International Journal of Science Academic Research
Vol. 06, Issue 06, pp.10251-10253, June, 2025
Available online at http://www.scienceijsar.com

ISSN: 2582-6425

Research Article

RARE OPHTHALMOLOGIC COMPLICATIONS DURING WISKOTT-ALDRICH SYNDROME IN A CHILD: CASE REPORT

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Received 15th April 2025; **Accepted** 10th May 2025; **Published online** 30th June 2025

Abstract

The particularity of this pathology is its rarity and poor prognosis. A 09-year-old patient with Wiskott-Aldrich syndrome treated by bone marrow allograft transplantation presented with a progressive decrease in visual acuity in a painless white eye. Ophthalmological examination revealed posterior pole and visual field abnormalities. The management consisted of surveillance and referral to a low-vision facility for further treatment.

Keywords: Wiskott-Aldrich, Auto-immune, Optic atrophy, Case report.

INTRODUCTION

Wiskott-Aldrich syndrome (WAS) is a primary immune deficiency characterized by micro-thrombocytopenia, eczema, infections and an increased risk of autoimmune and/or onco-hematological manifestations. It is a rare condition, with an estimated incidence of between 1 and 10 cases per million people worldwide. The ophthalmological manifestations are very rare, and were first described in 1975 and 1982 [1-2].We describe the case of a 09-year-old patient who developed ophthalmological complications during the management of Wiskott-Aldrich syndrome.

Patient and observation

This was a 09-year-old patient with a pathological history of Wiskott-Aldrich syndrome treated by bone marrow allograft transplantation, who consulted for a progressive drop in visual acuity in a painless white eye. The Ophthalmological examination revealed visual acuity of 8/10 in both eyes. Intraocular pressure was 16 mm hg in both eyes. Examination of the anterior segment and adnexa was normal in both eyes. The fundus showed a pale papilla in the temporal region, optic atrophy in both eyes, sheathed vessels, a white spot and poor macular reflex. The examination was completed by optical coherence tomography (OCT), visual field and fluorescein angiography. The OCT showed atrophy of the nerve fiber layer, with an estimated thickness of 7/10 C/D (figure 1-2-3), and a decrease in macular thickness. The visual field shows a diffuse deficit with tubular vision (figure 4-5).Angiography reveals vessel engorgement with a white spot on the vascular pathway with fluorescein uptake (Figure 6-7). A Goldman visual field was no performed. An MRI examination was performed to eliminate a neurological cause. The exam was normal. The management consists of monitoring lesions and referring patients to a low-vision therapy center. The patient consent has been obtained.

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DISCUSSION

Aldrich Wiskott syndrome was first described in 1937 and is defined as an X-linked immunodeficiency disorder. It is characterized by three main manifestations: micro cytopenia, eczema and infections [3-4-5]. Ophthalmological manifestations reported in the literature include scleritis, herpetic keratitis and retinal damage (retinal hemorrhage and retinal degeneration) [1-2-5]. In our patient, the ophthalmological complications were papillary and retinal. They are represented by papillary atrophy, pallor and glaucomatous excavation estimated at 7/10 in both eyes, with tubular vision, retinal white spots and macular atrophy. The complex etiopathogenesis of these disorders is not yet well understood. Treatment is essentially based on bone marrow transplantation. New gene therapies are also being considered, with relative efficacy. In the eye, treatment is essentially symptomatic, depending on the type of damage [6-7-8]. In our patient, management was based essentially on monitoring and referral to low-vision centers. Aldrich syndrome is a very rare condition with a guarded prognosis. Knowledge of its various manifestations, including those of the eye, is essential for early and comprehensive management.

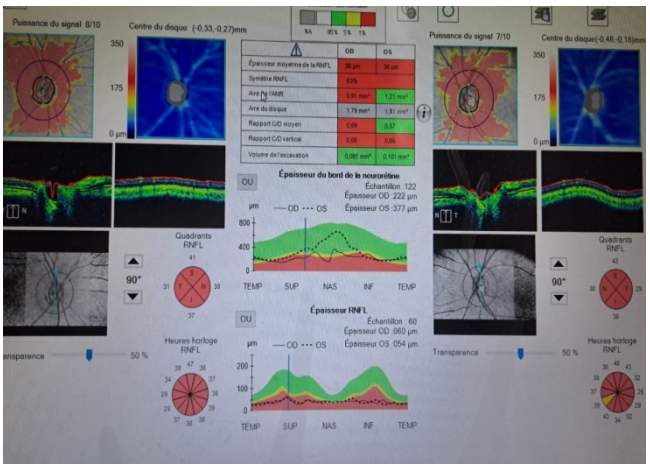

Figure 1. Papillary optic coherence tomography: atrophy of the nerve fiber layer, with an estimated thickness of 7/10 C/D

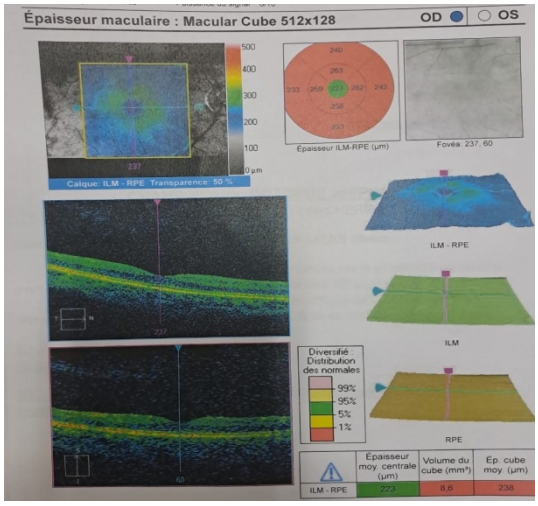

Figure 2. Macular optic coherence tomography of right eye: decrease in macular thickness

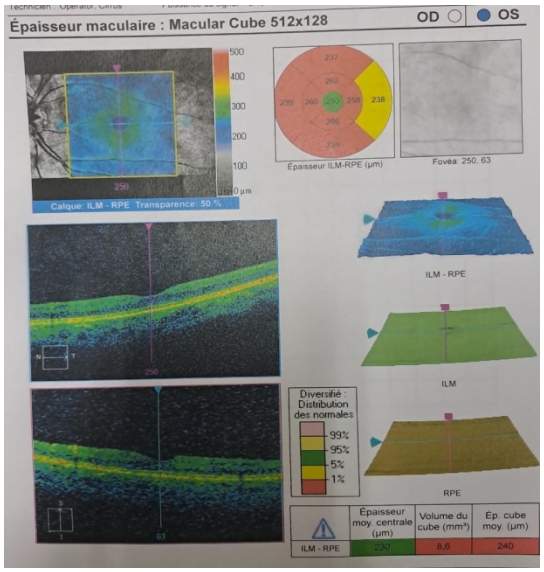

Figure 3. Macular optic coherence tomography of left eye: decrease in macular thickness

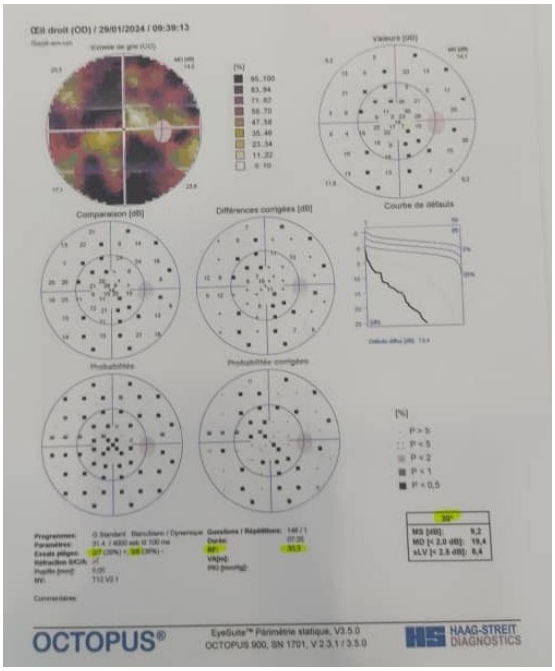

Figure 4. Visual field exam of right eye: The visual field shows a diffuse deficit with tubular vision

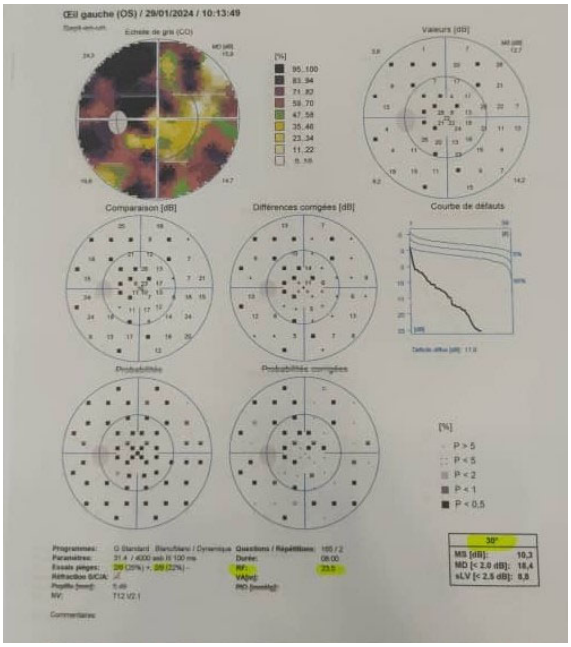

Figure 5. Visual field exam of left eye: The visual field shows a diffuse deficit with tubular vision

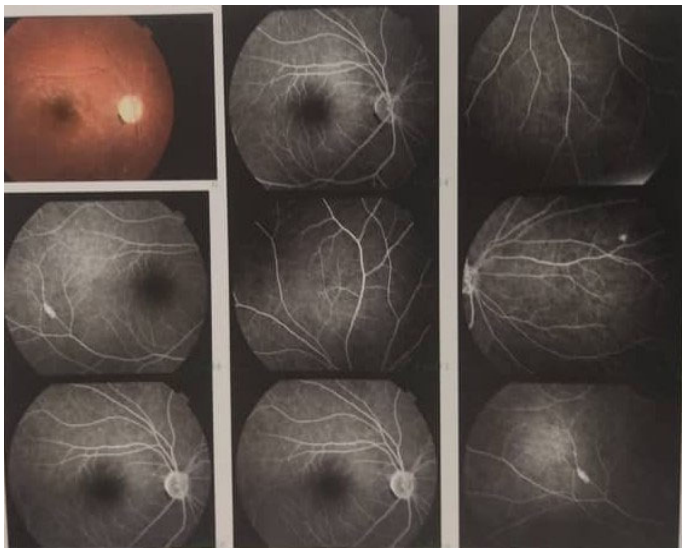

Figure 6. Angiography of right eye: pale papilla in the temporal region, optic atrophy in both eyes, sheathed vessels, a white spot and poor macular reflex.

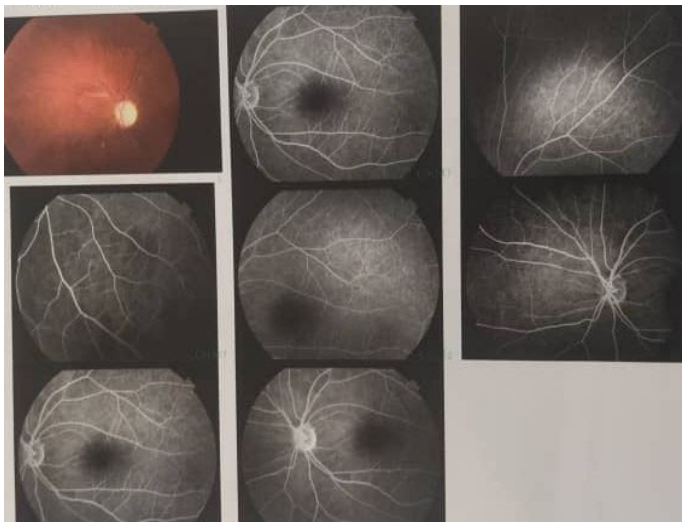

Figure 7. Angiography of left eye: pale papilla in the temporal region, optic atrophy in both eyes, sheathed vessels.

Conclusion

The ophthalmological manifestations of Aldrich syndrome are very rare, especially as the condition itself is rare. It is therefore often unknown to the general public and to some novice ophthalmologists. Our case study sheds light on some of the particularities of this syndrome and advises medical staff on the various manifestations.

Ethics approval and consent to participate: The patient consent has been obtained.

Consent for publication: The patient consent has been obtained.

Availability of data and materials: Data and materials are available

Competing interests: The authors declare no conflict of interest.

Funding: No funds received

Acknowledgements: Not applicable

Authors' information : Not applicable

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