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Late Incidental Diagnosis of Axenfeld–Rieger Syndrome without Glaucoma in A 59-Year-Old Patient: Multimodal Imaging Findings

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Abstract **Case Report**

Axenfeld–Rieger syndrome (ARS) is a rare developmental disorder characterized by anterior segment dysgenesis and frequently associated with glaucoma. We report a late incidental diagnosis of ARS in a 59-year-old patient without ocular hypertension or glaucomatous optic neuropathy. The patient presented with bilateral anterior segment abnormalities, including posterior embryotoxon, iris stromal hypoplasia, and iridocorneal angle dysgenesis, associated with craniofacial anomalies and severe bilateral sensorineural hearing loss. Multimodal imaging using anterior segment optical coherence tomography and ultrasound biomicroscopy revealed significantly narrowed angles with reduced angle opening distance (AOD500), confirming structural dysgenesis. Despite these marked anatomical abnormalities, intraocular pressure and optic nerve evaluation remained within normal limits. This case illustrates the phenotypic variability of ARS and demonstrates that significant structural abnormalities may exist without glaucoma, even in late adulthood. Lifelong ophthalmologic follow-up remains essential due to the potential risk of delayed glaucoma onset.

Keywords: Axenfeld–Rieger syndrome, anterior segment dysgenesis, iridocorneal angle, ultrasound biomicroscopy, multimodal imaging.

INTRODUCTION

Axenfeld–Rieger syndrome (ARS) is a rare congenital disorder belonging to the spectrum of anterior segment dysgenesis, resulting from abnormal migration and differentiation of neural crest cells during embryogenesis (Shields, 1983). These cells contribute to the formation of multiple ocular structures, including the iris stroma, trabecular meshwork, Schlemm's canal, and corneal endothelium. Their developmental disruption leads to a wide range of structural abnormalities involving the anterior chamber angle and iris morphology.

Initially described by Axenfeld as posterior embryotoxon associated with iridocorneal strands, the condition was later expanded by Rieger to include additional iris anomalies and systemic features such as craniofacial and dental abnormalities (Tümer & Bach-Holm, 2009).

Genetic mutations in *PITX2* and *FOXC1* genes have been identified in a significant proportion of cases and play a crucial role in ocular and craniofacial morphogenesis (Alward, 2000; Tümer & Bach-Holm, 2009). Glaucoma represents the most common and severe complication, occurring in approximately 50% of patients and often developing during childhood or early adulthood (Idrees et al., 2006).

We report an unusual case of ARS incidentally diagnosed in a 59-year-old patient without evidence of glaucoma, highlighting the variability of clinical presentation.

CASE REPORT

A 59-year-old patient was referred for ophthalmologic evaluation due to progressive bilateral visual impairment. The patient had no known cardiovascular or metabolic risk factors. Medical history revealed severe bilateral sensorineural hearing loss since childhood, associated with delayed language acquisition. The patient had previously undergone cataract surgery in the left eye. General examination revealed marked craniofacial dysmorphism.

Best-corrected visual acuity was counting fingers at one meter in the right eye and 5/10 in the left eye. Intraocular pressure measured by Goldmann applanation tonometry was 15 mmHg in both eyes.

Slit-lamp examination revealed a shallow anterior chamber bilaterally (Van Herick grade II) with diffuse iris stromal hypoplasia. The right eye presented a dense total brown cataract, while the left eye was pseudophakic. Fundus examination showed normal optic discs with a cup-to-disc ratio of approximately 0.3, without signs of glaucomatous optic neuropathy. Macular examination was unremarkable.

Gonioscopy demonstrated narrow iridocorneal angles with extensive anterior synechiae and a prominent anteriorly displaced Schwalbe line consistent with posterior embryotoxon. The trabecular meshwork remained partially visible.

Corneal topography revealed high regular astigmatism (6.6 diopters in the right eye and 6.9 diopters in the left eye), suggestive of abnormal corneal morphogenesis.

Anterior segment optical coherence tomography confirmed a narrow but open iridocorneal angle without retrocorneal membrane.

Ultrasound biomicroscopy demonstrated markedly reduced angle opening distance (AOD500) values of 118 µm in the right eye and 132 µm in the left eye, significantly below normal reference values (~250 µm). Trabecular angles measured 14° and 16°, respectively. Additional findings included iridotrabecular strands, anterior positioning of the ciliary processes, reduced iris thickness, and shallow anterior chamber depth (2.3 mm OD and 2.5 mm OS), confirming significant structural dysgenesis.

Optical coherence tomography of the optic nerve head showed normal retinal nerve fiber layer thickness without glaucomatous damage. Macular OCT revealed preserved retinal architecture.

Audiometric evaluation confirmed severe bilateral sensorineural hearing loss. Electrophysiological testing demonstrated reduced visual evoked potential amplitudes with prolonged P100 latency, and multifocal electroretinography showed decreased macular responses.

Maxillofacial examination revealed maxillary hypoplasia, telecanthus, and dental anomalies consistent with ARS.

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Figure 1. Anterior segment optical coherence tomography confirmed a narrow iridocorneal angle (Right eye)

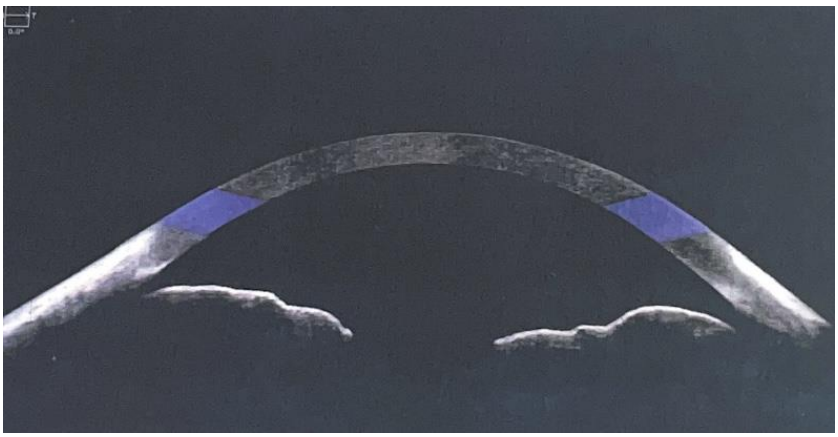


Figure 2. Ultrasound biomicroscopy showing narrow iridocorneal angle and reduced AOD500 (Left eye)

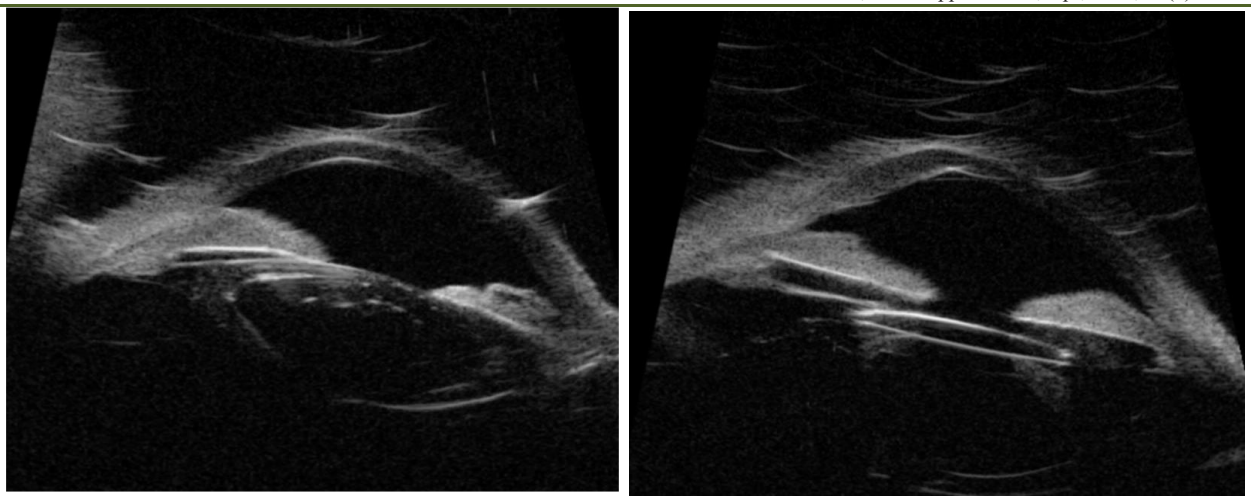
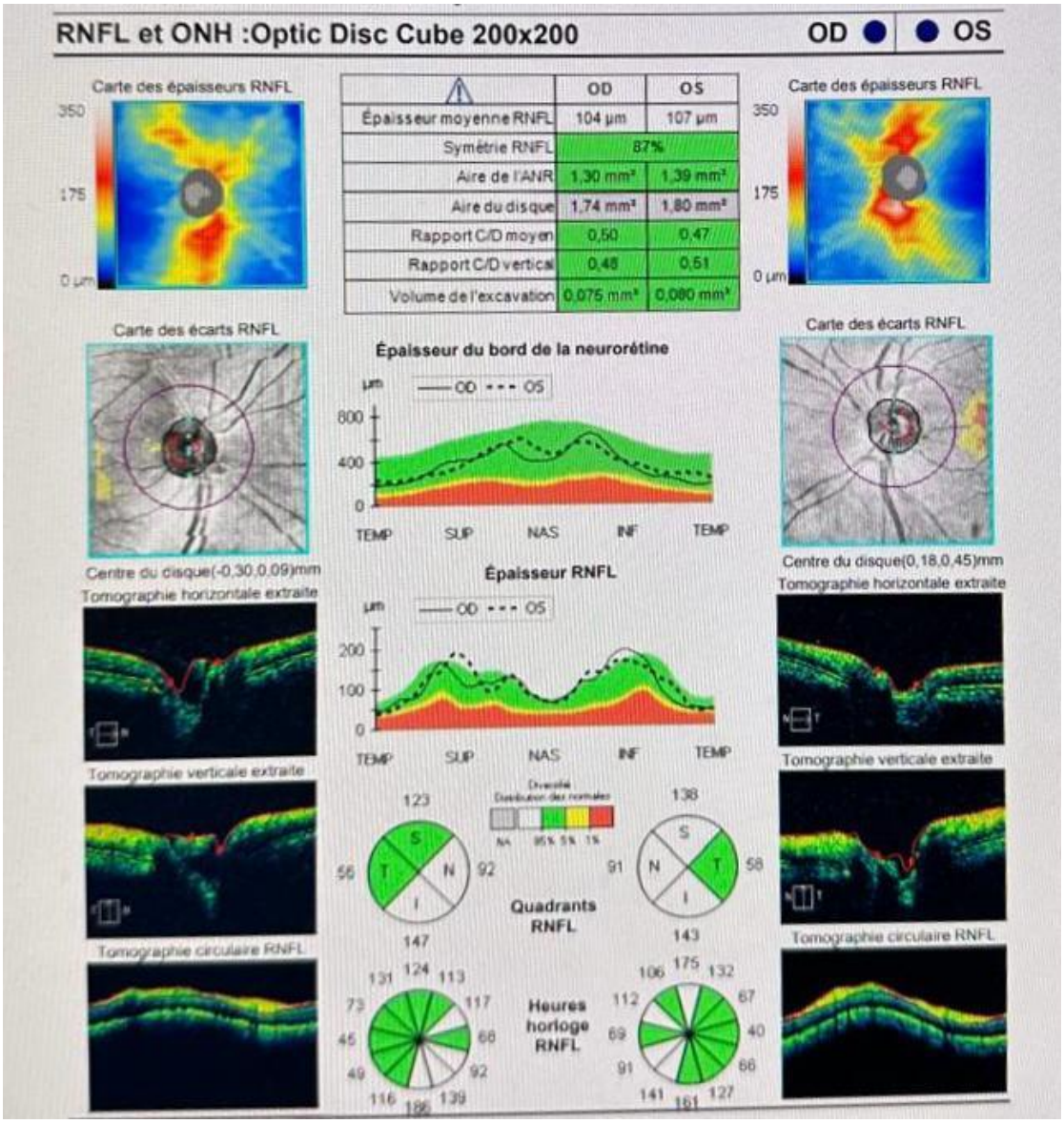


Figure 3. Optical coherence tomography of the optic nerve head showing normal retinal nerve fiber layer thickness



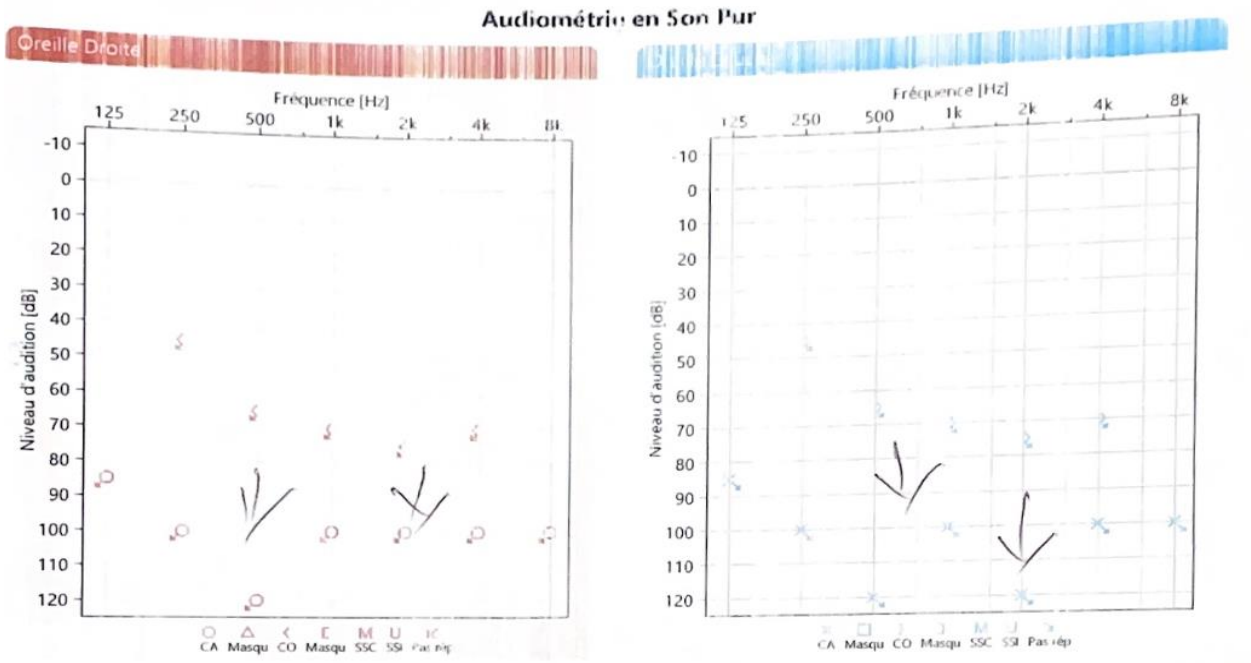


Figure 4: Audiometric evaluation confirmed severe bilateral sensorineural hearing loss

DISCUSSION

Axenfeld–Rieger syndrome is part of the anterior segment dysgenesis spectrum caused by abnormal neural crest cell development (Shields, 1983). These cells are essential for the formation of key anterior segment structures, including the trabecular meshwork and Schlemm's canal, explaining the frequent association with glaucoma.

Mutations in *PITX2* and *FOXC1* genes are the most commonly identified genetic causes and are responsible for defects in ocular and craniofacial development (Alward, 2000; Tümer & Bach-Holm, 2009). These abnormalities impair aqueous humor outflow and predispose patients to glaucoma.

However, ARS demonstrates significant phenotypic variability, with incomplete penetrance and variable expressivity. Patients with similar genetic mutations may exhibit markedly different clinical presentations (Reis & Semina, 2011).

In the present case, despite significant structural abnormalities of the anterior chamber angle demonstrated by ultrasound biomicroscopy, intraocular pressure and optic nerve morphology remained normal. This suggests preserved aqueous humor dynamics and highlights that glaucoma does not necessarily develop in all patients, even at advanced age.

Ultrasound biomicroscopy provided valuable quantitative assessment, particularly through reduced AOD500 values and narrow trabecular angles, confirming anterior segment dysgenesis. Multimodal imaging is therefore essential in the evaluation and follow-up of such patients.

The presence of high regular astigmatism may reflect abnormal corneal development linked to neural crest cell dysfunction. Systemic manifestations such as craniofacial dysmorphism and dental anomalies are well described in ARS. The association with severe sensorineural hearing loss, although uncommon, may be explained by broader embryological involvement of neural crest derivatives.

Differential diagnoses include iridocorneal endothelial syndrome, partial aniridia, and other anterior segment dysgenesis disorders. However, the bilateral presentation and associated systemic features strongly support the diagnosis of ARS.

This case highlights the possibility of late diagnosis and emphasizes that significant anatomical abnormalities do not necessarily result in glaucomatous damage. Nevertheless, lifelong monitoring remains crucial due to the risk of delayed glaucoma onset.

CONCLUSION

This case illustrates the phenotypic variability of Axenfeld–Rieger syndrome and demonstrates that significant anterior segment abnormalities may be present without glaucoma, even in late adulthood. Regular long-term ophthalmologic follow-up is essential to detect potential delayed onset of glaucoma and to optimize patient management.

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