yuri Sacai indicate the problem is at the light energy

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collection by the cone cells. The extent of abnormal color perception varies, some individuals have nearly normal trichromacy.

4.2. Dichromacy

Dichromacy is more severe than anomalous trichromacy, since only two primary colors are available, so the number of colors that can be perceived is limited. According to the type of cone cell that is absent or nonfunctional, this situation will be classified as protanopia, deuteranopia, or tritanopia. Protanopia is due to no functional L cone (long wavelength sensitive) on the X chromosome, leading to a lack of red color [16]. Deuteranopia is due to the lack of a functional M cone on the X chromosome, leading to a lack of green color. These two diseases are recessive on the X chromosome.

There are two different theories about the mechanism of this situation. One theory suggests that another functional normal cone cell replaces the type of mutated cone cell. Another theory suggests that the mutated cone cell stays at the original location but is not functional. After investigation on one individual with non-functional cone cells and one individual with functional alternative cone cells, non-functional cone cells occur when the first two genes in the X-chromosome array are replaced by a gene that encodes a non-functional pigment, and replacement of functional cone cells occurs when the first two genes code for the same type of cone cells [18].

Ritanomaly is scarce, about 1 in 65,000, compared to protanopia and deuteranopia, about 1 in 13,000 [19]. It is due to a mutation of the gene sequence on chromosome 7 that codes for opsin in S cone (short wavelength sensitive), which will lead to abnormal color perception of blue color. L. N. Went and N. Pronk investigate the inheritance sequence of tritan disturbances and find that it is dominant inheritance on autosomes [20].

4.3. Monochromatism

Monochromatism includes S cone and rod types, with the former, caused by Xq28 mutations affecting L and M cones, resulting in blue-only primary color function, leading to yellow-blue but not red-green color vision [8]. Individuals with this situation also have other symptoms: low vision, photophobia, scotopic responses that are slow and absent to dim light, and photopic responses that are slow in bright light [16].

Rod monochromatism is also called achromatopsia; it is a sporadic disease, with a probability of about 1 in 30,000–50,000 and has an autosomal recessive inheritance pattern [21]. The defining characteristic is the complete absence of cone cell function, resulting in images formed solely from rod cell signals. This restricts vision to black-and-white discrimination, causing total color blindness [22]. Achromatopsia always leads to inferior color vision, poor vision and photophobia.

5. Conclusion

This paper reviews past theories on trichromacy, primate color vision evolution, and congenital color vision deficiency. While prior research on color vision mechanisms was conflicting, the 2011 compromise theory reconciles these views. By examining the evolutionary history and trichromacy inheritance patterns of New and Old World monkeys, this work supports the recent duplication origin of L and M cone diversity. A growing number of studies about color vision deficiency provide basic theories for therapy for these diseases. This essay classified the color vision deficient into anomalous trichromacy, dichromacy and monochromacy. After reviewing relevant literature, it is

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shown that the causes of anomalous trichromacy and dichromacy still lack consensus, and there are arguments about the basic mechanisms behind different classifications of colour vision deficiency.

Unfortunately, the paper omits discussion on why some individuals have dichromatic or monochromatic vision, considering the evolutionary advantage of red-green color vision for food acquisition in Old World monkey ancestors. This study summarizes the causes of color vision deficiency to aid in developing therapies that enhance the quality of life for those with impaired color vision.

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