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A RARE ASSOCIATION OF BILATERAL STARGARDT'S DISEASE WITH BILATERAL KERATOCONUS

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ABSTRACT **PURPOSE**: To report a case of rare association of bilateral stargardt's disease with bilateral keratoconus
Methods: A 33year old male presented with bilateral progressive vision loss with BCVA of counting fingers at 3 metres in right eye and at 1 metre in left eye.On examination corneal ectasia with thinning,vogt'sstriae and munson's sign were observed. Scissoring reflex was seen on retinoscopy.Hence corneal topography was done with k values 57.3x59˚ in right eye and 59x149˚ in the left eye and pupils were round,regular,reacting to slight. On fundus examination,retinal pigment epithelial changes of Beaten Bronze appearance were observed characteristic of stargardt's disease
Results:High k values on corneal topography suggestive of Keratoconus and retinal pigment epithelial changes suggestive of stargardt's disease
Conclusion: After an intense search, the association of keratoconus and stargardt's has not yet reported so far, hence it is a rare association

KEYWORDS : Keratoconus,stargardt's Disease, Common Associations, Rare Association

INTRODUCTION:

Keratoconus is a sporadic, bilateral(usually asymmetric), progressive, non inflammatorycorneal ectasia with prevalence of 1 in 2000 people[1].Stargardt's disease is the most common autosomal recessive inherited macular dystrophy with prevalence of 1 in 8000-10000 population.It accounts for about 7% of all retinal degenerations and manifests with defective central vision,impaired colour vision.

Case Report: A 33 year old male presented with bilateral progressive loss of vision.Patient had no history of trauma,redness,pain,watering or any other systemic illness.Family history was not significant . On examination face was bilaterally symmetrical with normal head posture and chin was in midline. He had a history of frequent change of glasses. At the time of presentation,his BCVA was counting fingers at 3 metres in right eye and at 1 metre in left eye.On ocular examination,cornea thinning, ectasia along withvogt'sstriae(figure 2),Fleischer ring at the base of cone were observed.Pupils were round,regular,reacting and rest of the anterior segment was normal.Munson's sign(figure1) was noted on down gaze of both eyes.Scissoring reflex was seen on retinoscopy.On corneal topography K value in right eye was 57.3x59˚ and in left eye was 59x149˚.On fundus examination,central macular atrophic lesion with Retinal pigment epithelial changes of beaten bronze appearance (figure 3,4)suggestive of stargardt's disease was seen.Optic disc and rest of the peripheral retina was normal.

Figure 1-MUNSON'S SIGN

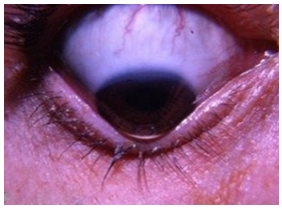


Figure 2-VOGT'S STRIAE

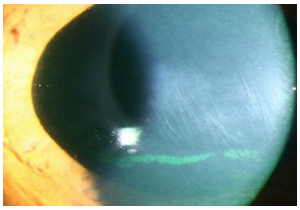
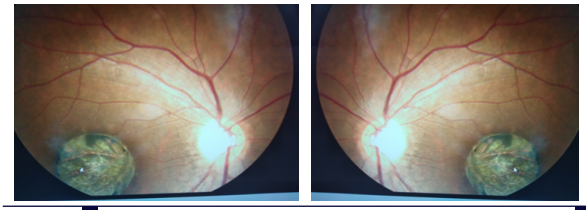


Figure 3 and 4- Fundus photograph showing macular atrophic BEATEN BRONZE APPEARANCE in right and left eye



DISCUSSION:

Keratoconus is anon inflammatory corneal ectasia characterised by progressive thinning of cornea and apical protrusion.It mainly occurs due to dysregulation of matrix metalloproteinases which cause degradation of collagen and extracellular matrix proteins of cornea which results incentral or paracentralstromal thinning and weakening.It leads to ectasia withvisual impairment ,mainly irregular astigmatism and high myopia.It is reported to have bilateral involvement in 90% of patients with asymmetric presentation .Its typical onset at puberty and progress upto 30-40 years.Positive family history can be detected in 6–8% of patients.It is a multifactorial disease resulting from the interaction of environmental, behavioural and genetic factors[2]. Risk factors include history of atopy ,contact lens wear and constant eye rubbing.In its earliest stages, keratoconus causes slight blurring, distortion of vision and increased sensitivity to glare. These symptoms usually appear in the late teens or early 20s. Keratoconus may progress for 10-20 years and then slow in its progression. Each eye may be affected differently. As keratoconus progresses, the cornea bulges more and vision may become more distorted.Characteristic signs include Munson's,Rizzutti's sign,Vogt's striae,Fleischer ring,prominent corneal nerves and other retroillumination signs like scissoring reflex on retinoscopy,charleaux sign.Usually it is an isolated ocular condition and sometimes coexists with other ocular and systemic diseases. Systemic associations are Down's syndrome, atopic dermatitis, EhlerDanlos, Marfan' ssyndromes[3], osteogenesisimperfecta. Common ocular associations of keratoconus are leber's congenital amaurosis,macular coloboma , central serous chorioretinopathy,Retinitis pigmentosa[4]

Stargardts disease is themost common inherited single-gene retinal disease.It is a juvenile macular dystrophy due to ABCA4 gene mutation[8] on chromosome 1p21 resulting in the accumulation of visual cycle kinetics derived byproducts in retinal pigment epithelium with secondary photoreceptor dysfunction and death[5]. Usuallyinherted in autosomal recessive mode and rarely by autosomal dominant type[6].It is the most commonly inherted childhood and adultmaculopathy.As with all autosomal conditions, males and females are equally affected.Symptoms and signs typically appear in late childhood to early adulthood and worsen over time.Initial symptoms include bilateral central vision loss,sensitivity toglare,delayed dark adaptation,central scotomas, and/or dyschromatopsia. Main symptomvision loss is due to accumulation of lipofuschin in the cells of macula leading to impairment of central vision.Peripheral vision is usually less affected than central vision.In up to two-thirds of patients, the macula is classically described as “beaten bronze” appearance with yellowish, “fish-tail” or pisciform lesions .These classic flecks are absent in approximately 30% of

patients with childhood onset disease[7]. It then progresses slowly to a characteristic atrophic macular degeneration classically resulting in a "bull's eye"pattern.Common ocular associations are retinitis pigmentosa,cone dystrophy, cone rod dystrophy[9]

CONCLUSION:

High K values in both eyes with corneal thinning and other signs suggestive of keratoconus and characteristic macular lesion with retinal pigment epithelial changes suggestive of stargardts disease.After an intense search, the association of keratoconus and stargardt's disease is not yet reported so far, which is a rare entity.

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