Type II Congenital Pyloric Atresia With Desquamative Enteropathy: A Case Report

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BACKGROUND

- Congenital pyloric atresia (CPA) is an exceedingly rare form of intestinal atresia with a mortality rate up to 50%[1,2]
  - **Type I**: Pyloric webs
  - **Type II**: Solid tissue replaces pylorus
  - **Type III**: Gap between stomach and duodenum
- CPA classically managed with surgical repair via pyloroplasty for types I and II, or gastroduodenostomy for type III
- CPA has been commonly associated with the following abnormalities among others:
  - Epidermolysis bullosa (up to 40%, often fatal)[2,3,4]
  - Additional gastrointestinal atresias (up to 25%)[2]
  - Aplasia cutis congenita[2]
- **Desquamative enteropathy** is rarely associated with CPA, and typically occurs along with epidermolysis bullosa (EB)[5]
  - Protein-losing enteropathy with severe diarrhea causing life-threatening illness
  - Mutation typically also contributes to EB, but cases have been described in the absence of skin findings

CASE REPORT

History of Present Illness

- Infant born at 37w4d via inducted SVD
- Echogenic bowels and late onset fetal growth restriction and mild polyhydramnios on prenatal ultrasounds
  - Resolved by week 36
- Weight down 17% from birth at 4-day follow-up

Initial Workup

- Pyloric ultrasound revealed no muscular wall thickening
- Labs: high-normal sodium, hyperchloremia
- Imaging demonstrated no gastric emptying
- Findings consistent with type I or type II CPA

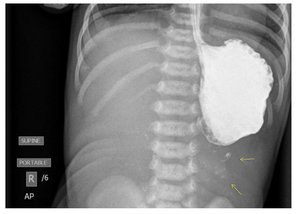

Figure 1: Upper GI series showing contrast retention in the stomach five hours after administration with reflux into the distal esophagus

CASE REPORT

Operative Management

- Heineke-Mikulicz pyloroplasty with pyloric web excision
- TPN postoperatively for nutritional support
- Started feeds on postoperative day 7

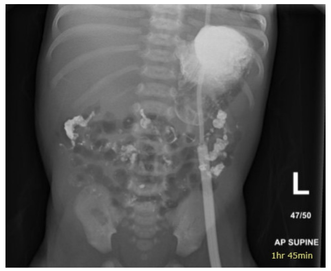

Figure 2: Upper GI series on POD#8 showing poor gastric emptying into nondilated small bowel with no evidence of obstruction

Postoperative Course

- Did not tolerate increasing feed volumes, prompting further workup
- Excessive high output diarrhea with largely unremarkable workup prompted EGD and colonoscopy
- Stool was hemoccult positive and patient required multiple packed RBC transfusions
- Stool studies revealed elevated calprotectin with no protein losses or steatorrhea
- Concern for desquamative enteropathy associated with CPA and EB
- Intravenous immunoglobulin was trialed with no significant clinical improvement

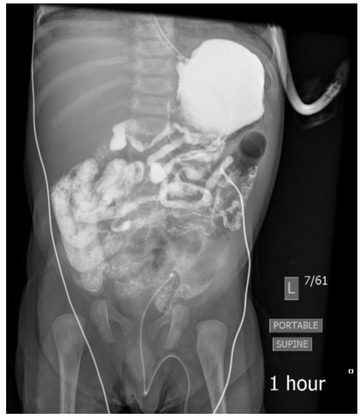

Figure 3: Upper GI series two months done postoperatively showing prompt gastric emptying with small caliber of duodenum and proximal small bowel though normal transit time without evidence of obstruction

CASE REPORT

Postoperative Course (continued)

- Genetic workup
  - TTC7A gene with two alterations
  - Unknown significance though predicted to be pathologic
- Octreotide resulted in initial improvement in stool output but then returned to previous high levels
- Patient required transfer to quaternary care center for management of her desquamative enteropathy

CONCLUSION

- This report demonstrates the association between CPA and desquamative enteropathy without findings of EB
  - EB is frequently associated with CPA
  - Desquamative enteropathy with CPA without findings of EB is seldomly documented in the literature
- This case emphasizes the importance of considering EPA as a differential diagnosis for neonates presenting with nonbilious emesis
- Additionally, the case contributes to the validation of the Heineke-Mikulicz pyloroplasty as an appropriate selection for management of these patients[1-5]

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