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A case Report on abnormal presentation of wilson's disease

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I. INTRODUCTION:

Wilson's disease is a rare autosomal recessive disorder characterised by the accumulation of copper in the liver, brain, cornea and kidneys. Wilson's disease occurs in about 1 in 30,000 people.[1] Children with WD are usually normal at birth and may remain healthy for a variable period of time; most cases present in the second and third decade of life[2]. Males and females are equally affected. It is a progressive disease and could be fatal if left untreated, but the timely diagnosis remains a challenge [3].

II. CASE SUMMARY:

An Twelve-year-old female girl first orderborn to second degree consanguineous marriage was presented to the paediatric department with severe abdominal pain and burning micturition and seizures ,dystonic movements , with abnormal gait and difficulty in speech. Routine blood investigations were normal,liver function tests were normal,no organomegaly detected.Usg showed multiple stones with dense concretions in pelvic cavity.MRI was done for dystonic movements showed hyperintensities in basal ganglia and caudate nucleus in FLAIR sequences suggesting of wilsons disease. Then on further evaluation slit lamp examinationshowed Kayser-Fliescher rings noted in both eyes.ceruloplasmin, urine copper were also low hence confirming the diagnosis of Wilson's disease. She was treated with copper chelators (D-penicillamine & Zinc) and anti convulsants and pacitane. Gradually she showed improvement in clinical signs.Later all the 4 siblings of the patient are also screened and 3 out of 4 siblings showed lower ceruloplasmin levels but asymptomatic thereby started on copper chelators.

III. DISSCUSION:

Wilson's disease is a autosomal recessive condition due to mutation in the disease protein gene ATP7B gene.Its a genetic disorder in which copper builds up in the body. There are three different types of Wilson disease based upon the genetic variability. The juvenile type appear before 16-years of age and is predominantly a liver disease before 5 years. The Salvic type appear after 16 years of age and is predominantly a neurological disease. The serum ceruloplasmin remain normal to these two types of the disease.[4]The third type, classified as atypical Wilson disease is characterised by low serum ceruloplasmin level and clinical picture similar to those of juvenile type[5] The patient reported has atypical form of the disease with low ceruloplasmin level.

Similarly in 2003 a patient presented with only psychiatric symptoms,diagnosed first as anxiety with psychotic disorder but symptoms didn't subsided with treatment on further evaluation diagnosed as wilsons disease and treated.It was published in indian journal of psychiatry.[6]

The basic biochemical abnormality in Wilson disease is the large deposit and accumulation of copper in the liver and tissues with accompanying cupri- uria. Non-ceruloplasmin bound copper concentration is increased in plasma, ceruloplasmin level is usually low but is often normal. There may be uric aciduria and amine aciduria as copper probably interferes with renal tubular function causing renal stones.

Liver transplantation is an effective cure for Wilson's disease but is used only inparticular scenarios because of the risks and complications associated with the procedure. It is used mainly in people with fulminant liver failure who fail to respond to medical treatment or in those with advanced chronic liver disease. Liver transplantation is avoided in severe neuropsychiatric illness, in which its benefit has not been demonstrated.[7]

IV. CONCLUSION:

Wilson's disease is an autosomal recessive inherited metabolic disorder.Not every patient has the same spectrum of symptoms. Early diagnosis and appropriate management help to prevent the

systemic complications Siblings needed to be screened to prevent manifestations.

REFERENCES:

[1]. "Wilson Disease". NIDDK. July 2014. Archived from the original on 2016-10-04. Retrieved 2016-11-06.
[2]. Saito T: Presenting symptoms and natural history of Wilson disease. Eur J Pediatrics. 1987, 146 (3): 261-265. 10.1007/BF00716470.
[3]. http://www.rarediseasesindia.org/wilsons.
[4]. Swaiman KF, Meknes JH , Devivo Dc, Prensky AL -" Metabolic disorders of the central nervous system" . In the practice of paediatric neurology. 2nd Ed. Vol. I Ed. Swaiman KF and Wright F.S. The C.V. Mosby Co. London. 1982. P. 575
[5]. Misra M. Rath S- Hepato lenticular degeneration. Orissa Medical Journal, 1988 (In press).
[6]. INDIAN JOURNAL OF PSYCHIATRY, 2003,45 (IV), 253-254
[7]. Ala A, Walker AP, Ashkan K, Dooley JS, Schilsky ML (2007). "Wilson's disease". Lancet. **369** (9559): 397–408. doi:10.1016/S0140-6736(07)60196-2. PMID 17276780