ORIGINAL ARTICLE

CLINICAL AND FAMILY PROFILE OF PATIENTS DIAGNOSED WITH KERATOCONUS

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ABSTRACT: PURPOSE: To screen the first degree relatives of patient diagnosed with keratoconus. **MATERIALS AND METHODS**: All the patients included in the study, patient details were taken which included - name, age, gender, hospital number, address and family history [pedigree tree]. All the patients underwent refraction, best corrected visual acuity, colour vision, keratometry, intraocular pressure measurement and corneal topography [orbscan]. Family screening was done, which included- refraction, best corrected visual acuity, colour vision, keratometry, intraocular pressure and corneal topography [orbscan]. **RESULTS:** In this study of 40 eyes, 24 eyes [Right eyes -12 & Left eyes-12] i.e. 60% have keratoconus, 3 eyes [2-Right eyes & 1-Left eye] i.e.15% have advanced keratoconus, 12 eyes [Right eye-6 & Left eye-6] i.e. 30% have VKC & Keratoconus, 1 eye have acute hydrops. Total family members screened 55, out of that 17 members are fathers, 20 members are mothers, 12 members are brothers and 6 are sisters
KEYMESSAGE: It seems reasonable to tell them the chance that a first-degree relative would be found to have symptoms of the disease is significantly greater than general population. Hence screening is mandatory for first degree relatives.
KEYWORDS: Keratoconus, first degree relative, screening.

INTRODUCTION: Keratoconus is not uncommon disorder with diverse etiology. Studies have shown that both hereditary and environmental factors play significant role in the formation of keratoconus. The frequency of hereditary inheritance is 6%. McMahon et al.[1] described two pairs of discordant monozygotic twins in which one of each pair had keratoconus and the others had normal corneal topography in both eyes. It was suggested that an environmental trigger might be necessary in addition to a genetic predisposition even in monozygotic twins. In a study on New Zealand population shows that Sporadic KC is the most common presentation; however, a positive familial history has been reported in at least 6–10% of patients. Twin studies performed since the advent of modern computerised videokeratoscopy have reported four monozygotic pairs concordant for KC and two monozygotic pairs which were discordant[2–5] suggesting a genetic component for KC. Keratoconus prevalence in first degree relatives of KC patients is significantly higher than in the general population, demonstrating familial aggregation of the trait.[6]

Most published studies have suggested autosomal dominant inheritance of KC with incomplete penetrance or variable expression.[7] Autosomal recessive inheritance as well as rare cases of X linked inheritance have also been described.[8]

AIMS AND OBJECTIVES:

1. To screen the first degree relatives of patient with keratoconus.
2. To study the association of keratoconus with other factors like consanguinity, allergy and other associated eye conditions.

1. **PERIOD OF STUDY:** January 2014 – December 2014.
2. **STUDY DESIGN**: Cross sectional study.

INCLUSION CRITERIA:

1. Newly diagnosed keratoconus patients.
2. Patients using contact lens.
3. Patients who have undergone C3R, keratoplasty.

EXCLUSION CRITERIA: Patient with other ocular conditions like glaucoma, retinal pathology, other corneal pathologies.

For all the patients included in the study, patient details were taken which included - name, age, gender, hospital number, address and family history [pedigree tree]. All the patients underwent refraction, best corrected visual acuity, colour vision, keratometry, intraocular pressure measurement and corneal topography [orbscan].

Family screening was done, which included- refraction, best corrected visual acuity, colour vision, keratometry, intraocular pressure and corneal topography [orbscan].

RESULTS: Table 1 shows the age distribution of the patients in the study. In our study of 20 patients, the youngest patient was of 11 years of age and the oldest patient was of 22 years of age. Majority of the patient's age group was 16-20 years (26.7%). Mean age was18.0 years.

In this study,there were 16 (80%) male & 4 (20%) female. M:F 4:1

Table 2 shows the complaints of the patients. In our study 17 patient[85%] was main chief complains was diminution of vision.3 patients[15%] was itching and 2 [10%] headache.Some patients had more than one complaints.

Table 3 gives the data of families of patients with family history of allergy. In our study, 3 family members [2 fathers &1 mother] i.e 15% have a asthama and other 3 family members [1 sister & 2 brothers] i.e 15% have aVKC

In this study, 45% [9 of the family members] have Consanguinity marriage and rest 55%[11 family members] not having Consanguinity marriage. That means consanguinity marriage important role in keratoconus.

Table 4 shows diagnosis of patients. In this study of 40 eyes, 24 eyes [Right eyes -12 & Left eyes-12] i.e. 60% have keratoconus, 3 eyes [2-Right eyes & 1-Left eye] i.e.15% have advanced keratoconus,12 eyes [Right eye-6 & Left eye-6] i.e. 30% have VKC & Keratoconus, 1 eye have acute hydrops.

Table 5 shows pachymetry of patients. In this study right eye pachymetry results were between 345-534 and left eye pachymetry were between 336-551.Mean in right eye was 430 & left eye was 429.2. S.D 62.1 & 64.9 respectively.

Table 6 shows data on family members screened. Total family members screened 55, out of that 17 members are fathers, 20 members are mothers, 12 members are brothers and 6 are sisters. [*In two families, two sisters each were screened and in another two families, two brothers each were screened].

Table 7 and table 8 shows spectrum of diagnosis for right eye and left eye respectively of screened family members.

In this study of right eye of 55 family members, 1 brother had KC[1.8%] and 1 had KC and VKC[1.8%].KC suspect were found to be - 1 mother and 2 brothers,[5.3%], but no hydrops in the family members. Total affected members were 5, 8.8% [1 mother,4 brothers and no sister].

In this study of left eye of family members of 55, 1 brother is having KC [1.8%] and KC and VKC [1.8%]. kc suspect 1 mother and 1 brother [3.7%] and 1 brother have hydrops [1.8%] in the family members.

Total abnormal was 5 family members 8.8% [4 brothers, 1 mother, none sisters]

DISCUSSION: Our study showed that keratoconus was prevalent in age range of 11 to 22 years with 65% age distribution in the range 16 -20 years. Most of the study showed that the onset of keratoconus occurs at about the age of puberty. The cornea begins to thin and protrude, resulting in irregular astigmatism with what is usually a steep curvature. Typically, over a period of 10 to 20 years the process continues until the progression gradually stops. The rate of progression is variable. The severity of the disorder at the time progression stops can range from very mild irregular astigmatism to severe thinning, protrusion, and scarring requiring keratoplasty.

Our study showed male predominance i.e. 80% which is usually bilateral and asymmetric. Most of the studies showed that keratoconus usually occurs bilaterally. Unilateral cases occur. However, it has been convincingly shown[9–11] that when diagnostic criteria and computer-assisted topographical analysis allow the detection of very early keratoconus in the fellow eye the incidence of unilateral involvement is probably in the range of 2–4%.

The most common chief complaint was dimness of vision [85%] followed by itching [15%] and head ache. Association of family history of allergy and consanguinity was evaluated. It was found that three families (15%) were found to have history of asthma 15% having VKC.

Association of consanguinity with keratoconus was found to be in 45% of the families, however no ocular problem was found in any of the family members. The largest controlled study[4],[5] found a positive history of atopic disease in 35% of 182 keratoconus patients as compared to 12% of 100 normal control patients.

The relationship between vernal conjunctivitis and keratoconus has been widely reported.[1],[12] Totan and his associates[13] evaluated 82 vernal patients with videokeratography and found evidence of keratoconus in 26.8%. Only 8.5% were diagnosed using biomicroscopy. Tuft et al.[14] Found keratoconus to be an important cause of vision loss in a study of 37 patients with atopic keratoconjunctivitis.

Our study showed that keratoconus was present in 60% of patients, advanced keratoconus in 10%, VKC with keratoconus was seen in 30% and acute hydrops was seen in 5%.

The patient with acute hydrops was mentally sound. Rados[6] was the first to report an association between Down's syndrome and keratoconus. Most series report the incidence between 5.5% and 15%.[6–8] Keratoconus also occurs with increased frequency among developmentally delayed individuals without Down's syndrome, and the incidence of unilateral disease may be substantially higher in this group compared with the general population.[15]

Two plausible explanations for this association with keratoconus are that genetic abnormalities induce structural or biochemical changes resulting in the well-recognized phenotype or that eye rubbing causes the condition. Corneal hydrops occurs with increased frequency in patients with Down's syndrome or other forms of intellectual impairment and this may also result from habitual ocular massage.

Mean pachymetry was 430microns in [RE] and 427.2 microns in [LE].

Contact lens-intolerant keratoconus patients without central scarring, who have mild or moderate disease, are candidates for intrastromal ring segment insertion. The ideal candidates also have low spherical equivalents and average keratometry readings of less than 53 D. The procedure improves visual acuity by flattening the central cornea, reducing astigmatism and centering the cone.[16-19]

Patients treated with RGP contact lens were 5% [RE] and 5% [LE]. Contact lenses were tailored to the individual's visual needs and comfort tolerance. Contact lens wear improves visual function by creating a new anterior refractive surface. Contact lenses do not prevent progression of corneal ectasia.

In this study of 55 family members, 1 brother had KC [1.8%] and 1 had KC and VKC [1.8%]. KC suspects were found to be - 1 mother and 2 brothers, [5.3%], but no hydrops in the family members. Total abnormal were 5 members 8.8% [1 mother and 4 brothers.]

Sporadic KC is the most common presentation; however, a positive familial history has been reported in at least 6–10% of patients. Twin studies performed since the advent of modern computerised videokeratoscopy have reported four monozygotic pairs concordant for KC and two monozygotic pairs which were discordant,[2–5] suggesting a genetic component for KC.

Keratoconus prevalence in first degree relatives of KC patients is significantly higher than in the general population, demonstrating familial aggregation of the trait.[6]

Most published studies have suggested autosomal dominant inheritance of KC with incomplete penetrance or variable expression.[1,7] Autosomal recessive inheritance aswell as rare cases of X linked inheritance have also been described.[6,8]

CONCLUSION: This study shows that keratoconus is more prevalent in males, in the first decade of life. Association was found between asthma and VKC. Patients commonly ask if keratoconus is inherited and if their children will develop the disorder. Using clinical evaluation and three keratographic indices, Wang et al found the keratoconus prevalence of first-degree relatives to be 3.34%, which is up to 68 times that found in the general population.

It seems reasonable to tell them the chance that a first-degree relative would be found to have symptoms of the disease, while significantly greater than the general population, is still less than 1 in 20. Hence screening is mandatory for first degree relatives. Our study found the keratoconus prevalence in first-degree relatives to be 8.8%. Consanguineous marriage was also

associated with high prevalence of keratoconus. Thus our study concludes that screening of first degree family members is essential for their early diagnosis and treatment.

During the course of our study we found that many of the family members were not willing to come for ophthalmic examination because of various reasons of their own. However, it is up to the ophthalmologist to emphasize upon the need for screening and eye examination of all the first degree relatives.[20] This will not only help in early diagnosis and treatment, but will also reduce the ocular morbidity.

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Age group	Cases	
	No	%
Upto 10 years	-	-
11-15 years	3	15
16-20 years	13	65
>20 years	4	20
Total	**20**	**100**
Range	11-22 years	
Mean	18.0 years	
SD	2.91 years	

Table 1: Age distribution of patients

Complaints of Patients	Cases	
	No	%
DOV	17	85
Itching	3	15
Headache	2	10
Total	**20***	**100**

Table 2: Ocular complaints of patients

Family history of Allergy	Cases	
	No	%
Father – Asthma	2	10
Mother – Asthma	1	5

Brother –Vernal Kerato conjunctivitis	2	10
Sister–Vernal Kerato Conjunctivitis	1	5
Nil	14	70
Total	**20**	**100**

Table 3: Family history of allergy

History of illness	**Diagnosis for Patients**			
	Right eye		**Left eye**	
	No	**%**	**No**	**%**
Keratoconus	12	60	12	60
Advanced Keratoconus	2	10	1	5
Vernal Kerato conjunctivitis & Keratoconus	6	30	6	30
Acute Hydrops	-		1	5
Total	20	100	20	100

Table 4: Diagnosis of patients

Patchy value	**Pachy value for Patients**			
	Right eye		**Left eye**	
	No	**%**	**No**	**%**
Upto 446	11	55	12	60
447 – 506	3	15	4	20
>506	3	15	2	10
Not recorded	3	15	2	10
Total	**20**	**100**	**20**	**100**
Range	345 – 534		336 – 551	
Mean	430		429.2	
S.D.	62.1		64.9	

Table 5: Pachymetry values of patients

Family members screened	**Cases**	
	No	**%**
Father	17	85
Mother	20	100
Brother	10+2*	50*
Sister	4+2*	20*

Table 6: Family members screened

Diagnosis	Father		Mother		Brother		Sister		Total	
	No.	%	No	%	No	%	No.	%	No	%
Keratoconus	-	-	-	-	1	8.3	-	-	1	1.8
Vernal Keratoconjectivitis & Keratoconus	-	-	-	-	1	8.3	-	-	1	1.8
Keratoconus suspect	-	-	1	5	2	16.7	-	-	3	5.3
Hydrops	-	-	-	-	-	-	-	-	-	-
Total abnormal	-	-	1	5	4	33.3	-	-	5	8.8
Total normal	17	100	19	95	8	66.7	6	100	50	91.2

Table 7: Diagnosis for family members – Right eye

Diagnosis	Father		Mother		Brother		Sister		Total	
	No.	%	No	%	No	%	No.	%	No	%
Keratoconus	-	-	-	-	1	8.3	-	-	1	1.8
Vernal Keratoconjectivitis & Keratoconus	-	-	-	-	1	8.3	-	-	1	1.8
Keratoconus suspect	-	-	1	5	1	8.3	-	-	2	3.7
Acute Hydrops	-	-	-	-	1	8.3	-	-	1	1.8
Total abnormal	-	-	1	5	4	33.3	-	-	5	8.8
Total normal	17	100	19	95	8	66.7	6	100	50	91.2

Table 8: Diagnosis for family members for left eye

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