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Case Report

Sunflower cataract: A rare diagnostic marker in Wilson disease

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ABSTRACT:
The occurrence of sunflower cataracts (SCs) is rarely reported in Wilson's disease (WD). We present a case of a 13-year-old female patient who was transferred from a general medical OPD to an Ophthalmology OPD with a preliminary diagnosis of Wilson's disease. The patient was asymptomatic. The patient was complaining about hemiparesis on the right side,slurring of speech, tingling and numbness on the right side of the arm/face 4 months ago and was progressive in nature. During the examination, unaided vision was 6/6 in both eyes, and pupils were central, cyclical and reactive to light. During the slit lamp examination, the conjunctiva and pupils were normal in both eyes. Sunflower cataract and a 3 mm Kayser Fleischer (KF) ring around the limbus were found in both eyes. On dilated fundus examination, both eyes were 0.4 CDR, circular, hyperemic disc, FR present. Laboratory tests included SGPT 36U/L, SGOT 199U/L, S. Ceruloplasmin <3 mg/dL, 24 hours urinary copper 470.28 micrograms, rest normal and negative. MRI brain shows giant panda face signs (bilateral thalamic hyperintensity). Based on clinical investigation and examination, we provided a diagnosis of sunflower cataracts in Wilson's disease.

Keywords: KF Ring, Sunflower cataract, Wilson's disease

INTRODUCTION:

Wilson disease, also known as Hepatolenticular degeneration or Westphal Strumpell disease, is an autosomal recessive disorder. It is inborn error of copper metabolism. The underlying cause of Wilson's disease is a mutation in ATP7B on chromosome 13. There is copper deposits in various parts of the body, such as the brain, liver, kidneys, and eyes[1]. The most common sign of Wilson's disease is the Kayser Fleischer (KF) ring, a copper deposit in the cornea. Sunflower cataracts are another sign, but less common than KF ring. This is characterized by a paradoxical ceruloplasmin deficiency. The copper binding sites are saturated, leading to excess copper accumulation. This case reports highlight the importance of early diagnosis of Wilson's disease and appropriate treatment for patients.

CASE REPORT:

A 13 year old female patient was referred from General medicine OPD to Opthalmology OPD for ocular examination, suspecting Wilson's disease. Patient was asymptomatic at present. On ocular examination, patient had no ocular complains like blurring of vision , photophobia, diplopia, headache, watering, itching, burning, foreign body sensation. There was no history of ocular trauma or ocular surgery. Patient had no history of spectacle use.

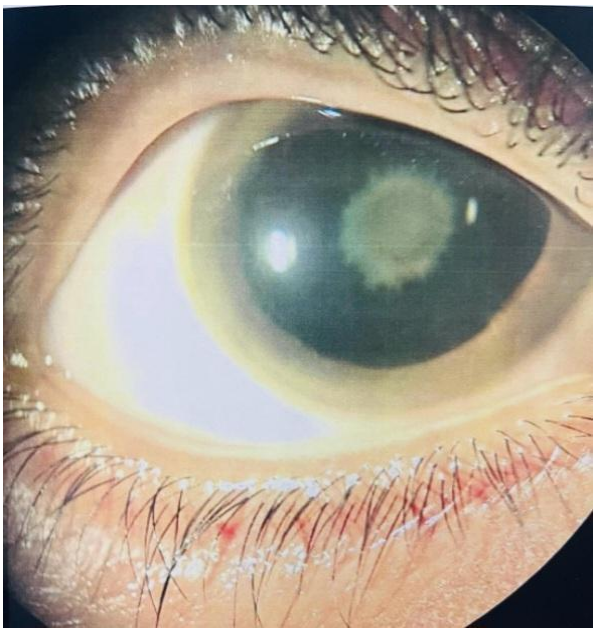
The patient had history of right sided hemiparesis, slurring of speech, tingling, numbness in right aspect of arm / face was on/off and non progressive in nature 4 months ago.

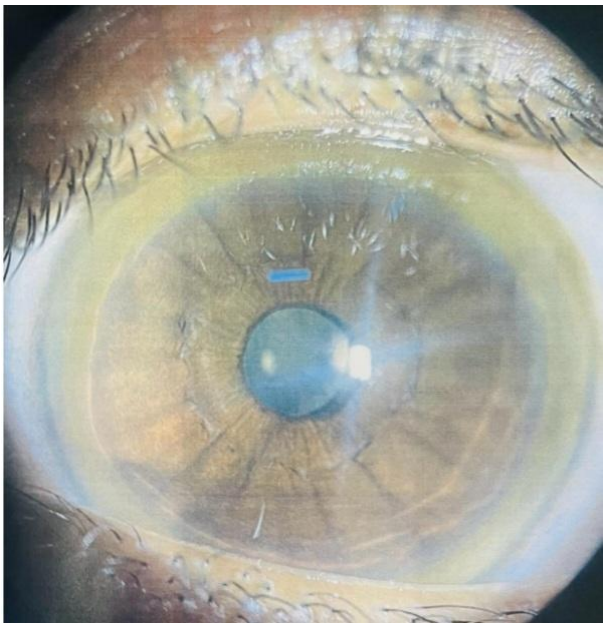
OCULAR EXAMINATION:

	OD	OS
UNAIDED VISION	6/6	6/6
SLIT LAMP EXAMINATION	LIDS, CONJUNCTIVA, PUPIL,	LIDS, CONJUNCTIVA, PUPIL,

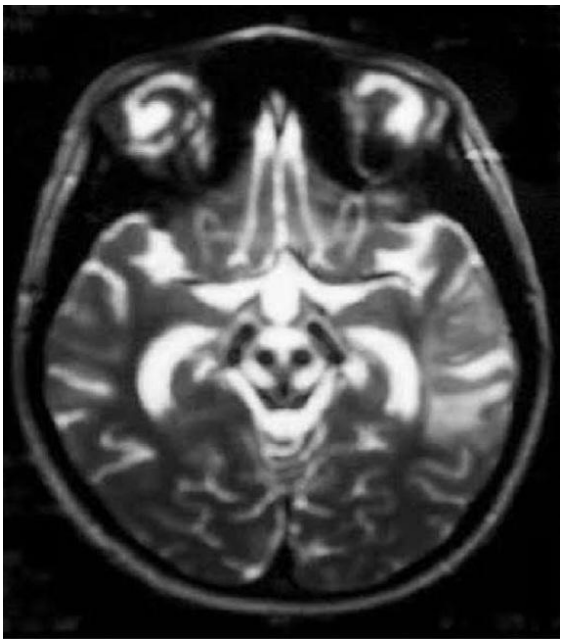
	IRIS NORMAL LENS - SUNFLOWER CATARACT CORNEA - KF RING OF 3mm AROUND LIMBUS NOTED	IRIS NORMAL LENS - SUNFLOWER CATARACT CORNEA - KF RING OF 3mm AROUND LIMBUS NOTED
FUNDUS EXAMINATION	0.4 CDR, CIRCULAR, HYPEREMIC DISC, FR PRESENT	0.4 CDR, CIRCULAR, HYPEREMIC DISC, FR PRESENT

On Slit lamp examination sunflower cataract along with KF ring noted in both eyes





MRI Brain was done. The T2 weighted image was suggestive of bilateral thalamic hyperintensity - Giant Panda face Sign. Based on MRI Brain suggestive of encephalitis was suspected and patient was started on Injection Optineuron IVC1A / 100cc NS.USG Abdomen showed Altered echotexture of liver and urinary bladder was mildly irregular and shows few dependent debris within.



LABORATORY INVESTIGATIONS:

PARAMETERS	LEVELS
SGPT	RAISED (36U/L)
SGOT	RAISED (199U/L)
SERUM COPPER	RAISED (200 MICROGRAM / L)
SERUM CERULOPLASMIN	LOW (<3 MG/DL)
	NORMAL 20.5 - 40.2 MG/DL
24 HOUR URINARY COPPER	470.28 MICROGRAM / 24 HOURS NORMAL 15-60 MICROGRAM / 24 HOURS
URINE ROUTINE MICROSCOPY	TRACE PROTEINS PRESENT, TRACE BLOOD PRESENT, RED BLOOD CELLS PRESENT, PUS CELLS 1-2, EPITHELIAL CELLS 0-2, CRYSTAL CALCIUM OXALATE PRESENT, BUDDING YEAST CELLS PRESENT
REST: ANA, ANCA, DENGUE, HHH, ASMA, ECG, CHEST X RAY	NEGATIVE WITHIN NORMAL LIMITS

Based on various investigation, ocular examination and neurological manifestations, diagnosis of Wilson's disease was made. Patient was discharged and started on chelating agent T. Penicillamine 250 mg 1-0-1 per oral (no food for 2 hours before and after) and T. Pyridoxine 20mg 0-1-0 per oral. Patient was advised regular follow up (3 month) in Gastro as well as Ophthalmology side to look for deterioration or development of any new signs and symptoms.

DISCUSSION:

Wilson's disease (hepatolenticular degeneration) is a rare autosomal recessive disorder caused by the abnormal and excess accumulation of copper in the body, and is particularly involved in the brain, liver and cornea. It affects 1 out of 30,000 people with a career frequency of 1 in 90. Some population groups have a higher incidence of Wilson disease due to more consanguineous marriage. It affects men and women equally[1]. The usual presentation age is between 4 and 40 years, but this disorder can be found in children aged 3 and adults up to 70 years old.

ATP7B gene defect causes excess accumulation of copper, leading to toxicity in various organs and tissues. Liver damage is often one of the first signs, with fatigue, nausea, weight loss, and jaundice as common symptoms. As the disease progresses without treatment, this can lead to severe liver failure. Neurological symptoms can occur as tremor, poor coordination, or changes in behavior or cognitive function. Psychological symptoms such as anxiety, depression, and

psychosis can also be observed. The build up of copper leads to oxidative stress which damages cells and causes liver failure, neurological symptoms (such as motor disorders and psychiatric changes), and KF ring in cornea. Copper enters the body through digestive tract by Copper transporter 1 (Ctr1) and is processed in the liver and copper levels are regulated by ATP7A and ATP7B. In Wilson's disease, ATP7A and ATP7B are defective, leading to accumulation of copper in the liver and spilling it into the bloodstream[2]. This leads to oxidative damage to the liver, leading to diseases such as chronic hepatitis and cirrhosis. Free copper is then deposited in other organs, particularly in areas responsible for movement and mood regulation of the brain, causing neurological and psychiatric symptoms of Wilson's disease.

Histological properties in Wilson's disease may resemble chronic active hepatitis. Even though copper levels are elevated in the liver, early in the disease when copper is located in the cytoplasm and may not be apparent with rhodamine staining. KF Ring is a pathological sign in WD (Wilson's disease) due to the deposition of copper in descemet's membrane of cornea. KF ring is seen on slit lamp examination , and sometimes can be seen through naked eyes. The KF ring does not interfere with vision. KF rings can be observed in 95% of patients with neurological symptoms and 65% of patients with liver dysfunction. The KF ring is a reversible ocular manifestation. It starts from the Schwalbe's line and extends into the surface of the cornea. It starts at the superior pole as an arc from 10 to 2O' clock, followed by a similar arc on inferior pole that finally surrounds the cornea. The free copper which is loosely bound to albumin, then enters aqueous humor and into the Descemet membrane. KF rings can also be observed in chalosis and primary bilient cirrhosis. Due to iron separation of the corneal limbus, a similar ring called Fleicher ring is also observed in keratoconus. Larger KF ring sizes may correlate with disease severity, but not necessarily with copper excretion magnitude.

Sunflower cataracts (SCs) are rare ocular manifestation in WD due to copper deposits in anterior lens capsule. The SC consists of acentral disk with radiating folds at periphery. The radiating folds is thought to be due to the impression of posterior surface of iris on anterior lens capsule.

SC may or may not regress after treatment with chelating agents and may sometimes obscure vision or cause any other symptoms[2,3] Although no reliable tests are known in the diagnosis of WD, low serum ceruloplasmin, high serum copper, high 24 hour urine protein (>100 micrograms/24 hours confirmatory and > 40 micrograms/24 hours strongly indicative) . The gold standard for diagnosis of WD is a liver biopsy.If other investigations are suggestive of WD, a liver biopsy is performed. Liver tissue has been evaluated for the degree of steatosis and cirrhosis and the degree of copper accumulation. Level of > 250 micrograms of copper per gram of dry liver tissue is confirmatory for WD.

Low ceruloplasmin levels are seen in 85-90% of cases. However, in the case of Menke's disease and aceroplasmemia, ceruloplasmin is also low.[4]

High urinary are also observed in autoimmune inflammation and cholestasis.

Genetic testing for ATP7B genes and other genes associated with copper accumulation can be tested.[4]

Low copper containing food is recommended as part of the treatment and avoid mushroom, nuts, chocolate, dried fruit, sesame seeds and oils.

Treatment is used to remove copper from the body and prevent the absorption of copper from nutrients. The first treatment is penicillamine. It binds to copper, leading to the excretion of copper in the urine. There are side effects in some people, such as drug-induced lupus and myasthenia. In these circumstances, trientine hydrochloride can be used in place of penicillamine. Also zinc acetate can be used which stimulates metallothionine, an enterocyte protein that binds copper to prevent absorption.

Copper treatment can take six months for work to begin. In addition to medical therapy, physiotherapy and occupational therapy are also used. Liver transplants are used in people who do not respond to treatment.[5] If left untreated, WD will worsen and ultimately be fatal.

CONCLUSION:

Association of KF ring and Sunflower cataract in WD with neurological manifestation is seen in 95% cases than with hepatic manifestation seen in 65% cases. Incidence of KF ring if 67-80% in WD, is a reversible sign, donot obstruct vision and more commonly in 80-90% cases fade or disappear on treatment. Incidence of Sunflower cataract is 1.7-20% in WD, regression is not common, may obstruct vision later and may require cataract surgery later for the same. Patients respond well to chelating agents. Relapses are possible on discontinuation of treatment.

FINANCIAL SUPPORT OR SPONSORSHIP: Nil

CONFLICT OF INTEREST: Nil

REFFERENCES:

1. https://eyewiki.org/Wilson%27s_Disease/Kayser_Fleischer_Ring#:~:text=4%20Referenc es-,Detailed%20Description,results%20in%20the%20disease%20process.
2. https://www.ncbi.nlm.nih.gov/books/NBK459187/#:~:text=Deterrence%20and%20Patient%20Education,resonance%20imaging%20of%20the%20brain.
3. https://pmc.ncbi.nlm.nih.gov/articles/PMC4989003/#:~:text=Introduction,1).
4. https://pmc.ncbi.nlm.nih.gov/articles/PMC4989003/
5. https://pubmed.ncbi.nlm.nih.gov/24809467/