Ocular signs in Wilson disease

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A known patient of Wilson disease was admitted for fever. He was not on regular therapy for the same. His brother also suffered from the same illness.

On examination, he had bilateral sunflower cataracts and Kayser-Fleischer (KF) rings [Figure 1]. He also had bilateral rigidity and batwing tremor.

Wilson disease is an autosomal-recessive disorder of copper metabolism due to mutation of the ATP7B gene in chromosome 13 (13q14.3). Ocular signs of Wilson disease are sunflower cataract and KF rings.

Sunflower cataract was first described by Siemerling and Oloff in 1922.[1] It occurs due to copper deposition in the anterior capsule. Posterior capsule deposition appears to be uniform. The sunflower consists of a central disc with radiating folds at the periphery. The radiating folds are thought to be due to the impression of the posterior surface of the iris on the anterior lens capsule, as proposed by Duke-Elder. The shape of the sunflower may be distorted if there is pupillary abnormality. This mechanism may not fully explain the sunflower shape as this appearance can also occur in the posterior capsule. Sunflower cataract has been reported to occur in other conditions like intraocular copper bodies (chalcosis) and primary biliary cirrhosis, and have been shown to regress either partially or completely following penicillamine therapy. Hereditary hyperferritinemia cataract syndrome (HHCS) is a newly described autosomal-dominant disorder characterized by early-onset cataracts and increased serum L-ferritin concentration, but without iron overload. These cataracts have a characteristic sunflower-like appearance and are due to crystalline deposition of ferritin.[2]

KF was described by the German ophthalmologists Bernhard Kayser in 1902 and Bruno Fleischer in 1903.[3] This is not to be confused with Fleicher ring, seen in Keratoconus, which is due to iron deposition in the corneal limbus. KF ring is due to copper deposition in the descemet membrane of the cornea at the limbus. It 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 the 2 o'clock position, followed by a similar arc in the inferior pole, and finally encircles the cornea.[4] Free copper loosely bound to albumin enters the aqueous humor and then enters the descemet membrane. The peripheral deposition is attributed to the direction of the aqueous humor.[5] KF rings are usually detected by slit lamp examination, but can also be visible to the naked eye and do not obstruct vision. KF ring is seen in 95% of the patients with neurological manifestations and 65% with liver dysfunction. KF rings can also occur in intraocular copper bodies (chalcosis) and primary biliary cirrhosis. The ring resolves in the reverse order either completely or partially following penicillamine therapy[5] and after liver transplant.

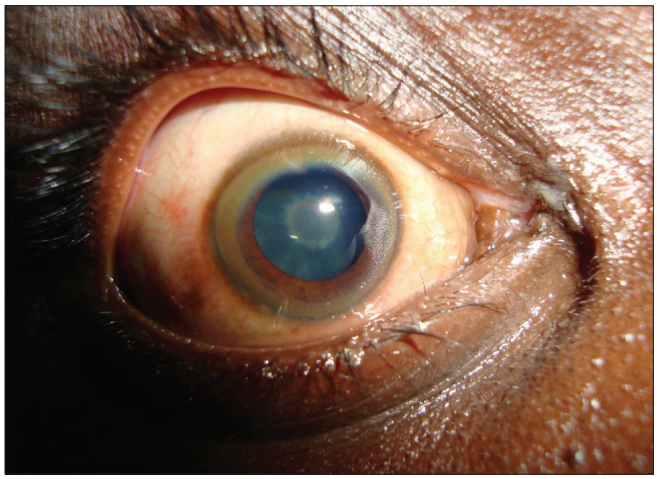

Figure 1: Sunflower cataract and Kayser-Fleischer ring

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References

1. Cairns JE, Williams HP, Walshe JM. Sunflower cataract in Wilson's disease. Br Med J 1969;3:95-6.
2. Mumford AD, Cree IA, Arnold JD, Hagan MC, Rixon KC, Harding JJ. The lens in hereditary hyperferritinaemia cataract syndrome contains crystalline deposits of L-ferritin. Br J Ophthalmol

2000;84:697-700.
3. Suvarna JC. Kayser-Fleischer ring. J Postgrad Med 2008;54: 238-40.
4. Datta S, Datta H. Kayser Fleischer ring. Indian Pediatr 2004;41:744.
5. Harry J, Tripathi R. Kayser-Fleischer ring: A pathological study. Br J Ophthalmol 1970;54:794-800.

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