

A 5-month-old girl is brought to the physician for a weight check. The patient has been evaluated several times for poor weight gain. She takes 6 ounces of regular formula every 4 hours. Increasing the caloric density of her formula has not improved her growth. The patient has no diarrhea or vomiting. There is a family history of nephrolithiasis. She was born full term without complications. The patient's birth weight was 3.6 kg (8 lb, 50th percentile). Current weight is <5th percentile; length and head circumference have been tracking along the 25th percentile. The infant appears thin, but the remainder of the physical examination is unremarkable. Newborn screening results were normal. Laboratory results are as follows:

Serum chemistry

Sodium	140 mEq/L
Potassium	3 mEq/L
Chloride	121 mEq/L
Blood urea nitrogen	10 mg/dL
Creatinine	0.5 mg/dL
Calcium	9 mg/dL
Glucose	98 mg/dL

Arterial blood gases

pH	7.21
PaCO ₂	31 mm Hg
Bicarbonate	14 mEq/L

Urinalysis

pH	7.9
Potassium	Normal
Sodium	Normal

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pH	7.9
Potassium	Normal
Sodium	Normal

Which of the following is the most likely cause of this patient's failure to thrive?

- ☐ A. Cystic fibrosis
- ☐ B. Gastroesophageal reflux
- ☐ C. Insufficient caloric intake
- ☐ D. Lactic acidosis
- ☐ E. Renal tubular acidosis

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Urinalysis

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Potassium	Normal
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Bicarbonate 14 mEq/L

Urinalysis

pH 7.9

Potassium Normal

Sodium Normal

Which of the following is the most likely cause of this patient's failure to thrive?

- ☐ A. Cystic fibrosis [11%]
- ☐ B. Gastroesophageal reflux [1%]
- ☐ C. Insufficient caloric intake [4%]
- ☐ D. Lactic acidosis [11%]
- ☒ E. Renal tubular acidosis [73%]

Proceed to Next Item

Explanation:

User Id: [redacted]

Renal tubular acidosis			
Type	1 (Distal)	2 (Proximal)	4
Primary defect	Poor hydrogen secretion into urine	Poor bicarbonate resorption	Aldosterone resistance
Urine pH	≥5.5	<5.5	<5.5
Serum potassium	Low-normal	Low-normal	High
	<ul style="list-style-type: none">Genetic disordersMedication toxicity	Fanconi syndrome (glucosuria, phosphaturia)	<ul style="list-style-type: none">Obstructive uropathy

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Causes	<ul style="list-style-type: none">Genetic disordersMedication toxicityAutoimmune disorders (eg, Sjögren syndrome, rheumatoid arthritis)	Fanconi syndrome (glucosuria, phosphaturia, aminoaciduria)	<ul style="list-style-type: none">Obstructive uropathyCongenital adrenal hyperplasia

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This infant has evidence of a **normal anion gap acidosis** and **failure to thrive**. In the absence of an anion gap, a renal or gastrointestinal etiology of the acidosis is most likely. This child has no diarrhea but does have markedly alkalotic urine. These findings are suggestive of **renal tubular acidosis (RTA)**. RTA is caused by a defect in the ability of the renal tubules to reabsorb bicarbonate (type 2 RTA) or excrete hydrogen (type 1 RTA). Type 1 RTA is often a genetic disorder and is commonly associated with nephrolithiasis. Type 2 RTA may be isolated but is more commonly a component of **Fanconi syndrome** (glucosuria, aminoaciduria, and phosphaturia are also present). Type 4 RTA is caused by a defect in the sodium/potassium exchange in the distal tubule, which results in hyperkalemic, hyperchloremic metabolic acidosis. In children, obstructive uropathy and aldosterone insufficiency are common causes.

All types of RTA can present as **growth failure** (due to poor cellular growth and division in acidic conditions). Screening laboratory results will show a **low serum bicarbonate** level and hyperchloremia, which lead to a normal anion gap metabolic acidosis.

Evaluation of urine pH and urine electrolytes can help distinguish between the types of

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(Choice A) Cystic fibrosis can present with failure to thrive due to malabsorption, chronic diarrhea, and frequent sinopulmonary infections. This diagnosis is unlikely given the lack of gastrointestinal and pulmonary symptoms.

(Choice B) Gastroesophageal reflux is common in infants and can cause failure to thrive if severe. However, this infant does not have a history of spitting up or vomiting.

(Choice C) This infant is taking 36 ounces of formula a day, which is above the normal caloric intake for an infant (up to 32 ounces a day).

(Choice D) Lactic acidosis causes a high anion gap metabolic acidosis.

Educational objective:

Renal tubular acidosis is caused by a defect in either hydrogen excretion or bicarbonate resorption in the kidney. In infancy, it most commonly presents with failure to thrive due to a chronic, normal anion gap metabolic acidosis. Treatment consists of oral bicarbonate replacement.

References:

1. **Bicarbonate therapy improves growth in children with incomplete distal renal tubular acidosis.**

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References:

1. [Bicarbonate therapy improves growth in children with incomplete distal renal tubular acidosis.](#)
2. [Clinical profile and outcome of renal tubular disorders in children: A single center experience.](#)