

Duodenal Web Presenting As Pseudohypoaldosteronism In Infancy



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Abstract

A 5-month-old girl born to first-cousin parents was referred to endocrinology for evaluation following two hospitalizations for vomiting and dehydration with severe hyponatremia and hyperkalemia. She had a history of recurrent emesis and poor weight gain, with a reportedly normal abdominal and renal ultrasound.

Initial evaluation showed hyponatremia with elevated renin and aldosterone. The suspected diagnosis was pseudohypoaldosteronism type 1 (PHA).

Treatment was initiated with sodium supplementation and she subsequently maintained normal sodium and potassium levels with relative improvement in weight gain and less frequent emesis.

She re-presented with severe bilious emesis. Abdominal X-ray and ultrasound indicated obstruction. This was further confirmed by an upper-GI fluoroscopic study with findings suggestive of a duodenal web or duodenal stenosis/partial atresia. At laparotomy, a duodenal web was found, and the patient underwent duodenojejunostomy.

Following surgical correction, the patient had complete resolution of emesis with normalization of the electrolytes, renin and aldosterone. She no longer required sodium supplementation. Genetic testing for PHA was negative.

This rare case highlights the presentation of transient PHA secondary to intestinal obstruction.

Introduction

The patient is a 5-month-old girl born full-term to first-cousin parents. She was born SGA (BW 2.0 Kg) with subsequent failure to thrive and recurrent emesis. She had two prior hospitalizations for vomiting and dehydration with severe hyponatremia and hyperkalemia. Records obtained from her second hospitalization indicate that she developed gastroenteritis with two episodes of vomiting and one episode of watery diarrhea, decreased oral intake and decreased urine output, which prompted admission to a local hospital.

Initial labs showed severe hyponatremia, Na 114 mmol/L, with hyponatremia, Cl 77 mmol/L, hyperkalemia, K 7.3 mmol/L and a normal HCO₃ of 23 mmol/L. She had a reported normal abdominal and renal ultrasound. She received hypertonic saline and salbutamol to correct her electrolyte imbalance and was maintained on intravenous fluids. She rapidly improved and was discharged home with a referral to endocrinology.

Clinical Course

At the initial clinic visit the patient was in good health, and had a normal physical examination including normal female genitalia with no virilization. Lab evaluation showed hyponatremia, Na 129 mmol/L and K 3.9 mmol/L, with elevated plasma renin activity 170 ng/ml/hr (NR 2-37 ng/ml/hr) and aldosterone 275 ng/dl (NR 5-90 ng/dl). Cortisol and 17-hydroxyprogesterone were normal.

The suspected diagnosis was pseudohypoaldosteronism type 1 (PHA). Treatment was initiated with sodium supplementation and she subsequently maintained normal sodium and potassium levels with relative improvement in weight gain and less frequent emesis.

Three months' later, she re-presented with severe bilious emesis. Abdominal X-ray indicated obstruction. Urgent ultrasound showed grossly distended, fluid-filled hyper peristaltic stomach and duodenum, indicating obstruction distal to the third part of the duodenum. An upper-GI fluoroscopic study (fig. 1) revealed distention of the duodenal bulb and descending loop. The third part of the duodenum showed funnel-shaped narrowing and obstruction to the flow of contrast. These findings suggested a duodenal web or duodenal stenosis/partial atresia.

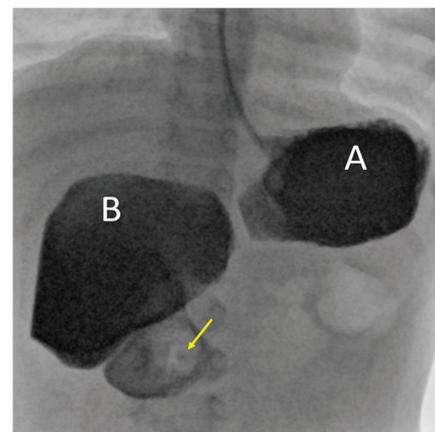


Figure 1: Fluoroscopy study showing obstruction of contrast flow distal to the third part of the duodenum.

A: Distended body and pylorus of the stomach with irregularity of the mucosal folds
B: Grossly distended duodenal bulb and loop with no distinction between the parts
Arrow: Windsock sign



Treatment

The patient had an urgent laparotomy. A duodenal web was found, and she underwent a duodenojejunostomy.

Following surgical correction, the patient had complete resolution of emesis with normalization of the electrolytes, renin and aldosterone. She no longer required sodium supplementation. Genetic testing for PHA was negative.

Figure 2: Relative weight improvement following sodium supplementation, and significant improvement post surgical correction of duodenal web

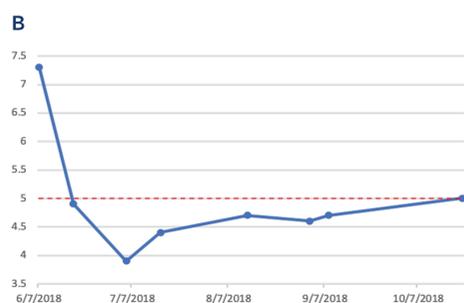
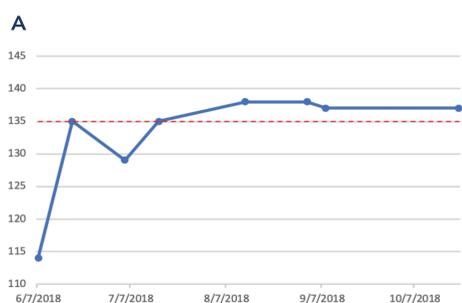
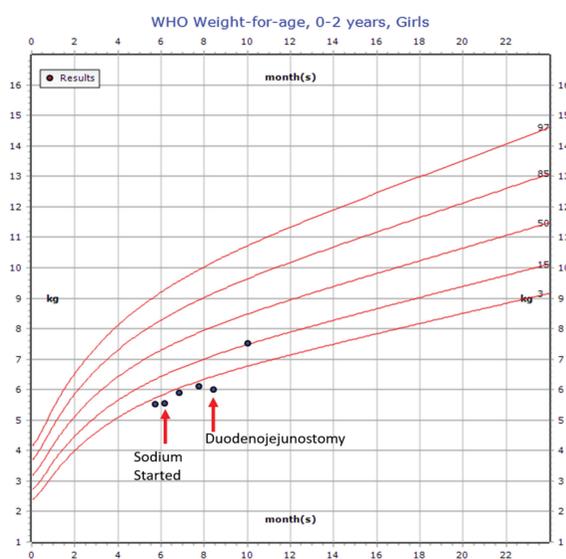
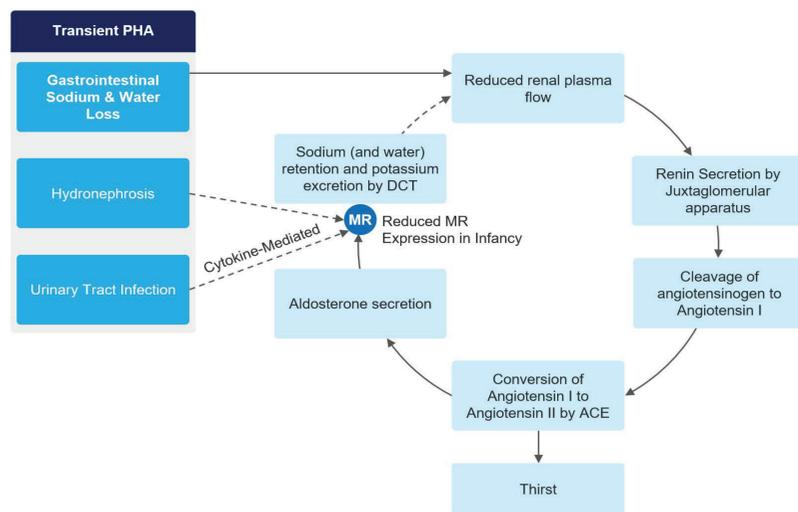


Figure 3: A) Severe hyponatremia at initial presentation, followed by normalization after starting Na supplementation and maintaining normal Na post surgery and off supplementation
B) Severe hyperkalemia at presentation with subsequent normalization even after stopping Na supplementation post-operatively

Discussion

Only one similar case of transient PHA secondary to intestinal obstruction has been previously described in an infant¹. Urinary tract infection or obstruction are the most commonly found causes of transient PHA of infancy (also known as type 3 PHA)². In the scenario we describe, PHA is due to gastrointestinal losses of sodium and water resulting in decreased renal perfusion from dehydration and consequent rise in renin and aldosterone, giving the biochemical picture of PHA.



References

1. Nissen, M., et al., Congenital Jejunal Membrane Causing Transient Pseudohypoaldosteronism and Hypoprothrombinemia in a 7-Week-Old Infant. *Klin Padiatr*, 2017. 229(5): p. 302-303.
2. Watanabe, T., Renal Mineralocorticoid Receptor Expression in Early Infancy and Secondary Pseudohypoaldosteronism Type 1. *Austin Padiatr*, 2017. 4(1): 1050