



Original Research Article

Demography of corneal dystrophies at a tertiary eye care centre in South India

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ABSTRACT

Purpose: To report the frequency of corneal dystrophies.**Materials and Methods:** All patients who presented to the out-patient department were examined for signs of corneal dystrophy from January 2017 to December 2019. All patients underwent a thorough evaluation for demographics, clinical presentation, history of consanguinity, family medical history, and clinical outcome of keratoplasty (in those who required it) and a clinicopathologic correlation was attempted. Complete ophthalmological examination was conducted for all patients which included vision (distance & near), slit lamp examination, retinoscopy, auto refractometer readings, dilated fundus examination, pentacam as well as anterior segment OCT. Photographic documentation was done for all dystrophy patients.**Results:** During this time period we had 118 patients who were included in the study. History of consanguineous marriage was noted in 62% of the study population. History of a similar problem in siblings and other members was elicited in 27 (22.88%) and 15 (12.71%) patients, respectively. 23 patients required keratoplasty. The average follow-up of each keratoplasty case was 36±14 months. Among these cases, only 2 groups had graft failure, viz, CHED and macular dystrophy.**Conclusion:** The higher frequency of dystrophies could be due to higher consanguineous marriage, which indicates a need for counselling. Also, there is an increased need for thorough work-up before planning intraocular procedures.© This is an open access article distributed under the terms of the Creative Commons Attribution License (<https://creativecommons.org/licenses/by/4.0/>) which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

1. Introduction

Corneal dystrophies are defined as primary, inherited, bilateral, hereditary disorders of the cornea affecting transparency, leading to varying degrees of visual disturbances.¹ The American Academy of Ophthalmology defines corneal dystrophies as "a group of inherited corneal diseases that are typically bilateral, symmetric, slowly progressive and without relationship to environmental or systemic factors."²

They are generally early onset, axially symmetric, slowly progressive or stationary, free from vascularization, and not usually associated with other systemic conditions. Some guidelines for a clinical diagnosis of a corneal dystrophy are described, but there are exceptions to these characteristics, such as unilateral presentation in lattice dystrophy, delayed presentation in Fuchs dystrophy and vascularization in gelatinous drop-like keratopathy.^{1,3}

The dystrophies have been classically classified according to the primary layer of involvement into anterior membrane dystrophies (epithelium, epithelial basement membrane, and Bowman layer dystrophies), stromal

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dystrophies, and endothelial dystrophies (endothelium and Descemet membrane). But recent studies of genetics of corneal dystrophies have changed the concept of dystrophies.^{1,3} The phenotypic description, based on clinical and histological findings, provides the most omnibus clinical picture.

The frequency of dystrophies requiring penetrating keratoplasty in India is 8.4%,⁴ and in other series it ranges from 4% to 24%.^{5–8}

The observation of atypical presentations, inflammation, and associated spheroidal degeneration was the catalyst for us to review the frequency, distribution, and the outcome of keratoplasty in corneal dystrophies at a tertiary eye care centre in South India. We present here a series of corneal dystrophy cases that presented to a tertiary care centre in Bangalore.

2. Materials and Methods

All patients who presented to the out-patient department were examined for signs of corneal dystrophy from 2017 - 2019. The following corneal dystrophies were identified, based on International Classification of Diseases 3 classification: epithelial dystrophy (Meesman corneal dystrophy), epithelial – basement membrane dystrophy, granular corneal dystrophy, lattice corneal dystrophy, macular corneal dystrophy, other stromal dystrophies and endothelial corneal dystrophy. Complete ophthalmological examination was conducted for all patients which included vision (distance & near), slit lamp examination, retinoscopy, auto refractometer readings, dilated fundus examination, pentacam as well as anterior segment OCT. Photographic documentation was done for all dystrophy patients.

3. Results

Our study period was from January 2017 to December 2019. During this time period we had 118 patients who were included in the study. History of consanguineous marriage was noted in 62% of the study population. History of a similar problem in siblings and other members was elicited in 27 (22.88%) and 15 (12.71%) patients, respectively. The predominant presenting symptom was decreased vision in 51 (43.22%) eyes, followed by pain in 23 (19.49%), photophobia in 11 (9.32%), and redness in 9 (7.62%) eyes. The clinical profile and the frequency of each dystrophy is given in Table 1.

In our study, out of 118 patients 23 required keratoplasty. The average follow-up of each keratoplasty case was 36±14 months. Among these cases, only 2 groups had graft failure, viz, CHED and macular dystrophy. Among the CHED patients 2(8.33%) of them were non-compliant to treatment and lost to follow-up for months at a time. 1(4%) CHED patient suffered blunt trauma to the operated eye, which ultimately led to failed graft. Among patients diagnosed

with macular dystrophy, 5(25%) had a recurrence as DALK was done for them. 3(15%) macular dystrophy patients had a failed graft.

4. Discussion

Dystrophy is derived from Greek (dys = wrong, difficult; trope = nourishment)⁹ and was introduced into ophthalmology by Arthur Groenouw in 1890 when he published his classic paper “Noduli Corneae.”¹⁰ The anterior corneal dystrophies include disorders that involve the corneal epithelium only, the epithelium and epithelial basement membrane, and Bowman’s membrane.

Meesmann juvenile epithelial dystrophy is a rare, autosomal dominant, bilaterally symmetric, hereditary epithelial dystrophy.¹¹ Its prevalence is unknown, but has been recognized in Denmark, Germany, Japan, USA, Saudi Arabia and Poland.¹²

GDCD has been reported from India, Tunisia, Vietnam, Turkey, the USA and other countries, but most cases seem to be in Japan where the disorder is estimated to 1 in ~300,000 persons¹³ Gelatinous drop-like corneal dystrophy epithelial and sub-epithelial lesions similar to band keratopathy appear early, followed by small epithelial and later larger nodules, and eventually coalescent opacities with massive amyloid epithelial and sub-epithelial deposits. In our study, we found the prevalence to be 8(47%).¹⁰ Mean age of presentation was 7±1.41 years. They did not need any active intervention at the time of presentation.

Epithelial basement membrane dystrophy (EBMD) is the most common type of corneal dystrophy, affecting 2% of the population.¹⁴

Granular stromal dystrophy prevalence estimate is 11.5/10,000 persons. According to a survey in the United States granular stromal dystrophy prevalence was found to be 4%.¹⁵ GCD has been extensively studied in Denmark by Møller. GCD1 seems to be most prevalent in Europe, but GCD2 is more common in Japan, Korea and the USA and is estimated to have prevalence of at least 5.52 affected persons/10,000 individuals in Korea.

The antecedents of some families with GCD2 have been traced to the Avellino district of Italy (hence the synonym Avellino corneal dystrophy). In our study, 29 (23.31%) patients all aged between 30-50 years presented with GCD type I, 8(8.12%) patients presented with GCD type II. Mean age of presentation of GCD types I and II were 43.2±4.5 and 20.75±5.71 years respectively. 9 GCD I patients underwent optical penetrating keratoplasty, all of them have been on regular follow-up and none have graft failure till date. 2 GCD II patients underwent penetrating keratoplasty and none of them have reported any complications till date.

According to the United States survey, both macular as well as lattice corneal dystrophy prevalence was found to be 2% each.¹⁵ MCD has been identified throughout the world, but in most populations it is rare. It is most commonly

Table 1:

	Frequency (%)	No. of patients	Age of presentation	Mean Age
Gelatinous Drop-like CD	8.47	10	< 10 years	7 ± 1.41
Reis–Bucklers CD	3.38	4	10 years – 20 years	13.5 ± 3.96
Lattice Corneal Dystrophy	5.08	6	30-50 years	39.83 ± 6.17
Granular CD, Type 1	23.31	29	30-50 years	43.2 ± 4.5
Granular CD, Type 2	8.12	8	10-30 years	20.75 ± 5.71
Macular CD	16.94	20	15-60 years	24.9 ± 8.03
Congenital Stromal CD	4.23	5	10-40 years	24.9 ± 8.03
Fuchs Endothelial CD	10.16	12	45-70 years	57.58 ± 7.05
Congenital Hereditary endothelial CD	20.3	24	< 10 years	6.29 ± 1.92
Total	100	118		

noticed in India, Saudi Arabia, Iceland and parts of the USA. At one point in time MCD was the most frequent indication for a penetrating keratoplasty in Iceland.¹⁶ In our study we found the prevalence of Macular dystrophy to be 16.94% (20), a little less compared to other studies. The age of presentation of all patients presented to our OPD was 30-60 years, averaging 24.9 ± 8.03 years. Among the macular dystrophy patients, 5(25%) had a recurrence as DALK was done for them. 3(15%) other macular dystrophy patients had a graft failure, as is well known in macular variant of corneal dystrophy.

LCD1 is one of the more common corneal dystrophies in the Western world, but cases have been recognized throughout the world including Bulgaria,¹⁷ Spain¹⁸ and China.¹⁹ It is most common in Finland, where the disease was first discovered and most extensively studied.²⁰ In our study, we found the prevalence of LCD to be 5.08%, mostly aged between 30 – 50 years. The average age of presentation of LCD was found to be 39.83 ± 6.17 years. All the patients had early lattice findings which was not hampering their vision, hence none of them underwent keratoplasty. Prevalence of other stromal dystrophies such as Schnyder, Fleck, central cloudy dystrophy of Francois was found to be <1%, hence insignificant.¹⁵

Congenital Stromal Corneal Dystrophy is extremely rare; only 4 families have been reported. Affected individuals with CSCD have been extensively studied in large French and Norwegian families.¹⁵ In keeping with the statistics of other studies, we found 5 (4.23%) patients with CSCD in our centre. The age of presentation was between 10-40 years, averaging 24.9 ± 8.03 . 2 patients underwent Penetrating Keratoplasty and have been on regular follow-up, with no complications or failure till date. They were all symptomatically treated and are on regular follow-up. Families with PACD have been reported in the USA and isolated cases have documented in Britain²¹ and Turkey.²²

The prevalence of FECD differs markedly in different parts of the world. It is common and the most prevalent corneal dystrophy in the USA. Approximately 4% of the population over the age of 40 years are affected.²³ In

the USA, FECD was a leading indication for penetrating keratoplasty and this surgical procedure accounted for 10–25% of all corneal transplants in different series. This is a significant number considering that the annual number of corneal transplants in the USA is > 32,000.¹⁵ It is also common in other developed countries. FECD is much more common and more severe in women than in men (3–4:1). FECD is uncommon in Saudi Arabia and in the Chinese of Singapore²⁴ and FECD is extremely rare in Japan⁶ The most common dystrophy they recorded was Fuchs endothelial dystrophy according to the Eye Bank Association of America. Fuchs endothelial dystrophy also predominated (84%) according to the Australian Corneal Graft Registry. In our study, we had 12 (10.16%) patients diagnosed with FECD. The average age of presentation was 57.58 ± 7.05 . Patients were mostly aged between 45-70 years. 8(66%) of them were female. 4 patients underwent DSEK and are doing well with no complications.

Both PPCD and CHED are rare, but CHED2 is more common than CHED1. Most cases of CHED2 have been identified in children of consanguineous parents from Saudi Arabia²⁵ India^{26–29} Pakistan²⁹ Myanmar (Burma)²⁹ and Ireland.^{30,31} It has also been recognized in Bosnia²⁵ the United Kingdom²⁷ and in an American of Chinese ancestry.³² To date, the Harboyan syndrome has been reported in 11 different families from different ethnic groups (Asian Indian, South American Indian, Sephardi Jewish, Brazilian Portuguese, Dutch, Gypsy, Moroccan and Dominican).³³ In our study, we had 24(20.3%) patients with CHED, all presenting below 10 years of age. Mean age was 6.29 ± 1.92 years. 3 of the operated cases had failed graft as mentioned above.

5. Conclusion

In summary, there is a high frequency of macular dystrophy and CHED in southern India, possibly related to consanguineous marriages, suggesting a need for genetic counselling. Recurrence was observed among the Macular dystrophy, in keeping with its previous findings. Around 30.5% of dystrophies were endothelial which shows that

there is an increased need for regular thorough screening before planning any intraocular procedures. 57.62% of the whole group were stromal dystrophies which indicate the high prevalence of this disease. Variations in frequencies of keratoplasty are likely to be reflective of these corneal dystrophies' relative severity. These data can serve as a basis for estimating the cost of care and future demand for intervention and donor corneal buttons as the population ages.

6. Source of Funding

None.

7. Conflict of Interest

None.

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