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Case Report

Paediatric Wilson's Disease: A Case Series.

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Abstract

Introduction: Wilson's disease (WD) is a potentially fatal rare copper metabolism disorder which leads to copper accumulation in organs such as liver, brain, and cornea. Prompt diagnoses and treatments help prevent complications, improve patients' quality of life, and ensure a normal life expectancy. The clinical presentations and outcomes of WD can vary within a single family.

First Case: We describe a twelve-year-old boy who presented with jaundice, haemolytic anemia, and advanced fibrosis (fibroscan 56.4 kPa) and on detailed investigations was diagnosed with exome confirmed Wilson's disease. He had recurrent episodes of decompensation but well-planned, eighteen-month multimodal therapy including D-penicillamine, zinc, carvedilol, and plasma exchange yielded dramatic improvement which was evidenced by symptomatic recovery as well as substantial decrease in fibroscan score from 56.4 kPa to 9.3 kPa, with normalized bilirubin and near-normal transaminases. The family history was significant and his elder sister died of WD, few years back but no family screening was done at that point of time, highlighting screening gaps. The parents are asymptomatic but yet to be screened for WD due to financial constraints. This case demonstrates fibrosis reversibility in children with aggressive early intervention.

Second Case: A sixteen-year-old male presented with backache, generalized weakness and low mood level for last three months. He was seen by pain physician first who advised for routine labs and ultrasonogram abdomen which revealed altered echotexture of liver with multiple regenerative nodules. Thus, on detailed evaluation for cirrhosis liver was done and on same he was diagnosed to be having Wilson's disease, on basis of increased 24- hour urinary copper excretion, low serum ceruloplasmin levels and liver biopsy. The upper gastro-intestinal endoscopy showed low grade oesophageal varices and Fibroscan score was 31 Kpa, suggestive of cirrhotic pattern. He was started on D-penicillamine and Zinc and was symptomatically better on follow up after two months.

Conclusion: Wilson's disease can have varied manifestations and can be easily missed in early stages. The timely diagnosis and initiation of management can lead to significant fibrosis reversibility. Early diagnosis and family awareness can prevent fatal outcomes.

Keywords: Wilson's disease, Pediatric, Fibrosis regression, D- penicillamine, Kayser-Fleischer rings, Exome sequencing.

INTRODUCTION

Wilson's disease (WD) is caused by an inborn error of copper metabolism. The estimated prevalence in Europe, Asia, and the United States varies between 1:30000 to 1:50000 inhabitants but certain regions show higher estimated prevalence due to increased frequencies of disease-causing mutations and consanguinity [1]. WD is a monogenic autosomal recessive disorder, with a genetic defect on chromosome 13q14.3. More than a thousand mutations of the ATP7B gene are there which cause alteration of the copper-transporting ATPase beta protein, and modifying copper homeostasis [2]. Hepatic manifestations of WD range from asymptomatic presentations to acute liver failure and clinical findings include hepatomegaly, steatosis, or incidental elevations in aminotransferases and are common at the time of diagnosis. Neurological manifestations are varied and include dysarthria, gait disturbances, ataxia, dystonia, parkinsonism, and tremors. Psychiatric symptoms comprise personality and behavioural changes, depression, cognitive problems, and bipolar and psychotic disorders [3,4]. Kayser–Fleischer rings (K-F rings), observed upon slit lamp examination, are considered pathognomonic for WD; they result from copper deposition on Descemet's membrane, and are visualized as a brown or golden pigment [4-6]. Diagnostic and therapeutic delays are common, leading to neurological disability and hepatic complications. The diagnosis, on the basis of the Leipzig scoring criteria, involves both clinical and laboratory examination results. Brain magnetic resonance imaging (MRI) is valuable for visualizing copper deposition on damaged areas especially the basal ganglia. The 'face of the giant panda' sign is another finding considered pathognomonic for WD [3-6]. WD requires a lifelong treatment with a combination of oral pharmacotherapy and a copper-restricted diet, recommended for both symptomatic and asymptomatic patients. Treatment with chelating agents, such as D-penicillamine (DPA) or trientine, is recommended initially for patients with active disease, and an oral chelator

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at a reduced dose or zinc may be indicated for maintenance therapy [4]. Liver transplantation is reserved for specific cases, such as patients with progressive chronic liver failure resistant to pharmacotherapy and acute liver failure [4]. Children often present with acute liver injury or cirrhosis. Fibrosis regression is possible with prompt chelation, yet family history of early death emphasizes screening urgency. We report a genetically confirmed pediatric case with serial imaging and long-term outcome.

FIRST CASE REPORT

A twelve-year-old boy, first noticed scleral icterus two years back, for which on advice of local private practitioner ultrasonogram abdomen was done which showed some doubtful changes. Later he developed increased jaundice and abdominal distension for which indoor admission and detailed evaluation was done. He had significant anemia with haemoglobin level of 8.1 g/dL which on evaluation was confirmed to be Coombs-negative haemolytic anemia. The total serum bilirubin was3.7 mg/dL with AST 139 U/L and ALT 85 U/L, a ratio reversal which was suggestive of cirrhotic pattern. The alkaline phosphatase level was 209 U/L. The viral screen and autoimmune profile were negative. The ultrasonogram abdomen showed heterogeneous liver with mild ascites and normal spleen. As other aetiological factors were ruled out, thus patient was investigated on lines of WD. The 24-h urinary copper was significantly increased to 2800 µg/day and serum ceruloplasmin level was low with level of 11 mg/dL. The slit-lamp examination of eyes confirmed Kayser-Fleischer rings. The FibroScan score was 56.4 kPa with Leipzig score of 6 suggestive of WD. On genetic exome: ATP7B mutation was confirmed (WD proven). The endoscopy showed low grade esophageal varices. He was put on D-penicillamine (20 mg/kg/day), zinc 50 mg/day, carvedilol 3.125 mg twice daily, and three sessions of therapeutic plasma exchange were done. He got stabilized and was discharged on above therapy. After, a gap of six months, despite being on regular treatment, he again decompensated with development of increased jaundice and ascites. He was coagulopathic with INR of 2.66, was again subjected to four sessions of therapeutic plasma exchange and D-penicillamine was increased up to 25 mg/kg/day. The endoscopy again showed small esophageal varices with HVPG of 13 mm Hg. He was again stabilized and discharged on above treatment. Later on, he developed gross haematuria from right ureteric calculus which was managed conservatively. At this point of time his 24-h urinary copper level have substantially decreased to 196.43 µg/day (marked decline). The total serum bilirubin is completely normal with almost normal AST/ALT level of 56 U/L and 50 U/L respectively. The FibroScan score has got reduced to 9.3 kPa. The current therapy includes D-penicillamine (25 mg/kg/day), zinc 50 mg OD, pyridoxine 40 mg OD, carvedilol 3.125 mg BD and he is totally asymptomatic on the same. He has two elder sisters, the eldest died from Wilson's disease. The second eldest is asymptomatic with no testing. Both parents are clinically normal; carrier status unconfirmed.

SECOND CASE REPORT

A sixteen-year-old male presented with backache, generalized weakness and low mood level for last three months. He was seen by pain physician first who advised for routine labs and ultrasonogram abdomen which revealed altered echotexture of liver with multiple regenerative nodules. Thus, on detailed evaluation for cirrhosis liver was done and on same he was diagnosed to be having Wilson's disease, on basis of increased 24- hour urinary copper excretion (322.70 ug/24 hrs), low serum ceruloplasmin levels (<70) and liver biopsy. The upper gastro-intestinal endoscopy showed low grade oesophageal varices and Fibroscan score was 31 Kpa, suggestive of cirrhotic pattern. The liver function tests were normal except mild transaminitis with ratio reversal i.e. AST/ALT level of 64 U/L and 55 U/L respectively. MRCP revealed changes of chronic liver disease with splenomegaly with multiple nodules (siderotic) and regenerative nodules in the liver parenchyma with distended gall bladder with pericholecystic fluid and oedema and central ducts showing irregularity and beaded appearance. The liver biopsy showed features of chronic hepatitis with steatohepatitis and mild activity with advanced fibrosis and peri-cellular fibrosis. There was pan zonal hepatic copper deposition. The slit lamp examination showed KF rings. The MRI brain revealed bilateral basal ganglionic changes along with hyperintense signal intensity in the pons. The nerve conduction study revealed changes of mild left carpal tunnel syndrome. The Leipzig score was 9, confirmatory of WD. He was started on D-penicillamine and Zinc and was symptomatically better on follow up after two months.

Figure 1. MRCP Showing Multiple Regenerative Nodules in Live.

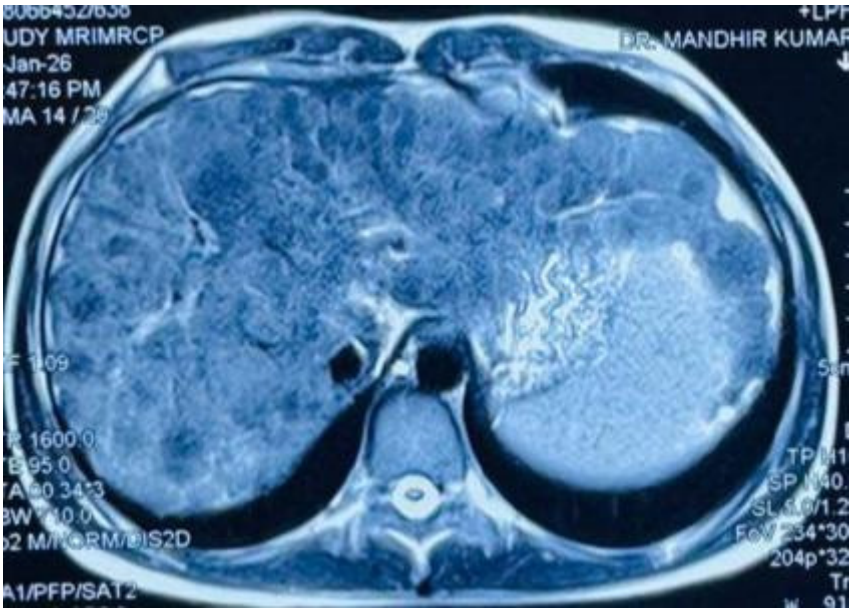
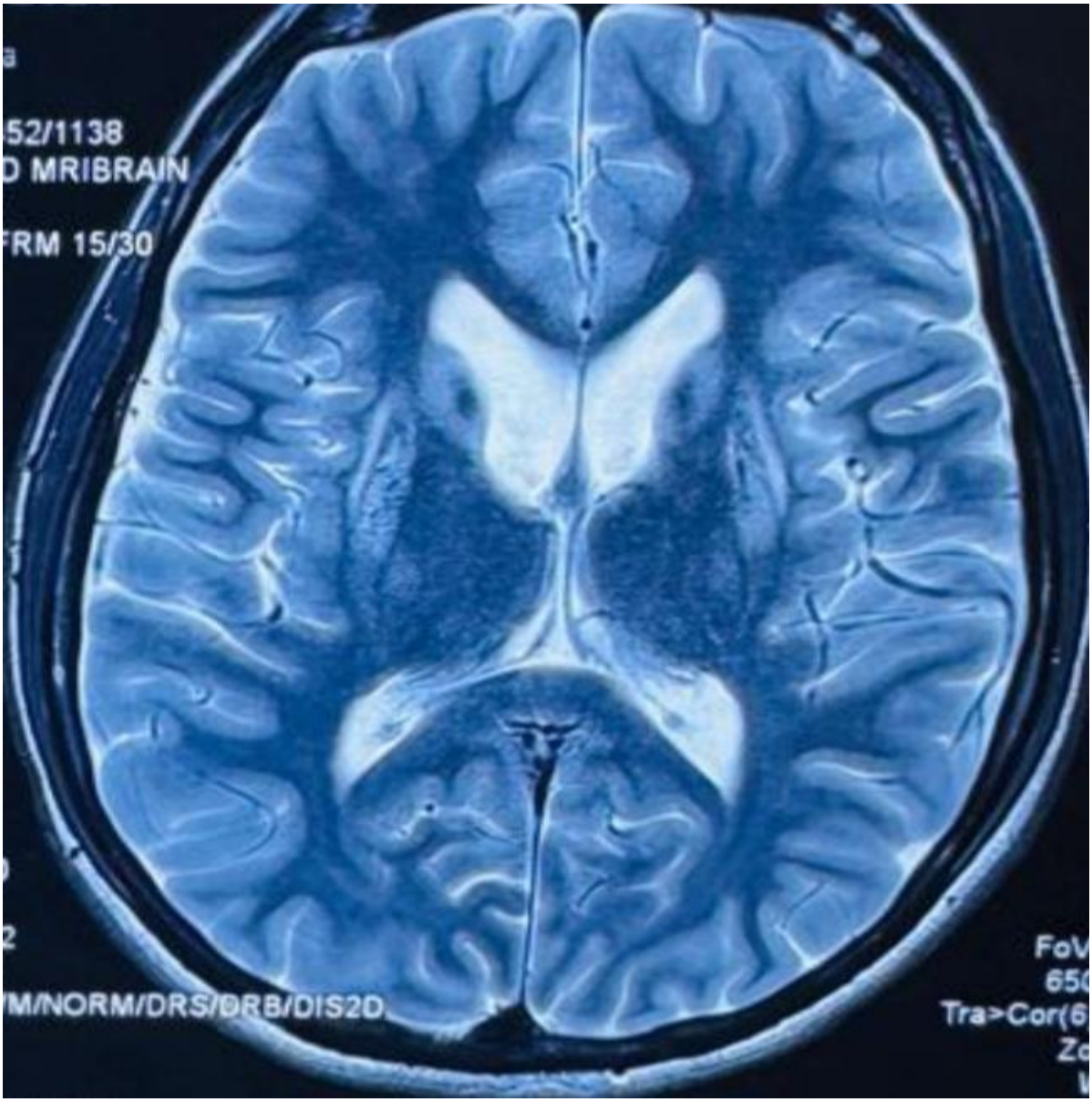


Figure 2. MRI Brain Showing Bilateral Basal Ganglia Calcification (Panda Sign).



DISCUSSION

Our cases highlight the varied presentation of Wilson's disease, importance of family screening and effectivity of multimodal approach, even in decompensated stage. In first case, presentation was with jaundice and decompensation but had there been family screening, at time of death of elder sister with WD, then the younger brother could have diagnosed early, without going into decompensated stage. Early haemolytic crisis, portal hypertension, and recurrent ascites reflect severe WD, yet copper excretion normalized and fibrosis regressed strikingly from >50 kPa) to <10 kPa, with chelation and supportive care. Plasma exchange bridged coagulopathy; zinc and pyridoxine mitigated toxicity. Family history of sibling death underscores urgent genetic screening in relatives. Long-term neurological monitoring remains essential. The second case presented with atypical features of backache, generalized fatiguability and low mood which is difficult to be attributed to WD but ultrasonogram abdomen revealed findings of chronic liver disease which ultimately led to diagnosis of WD on detailed evaluation. Hence, the treating Physicians or Gastroenterologist or Hepatologist should have broader vision for timely and proper evaluation of both typical and atypical findings of WD.

CONCLUSION

Wilson's disease can have varied manifestations and can be easily missed in early stages. The timely diagnosis and initiation of management can lead to significant fibrosis reversibility. Early diagnosis and family awareness can prevent fatal outcomes.

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