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(CASE REPORT)

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Clinical and tomographic features of a presumed macular corneal dystrophy in a Moroccan sibling cohort

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Abstract

Corneal dystrophies are a group of rare inherited disorders affecting the cornea. We report four affected siblings from a consanguineous Moroccan family with clinical and anterior segment optical coherence tomography (AS-OCT) findings consistent with presumed macular corneal dystrophy. All patients underwent comprehensive ophthalmological examination and AS-OCT imaging, including central corneal thickness measurements. Symptoms began between 11 and 14 years of age and included progressive visual impairment, photophobia, and tearing. Slit-lamp examination revealed bilateral grayish-white stromal deposits with poorly defined borders and diffuse stromal haze of variable severity, while AS-OCT demonstrated subepithelial hyperreflective deposits associated with diffuse stromal hyperreflectivity and reduced central corneal thickness. Marked intrafamilial phenotypic variability was observed, likely reflecting differences in disease stage and progression. Genetic testing was unavailable. This case series highlights the importance of careful clinical examination combined with AS-OCT for the diagnosis of corneal dystrophies, particularly in settings where genetic testing is not readily accessible.

Keywords: Corneal dystrophy; Stromal corneal dystrophy; Macular corneal dystrophy; Autosomal recessive inheritance; Anterior Segment Optical Coherence Tomography

1. Introduction

Corneal dystrophies include a group of hereditary or epigenetically influenced disorders that affect one or more layers of the cornea (1). These disorders are relatively rare, with an estimated prevalence of about 0.09% in the general population (2). Corneal dystrophies are typically classified based on both phenotypic and genotypic characteristics, as outlined in the international classification system developed by the International Committee for Classification of Corneal Dystrophies (IC3D) (3–5).

The clinical presentation is highly variable, ranging from asymptomatic cases to severe visual impairment. Common symptoms include epithelial erosions, reduced visual acuity, photophobia, and foreign body sensations (6).

Treatment is primarily surgical and is tailored to the severity and specific type of dystrophy. Minimally invasive techniques, such as phototherapeutic keratectomy (PTK) and lamellar keratoplasty, are often employed to manage less severe cases. In more advanced stages, more invasive techniques may be necessary, such as penetrating keratoplasty (7).

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In this case series, we present four siblings affected by a specific type of stromal corneal dystrophy. We describe their clinical presentation and anterior segment optical coherence tomography (AS-OCT) findings, highlighting the role of both slit-lamp and imaging characteristics in establishing the diagnosis in the absence of genetic testing.

2. Case Presentation

This case series involved six family members across two generations: two unaffected parents and four affected siblings. The parents exhibited 4th-degree consanguinity. Notably, the mother's two uncles and a cousin from the father's side had similar conditions, but they were not included in this study.

The affected siblings were aged 25, 24, 18, and 11 years, respectively. None of them showed extra-ophthalmological symptoms. Ophthalmological symptoms began to manifest between the ages of 11 and 14, marking the onset of the second decade of life. Progressive visual impairment was the most common symptom, accompanied by photophobia, tearing, and foreign body sensation. These symptoms were consistently reported by all patients except for the youngest. None of the patients presented symptoms of ocular pain, which usually results from recurrent corneal erosions **(Table 1).**

Table 1 Demographic and clinical characteristics of the study population

Sibling	Gender	Age	Age of onset	Gradual visual impairment	Photophobia	Tearing	Ocular pain	Foreign body sensation
S1	Female	25	14	Present	Present	Present	Absent	Present
S2	Female	24	12	Present	Present	Present	Absent	Present
S3	Male	18	13	Present	Present	Present	Absent	Present
S4	Female	11	11	Present	Absent	Absent	Absent	Absent

All patients underwent a comprehensive ophthalmological examination and AS-OCT, including central corneal thickness measurements. Visual acuity at the time of consultation ranged from 20/200 to 20/25, depending on the extent and density of the corneal deposits and the presence of corneal haze. The two eldest sisters had the lowest visual acuity. Corneal sensitivity was notably reduced in all patients except for the youngest. On slit-lamp examination, all patients showed corneal deposits and stromal haze, with the exception of the youngest sibling, who only exhibited central stromal haze. The deposits were grayish-white, had poorly defined edges, and appeared to be circular and at times confluent. They involved the subepithelial, anterior stromal, and posterior stromal layers without intervening clear cornea. The extent of these deposits increased with age. Both the epithelium and endothelium appeared intact across all patients. The two eldest sisters had the most extensive corneal damage, the middle sibling had intermediate damage, and the youngest sibling had minimal central stromal clouding **(Table 2).** Genetic testing was not performed.

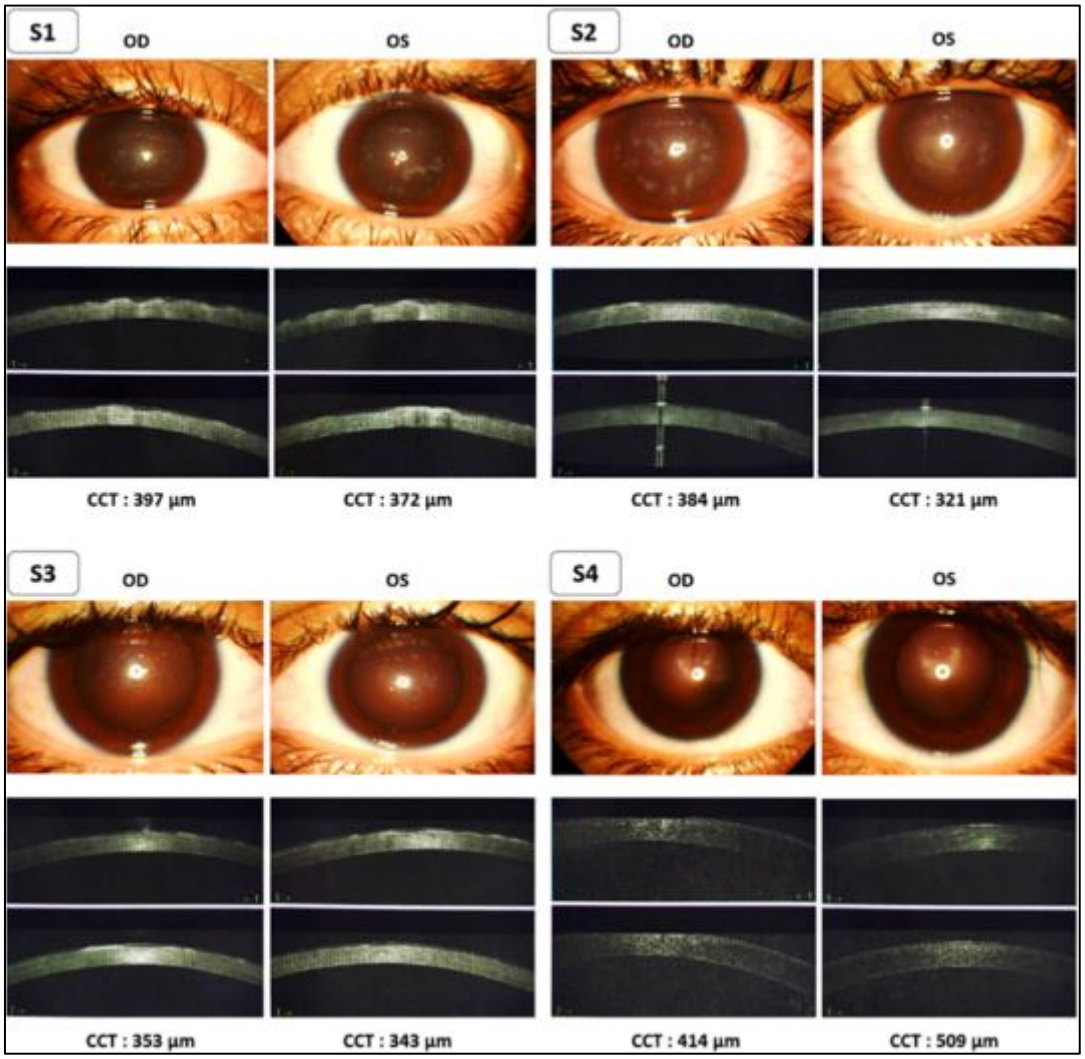
Table 2 Visual acuity and slit lamp examination results of the study population

Examination	S1	S2	S3	S4
VA OD	20/200	CF at 2 meters	20/200	20/32
VA OS	20/200	CF at 2 meters	20/200	20/32
AutoRef OD	NT	−8.75 (−2.00 × 96°)	−3.00 (−1.75 × 128°)	−0.25 (−0.25 × 130°)
AutoRef OS	NT	−5.50 (−1.00 × 64°)	−3.75 (−1.75 × 2°)	−0.25 (−0.25 × 2°)
BCVA OD	NA	20/200	20/40	20/25
BCVA OS	NA	20/200	20/32	20/25
Corneal sensitivity OU	Reduced	Reduced	Reduced	Maintained

Deposit localization OU	Stroma	Stroma	Stroma	NA
Deposit color OU	Grayish-white	Grayish-white	Grayish-white	NA
Deposit appearance OU	Dense macular granules	Dense macular granules	Dense macular granules	NA
Deposit extent OU	From axis to 2 mm to limbus	From axis to 3 mm to limbus	From axis to 4.5 mm to limbus	NA
Corneal erosions OU	Absent	Absent	Absent	Absent
Stromal haze OU	Present	Present	Present	Present

VA: visual acuity; AutoRef: Automated Refraction; BCVA: Best-corrected visual acuity; CF: Counting fingers; NT: No target; OD: Oculus dexter (right eye); OS: Oculus sinister (left eye); OU: Oculus uterque (both eyes)

AS-OCT demonstrated ill-defined hyperreflective subepithelial deposits associated with diffuse stromal hyperreflectivity. The epithelium and endothelium were intact. Central corneal thickness was reduced in all patients, ranging from 321 to 509 µm **(Figure 1).**



CCT: central corneal thickness; OD: Oculus dexter (right eye); OS: Oculus sinister (left eye); OU: Oculus uterque (both eyes)

Figure 1 Corneal characteristics and anterior segment OCT images of the affected members

3. Discussion

Corneal dystrophies represent a heterogeneous group of genetic disorders. They vary widely in age of onset, clinical appearance, impact on corneal transparency, inheritance patterns, and progression (1). The international classification proposed by the IC3D is currently the most widely used. It was established in 2008 by Weiss et al. based on the specific corneal layer concerned and subsequently revised in 2015 and 2024 to incorporate new clinical, histopathological and genetic data (3–5). Genetically, dystrophies are typically autosomal dominant, rarely autosomal recessive, or X-linked (1,6).

Among the different types of corneal dystrophies, macular corneal dystrophy (MCD) is one of the very few with autosomal recessive inheritance. Although genetic testing was unavailable in our cohort, the clinical, biomicroscopic, and AS-OCT findings strongly support the diagnosis of MCD. Molecular genetic studies have identified its genetic basis at a locus on chromosome 16. It results from more than a hundred mutations in the carbohydrate sulphotransferase (CHST6) gene, resulting in the accumulation of keratan sulfate deposits in each layer of the cornea (8–10). MCD has been identified throughout the world, but remains rare in most populations (11). Its higher prevalence in regions with elevated consanguinity, such as southern India (12), Saudi Arabia (13), and Iceland (14), suggests an important influence of genetic background on its distribution (15).

MCD typically manifests within the first two decades of life and progresses gradually over several years. Clinically, deposits are arranged in grayish-white, fuzzy-edged, dense, fibrogranular macules in the stroma initially, and may extend to all layers later. They first appear centrally and later spread across the entire corneal surface, extending to the limbus. Simultaneously, a progressive diffuse haze develops, involving the entire corneal stroma. Although the epithelium generally remains smooth, occasional erosions may occur (3,10,13). Additionally, the cornea is significantly thinner than normal (16,17). According to the IC3D classification, it falls under stromal corneal dystrophies with certainty category 1 (4).

The findings observed in our case series are consistent with the classical clinical presentation of MCD. All affected siblings developed symptoms during the second decade of life and showed bilateral stromal haze associated with poorly defined grayish-white stromal deposits, while the corneal epithelium and endothelium remained clinically intact. However, despite sharing the same presumed genetic background, the severity of corneal involvement varied considerably among the siblings. The two eldest sisters showed advanced disease with marked visual impairment, whereas the youngest sibling presented only mild central stromal haze without clinically evident stromal deposits. This intrafamilial variability reflects differences in disease stage and progression rather than differences in the underlying disorder. This shows the progressive nature of MCD.

Multimodal imaging plays an important role in the evaluation of corneal dystrophies. Investigations are common for all corneal dystrophies. Confocal microscopy typically reveals ill-defined deposits of hyper-reflective material in both the anterior and posterior stroma, with the loss of normal keratocytes (18). On AS-OCT, hyperreflectivity is seen throughout the corneal stroma, with diffuse hyperreflective opacities, particularly in Bowman's layer, and a reduction in central corneal thickness (19). Our AS-OCT findings closely mirrored these previously described features, demonstrating diffuse stromal hyperreflectivity associated with reduced central corneal thickness in all affected siblings, thereby supporting the clinical diagnosis despite the absence of genetic confirmation.

The management of MCD generally involves keratoplasty. Lamellar techniques, such as deep anterior lamellar keratoplasty (DALK), are preferred due to their less invasive nature and lower risk of complications compared to full-thickness transplants. In cases where the endothelium and Descemet's membrane are also affected, penetrating keratoplasty may be necessary (7,15).

Recurrence has been reported in the literature, occurring between 20 months and 30 years. These recurrences are often dense and obturating, requiring additional treatment (20).

4. Conclusion

Corneal dystrophies represent a diverse group of genetic disorders, each with distinct clinical features and progression patterns. Although the international classification system relies on molecular genetics for accurate diagnosis, comprehensive phenotypic analysis is crucial when genetic data is lacking. A detailed clinical evaluation combined with advanced imaging techniques such as confocal microscopy and AS-OCT, can strongly support the diagnosis and guide appropriate management. Genetic testing remains valuable for confirming the diagnosis whenever available.

Compliance with ethical standards

Acknowledgments

This work was previously presented as 'L'OCT du segment antérieur dans une dystrophie cornéenne familiale: une alternative diagnostique en l'absence de test génétique ?' at the 19th national Conference of the Moroccan Society of Refractive Surgery and Implantology (SAMIR), May 2025, Rabat, Morocco.

Disclosure of conflict of interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Statement of ethical approval

This case series was conducted in accordance with the ethical principles of the Declaration of Helsinki.

Statement of informed consent

Clear informed consent was obtained from all adult participants and from the parents of the minor participant for participation and for the publication of the clinical data and accompanying images.

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