

Wilson's Disease Presenting as Resistant Rickets

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Abstract

Wilson's disease is most common disorder of copper metabolism. It has varied clinical presentations. We report a 12 years old female child presenting with genu valgum progressed over 6 months. Careful examination, high index of suspicion and investigations confirmed Wilson's disease.

Keywords: Wilson's disease; Rickets

Introduction

The first reported case of Wilson's disease with rachitic presentation was in 1968 by Cavallino [1]. Since then only few pediatric cases of Wilson's disease with this rare presentation have been reported.

Case Report

A 12 years old female child was admitted with mild pain and deformity at both knee joints which was progressive over 6 months. There was no history of swelling or morning stiffness of joints, no history of repeated fractures or similar illness in family. She was admitted for jaundice 3 years back. She was given mega doses of vitamin D before she referred to our center.

On examination she was normally built and nourished

with height 138 cm and weight 27 kg, and vital signs and parameters were normal. On Local examination, knee joint was normal and there was genu valgum with bimalleolar distance of 13 cm; angulations were more in right than left (Fig. 1). Rest of systemic examination was found to be normal.

Investigations revealed Hb10.4 gm/dl, TLC $4.8 \times 10^3/\mu\text{L}$, PC $208 \times 10^3/\mu\text{L}$, ESR 25 mm/h, ALT 30 U/L, AST 22 U/L, bilirubin 0.8 mg/dl, Urea 28 mg/dL, Creatinine 1.1 mg/dL, Na 149 mEq/L, K 3.1 mEq/L, Ca 7.5 mg/dL, Phosphate 4.8 mg/dL (2.9 - 5.4), AlkPO₄ 203 units/L (105 - 420). X-ray local part (knee) was suggestive of mild osteoporotic changes.

Urine examination showed urine pH 7.5, urine anion gap +26, urinary calcium 700 mg (100 - 250) and urinary phosphate 300mg (900 - 1300). ABG revealed pH 7.29, PO₂ 108 mmHg, PCO₂ 36 mmHg, and HCO₃ 17 mEq/L with anion gap of 14. Parathormone level was 25.5 pg/ml (12 - 95) and Vitamin D₃ level was 27 ng/ml (5.9 - 59).



Figure 1. Genu valgum with increased bimalleolar distance.

Manuscript accepted for publication November 23, 2010

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doi:10.4021/gr262w

We suspected Wilson's disease in this unexplained bone disease. Slit lamp examination of her eyes revealed Kayser-Fleisher ring. Ceruloplasmin level was 2.81 mg/dL (25 - 60). Urinary copper excretion was 1140 µg/day. USG abdomen revealed diffuse fatty infiltration of liver. MRI brain showed copper deposits in caudate nucleus and putamina. So diagnosis of Wilson's disease with rickets due to renal tubular acidosis was made. She was given zinc acetate, D-penicillamine, sohl solution and oral calcium; and asked to follow up in OPD to observe improvement.

Discussion

Wilson's disease (WD; Hepatolenticular degeneration) is a rare inherited autosomal recessive inborn error of copper metabolism characterized by toxic accumulation of copper in liver, brain, cornea and other tissues [2]. The genetic basis for WD is linked to one of the transport proteins. It was linked to the long arm of chromosome 13 by identifying the association of WD with esterase D deficiency in an inbred Arab family [3]. Subsequent analysis revealed a defect in a P-type adenosine triphosphatase (ATPase) involved in the transport of copper across the trans-Golgi and into transport vesicles [4]. The interesting feature of WD is its wide variety of phenotypic presentations which is also influenced by race and region. This may be due to genetic heterogeneity and different types of mutations involved.

Clinical presentation of WD in India does not always follow the pattern as is usually seen in the Western world [5, 6]. Wadia et al [7] first drew attention in this regard and observed that generalized osteoporosis, rickets, or even renal rickets could be the striking manifestations, at least in 8 out of 23 patients. Rickets and osteoporosis as presenting feature is also reported by few other Indian authors [8, 9].

In our case we also found rickets as only presentation. The child had no symptoms and signs related to liver and CNS involvement although involvement was detected through MRI brain and USG abdomen. There are many mechanisms implicated for rickets in WD. It may be due to hypoparathyroidism, and renal rickets may be due to fanconi disease. In our case, we ruled out hypoparathyroidism, and

rickets was probably due to renal tubular acidosis.

Authors' Contribution

JPG and SSR were involved in diagnosis and management of patient. NK wrote manuscript which was critically reviewed by JPG. JPG will act as guarantor.

Conflict of Interest

Authors declare no competing interest.

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