

RESEARCH ARTICLE

Hereditary Dyschromatopsia: Two Cases at an Ophthalmology Centre in Guadeloupe

Baldé Ak¹, Baldé AI^{1,2}, Sylla A^{1,2}, Sovogui MD^{1,3}, Bah TM¹, Magassouba A¹, Hann F¹, Diop MS⁴

¹Faculty of Health Sciences and Technology (FSTS).

²Camp Samory Touré Military Hospital.

³Bartimé Hospital.

⁴Labé Regional Ophthalmology Centre.

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Corresponding Authors: Baldé Ak, Faculty of Health Sciences and Technology (FSTS).

Abstract

Introduction: Colour blindness is a genetic vision disorder that alters colour perception. It is caused by cones, normally located on the retina and responsible for receiving colours, which are absent in people with this condition. It affects 9% of men and 0.5% of women.

Materials and methods: This was a descriptive cross-sectional study covering a six-month period from 1 August 2023 to 31 January 2024.

Results: The 100 HUE test was completely distorted. The notion of dyschromatopsia in the family, the results of the Ichihara tests and the 100HUE test confirmed the diagnosis of colour blindness. Treatment consisted of vitamin therapy, photochromic lenses and advice on careers he could pursue later in life for better social integration.

Conclusion: Gene therapy offers new perspectives in treatment. Colour-filtering contact lenses have been successfully synthesised using gold nanoparticles with HEMA and EGDMA as the base polymer and cross-linking agent, respectively. These lenses can significantly improve the situation for people with colour blindness.

Keywords: Dyschromatopsia, Hereditary, Colour Blindness, Guadeloupe.

1. Introduction

Colour vision disorders are grouped under the term «dyschromatopsia». These deficiencies can be hereditary or acquired.

Colour blindness is a genetic vision disorder that alters colour perception. It is caused by cones normally located on the retina and responsible for receiving colours that are absent in colour-blind individuals [1]. Hereditary dyschromatopsia is present in 9% of men and 0.5% of women [2].

Although hereditary dyschromatopsia cannot be treated, it must always be screened for and assessed,

as its presence can lead to restrictions or professional incapacity.

The most common types of colour blindness are red-green colour blindness, deuteranopia and tritanopia, which are rarer.

Nowadays, there are ways to improve the vision of people with colour blindness, such as visual aids that enable them to better identify colours, including coloured lenses and special glasses. However, these aids are not accessible to everyone due to their high cost and do not enable colour-blind people to see as many colours as people with normal vision.

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The lack of previous studies, the rarity of this condition and the impact of dyschromatopsia on daily life motivated this study.

To carry out this work, we set ourselves the objective of studying the profile of colour-blind patients at the ophthalmic centre and reviewing the literature.

2. Methodology

An ophthalmology centre in Guadeloupe served as the setting for this study. This was a descriptive cross-sectional study covering a six-month period from 1 August 2024 to 31 January 2025.



Figure 1. Six-year-old boy with colour vision disorders

The parents had noticed that he had been confusing colours since he was very young, so they decided to seek medical advice to ensure he received the best possible care.

3.2 Personal History

3.2.1 Medical

Child born at term, no cardiac abnormalities found, no neurological problems (never had an epileptic seizure), no known drug allergies, no ongoing treatment.

3.2.2 Ophthalmological

No history of eye surgery or eye trauma, does not wear corrective lenses.

3.2.3 Surgical

Never had surgery.

3.2.4 Family History

History of dyschromatopsia in grandfather and throughout maternal line.

3.3 Clinical examination

3.3.1 Physical Examination

- Measurement of distance visual acuity: OD: 10/10, OG: 10/10.
- Near visual acuity: reads P2 ODG.
- We performed the Ichihara test to assess colour vision impairment. This test involves arranging colours according to the individual's perception (easier in children).

This study involved all patients who attended consultations, underwent a comprehensive ophthalmological examination, and were diagnosed with colour blindness.

3. Results

3.1 Clinical Case

This was a six-year-old boy whose parents had consulted the ophthalmology centre after noticing that their child was confused by colours.

The results of this test showed more than 16 incorrect plates out of 38, leading to a diagnosis of colour vision deficiency without determining its severity.

- The other test that allowed us to assess the severity of the disorder was the 100 HUE test, which was completely distorted in terms of wavelength (L).
- Slit lamp examination: OD: normal, OG: normal.
- Intraocular pressure: OD: 12 mmgh, OG: 12 mmgh.
- Fundus examination: normal in both eyes.

3.4 Summary Of Ophthalmological Examination

6-year-old male child, referred for colour vision disorder reported by parents, present since early childhood. Personal medical history: full-term birth, no heart problems or epilepsy, no drug allergies, no diabetes or hypertension, No ophthalmological history, no history of eye surgery or eye trauma.

3.5 Family History

Dyschromatopsia was noted in the grandfather and throughout the maternal line.

The bilateral and comparative ophthalmological examination revealed.

Normal visual acuity in both eyes at 10/10 distance and P2 reading, no oculomotor paralysis, no redness of the conjunctiva.

The slit lamp examination did not reveal any notable abnormalities.

Normal intraocular pressure at 12 mmHg ODG.

Fundus examination was normal in both eyes.

The Ichihara test showed a reading of 16 false plates out of 38.

We performed the 100 HUE test, which was completely distorted.



Figure 2. Retinography of the right eye: view of the right posterior pole, completely normal.



Figure 3. Retinography of the left eye: view from the left pole, completely normal.

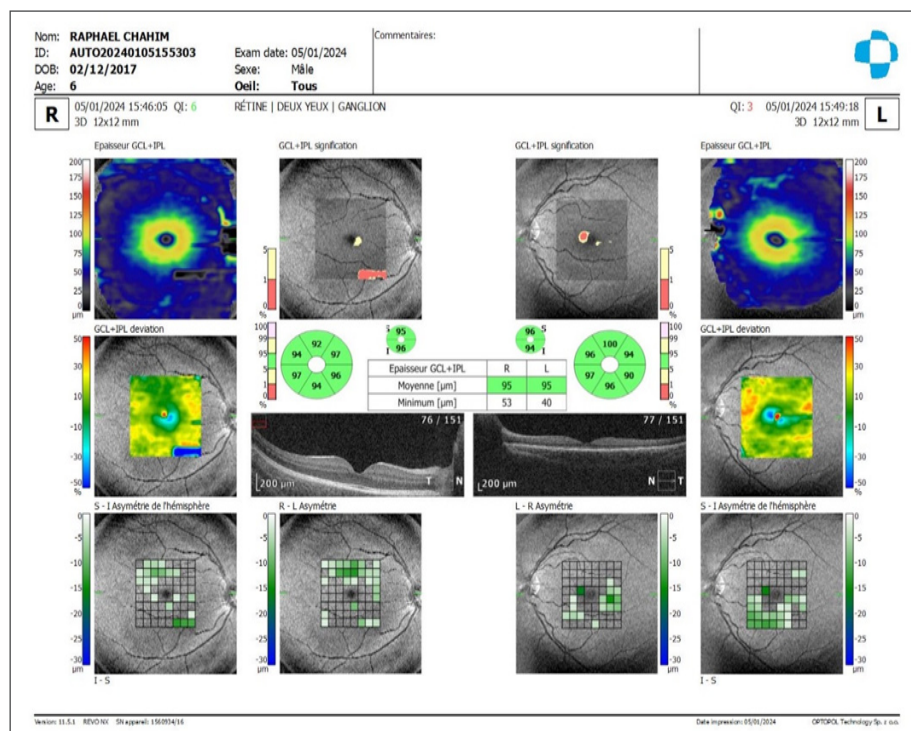


Figure 4. Macular optical coherence tomography (OCT) ODG was normal.

3.5.1 Diagnostic Criteria

Based on the results of the above tests, a diagnosis of colour blindness was made.

3.5.2 Medical Treatment

❖ **Objective:** To improve colour vision quality.

❖ Method:

- Physical: photochromic lenses, which are tinted lenses with filters that absorb the problematic wavelength, so that the red-green band is not stimulated and perception is significantly improved.

- Advice: It is recommended to guide the patient and explain the limitations that this disorder can cause in daily life, as occupations where colour discrimination is important will not be accessible to them.

3.6 Clinical Case

This was an 8-year-old boy who was referred to the department for suspected colour vision deficiency. His mother preferred to seek consultation for better care.

It was noted that he was being monitored for an orthoptic assessment requested by a speech therapist in the context of severe learning difficulties and that he was being home-schooled.

3.7 Personal History

3.7.1 Medical

Child born at term with no heart problems and a known drug allergy to amoxicillin.

3.7.2 Ophthalmological

Wears corrective lenses, no history of eye trauma or eye surgery, myopia.

3.7.3 Surgical

Never operated on.

3.8 Family History

3.8.1 Father Medical

No heart problems, no neurological problems, no diabetes or high blood pressure.

3.8.2 Ophthalmological

No ophthalmological pathology found.

3.8.3 Surgical

Never had surgery.

3.9 Mother

3.9.1 Medical

No heart or neurological problems, no diabetes or high blood pressure.

3.9.2 Ophthalmological

No ophthalmological pathology found.

3.9.3 Surgical

Never had surgery.

3.9.4 Collateral History

History of hereditary dyschromatopsia found in the maternal line.

3.10 Physical Examination

- Visual acuity test: OD 7/10, OG 7/10.
- Distance visual acuity: OD 10/10, OG 10/10.
- The Ichihara test was performed to assess colour vision deficiency. This test consists of arranging colours according to the individual's perception (easier in children). He had misidentified all the plates.
- The test that allowed us to assess the severity of the disorder was the 100 HUE test, which revealed protanopia in both eyes.
- Slit lamp examination: OD: normal, OG: normal.
- Intraocular pressure: Normal intraocular pressure (12 mmHg)
- Fundus examination: normal

3.11 Summary of the Ophthalmological Examination

This was an 8-year-old male child who came in with suspected colour vision disorder, a history of dyslexia and dysorthographia, and a drug allergy to amoxicillin. The interview and physical examination revealed average visual acuity, normal distance visual acuity, low near visual acuity, conjunctival redness, no oculomotor paralysis, and normal intraocular pressure (12 mmHg). The slit lamp examination revealed no abnormalities and the fundus was normal.

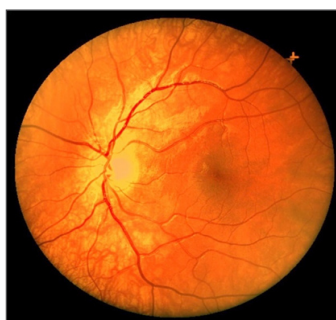


Figure 5. Retinography of the left eye: viewed from the left posterior pole, it was completely normal.

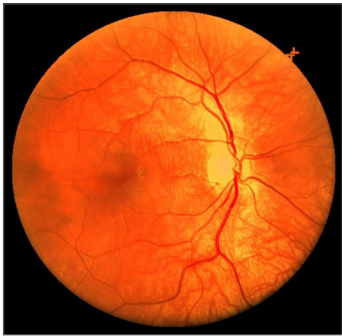


Figure 6. Retinography of the right eye: viewed from the right posterior pole, no abnormalities were found.

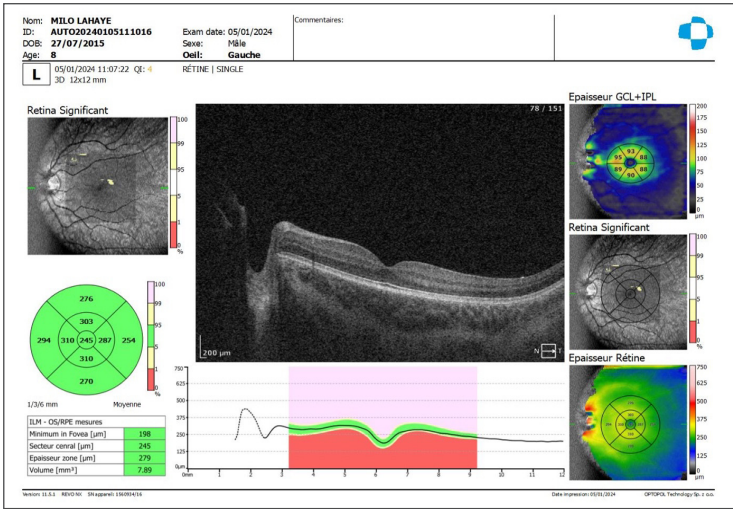


Figure 7. Optical coherence tomography (OCT): Normal.

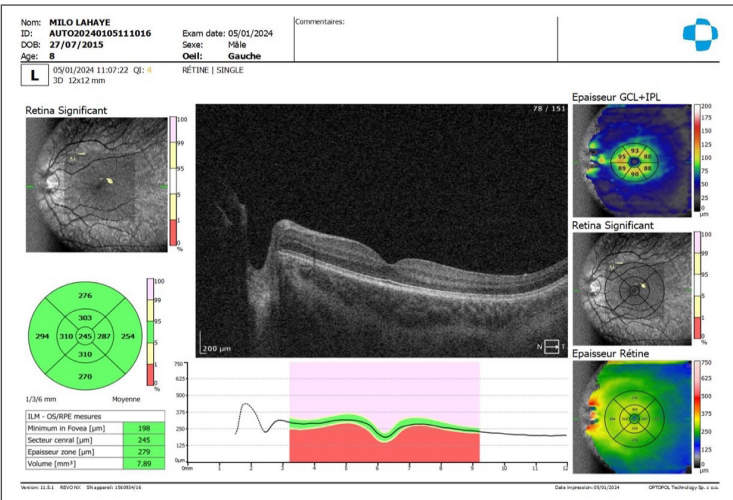


Figure 8. Optical coherence tomography (OCT) of the right macula: was unremarkable.

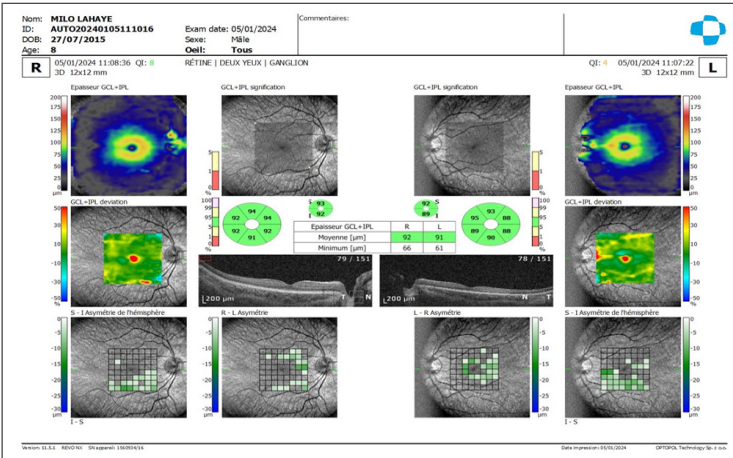


Figure 9. Optical coherence tomography (OCT) of the left macula was normal.

- ❖ No neurovisual assessment was performed.
- **Diagnostic Criteria:** Based on the results of the above tests, a diagnosis of colour blindness was made.
- **Medical Treatment:**
 - ❖ Objective: To improve colour vision and advise the patient on certain occupations that he would be unable to perform.
 - ❖ Means:
 - Physical: photochromic lenses are glasses that improve the contrast of certain colours, making colour vision closer to normal vision.
 - Advice: patients need to be aware that not all occupations will be accessible to them, particularly those where colour discrimination is very important (drivers, the army, aviation, firefighters, the paint industry), to name but a few.

4. Discussion

This was a descriptive cross-sectional study covering a six-month period from 1 August 2023 to 31 January 2024, with the overall objective of observing colour-blind patients.

Colour vision deficiency (CVD) or colour blindness is an eye disorder that prevents patients from distinguishing between certain colours.

In terms of age, in our study, our two cases were aged 6 and 8 years, which could be comparable to the average age found by Ezagono N. , et al. in Cameroon in 2021 [3], who reported an average age of 8.7+/- 1.93 years. This confirms the data in the literature that DH should be diagnosed early in order to limit its impact on learning and acquisition on the one hand, and to limit disqualifications, academic disappointment and even enrolment in an inappropriate course of study with predictable socio-professional consequences sooner or later on the other.

According to the symmetry of the disorder, in our series the diagnosis of red-green colour blindness was made in both eyes. Our results are comparable to those of Salih et al [4], who reported that red-green colour blindness was the most common disorder, accounting for 95% of all colour vision disorders.

According to the frequency by gender in our study, our cases involved two male subjects, which is consistent with the results of Marechal M. et al. in France in 2019[2] , who reported that DH

is present in 9% of men and only 0.5% of women. This could be explained by the fact that this condition is X-linked recessive and sex-linked, and therefore much more prevalent in men than in women.

According to diagnostic tests, our results are comparable to those of Kartika et al[5] in 2021, in whom colour vision tests using Ichihara plates showed normal results for each eye, while a Roth test revealed unspecified colour vision deficiency in the right eye and deuteranopia in the left eye. These results show us that Ichihara plates are only sensitive in identifying those with red-green deficiency. The Farnsworth-Munsell and Roth tests are more accurate in identifying the type of DVC. The principle is to discriminate between coloured tablets based on the most adjacent shades, so the patient is asked to arrange the tablets with the most similar shades next to each other.

Rogosic et al[6] in Croatia used Ichihara pseudoisochromatic plates, the Nagel II anomaloscope and the Lanthony Hue colour test as diagnostic tools in their study, which enabled them to confirm the diagnosis of colour blindness. In our series, we used Ichihara plates and the Farnsworth Munsell test. These tests enabled us to make the diagnosis.

With regard to treatment, certain methods have been developed to help improve colour discrimination, in particular the use of visual aids with coloured filters. In our series, we used photochromic lenses as a means of treatment for our patients. These are tinted lenses that greatly increase contrast.

In addition, some of the available options include tinted contact lenses. These devices have filters that absorb specific problematic wavelengths (520-580 nm), so that the red-green band is not stimulated and colour perception is improved[7] . Sodhi et al[8] in 2023 in India reported in their study that 70 eyes that read both digits on plates 22 to 25 and appeared to have normal colour vision at baseline, this number increased to 99 eyes after using red-tinted lenses. This shows an improvement in vision through the use of red-tinted lenses.

Previous studies have shown that these types of filters in glasses can improve colour contrast and perception for people with colour vision deficiency. However, glasses are bulky and impractical for use in everyday activities, which is why tinted lenses have been developed to overcome the limitations of glasses.

5. Conclusion

Our study shows that colour blindness is a hereditary disorder that alters colour perception. It is much more common in men than in women because it is transmitted through the X chromosome. It is essential to recognise and classify hereditary dyschromatopsia according to its type and severity in order to avoid misinterpretation of results and the resulting unjustified limitations imposed on colour-blind people in their daily lives. The Ichihara test and the 100 HUE test are ways of confirming the diagnosis. Gene therapy, although still in the experimental stage, offers new prospects for curative treatment. In addition, special contact lenses and glasses that filter colours have been successfully developed. These lenses can significantly improve the situation for people with colour blindness. However, these lenses are not affordable for everyone, which means that some people with colour blindness will still need special monitoring and support in order to enjoy everyday life to the full.

6. References

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