Renal tubular acidosis causing severe growth delay and rickets in two siblings in Haiti

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BACKGROUND

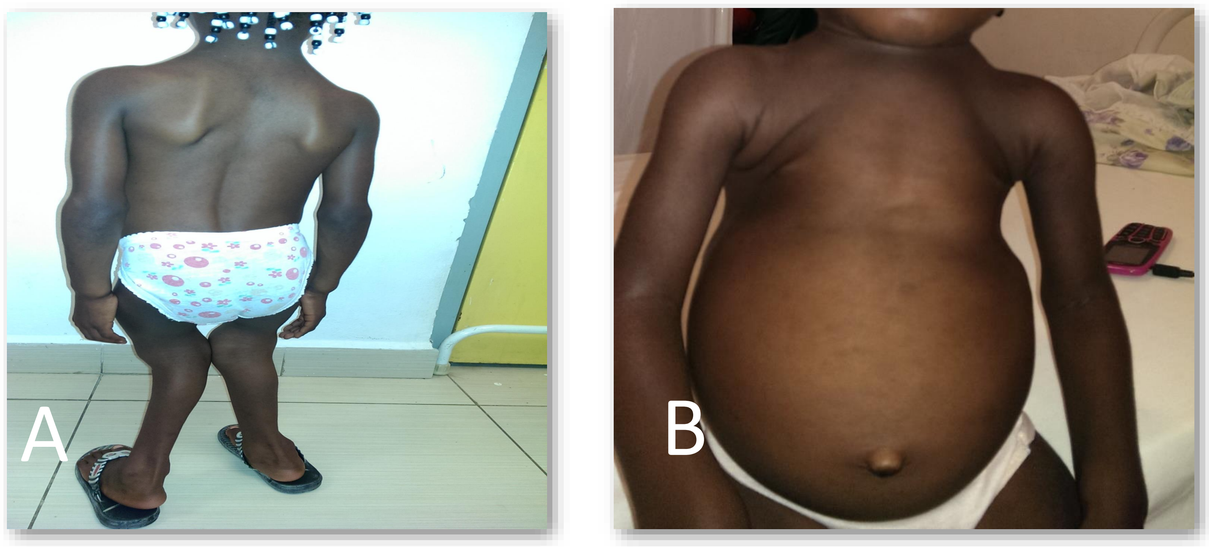
Worldwide, rickets is the most common form of metabolic bone disease in children. Many cases are due to poor vitamin D intake or calcium deficient diet, however some cases are refractory to vitamin D therapy and are related to renal defects, these include rickets of renal tubular acidosis.

CASE DESCRIPTION

An 8-year-old girl born to non-consanguineous parents was evaluated for a 6 year history of bony deformations, short stature, and abdominal pain. Her 2.5 year-old sister presented similar symptoms, associated with inability to walk for the past six months. Neuro-cognitive development was described as normal in both children. There was no history of bone deformation in the family. Both siblings were exclusively breastfed for six months and were since consuming a balanced diet. Neither was taking any medications and they were adequately exposed to sunlight

EXAMINATION

On exam, weight and height were 17.2 kg (-3 standard deviation scores [SDS]) and 99 cm (-5.6 SDS) in the older, and 9.8 kg (-3 SDS) and 77 cm (-3 SDS) in the younger sibling. Mild Kussmaul breathing was evident. Both had a sawtooth enamel and multiple dental caries, asymmetrical chests, rachitic rosary, distended abdomen, wide wrist, epiphyseal beads, multiple bone deformity, the older had a waddling gait and bilateral genu valgus.



Photographs of the 1st (A) and the 2nd (A) sibling

INVESTIGATIONS

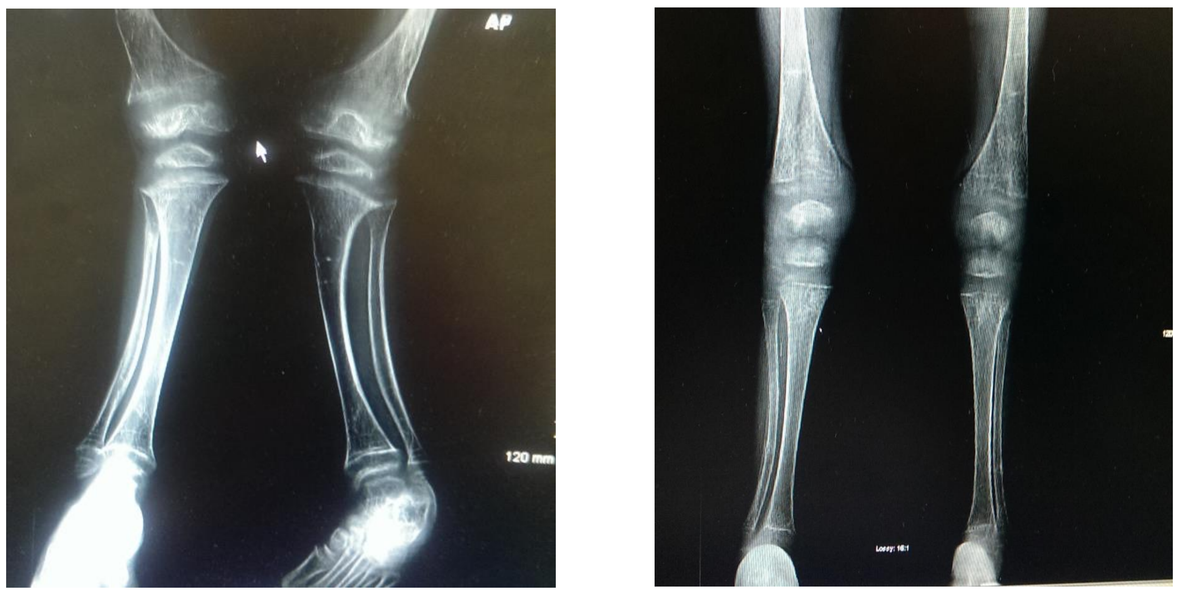
Abdominal Ultrasound:
No evidence of lithiasis nor nephrocalcinosis.

Table 1: Initial laboratory investigations in both siblings:

Lab test	1st sibling	2nd sibling	Reference
Bicarbonate	9	10	23-30 meq/L
Phosphate	2.8	2.5	2.5-4.8 mg/dl
Calcium (total)	8.8	8.7	8.4-10.2 mg/dl
Chloride	124	121	98-107 meq/l
Anion gap	16	15	8-16 meq/L
Alkaline Phosphatase	880	1047	38-126 U/L
Urinary pH	9	8.5	4.5 - 8
PTH	22.3	17.9	8.3-68.0 pg/ml
25-OH-Vitamin D	60.6	59.2	30-100 ng/ml

INVESTIGATIONS cont'd

Knee X-rays of sibling 1 (left) and 2 (right) at presentation



DIAGNOSIS & TREATMENT

Diagnosis: Hypophosphatemic rickets complicating renal tubular acidosis (RTA).
Treatment: Sodium bicarbonate PO 6meq/kg/day (given as baking soda)

FOLLOW-UP

About one month after the introduction of sodium bicarbonate both girls developed edema such that treatment was held. Physical exam was unchanged. Blood chemistries were surprisingly normal in one laboratory, and upon repeat 10 days later in another laboratory were similar to baseline (table 2). Plan to restart sodium bicarbonate at a lower dose, and add vitamin D and oral phosphate was delayed due to significant barriers to follow-up, including family's remote location and availability of laboratory testing and access to clinical care closer to home. Four months later there has been no follow-up despite multiple attempts at scheduling clinic visits.

Table 2: Follow-up laboratory investigations in both siblings:

Lab test	1st / 2nd sibling	Repeat 1st / 2nd sibling	Reference
Bicarbonate	31 / 28	13 / 11	23-30 mEq/L
Phosphate	3.7 / 4.2	2.4 / 2.5	2.5-4.8 mg/dl
Calcium (total)	8.5 / 8.4	8.9 / 9.3	8.5-10.7 mg/dl
Chloride	101 / 101	117 / 112	98-108 mEq/L
Potassium	3.9 / 4.5	3.2 / 3.1	3.5-5.4 mEq/L
Anion Gap	6 / 9	11 / 16	8-16 meq/L
Urinary pH	--	7.0 / 7.0	4.5 - 8

DISCUSSION

Rickets in the world mostly stems from nutritional insufficiency, but some cases relate to renal disease. In our patients, the initial diagnosis of rickets was based on clinical and radiologic findings. The primary diagnosis of RTA was identified after further evaluation: hyperchloremic non-anion gap metabolic acidosis, hypophosphatemia and alkaline urine. Unfortunately, urine bicarbonate and phosphorus measurements, ammonium chloride challenge test to determine the type of RTA, and genetic studies to identify the genetic alteration accompanying the RTA were not available, and clinical follow-up has remained a challenge.

CONCLUSION

Index of suspicion for causes of rickets other than nutritional deficiency should be raised in familial or severe cases with atypical co-symptomatology – even in settings with high rates of malnutrition. Diagnostic and therapeutic availabilities remain unacceptably limited in resource-constrained settings.

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