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Case Report
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A COMPREHENSIVE CASE REPORT ON PEDIATRIC PRESENTATION OF WILSON'S DISEASE

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ABSTRACT

Wilson's disease is a rare genetic illness characterized by defective hepatic transport indicating towards accumulation of copper in different organs, mostly in the brain, liver, eye, among various organs. It is also referred as hepatolenticular degeneration in which improper copper metabolism takes place. Although copper is an basic element for normal physiologic function, If there is too much copper in organs of the body leads to toxic effects and life-threatening conditions, especially liver and brain. Early recognition and diagnosis are essential to prevent irreversible organ damage. We present a case of pediatric patients with Wilson disease where the patient presented with hepatic symptoms and neurological symptoms. This case report offers clinical manifestation, diagnostic challenge, and therapeutic management of a 10-year-old male patient diagnosed with Wilson disease. This case report emphasizes the significance of prompt recognition, diagnosis, and intervention to avert life-threatening consequences.

KEYWORD: Wilson's Disease, Hepatolenticular, Copper toxicity, Neurological symptoms, ATP7B Gene.

INTRODUCTION

Wilson's disease is a rare autosomal recessive genetic disorder also called hepatolenticular degeneration liver disorder characterized by mainly hepatic and neurological manifestations.[1] Due to mutation in the gene ATP7B that encodes a copper transport protein on chromosome 13[2,3] leads to the deposition of copper into various organs, mostly in liver, eye, brain, and other different tissue which result in numerous clinical symptoms mostly neurological, hepatic, as well as psychiatric symptoms along with this presence of kaiser-Fleisher ring on the cornea.[5,6,7]

Copper is an essential element for normal physiologic functions like the development of healthy nerves, bones, collagen, and pigment melanin to color skin, hair, and eyes, the formation of RBC, neuron activation, and helps in iron absorption. Normally copper is absorbed from the daily diet and excess copper is excreted through bile. But for some peoples with Wilson disease, the elimination of copper doesn't take place leading to the accumulation of copper in various organs which cause a life-threatening condition that needs early recognition and diagnosis is crucial to avoid irreversible organ damage and other difficulties. Prevalence is around in 1 case per 30 thousand life births.[4] In this case report the treatment is mainly focused on removing the amount of copper that has accumulated in the body and blocking the absorption of copper in the intestine.

CASE REPORT

A 10-year-old boy was hospitalized in department of pediatrics in tertiary care hospital of Rajahmundry, Andhra Pradesh with main complaint of upper limb finger folding unknowingly for 45 days with speech disturbance and difficulty in swallowing of food for 10 days. Also, the involuntary movements which are intentional and spontaneous, progressive in nature from 20 days.

He had a history of jaundice 1 year back and was relieved after taking medication again reoccurrence occurred after 3 weeks and he had a history of epistaxis. His mother had a history of antenatal and had taken 2 inj-Tetanus Toxoid dose.

OBSERVATION

In typical physical inspection, blood pressure was determined to be 110/80mmHg, respiratory rate 22/min, pulse rate 88bpm, and temperature was determined to be afebrile. Systemic investigation was determined to be normal except Central Nervous System (CNS) examination some neurological symptoms were observed speech disturbance, difficulty in swallowing, involuntary movement, and the existence of a Kaiser-Fleisher ring on cornea. Also, the Finger-Flapping Hand Stereotype with vitiligo was observed in limbs with difficulty in balancing present.

Laboratory examinations depicted WBC count is 8600 cells/cumm, Hb level of 12.6gm/dl%, urine ketone bodies were normal, serum creatinine 0.9mg/dl, serum potassium 3.7meq/L, serum sodium 143meq/L, elevated level of liver enzyme of AST and ALT was observed with increase in total bilirubin, serum copper level was elevated 150mcg/dl, liver biopsy report (fig. A and fig. B) was 300 microgram/gram of liver tissue observed and urine examination more copper level was found then normal By considering the general physical examination, laboratory report with liver biopsy reports the final diagnosis was made as Wilson disease. After confirmation of diagnosis, the standard treatment to maintain normal copper levels was started with copper chelation (tab-D-Penicillamine-250 mg/po/bd), zinc Supplement (tab-Zinc 25 mg/po/tid) with Dietary Supplement (tab-pyridoxine35mg/po/od in the morning) along with the antipsychotic drug (tab-haloperidol-1025mg/po/bd) and antimuscarinic drug (tab-trihexyphenidyl-2mg/po/od). After treatment, a significant improvement was observed in the patient's symptoms with a decrease in serum copper level.

LIVER BIOPSY REPORT

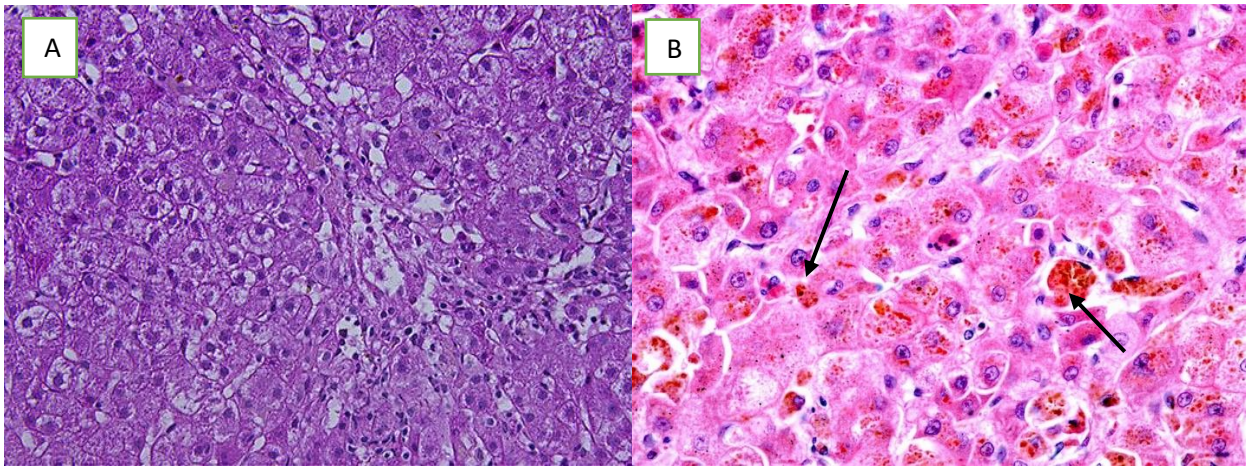

Fig. A (Normal liver cells), fig. B (Concentration of copper stain(rhodamine) in liver cells) And give a confirmatory diagnosis of Wilson's Disease.[8]

TREATMENT PROVIDED

A 10-year-old patient was admitted to pediatric ward and diagnosed with Wilson's disease. As It is life life-threatening disease, prompt recognition and diagnosis are critical to early management. The systemic treatment was given After confirmation of diagnosis, the standard treatment to maintain normal copper levels was started with copper chelation (tab-D-Penicillamine-250 mg/po/bd), zinc Supplement (tab-Zinc 25 mg/po/tid) with Dietary Supplement (tab-pyridoxine35mg/po/od in the morning) along with the antipsychotic drug (tab-haloperidol-1.25mg/po/bd) and antimuscarinic drug (tab-trihexyphenidyl-2mg/po/od). After treatment, a significant improvement was observed in the patient's symptoms with a decrease in serum copper level.

He was discharged after 15th days with oral medicine of copper chelation (tab-D-Penicillamine-250 mg/po/bd), zinc Supplement (tab-Zinc 25 mg/po/tid) with Dietary Supplement (tab-pyridoxine35mg/po/od in the morning and with the antipsychotic drug (tab-haloperidol-1.25mg/po/od) and follow-up after 8 days was suggested. He was substantially recovering with improvement in patient symptoms with a decrease in serum copper level also the neurological symptoms were relived. Although the condition gets better with medicine for management,

life-long should be taken with time-to-time follow-up recommended.

DISCUSSION

This case presents a 10-year-old boy who was hospitalized in department of Pediatrics with main complaint of upper limb finger folding unknowingly for 45 days with speech disturbance and difficulty in swallowing of food for 10 days. Also, the involuntary movements which are intentional and spontaneous, progressive in nature from 20 days. The patient had a history of jaundice 1 year back and was relieved after taking medication again reoccurrence occurred after 3 weeks and he had a history of epistaxis. His mother had a history of antenatal and had taken 2 inj- Tetanus Toxoid dose.

Diagnosis of Wilson disease was made by clinical presentation, serum copper level and liver biopsy report. After confirmation of diagnosis, the standard treatment to maintain normal copper levels was started with copper chelation (tab-D-Penicillamine-250 mg/po/bd), zinc Supplement (tab-Zinc 25 mg/po/tid) with Dietary Supplement (tab-pyridoxine35mg/po/od in the morning) along with the antipsychotic drug (tab-haloperidol-1025mg/po/bd) and antimuscarinic drug (tab-trihexyphenidyl-2mg/po/od). After treatment, a significant improvement was observed in the patient's symptoms with a decrease in serum copper level. It is a rare genetic condition that is significantly seen in pediatrics and reported as a prevalence of 1 in 30,000 births. The clinical presentation of Wilson disease hepatic symptoms, neurological symptoms, other symptoms like renal dysfunction, hemolysis, joint pain, and Kaiser-Fleisher ring on the cornea.

As it is a non-curable disease several studies recommended management of Wilson's disease using copper chelation, zinc supplements, dietary change and in some cases liver transplantations is necessary.

CONCLUSION

Due to mutation in the gene ATP7B that encodes a copper transport protein on chromosome 13 leads to the deposition of copper into various organs, mostly in the liver, eye, brain, and other different tissue which result in several clinical symptoms mostly neurological, hepatic, as well as psychiatric symptoms along with this the presence kaiser-Fleisher ring on cornea. In Wilson disease, elimination of copper doesn’t take place leading to the accumulation of copper in various organs which causes a life-threatening condition that needs early recognition with diagnosis. This is crucial to avoid irreversible organ damage and other hurdles. The diagnosis was made by clinical presentation, serum copper level, and liver biopsy report. After confirmation of diagnosis, the standard treatment to maintain normal copper levels was started with copper chelation, zinc Supplement with Dietary Supplement along with the antipsychotic drug and antimuscarinic drug. After treatment, a significant improvement was observed in the patient's symptoms with a decrease in serum copper level and time to time follow-up was recommended. Early recognition and diagnosis are crucial to prevent irreversible organ damage and other complications in Wilson's disease.

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ABBREVIATIONS

WD: Wilson Disease
ATP7B: ATPase Copper Transporting Beta
KFR: Kaiser-Fleisher Ring
PO: Per Oral
OPD: Out Patient Department
FFHS: Finger-Flapping Hand Stereotype
CNS: Central Nervous System

FUNDING

No funds were obtained.

CONFLICT OF INTEREST

All authors declared the absence of any conflict of interest.

INFORMED CONSENT

Informed consent was obtained from the parents of patient.

AUTHOR CONTRIBUTION

All authors contributed equally.

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