

Wilson's disease and the eye

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Abstract

Wilson's disease is known for its rarity. It is inherited as an autosomal recessive trait and characterised by excessive accumulation of free copper in the body particularly the liver, brain, cornea and kidney. We report below a case of Wilson's disease with well documented ocular findings. Incidental detection of Kayser-Fleischer ring and Sunflower cataract by ophthalmologist in slit lamp examination in pre-symptomatic cases of Wilson's disease may lead to early diagnosis and prompt management.

Key words: Sunflower cataract, Kayser-Fleischer ring, Wilson's disease

Introduction

Wilson's disease is the clinical condition resulting from mutations in the chromosome 13q14 in the region coding for the protein product *ATP7B*, and occurs in a sporadic fashion as well as inherited as an autosomal recessive disease [1]. Many cases of Wilson's disease have been reported in India ever since it was first published by Samuel Alexander Kinnier Wilson's entitled "Progressive lenticular degeneration: a familial nervous disease associated with cirrhosis of the liver" [2] where he elaborated cardinal features of this

condition including the hereditary nature, co-occurrence of liver cirrhosis with neurological deficits, and the predominantly extrapyramidal nature of signs and symptoms. Community-based incidence and prevalence rates are not available in India and incidences are limited to hospital based reports. Most often, the diagnosis is delayed. This case report highlights the importance of slit lamp examination in early detection of ocular signs of Wilson's disease hence prompt intervention.

Case Report

A 12 year old female child born of a consanguineous marriage in southern India presented to us on July 2017 for routine eye examination with complaints of headache and blurring of vision since 1 month. There was no history of spectacle usage. Father reported history of slightly delayed milestone and having noticed trembling of her hands while writing and poor school performance. Antenatal history and birth history was uneventful.

Her general examination showed normal built, mild cognitive delay, tremors of both hands specially noticed when asked to write and hold objects. Her unaided visual acuity was OU 6/24. Cycloplegic refraction showed a best corrected vision of OD 6/9 with -3.25 CX @ 180° and OS 6/9 with -3.0 CX @ 180°. Slit lamp examinations showed a brownish-yellow ring around limbus OU suggestive of Kayser - Fleischer ring (Fig: 01).

We also observed OU a centralized lens opacity lying just beneath the anterior capsule surrounded by addition lens opacification arranged in ray pattern simulating a sunflower with a central disc and surrounding petals (Fig:02). Fundus was normal OU. Based on our findings of Kayser Fleischer ring, sunflower cataract and hand tremors a clinical diagnosis of Wilson's disease was made and patient was referred to clinician for further systemic evaluation.

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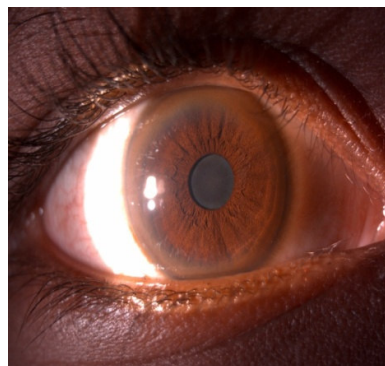


Fig 01: Kayser Fleischer ring in Wilson's disease

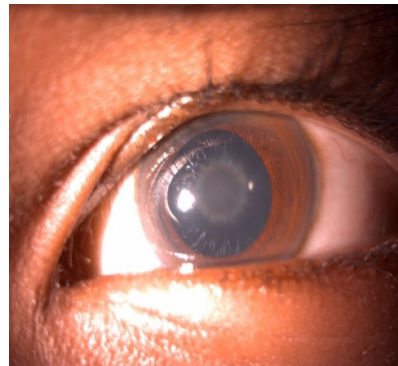


Fig 02: Sunflower cataract in Wilson's disease

Discussion

KF ring is due to copper deposition in the descemet's membrane of the cornea at the limbus. This was described by the German ophthalmologists Bernhard Kayser in 1902 and Bruno Fleischer in 1903 [3]. Fleischer ring is a different entity seen in Keratoconus, which is due to iron deposition from the tear film. Deposition of free copper starts from the Schwalbe line and extends till 5 mm on the corneal surface.

It starts in the superior pole as an arc from the 10 to 02 clock hours position, followed by a similar arc in the inferior pole, and finally encircles the cornea [3].

Free copper loosely bound to albumin enters the aqueous humor and then enters the descemet's membrane. KF rings are easily identified in slit lamp examination. It does not disturb vision.

Other conditions where KF ring may be seen are chalcosis and primary biliary cirrhosis. The ring resolves in the reverse order totally or partially following penicillamine therapy [4] and after liver transplantation. Sunflower cataract was first described by Siemerling and Oloff in 1922 [5] Copper is deposited in anterior and posterior capsule. Sunflower name ascribed to the appearance of a central disc with radiating peripheral folds.

Duke Elder proposed that the radiating folds are because of impression of the posterior surface of the iris on the anterior lens capsule. The shape of the sunflower may vary in presence of pupillary abnormality.

Other conditions where Sunflower cataract has been reported are intraocular copper bodies (chalcosis) and primary biliary cirrhosis, and have been shown to regress either partially or completely following penicillamine therapy. Recently hereditary hyperferritinemia- cataract syndrome (HHCS) due to a 29-bp

deletion in L-ferritin gene is reported to have sunflower cataract due to crystalline deposition of ferritin [6].

Conclusion

Wilson's disease is more common than reported from India. A nationwide reporting of cases may reflect disease burden, improve awareness among medical communities and lead to early diagnosis and prompt treatment. Role of ophthalmologist in detecting ocular signs in pre symptomatic patients is valuable. Correlation of K-F ring to the disease severity, disappearance with proper treatment, reappearance with non-compliant treatment may aid in optimum management.

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Case Report

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