International Journal Dental and Medical Sciences Research
Volume 3, Issue 6, Nov-Dec 2021 pp 211- 213 www.ijdmsrjournal.com ISSN: 2582-6018

A Rare Case Report- Hypophosphataemic Rickets Due To Fanconis Syndrome Secondary To Wilsons Disease

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Submitted: 10-11-2021 Revised: 25-11-2021 Accepted: 28-11-2021

ABSTRACT
Renal tubular disorders are an important cause of refractory rickets. Wilson's disease, an inherited disorder of copper metabolism has varied presentations. We present a case of refractory rickets due to Fanconi's syndrome attributable to Wilson's disease. 11 year old cachexic looking child presented with complains of clumsy gait and slow unclear slurred speech for 6 months.On examination he had widening of bilateral wrist joint and genuvalgum,dysarthric speech, diplegic spastic gait, Hypertonia in lower limbs, exaggerated deep tendon reflexes in Bilateral lower limbs, Babinski's positive ,with liver span of 12 centimeters.Skeletal radiographs showed features suggestive of rickets at knee and wrist joints. Routine biochemistry was normal, 25-hydroxyvitamin D [25(OH)D] was adequate (57.1 ng/dL), with normal corrected calcium (9.24 mg/dL), low phosphate (2.50 mg/dL), elevated bone-specific alkaline phosphatase, and normal renal functions.Twenty four hour urine revealed proteinuria,phosphaturia, glucosuria with normal blood sugars. Blood gas analysis revealed normal anion gap metabolic acidosis with a urine pH of 7. A search for causes revealed Kayser-Fleischer rings. The diagnosis of Wilson's disease was confirmed by low serum ceruloplasmin levels(8.86 mg/dl) (N=22-61) with high 24-hour urine copper levels (720 ug/24hr (N=15-60). Treatment started was Depenicillamine ,osteocalcium , copper free diet, pyridoxine and zinc along with replacement of alkali. Rickets as a presenting feature of Wilson's disease has been reported rarely. Recognition of this entity is important, as treatment of the primary condition may improve tubular function as well.

KEYWORDS: Fanconi's syndrome, refractory rickets, Wilson's disease

INTRODUCTION:

The deficiency of Vitamin D is the most common cause of rickets with satisfactory response to treatment. In patients refractory to replacement of vitamin D, a detailed search for other aetiologies of rickets like hypophosphatemic rickets including renal tubular acidosis, renal osteodystrophy, and vitamin D-resistant rickets should be carried out.[1] Proximal renal tubular acidosis which when accompanied by other proximal tubular defects like glycosuria, aminoaciduria, uricosuria, and phosphaturia is referred to as Fanconi's syndrome[2] which is very rare but an important cause of short stature and hypophosphatemic rickets. Fanconi's syndrome can either be a due to a primary abnormality in the renal tubular cells or secondary to prerenal disorders in which toxic metabolic substances lead to the derangement of tubular functions like cystinosis, Wilson's disease, tyrosinemia, galactosemia, and Lowe's syndrome.[2] Wilsons disease (WD) also known as Hepatolenticular degeneration is an autosomal recessive disease which occurs due to abnormal over accumulation of copper in various organs of the body. One of the rare presentations is Fanconi's syndrome in which there is a defect in Proximal convoluted tubule (PCT) of kidney leading to inadequate reabsorption of various substances.
We present a child of 11 years presenting with rickets refractory to replacement of vitamin D due to Fanconi's syndrome secondary to Wilson's disease.

CASE REPORT: 11 year old cachexic looking child presented with complains of clumsy gait and slow unclear slurred speech for 6 month.On examination he had widening of bilateral wrist joint and genuvalgum,dysarthric speech, diplegic spastic

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gait, Hypertonia in lower limbs, exaggerated deep tendon reflexes in Bilateral lower limbs, Babinski's positive ,with liver span of 12 centimeters. MRI Brain showed Panda's sign(figure 2). Low Serum Ceruloplasmin ,Increased 24 hour urinary copper leveland KF rings on examination was confirmatory of WD.(Table 1)X-rays of Bilateral wrists and Bilateral knee joints showed decrease in bone density, widening of metaphysis and metaphyseal splaying and cupping.(figure 2) Decreased Serum phosphorous, increased Urinary phosphorous , increased Alkaline phosphate ,normal PTH with normal 25 hydroxy Vit D , normal corrected calcium and elevated alkaline phosphatase was confirmative of Hypophosphataemic Rickets (HR).(Table 1)Proteinuria with increase in urinary Electrolytes (sodium, potassium, chloride and phosphorous) along with glucosuria with normal blood sugars was confirmative of Fanconi's syndrome.(Table 1) Blood gas analysis revealed normal anion gap metabolic acidosis with a urine pH of 7Depenicillamine , osteocalcium , copper free diet, pyridoxine and zinc was started. His elder brother had low serum ceruloplasmin and increased 24 hour urinary copper with KF ring which was confirmative ofWD. On follow up there is improvement in gait & speech of the child.

DISCUSSION: There are varied presentations of WD which is an inherited disorder of Copper metabolism.FS is one of the rare presentations in which there is a defect in Proximal convoluted tubule (PCT) of kidney leading to inadequate reabsorption of various substances. Renal rickets as a presenting feature of WD has been reported rarely due to copper deposition in PCT. Features of FS include Acidosis, Electrolyte imbalance, Osteomalacia. Treatment includes treating the cause of FS along with replacement of various substances lost by malfunctioning of kidney. Though the majority of rickets is due to deficiency of Vitamin D, it is important to be vigilant of the cases which do not respond adequately to its replacement.Renal tubular diseases account for one third of the cases of refractory rickets in nonazotemic Indian patients .[5] Rickets and osteomalacia are more common in Fanconi's syndrome as compared to other renal tubular diseases.[2] Wilson's disease, though its exact prevalence is not known in our country, is believed to be under-reported and may have a slightly varied clinical profile in terms of presentations in the first or second decade, and the musculosketetal form is more common.[3] Wilson's disease is one of the few conditions, which on after treatment there is an improvement in renal function.So it is important to make an aetiological diagnosis in Fanconi's syndrome.

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TABLE 1: INVESTIGATIONS

24 hrs Urinary copper	720 ug/24hr (15-60)
Serum ceruloplasmin	8.86 mg/dl (22-61)
Parathyroid hormone levels	59.3 pg/ml (50-68)
Alkaline Phosphate	1245 U/L (104-345)

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25 hydroxy vit D	57.2 ng/ml (25-80)
S.Calcium	9.2 mg/dl (9-11)
Serum Phosphorous	2.50 mg/dl (4.5-6.5)
Urine R/M	Ph=7 , sugar and protein present
S. creatine	0.6
Urinary electrolytes	Na:194, Cl:185, K:60, Phosphorous:92.4

FIGURE 1:MRI BRAIN – PANDA'S SIGN

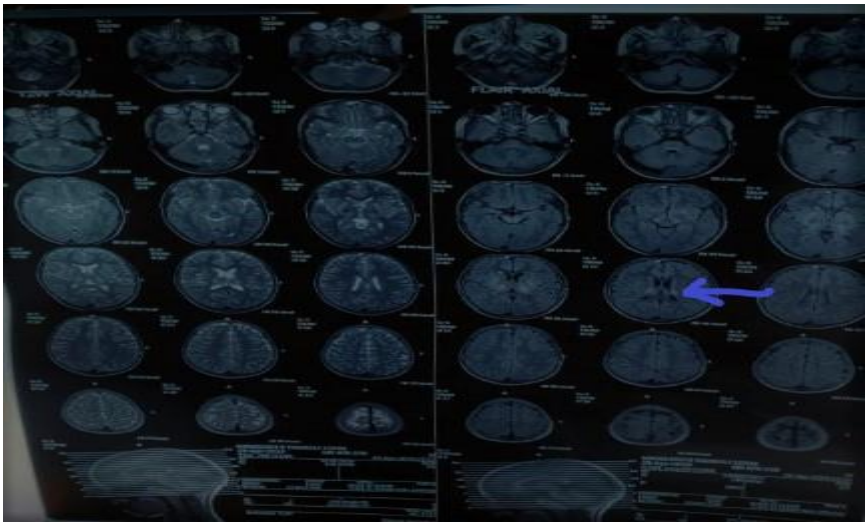

FIGURE 2: BILATERAL XRAY AND KNEE JOINT