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# Ophthalmologic Complications of Friedreich's Ataxia

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**Abstract** | **Case Report**

Friedreich's ataxia is an autosomal recessive neurodegenerative disorder caused by a GAA trinucleotide expansion in the *FXN* gene, leading to multisystem involvement including neurological, cardiac, and ophthalmologic manifestations. Visual impairment is mainly characterized by progressive optic neuropathy that may evolve toward severe optic atrophy. We report the case of a 34-year-old patient with genetically confirmed Friedreich's ataxia presenting with progressive bilateral visual loss. Ophthalmologic examination revealed severely decreased visual acuity, bilateral horizontal nystagmus, and diffuse optic disc pallor suggestive of optic atrophy. Optical coherence tomography (OCT) demonstrated diffuse thinning of the retinal nerve fiber layer and ganglion cell complex. Automated visual field testing revealed diffuse depression of retinal sensitivity. The overall clinical and paraclinical findings were consistent with chronic optic neuropathy associated with Friedreich's ataxia. Ophthalmologic complications of Friedreich's ataxia mainly result from frataxin deficiency leading to mitochondrial dysfunction and oxidative stress affecting retinal ganglion cells and optic nerve fibers. OCT represents an essential tool for the structural evaluation of optic involvement and shows good correlation with neurological severity. To date, no specific neuroprotective treatment has demonstrated efficacy for visual impairment associated with Friedreich's ataxia. Ophthalmologic involvement in Friedreich's ataxia is frequent and potentially severe. Regular ophthalmologic follow-up based on functional assessment and OCT imaging allows better evaluation of optic neuropathy progression and contributes to optimal multidisciplinary management.

**Keywords:** Friedreich's ataxia; optic neuropathy; OCT; optic atrophy; mitochondria; visual field.



## INTRODUCTION

Friedreich's ataxia is the most common form of hereditary autosomal recessive ataxia. In most cases, it is caused by a homozygous GAA trinucleotide expansion in the *FXN* gene, resulting in frataxin deficiency, a protein essential for mitochondrial function. [1]

The disease is characterized by progressive degeneration of the spinocerebellar tracts, posterior columns, and peripheral nerves, leading to severe multisystem neurological impairment. Extra-neurological manifestations notably include hypertrophic cardiomyopathy, diabetes mellitus, and sensory involvement, among which ophthalmologic complications occupy an important place. [2]

Ocular manifestations of Friedreich's ataxia are mainly dominated by progressive optic neuropathy associated with retinal ganglion cell involvement. These manifestations may present as decreased visual acuity, visual field abnormalities, dyschromatopsia, and optic atrophy visible on fundus examination. [3]

The aim of this work is to report a clinical observation illustrating the ophthalmologic complications of Friedreich's ataxia and to discuss the main pathophysiological, clinical, and paraclinical aspects described in the literature.

## CASE REPORT

We report the case of a 34-year-old man followed for genetically confirmed Friedreich's ataxia based on identification of a GAA expansion in the *FXN* gene, referred for progressive bilateral visual loss.

Best-corrected visual acuity was 2/10 in the right eye and counting fingers at 2 meters in the left eye. Oculomotor examination revealed bilateral horizontal nystagmus without limitation of ocular movements.

Intraocular pressure was within normal range, measured at 16 mmHg in the right eye and 17 mmHg in the left eye. Anterior segment examination was unremarkable.

Fundus examination demonstrated diffuse bilateral optic disc pallor predominantly involving the temporal sectors, associated with a cup-to-disc ratio estimated at 0.8, suggestive of advanced optic atrophy. The macula showed a preserved foveal reflex without visible pigmentary abnormalities.

Peripapillary optical coherence tomography (OCT) revealed diffuse thinning of the retinal nerve fiber layer (RNFL), predominantly involving the temporal and superior sectors. Ganglion cell complex (GCC) analysis demonstrated significant diffuse impairment consistent with chronic optic neuropathy. (Figures 1 and 2)

Automated 24-2 visual field testing demonstrated moderate-to-severe concentric constriction associated with diffuse depression of retinal sensitivity without strict respect of glaucomatous meridians. (Figure 3) Color vision testing did not reveal any significant abnormality. (Figure 4)

The overall clinical, functional, and structural findings were consistent with atrophic optic neuropathy secondary to Friedreich's ataxia. [4]

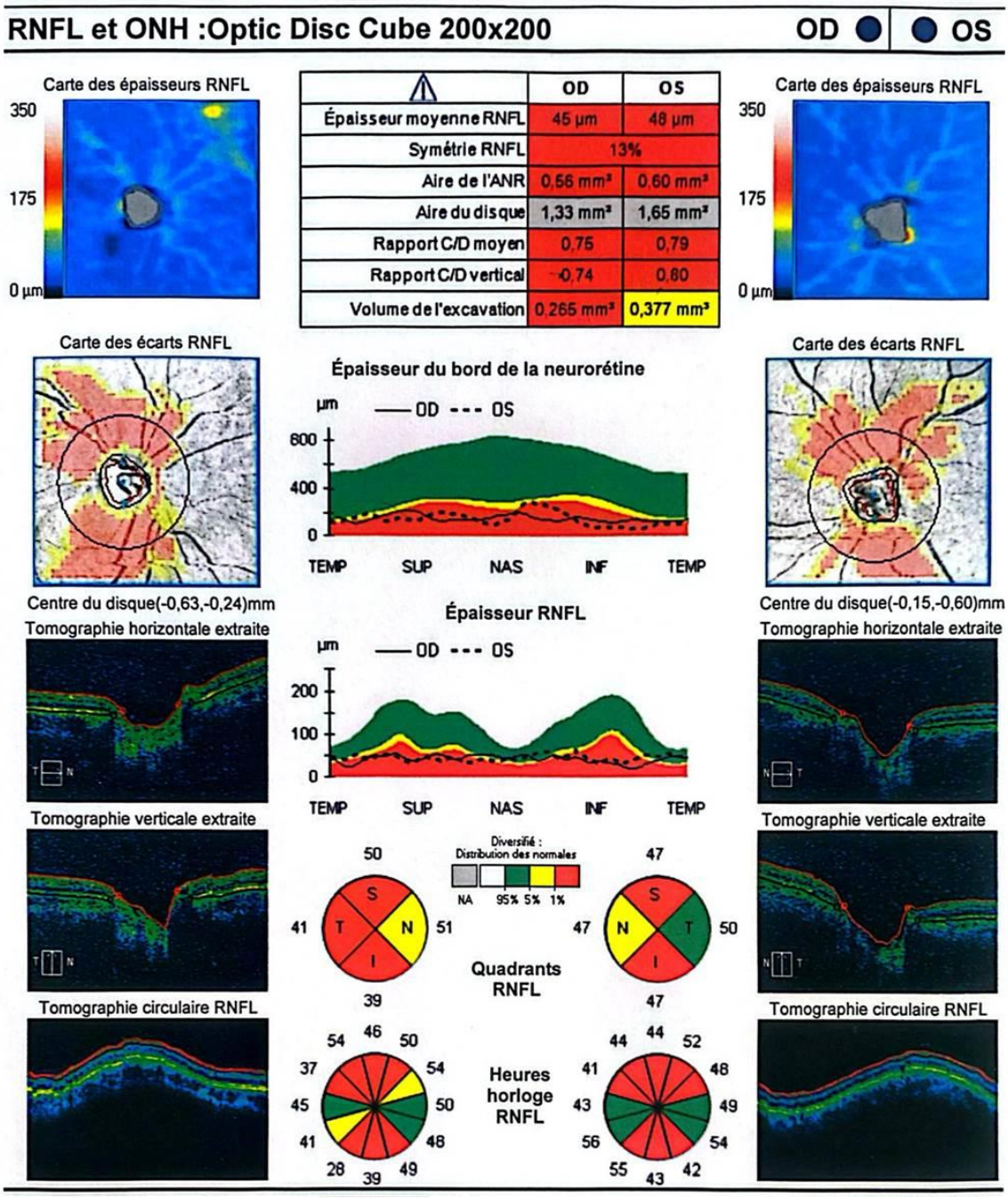



**Figure 1 : Peripapillary OCT**

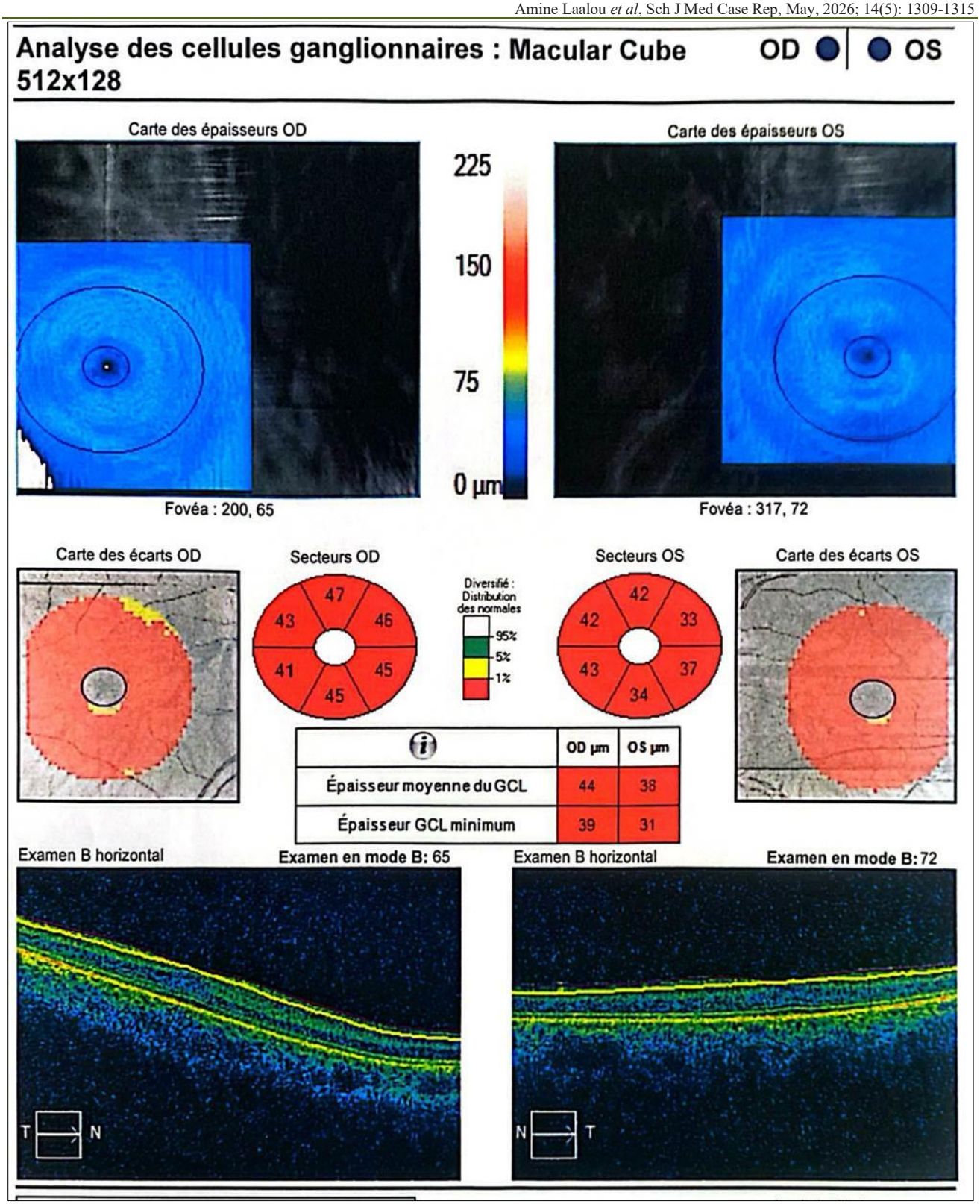
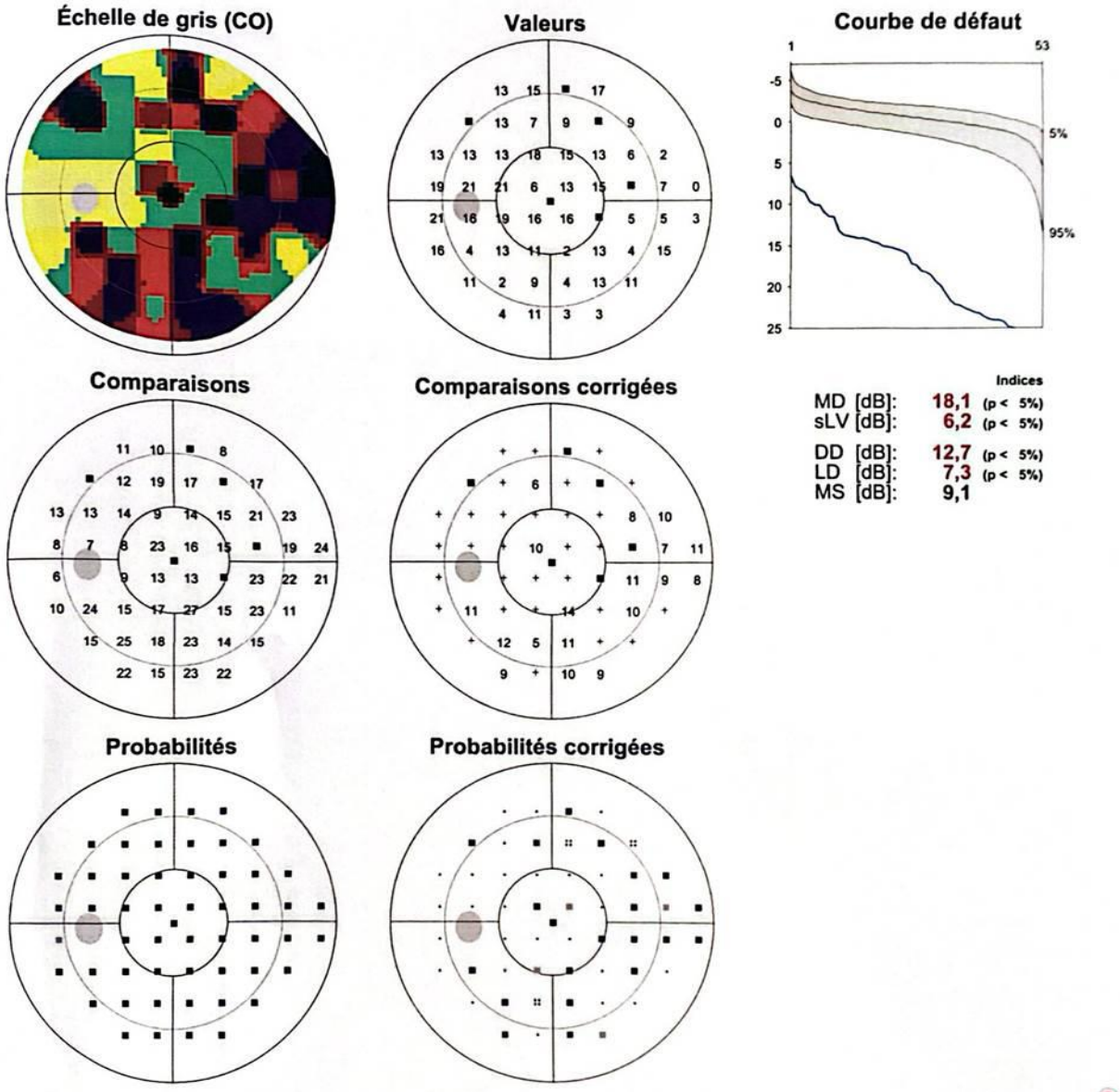



**Figure 2 : Ganglion cell complex**

# OS 31/12/2025 / 10:44

| | |
|---|---|
| Paramètres d'examen: | 24-2, Dynamique, B/B, Blanc/Blanc, III |
| Réfraction, verre d'essai (S/C/A), pupille: | -, +1.0/-/-, Non mesuré |
| Faux positifs/négatifs: | 0% (0/6), 33% (2/6) |
| Durée, questions/répétitions: | 06:32, 140/0 |
| Contrôle de fixation: | Arrêt |




Stimulus: Blanc, Standard, III, 4000 asb, 100 ms
Fond: Blanc/31.4 asb
Échelle de gris (CO) : défaut [% de normal] 100 0
Comparaison et comparaison corrigée + Défaut < 5 [dB] ■ Défaut absolu
Probabilités et probabilités corrigées · p > 5%; :: p < 5%; p < 2%; ■ p < 1%; ■ p < 0.5%

OCTOPUS 900 SN: 5190 Software: 4.7.0 EyeSuite I9.10.0.0 Norm value table: T15 V2.2 (2017-07-14) 31/12/2025, Page 1/1 HAAG-STREIT DIAGNOSTICS

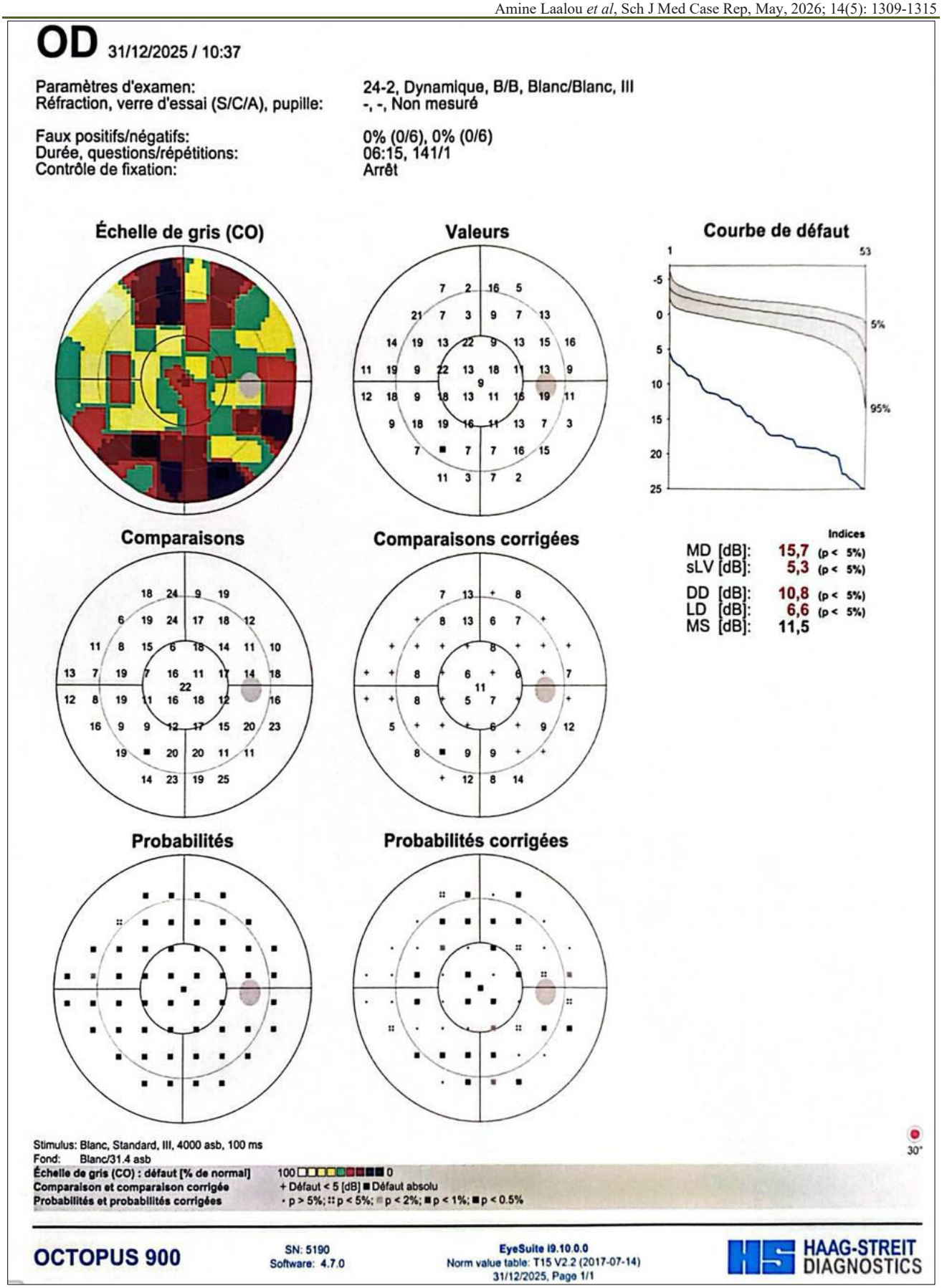



**Figure 3: Visual field**

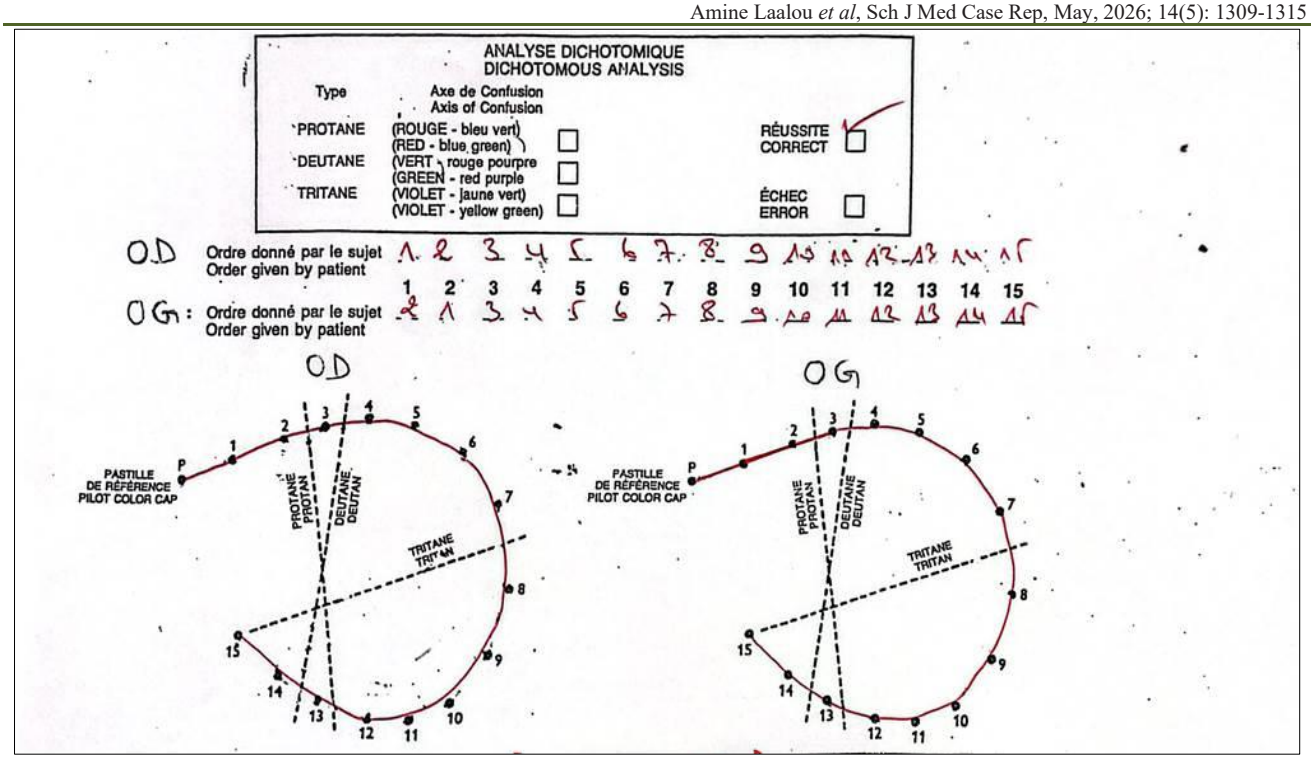



**Figure 4: Color vision test**

## DISCUSSION

Ophthalmologic complications of Friedreich's ataxia are mainly related to progressive optic neuropathy secondary to degeneration of retinal ganglion cells and optic nerve fibers. [5]

Frataxin deficiency induces impaired mitochondrial metabolism resulting in marked oxidative stress. Tissues with high energy demand, such as retinal ganglion cells, appear particularly vulnerable to mitochondrial dysfunction. [5,8]

Clinically, patients most commonly present with progressive bilateral visual loss associated with acquired dyschromatopsia and diffuse optic disc pallor. [6] Horizontal nystagmus observed in some patients reflects the underlying cerebellar involvement. [7]

OCT currently represents an essential tool for evaluating visual involvement in Friedreich's ataxia. Several studies have demonstrated significant reduction in RNFL and ganglion cell complex thickness correlated with overall neurological severity and disease duration. [6]

Visual field abnormalities are variable and generally non-specific, predominantly characterized by diffuse sensitivity depression or central scotoma reflecting papillomacular bundle involvement. [7]

To date, no specific treatment has demonstrated efficacy for optic neuropathy associated with Friedreich's ataxia. Management therefore remains mainly symptomatic and multidisciplinary, based on regular neurological, cardiological, and ophthalmologic follow-up. [2]

Ongoing research focusing on mitochondrial and neuroprotective therapies may open new therapeutic perspectives in the coming years.

## CONCLUSION

Ophthalmologic complications of Friedreich's ataxia are mainly dominated by progressive optic neuropathy that may lead to severe and irreversible visual impairment. OCT and automated visual field testing constitute essential tools for objective assessment of structural and functional optic nerve damage. Regular ophthalmologic surveillance integrated into multidisciplinary management is essential to optimize follow-up of patients with Friedreich's ataxia.

### Declarations

**Conflicts of Interest:** The authors declare no conflicts of interest.

### Authors' Contributions

All authors participated in patient management, manuscript drafting, and critical revision of the manuscript. All authors approved the final version of the manuscript.

### Patient Consent

Informed consent was obtained from the patient for publication of this case report and the associated imaging data.

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