v. 4, n. 15, 2024

International Journal of Health Science

BIOPHYSICAL-GENETIC ASPECTS OF CONGENITAL DYSCHROMATOPSIA: A DESCRIPTIVE REVIEW

Rayssa Almeida Nogueira
Centro Universitário Redentor, medicine course
Itaperuna – RJ
https://orcid.org/0000-0002-5998-6367

Robison Antônio Coelho Júnior
Centro Universitário Redentor, medicine course
Itaperuna – RJ
http://lattes.cnpq.br/7420174932492024

Natália Vinco Guarnier
Centro Universitário FAMINAS, medicine course
Muriaé – MG
http://lattes.cnpq.br/4100930978722376

Alice Moreira Silveira
Centro Universitário Redentor, medicine course
Itaperuna – RJ
https://orcid.org/0009-0002-4612-0406

Anna Carolina Fock Tasca
Centro Universitário Redentor, medicine course
Itaperuna – RJ
https://orcid.org/0009-0002-0128-3906

Alexandra de Carvalho Matos
Centro Universitário Redentor, medicine course
Itaperuna – RJ
https://orcid.org/0009-0007-7873-1523

All content in this magazine is licensed under a Creative Commons Attribution License. Attribution-Non-Commercial-Non-Derivatives 4.0 International (CC BY-NC-ND 4.0).

Luíza Coelho
Centro Universitário UNIFACIG, medicine course
Manhuaçu – MG
http://lattes.cnpq.br/9439127624982817

Tayná Sudré Viana
Universidade Nove de Julho, medicine course
São Bernardo do Campo - SP
http://lattes.cnpq.br/6486779561696394

Felipe Terra De Martin Galito
Centro Universitário Redentor, medicine course
Itaperuna – RJ
http://lattes.cnpq.br/1763004172818387

Samira Vitório Menezes Seródio
Faculdade Metropolitana São Carlos, medicine course
Bom Jesus do Itabapoana – RJ
http://lattes.cnpq.br/1969281294730922

Rodolfo da Silva Gonçalves
Centro Universitário Redentor, medicine course
Itaperuna – RJ
http://lattes.cnpq.br/1104817185397878

Felipe Tavares Souza
Faculdade Metropolitana São Carlos, medicine course
Bom Jesus do Itabapoana – RJ

Abstract: Congenital dyschromatopsia, better known as color blindness, is the name given to a defect in color vision, linked to gender, where there is difficulty in detecting and recognizing different colors. This condition, which can occur due to trauma, tumors, injuries and even retinal detachment, is predominantly hereditary, resulting from a deficit in the production of genes encoding photopsins. Therefore, the present study aims to analyze the biophysical and genetic aspects surrounding dyschromopsia, describing the related mechanisms and highlighting the influences of this dysfunction on the lives of patients affected by this condition. To develop the present study, original articles were used, written in English and Portuguese, published in the last 34 years, in the main bibliographic databases, such as SciELO and PubMed, in order to guarantee greater reliability of the information presented in the project. It was then realized that congenital dyschromatopsia is a dysfunction that mainly affects men, due to its recessive character linked to the X chromosome. Furthermore, it generates visual changes that manifest themselves in accordance with the characteristic of the dysfunction, which can be deuteranopia, trypanopia or protanopia. Although it has a strong associated genetic character, this dysfunction can currently be corrected, which contributes to improving the quality of life of patients with it, as well as their inclusion in the social environment, which is sometimes hampered by the visual alteration presented. This way, technology is a great ally in advances in visual health, through the creation of lenses, such as ColorMax and EnChroma, as well as inclusive projects, such as the Color Identification System for Color Blind, ColorADD and Feelipa, which expand access to healthcare for the affected population and restore visual quality, which was, until then, compromised by the disease.

Keywords: Color blindness; Congenital dyschromatopsia; Dyschromopsia; Genetics.

INTRODUCTION

The meaning of color encompasses denotative and connotative concepts. In a denotative sense, color is often used as a comment on some object, such as, for example, "the bicycle is red", "the sock is gray". Connotative meanings are the associations suggested by a color, for example, on a traffic sign the color red informs that the intersection is not free for movement. Circumstances in which connotative meanings are used include, in addition to traffic signs, electricity warnings and map statements (COLE, 2007).

Color blindness, also called dyschromatopsia or dyschromopsia, is a visual disorder responsible for difficulty in determining colors (BRUNA, 2015). The word "color blindness" is popularly used as a synonym for dyschromatopsia, in honor of the famous chemist and scientist John Dalton, who had protanopia (a type of dyschromatopsia) and was the first scientist to study and understand this disease. Currently, the main method used to diagnose this condition is the Ishihara test, which consists of discs of varying sizes and the same color, with more central circles with different colors, in order to evaluate the recognition and interpretation of colors, through the numbers formed by the joining of these discs (BRUNI; CRUZ, 2006).

In most cases, color blindness is caused by an inherited genetic condition, therefore, cases of color blindness that are caused by trauma to one of the structures of vision, tumors or lesions in the brain, or retinal detachment are rare (BRUNA, 2015).

Furthermore, color blindness has a biophysical aspect, since affected individuals do not have the number of cones, or the cones present some anomaly in their photoreceptors, which results in poor color differentiation (MOURA, 2019). Therefore, color vision is a complex process, which involves special photosensitive cells, the cones. In the human retina, there are around 5 million cones and each one has its own class of photopsin (red, green and blue) which is a protein with the function of modifying the light signal into an electrical signal (GORDON, 1998).

Congenital changes in color vision are a consequence of changes in the genes encoding photopsins and are divided into: anomalous trichromatism (when one of the three photopsins has its light absorption spectrum directed to another wavelength), dichromatism (when that there is a lack of one of the types of photopsins) and monochromatism (a very rare circumstance characterized by the presence of only one of the photopsins, usually blue) (COLE, 2007).

Furthermore, people with dyschromatopsia face difficulties in their daily lives, in simple activities, such as, for example, when they need to understand maps and graphs. Colors also play a fundamental role in a child's development, as essential symbols are taught at school for understanding colors as a form of universal identification (HRUBA, 2018; CUNHA; CRUZ, 2016). However, there are currently treatments that improve the quality of life of patients with color blindness, such as ColorMax and EnChroma lenses, and inclusive projects such as the Color Identification System for Color Blind, ColorADD and Feelipa (HENRIQUES et al., 2016).

Therefore, this article aims to address color blindness, discussing its genetic aspect, and how this condition alters the color perception of the affected individual in comparison to the vision of a normal person, through a biophysical analysis. Furthermore, it is also necessary to explain how the disease is identified in carrier patients, through tests, how this condition affects the social life of carriers and the technological means to adapt

to this condition, which aim to guarantee visual and improve patients' quality of life.

METHODOLOGY

To construct this article, exploratory research was used, through a bibliographical survey. Data collection took place from original articles, published in Portuguese, English and Spanish, published in the last 34 years, in the main databases, such as SciELO and PubMed, in order to guarantee the reliability of the data presented and the veracity from the project. As search criteria, the keywords "color blindness", "congenital dyschromatopsia", "dyschromopsia" and "genetics" were used".

BIBLIOGRAPHIC REVIEW

CLASSIFICATION

Dyschromatopsia can be classified into protanopia, achromatic, deuteranopia and tritanopia. Protanopia is when the individual has a change in the red pigment, either a decrease or its absence, or waves that are long in length. This way, he only sees shades of beige, brown, green or gray (BRUNA, 2015; XAVIER, 2012). Achromatic, in turn, is when the individual can only see the tones: white, gray and black (MOURA, 2019). Deuteranopia is when the individual has a change in the green pigment, be it a decrease or its absence, or waves that are of medium length. In the case of absence, the individual sees brown tones (BRUNA, 2015; XAVIER, 2012) And tritanopia, finally, is when the individual presents a change in blue and yellow pigments, or, in waves that have a short length. In this case, the blue and yellow pigments will be seen by the individual as pink tones (BRUNA, 2015; XAVIER, 2012).

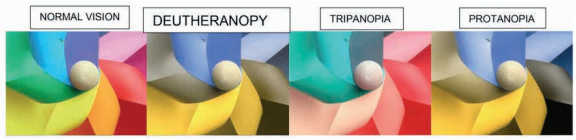

Figure 01. Types of Color Blindness.

Source: Neo Visão Ophthalmology (*online*).

BIOPHYSICAL ASPECT OF VISION AND COLOR BLINDNESS

Vision is one of the five special senses of human beings. It is linked to self-perception, which occurs through the capture of light stimuli by photoreceptor cells, which will later be processed by the brain to form images as we know them. This sense is of great importance, as, from it, it is possible to perceive colors and light intensities (COSTA, *online*).

The eye can be related, optically, to a common photographic camera, being composed of lenses, light regulation (pupil) and the retina corresponds to photographic film (GUYTON; HALL 2011). It functions as an organ for selectively converting light into action potentials, given that during their journey through the visual system, these electrical stimuli are selected until they generate a visual impression in the occipital cortex. This light coming from the observed object passes through the transparent media of the eye until it reaches the retina, and there, through mainly two cell types, the cones and rods, it is transduced into electrical impulses that are carried by the nerves and optic pathways to the cortex. occiput where it will be formed (RAMOS, 2016).

In the retina are located the cones that are responsible for color vision and the rods that are of great importance for seeing in dim situations, mainly forming black and white (GUYTON; HALL 2011). These cell types have chemical substances in their composition that are decomposed with exposure to light and during this decomposition the process of excitation of the optic nerve occurs. The

chemical substance present in rods is called rhodopsin, and, in the case of cones, they are called cone pigments or colored pigments, which are discretely different from rhodopsin. (GUYTON; HALL 2011).

The processing of color perception is said to be the result of a visual sensation, which is caused by the absorption of photons through the retina's cones and these colors, or wavelength, are mainly determined by three basic characteristics, which are tone, saturation and brightness (BRUNI; CRUZ, 2006).

Color vision is a complex mechanism, as it involves photosensitive cells and, based on the most accepted model, there are three colors that they can make alone and based on the variation in frequency and length of light waves, it is possible to achieve this. transduce the entire range of colors (MELO, 2014). When there is a problem related to any of the cones, red, green or blue, it causes that person to be unable to distinguish any color, and then they experience color blindness (GUYTON; HALL 2011).

People with normal vision have 3 types of cones, each of which corresponds to certain colors: L cones correspond to red, S cones correspond to blue and M cones correspond to green. The L means, in English, "long", which refers in Portuguese to long wavelengths; the S, "short", for short wavelengths and the M, "medium", for medium wavelengths. Therefore, if any of the cones does not work, or works partially, it leads to a decrease in the ability to perceive colors, that is, it leads to color blindness (ZEISS, 2017).

GENETIC ASPECT

Color blindness, in most cases, is a hereditary, recessive disorder linked to the sex chromosome (allosome) X. Due to being linked to the X chromosome, color blindness affects women less, since women have two X sexual chromosomes, and, therefore, for color blindness to be expressed in women, both X chromosomes must have the recessive gene for color blindness. Unlike what happens with men, who only have one X chromosome, if this chromosome carries the recessive gene, they will have color blindness (BRUNA, 2015; MOURA, 2019). However, although a woman has a normal phenotype, that is, she can differentiate between colors, she may or may not have an X chromosome that has the recessive gene for color blindness and, if she does, this gene can be transmitted to her their descendants (BRUNA, 2015; MOURA, 2019).

Below is a summary, in table form, of the aforementioned and which specifies the genotypes and phenotypes corresponding to each situation, in both genders:

Gender	Genotype	Phenotype
Feminine	X^DX^D	Normal
	X^DX^d	Normal
	X^dX^d	Colorblind
Masculine	X^DY	Normal
	X^dY	Colorblind

Figure 02. Phenotypes and Genotypes of Color Blindness.

Source: Adapted from SANTOS (online).

SOCIAL AND EDUCATIONAL ISSUES

People with dyschromatopsia may experience difficulties in their daily lives, for example, in decoding and understanding information from maps and graphs that require colors to be interpreted. It is also necessary to use colors at traffic lights; however, color blind people can identify due to the position of the light. And in activities considered simple and commonplace, such as sending a form, color-blind people experience obstacles when the only way to notify them that it is not filled out correctly is the red outline (HRUBA, 2018).

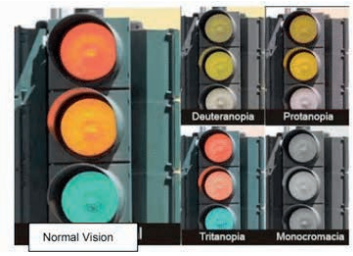

Figure 03. Simulation of color-blind conditions at a traffic light.

Source: Adapted from HRUBA, 2018.

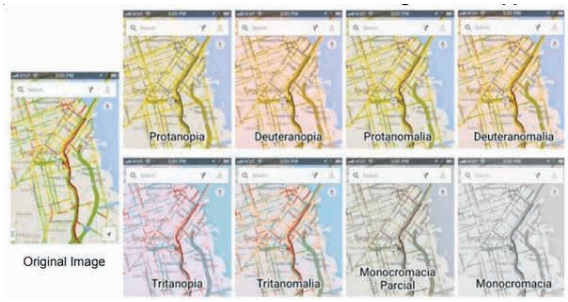

Figure 04. Color Blindness Simulation in the Google Traffic application.

Source: Adapted from WERB (2015) apud HRUBA (2018, p.26).

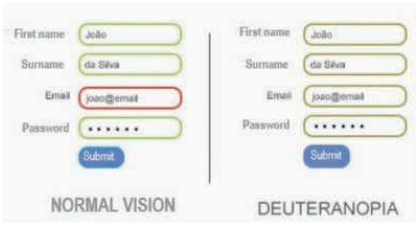

Figure 05. Example of a non-accessible form viewed by colorblind people.

Source: Adapted from GUSTAFSON (2015) apud HRUBA (2018, p.27).

Furthermore, the use of colors, in addition to being present in everything around us, has a direct link with the child's development. Therefore, it is up to the school to understand eye deficiency, reflect and develop pedagogical inclusion projects, since the suspicion of color blindness arises in childhood, between two and three years of age, however, many only take tests and identify it in adulthood.

However, it is important that the diagnosis occurs in childhood, when color learning occurs, so that inclusion can occur and not hinder the child's learning due to anomalies, such as color blindness. Therefore, with early identification, it is possible for schools to teach fundamental symbols for understanding colors, as in the universal identification system, ColorAdd, and this process needs to occur continuously and constantly, considering attention, observation, interaction and painting, providing social inclusion and facilitating everyday life in the present, as well as in the future of this color-blind child (CUNHA; CRUZ, 2016).

INTERVENTIONS

Due to the advancement of the printing industry and technology, the use of colors began to be used with greater frequency and flexibility, not only for aesthetic purposes, but also for organizational and informative purposes. Therefore, it was necessary for products and environments to take into consideration, the needs of individuals with color blindness, with strategies such as color representations via symbols and the choice of color palettes that do not cause confusion for the three most common types of this anomaly. And, based on studies, although color blindness has no cure, minimizing the discomfort of color blind people is possible, through ColorMax and EnChroma lenses and inclusive projects such as the Color Identification System for Color Blind, ColorADD and Feelipa (HENRIQUES et al, 2016).

Figura 06. ColorADD.

Source: Coloradd (*online*).

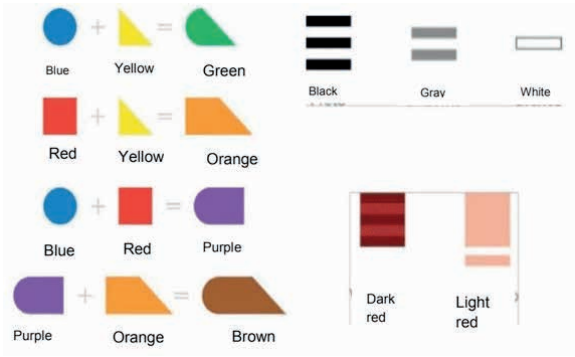

Figure 07. Graphic representation of colors and tactile feelipa code.

Source: Feelipa Color Code (*online*).

These adaptations for color blind people promote more autonomy and independence for those with the anomaly, therefore, ColorADD is already applied in some places, such as in the metro signaling system in the city of Porto, in Portugal, and can also be applied in colored pencils, paint packaging, emergency signage, labels, clothing and card games (HENRIQUES et al, 2016).

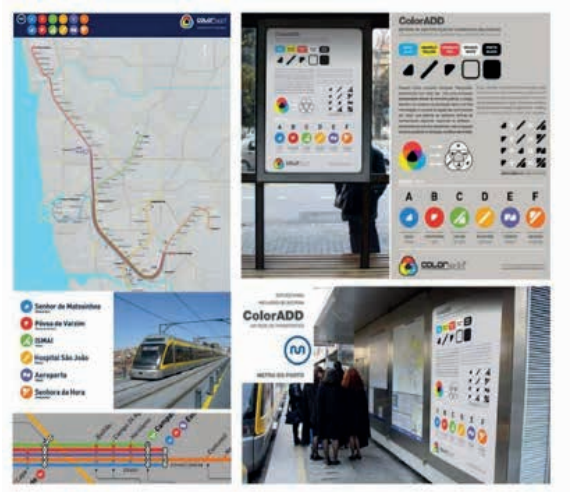

Figure 08. ColorADD in subway signage in Portugal.

Source: Coloradd (*online*).

TESTS

Due to the phenotypic variety of dyschromatopsia, evaluation is important and psychophysical methods become essential for its diagnosis. This way, four types of tests can be applied: ordering tests, which can measure and classify the degree of severity of the condition, and consist of making individuals group, in a sequence, colored pieces, according to a similarity parameter; equalization tests, which aim to identify and classify, through colored spectral projection, colors, with the individual having to equalize two different ones; measurement tests, which identify and measure the ability to appropriately discern colors in everyday activities, with the person having to name the color corresponding to a pre-established stimulus; and pseudoisochromatic figure tests, which also identify and classify, through groupings of circles with different luminances and sizes (FARIAS, 2015).

The most used and well-known test in the world for diagnosing dyschromatopsia is the Ishihara Test, which was published in 1906, and after that it had its variations and reproductions. Thus, nowadays, versions of 24 and 38 boards are available, and the most commonly used editions contain numbers and lines drawn as objects for identification. The Ishihara Test, after evaluations, proved to be the most effective test for rapid identification of congenital color vision disorders. However, this test does not provide a quantitative assessment of the disability, as there are no severity measurement boards (BRUNI; CRUZ, 2006).

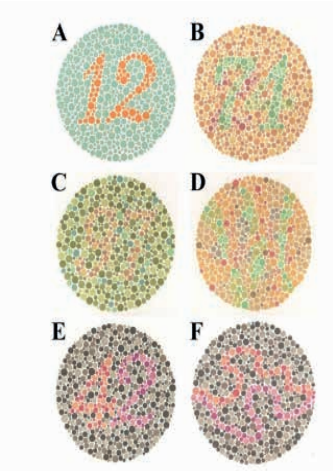

Figure 09. Pseudoisochromatic Ishihara boards. A - Introductory; B - Transformation type; C - Disappearance; D – Hidden digit type; E – Classification type; F – for illiterate individuals.

Source: Adapted from FARIAS (2015, p.44).

The introductory board can be seen by all individuals, while the transformation board can be viewed by trichromats (and only they see the disappearance board). Furthermore, for people with dyschromatopsia, another number is visualized (and only they see the hidden digit type board), and for some individuals, on the classification type boards, only one of the digits is seen, and there is also a variation used only for illiterate subjects (FARIAS, 2015).

FINAL CONSIDERATIONS

From this bibliographic research, it was possible to verify that color blindness is, in the majority of affected people, a hereditary genetic condition, originating from a recessive gene, linked to the X sex chromosome and, as a consequence, affected people have poor color differentiation., through a change or absence of cones. Although color blindness is a prevalent condition, there is no type of law or public policy that addresses this condition, which can lead to poor learning consequences for those with it, and also locomotor problems on public streets, as referenced in the literature. The use and improvement of technologies used for the social reintegration of people with color blindness are possibly strategies that will increasingly provide comfort and inclusion for these patients, in addition to enabling greater cognitive and intellectual development, as well as minimizing the negative impacts of dyschromatopsia on the quality of life of sufferers.

REFERENCES

BRUNA, Maria Helena Varella. **Doenças e Sintomas: Daltonismo**. Drauzio Varella, *online*. 2015. Disponível em: < https://drauziovarella.uol.com.br/doencas-e-sintomas/daltonismo/>. Acesso em 12 jun. 2020.

BRUNI, Lígia Fernanda; CRUZ, Antônio Augusto Velasco e. **Sentido cromático: tipos de defeitos e testes de avaliação clínica**. Arq Bras Oftalmol. v.69, n.5, 2006.

COLE, Barry L. **Assessment of inherited colour vision defects in clinical practice**. Clin Exp Optom, v. 90, n. 3, p. 157-175, 2007.

COLORADD. **CODE ColorADD.** *Online*. Disponivel em: <http://www.coloradd.net/code.asp>. Acesso em: 26 jun. 2020.

COSTA, Marcelo. **Biofísica da Visão.** (*online).* s/d. Disponível em: <http://repositorio.unicentro.br:8080/jspui/bitstream/123456789/1066/1/COSTA_Biof%C3%ADsica%20da%20vis%C3%A3o.pdf>.

CUNHA, Arielly Kizzy; CRUZ, José Anderson Santos. **Inclusão pedagógico cultural - daltonismo e o ensino de cores da educação infantil**. Rev. on line de Política e Gestão Educacional. v.20, n.3, 2016.

FARIAS, Letícia Miquilini de Arruda. **Correlação entre parâmetros estimados pelos testes Colour Assesment and Diagnosis and Cambridge Test na avaliação da discriminação de cores**. Belém, PA: Repositório UFPA. 2015.

FEELIPA COLOR CODE. **Código de cor: para deficientes visuais.** *Online*. Disponível em: <http://www.feelipa.com/pt/para-deficientes-visuais/>. Acesso em 26 jun. 2020.

GORDON, Neil. **Colour blindness**. Public Health, v. 112, n. 2, p. 81-84, 1998.

GUYTON, Arthur Clifton.; HALL, John E. **Tratado de fisiologia médica**. 12. ed. Rio de Janeiro: Elsevier, 2011.

HENRIQUES, Fernanda; GADOTTI, Marcella; IAMAGUTI, Mariana Shizue**. Democracia cromática: dispositivos e códigos de representação da cor para portadores de daltonismo e baixa visão**. Belo Horizonte: Congresso Brasileiro de Pesquisa e Design, Out, 2016.

HRUBA, Felipe Franchini. **Desenvolvimento de interfaces web adaptado para portadores de daltonismo.** Ponta Grossa: Repositório ROCA, 2018. Disponível em: <http://repositorio.roca.utfpr.edu.br/jspui/bitstream/1/10411/1/PG_COCIC_2018_2_01.pdf>. Acesso em: 03 jun.2020.

MELO, Débora Gusmão; GALON, José Eduardo Vitorino; FONTANELLA, Bruno José Barcellos. **Os" daltônicos" e suas dificuldades: condição negligenciada no Brasil?**. Physis: Revista de Saúde Coletiva, v. 24, n. 4, p. 1229-1253, 2014.

MOURA, Marcello. **Detetive das Cores: Aplicativo para identificação e assimilação das cores para crianças daltônicas**. Universidade Federal do Rio de Janeiro, Centro de Letras e Artes, Escola de Belas Artes e Departamento de Comunicação Visual BAV, 2019.

NEO VISÃO OFTALMOLOGIA. **O que é Daltonismo?**. *Online.* Disponível em: <http://www.neovisao.com/saude-ocular/daltonismo/>. Acesso em: 6 de junho de 2020.

RAMOS, André. **Fisiologia da visão**. Um Estudo Sobre o "Ver" e o "Enxergar". Análise do Simbólico no Discurso Visual, PUC (Universidade Católica do Rio de Janeiro), Brasil, 2006.

SANTOS, Vanessa Sardinha dos. **Herança ligada ao sexo**. *Online*. Disponível em: <https://alunosonline.uol.com.br/biologia/heranca-ligada-ao-sexo.html>. Acesso em: 13 jun. 2020.

XAVIER, Joaquim José. **Parecer conselheiro n° 010/2011: daltonismo**. Conselho Regional de Medicina do Estado do Rio Grande do Sul, 2012.

ZEISS. **Compreendendo a visão: Deficiência vermelho-verde, cegueira para vermelho e verde e cegueira total das cores. Quais são os tipos de cegueira e deficiências para cores? Como reconhecê-las?**. 2017. Disponível em: <https://www.zeiss.com.br/vision-care/melhor-visao/compreendendo-a-visao/daltonismo-deficiencia-vermelho-verde-cegueira-para-vermelho-e-verde-e-cegueira-total-das-cores.html>. Acesso em: 6 jun. 2020.