

# Kayser Fleischer Ring - An Early Indicator of Wilson's Disease - A Case Study

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**Abstract:** A 10 year old female child came to the OPD with complaints of decreased vision. She is a known case of lupus nephritis. Though her vision could be improved after refraction in both eyes, more shocking revelation was the deposition of a dense yellow brown deposit of Kayser Fleischer (KF) ring and a sunflower cataract. She was further carried out various diagnostic tests for Wilson's disease and was immediately treated for the same. Unfortunately the child died within two months due to hepatic encephalopathy and sepsis. Hence it is important to understand the significance of early detection of KF ring which is a definite and a long standing indicator of neurological Wilson disease.

**Keywords:** Kayser Fleischer ring, Wilson disease

## 1. Introduction

Wilson's disease also known as hepatic degeneration and progressive lenticular degeneration is a medical condition having a wide clinical spectrum. It tends to affect 1 in 30,000 people worldwide. It is a very rare genetic disorder where the liver fails to remove the excess copper, which eventually tends to build up in organs like the brain, liver and eye and cause various abnormalities. Kayser Fleischer ring is one of the earliest manifestations of Wilson's disease. Early and prompt clinical diagnose of the KF ring is extremely crucial and life saving for the patient.

## 2. Case Report

A 10 year old female child came to the eye OPD with complaints of decreased vision since 15 days. She was diagnosed as a case of lupus nephritis when she developed symptoms of hematuria, facial puffiness and pedal edema on and off. After blood investigations, urine routine examination and ultrasound of the kidney, ureter and bladder it was confirmed to be a case lupus nephritis with positive ANA.

Her uncorrected vision in her right and left eye were 6/36 and 6/60 respectively and she improved after refraction to 6/9 and 6 / 12 respectively in the right and left eye. On examination of the anterior segment, the cornea showed a very dense yellow brown KF ring in all the quadrants. After dilating the pupil, a central disc of golden brown deposits with spokes appearance over the anterior lens capsule suggested that of a sunflower cataract which could be the reason for her decrease in vision in both eyes. Fundus examination was done and it appeared normal in both the eyes.

Based on the above findings the child was assessed further for Wilson's disease and it showed a low ceruloplasmin level and a high urinary copper level. She was immediately treated with steroids, mycophenolate mofetil, penicillamine and calcium

Though initially in the first month the child responded well to the treatment, but gradually during the follow up there

were alterations in the blood parameters. The child started developing hypertension, behavioral changes, dystonia, dysphagia. She soon developed focal seizures which suggested significant hepatic encephalopathy and it eventually led to the death of the child.

## 3. Discussion

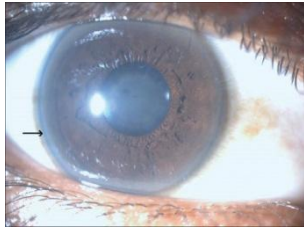
Wilson's disease is a rare inherited autosomal recessive disorder. It occurs due to mutation in the ATP7B gene that mainly encodes for copper transporting P type adenosine triphosphate expressed in the liver and kidney<sup>2</sup>. Copper which gets absorbed from food through albumin in the small intestine reaches the liver cells with the help of membrane transporters. The gene ATP7B helps in the binding of copper to ceruloplasmin and the excess of copper gets eliminated into the bile. Impairment in the biliary excretion leads to an improper accumulation of copper into the extra hepatic sites like the brain, kidney, cornea and heart which lead to further manifestations. Most people are diagnosed between the age group of 5 to 35 years.

Clinical features of Wilson's disease don't appear until the copper builds up in the brain liver and other organs. The features vary depending on the body part it is affected with. Neurological manifestations like dystonia, tremors, dysarthria are common<sup>4</sup>. They may also have fatigue, lack of appetite, abdominal pain, accumulation of fluid in the legs and abdomen. Uncontrolled movements and stiffness of the muscles, yellowish discoloration of the skin and the conjunctiva of the eye are also seen. Deposition of yellowish brown pigment around the cornea called the Kayser Fleischer ring along with sunflower cataract is a very common finding. Hemolytic anemia, thrombocytopenia, renal tubular dysfunction, hypercalciuria, hyperphosphaturia, hypokalemia, cardiovascular dysfunction are also seen.

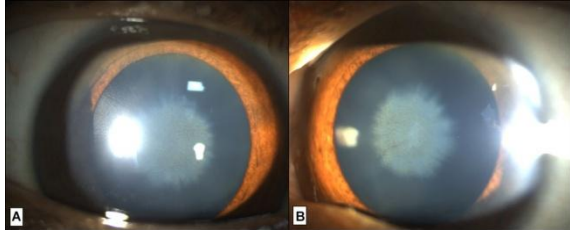
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**Figure 1:** KF ring



**Figure 2:** Sunflower Cataract

The diagnostic tests used for the diagnosis of Wilson disease include

- low serum ceruloplasmin levels (<0.20 g/l, normal is 0.20 -0.40 g/l)
- 24 hour urinary copper excretion (>100 $\mu$  g/day or 1.0mol/day)
- 24 hour urinary copper excretion after D penicillamine (>25mol/day)
- Hepatic copper level on liver biopsy (>250 $\mu$ g/g dry weight, normal is <50  $\mu$ g/g dry weight).

It is important for the early recognition of Wilson disease either by clinical, biochemical or genetic examinations in order to prevent the progression of disease.

Kayser Fleischer ring is a characteristic feature in Wilson disease. It is seen that KF rings are seen in 100 percent of the neurological involvement, 85% in hepatic involvements and about 54% in pre -symptomatic patients. KF ring was first described by a German ophthalmologist by name Bernhard Kayser in the year 1902 and Bruno Fleischer in the year 1903<sup>1</sup>. The ring occurs due to deposition of copper in the descemet's membrane of the cornea and limbus. The free copper which is loosely bound to the albumin enters the aqueous humor and then the descemet's membrane<sup>3</sup>. It starts from the Schwalbe's line and extends 5 mm on the corneal surface. It initially starts at the superior pole in the form of an arc from the 10 to 2 o'clock position followed a similar inferior pole before it finally encircles the cornea.

A study was done by Inne Et al reported a case of unilateral KF ring in a patient with WD. The patient had a reduced intraocular pressure with decreased aqueous production in the scarred eye. And hence the KF ring was not manifested. Thus it was postulated that copper deposition is through the aqueous rather than the limbal circulation which was well maintained in the scarred eye. Both passive diffusion and a cellular activity may be responsible for the copper granule deposition.

The KF ring tends to evolve either partially or completely once the patient is started on penicillamine treatment or following a liver transplant. Other disorders where KF ring can be seen are cholestasis, primary biliary

cirrhosis<sup>5</sup>. Anterior segment optical coherence tomography (AS-OCT) can be used to visualize the KF ring. It appears as an intense hyperreflectivity of the descemet membrane in the peripheral cornea. This is a very useful indicator; as if the KF cannot be captured using a slit lamp then a AS-OCT comes in handy.

Compared to the KF ring, sunflower cataracts are comparatively more rare. They are seen in nearly only 17 % of the patients with Wilson disease. It was first described by Siemerling and Oloff in the year 1922. It is mainly seen due to the deposition of copper on the anterior capsule. Posterior capsule tends to be more uniform. Cataract consists of a central disc with folds radiating at the periphery. As reported by Duke Elder the radiating folds are due to the impression of the posterior surface of the iris on the anterior capsule of the lens.

#### 4. Conclusion

This case was reported to understand the importance of KF rings and its significance. A young girl who was a case of lupus nephritis had undergone an ophthalmological examination as she complained of reduced vision during which the KF ring and sunflower cataract was diagnosed. It was a coincidental finding. Prompt referral by the pediatrician and early identification by the ophthalmologist is very crucial and life saving in a case of Wilson's disease. Though KF rings can occur in other conditions but it is most commonly seen in Wilson's disease and the necessary investigations and treatment should be started without any delay. In case of subclinical occurrence of KF ring, an AS -OCT should be done to confirm the above.

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