Harvard Medical School Exchange Clerkship Program

Case Report

Location: Boston Children's Hospital

Department: Pediatric Surgery

Student: Angel Pai (KMU BM 101)

Duration: Sep.28 ~ Oct 24, 2015

Brief Summary

Baby boy SM is an ex 39wks and 3 days LGA infant born on Sep.29, 2015, to a G5P(now)3 mother by vaginal delivery at BI. He had a prenatal diagnosis of Tetralogy of Fallot with pulmonary stenosis via ultrasound in June. His prenatal course was otherwise noted for self-resolving pericardial effusion, mild bilateral inguinal hernia, and left-sided ventriculomegaly. He has no known significant family history. His father (41y), mother (39y), and two brothers (age 5 and 3) have been healthy with no recorded operations.

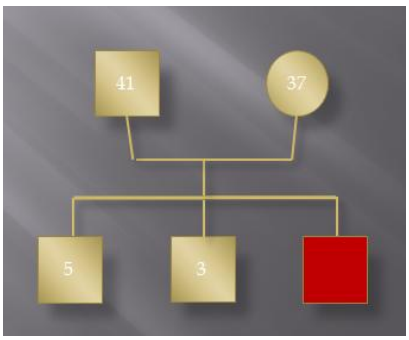
Upon delivery, he was active but ashen in appearance. His Apgar score at one minute was 7 and 8 at five minutes. He then demonstrated cyanosis and tachycardia (around 200 bpm). Since the O2 saturation on his right arm was 70%, they gave him blow by oxygen at FiO2 0.4. After demonstrating mild improvement, he was transitioned to CPAP FiO2 0.35, which significantly helped with the oxygenation. He was then transferred to NICU. There was no birth complication otherwise.

Due to his cardiac problems, the CV team was called for consult. In accordance with his condition, II-III/VI, low-pitched systolic murmur lasting throughout the majority of systole could be heard. An echocardiogram was also performed bedside, which revealed mild RVOTO, VSD with bidirectional flow, and no PDA. Overall, he appeared well and breathing comfortably with good oxygen saturation; there was no central, perioral, or peripheral cyanosis. If he continued to remain stable, surgical repair or patch closure and enlarging the RVOT will be scheduled when he reaches 3~6 months old for better heart growth. In the meantime, he would be monitored for signs of worsening pulmonary stenosis.

Since his prenatal US also showed asymmetrical cerebral ventricle size, with left-sided ventriculomegaly, a post-natal head US was also obtained. The ultrasound reported normal findings. Therefore, based on the infant's history and exam, the Neurology team did not have high concerns for any particular syndrome at that time. Genetic testing could be considered if needed in the future.

During the first night in NICU, he had an episode of bilious emesis. In addition, minimal meconium was passed, and his abdomen became increasingly distended. Therefore, we were consulted for further evaluation.

Patient Profile: weight 4060g (90%), HC 36cm (80%), L 52.5cm (80%)



Initial Assessment

Upon initial assessment, he was already weaned to room air and did not have any fever. His physical examination appeared normal except for soft but distended abdomen and explosive stool release with rectal exam.

PE: body temp 97.2, HR 130, BP 92/72, saturation 92%

Awake and alert

CV→ low pitch systolic murmur

Resp→ bilaterally clear

His laboratory data was unrevealing, with WBC of 9.8, Hct 41.7, BUN/Cre 9/.06, and Tbili 0.3. Since he did not show signs of infection or instability, he was sent to BCH for imaging studies.

Imaging Studies

UGI+ Contrast enema:

- Preliminary film of the abdomen shows moderate diffuse distension of bowel
- Contrast enema showed rectum and sigmoid are quite narrow. There is a transition zone in the sigmoid colon, which goes into a moderately dilated descending colon. The remainder of the visualized colon is also moderately dilated
- UGI via NG tube showed normal positioned duodenal jejuna junction

Impression: Findings are consistent with Hirschsprung's disease

Admission Plan

Besides TOF with pulmonary stenosis, this patient was possibly demonstrating Hirschsprung's disease based on physical and radiologic findings. On Sep.30, 2015, the baby, at DOL 1, was transferred to our care.

He would first need to be kept NPO while NG tube would be used for gastric decompression. Feeds could be restarted after the distension improved. Antibiotics did not seem necessary because he was afebrile with normal WBC level; we needed to be cautions of signs of enterocolitis. In the meantime, he would receive rectal

irrigation q6h with 10~20cc/kg normal saline. In addition to continuing follow-up with CV team, GI would be consulted for suction rectal mucosa biopsy.

Clinical Course

While he waited for his biopsy, we continued to support him with fluid and electrolyte and rectal irrigation. His vital signs remained stable, with no desaturations. With the aid of the irrigations, we noted bowel movements and scant mucous output from the NG tube.

On Oct. 1, 2015, he received a bedside suction biopsy, with the following result:
Two specimens (rectum at 3 cm and rectum at 2cm); both demonstrated features of aganglionosis.

- No ganglion cells seen in the superficial submucosal (Meissner's) plexus
- Hypertrophic submucosal nerves
- Absence of calretinin positive neuritis in the muscularis mucosae and lamina propria
- Focal mild acute colitis

The procedure was well tolerated, with no evidence of discomfort or bleeding.

After the diagnosis of Hirschsprung's was confirmed with the biopsy, SM was scheduled for surgery on Oct. 7, 2015. During the days leading up to the surgery, he was able to start on feeds (15cc, q3h). The rectal irrigations had been reduced to BID, and he was stooling normally. On his fifth day of life, his skin became a bit yellowish, with elevating bilirubin levels. However, after rechecking on the seventh day, the levels were already trending down (peaked at 8.3/0.2→ 7.3/0.3). He also received sodium replenishment once after a blood test revealed hyponatremia; it was efficiently corrected to normal level.

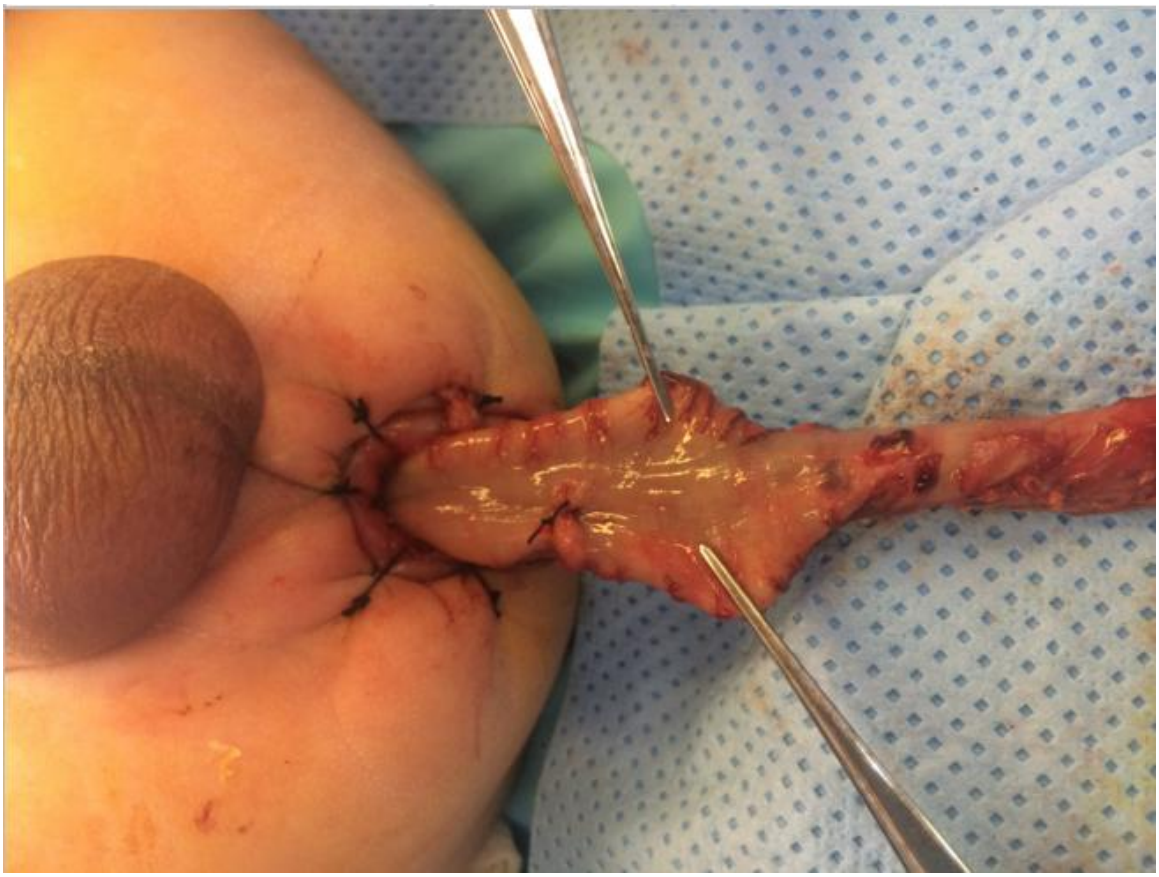
The cardiac team was also consulted about the risk of surgery in patients with this type of cardiac issues. He was currently hemodynamically stable with good respiratory status on room air and had normal starting hemoglobin. Because SM was showing good saturation and clinical stability, the CV team supported the decision of transferring him to our surgical floor for further management of the bowel issues.

On Oct 7, 2015, SM was taken to the OR and received laparoscopic-assisted Soave endorectal pull-through procedure with circumcision. He was then 9 days old and clinically stable. During the operation, we first performed an abdominal exploration. Given his cardiac condition, the abdomen was only insufflated to a pressure of 8. Finding no other abnormalities, we then identified the rectosigmoid transition zone; partial thickness biopsies on the antimesenteric border were then taken proximally and distally to the transition. While we sent the biopsies for frozen and waited for the

readings, we used Gomco clamp and chromic sutures to complete his circumcision (requested by the patient's father).

We then went to the pathology frozen section room to identify the specimen with the pathology colleagues. As we had expected, based on the anatomic transition point observed laparoscopically, there were ganglion cells in the proximal specimen while none were found in the distal specimen. With this confirmation, we went back to the OR and completed the rest of the bowel dissection. The abdomen was deflated, and the attention was shifted to the transanal portion of the procedure.

With careful maneuvers, the surgeons developed a muscular layer approximately 2 cm to the dentate line and then transitioned to a full thickness dissection. Full thickness bowel was delivered while they divided any branches of the mesentery that were not divided laparoscopically. After identifying the proximal extent of our marking, the colon was divded approximately 3cm proximal to the biopsy site. Finally, coloanal anastomosis was performed and we could calibrate it to a 14 Hegar. Before closing all the surgical wounds, the abdomen was rechecked laparoscopically to make sure there were no twisting of bowels and bleedings at any sites. The surgery was uncomplicated and the patient was started on pain control with fentanyl drip and prophylactic antibiotic (cefoxitin x48h).



He was successfully extubated the following day and received adequate pain relief with around the clock IV Tylenol and low-dose fentanyl drip. While he was in ICU, he also received regular blood draws for monitor and fluids at 120ml/kg/day. Bacitracin was applied to his circumcision site while his urine (from foley) and stool were also carefully monitored. He was clinically stable, with 100% saturation in room air. On his post-operative day 2, he was able to start on feed after the passage of stool. Base on the nutrition assessment, we started feeds of breast milk 20kcal/oz, with a goal of 120kcal/kg, while weaning IVF.

On Oct.10, 2015, due to stabilizing condition, SM was transferred from ICU to the floor. For the rest of his stay, he did not have many complications except the development of a diaper dermatitis, which was treated with desitin and Aveeno soaks. He did require multiple feeding consultations due to the difficulty of bringing up his feeds to goal volume. His irritability during feeding caused the volume of his feed to be limited and his weight decreasing.

Over the course of next couple of weeks, we concentrated on bringing up his feeds to goal. On Oct. 21, 2015, he finally received the green light to be discharged for home care. He was then on intermittent feeds of Enfamil Infants 26 kcals at 75cc/hr, q3h. Aggressive peri-care for his diaper rash needed to be continued to prevent his skin from breaking down. Dilation was performed to 10 Hegar to ensure adequate passage for stool. Regular follow-ups both in our department and Cardiology are indicated.

Genetics Consultation

Purpose: for a unifying diagnosis of his diseases
Material: chromosomal microarray
Result: negative
Discussion:

- Tetralogy of Fallot and Hirschsprung disease are both major anomalies which increase the chance of a syndrome, but they are not often seen together.
- Since there is a possibility that other symptoms of a syndrome might present later, follow-up is recommended.

Other points of discussion

1. **What are the differential diagnoses for the patient's initial presentation?**

   VOMITING
   More likely causes (pertinent to our patient)

- Gastrointestinal obstruction: pyloric stenosis, **malrotation**, intussusceptions,

intestinal duplication/stenosis/atresia, **Hirschsprung disease**, incarnated hernia
- Other GI causes: physiological gastroesophageal reflux or GERD, food allergy/intolerance, gastroenteritis, peptic ulcer disease, eosinophilic esphagitis, gastroparesis, pancreatitis

Less likely causes:
- Cardiac: heart failure
- Infectious: sepsis, UTI, pneumonia, otitis media, hepatitis, meningitis
- Others: metabolic/endocrine, neurologic, renal, toxic

DELAYED PASAGE OF MECONIUM
- Hirschsprung, intestinal obstruction, meconium ileus, meconium plug syndrome, functional ileus, small left colon, drug administered to mother before delivery, hypothyroidism
- Other causes of constipation: physiologic, neurogenic, endocrine and metabolic

SYMPTOMS OF GASTROINTESTINAL OBSTRUCTION/DISEASE
- Bilious vomiting
- Projectile vomiting in an infant three to six weeks of age
- Hematemesis
- Hematochezia (rectal bleeding)
- Marked abdominal distension and tenderness

In light of clinical status and physical exam, we were focusing on gastrointestinal causes. Especially in neonates, bilious vomiting often suggests intestinal obstruction or volvulus. The two main problems on the top of our list were malrotation and Hirschsprung disease.

2. What should be ordered for the initial assessment? Why?

It was important for us to be able to quickly make an initial diagnosis for one of our top two suspicions was more time-sensitive. Laboratory tests were routinely on the top of the list. This patient had all values within normal range, so we could first rule out infectious causes or necrotizing enterocolitis. For neonates up to one week old with the likelihood of gastrointestinal problems, we could order three main types of imaging—xray abdomen, upper GI series, and contrast enema. A babyogram could serve as an initial workup that would help determine further strategy. However, whether this radiograph was absolutely necessary was controversial. For this patient, the upper GI series and contrast enema were performed at the same time. We needed to be more cautious with injecting contrast if the patient was demonstrating any signs of contraindications. Moreover, since dye retention is one of the clues for Hirschsprung disease, performing UGI series first was probably better to rule out malforation.

Contrast enema served as a good initial diagnostic procedure for it allowed us to see a transition zone and measure the rectosigmoid index (normally >1). For our patient, a clear transition zone could be seen at the sigmoid colon. Therefore, along with his

physical examination, we were pretty certain of him having Hirschsprung disease, so a confirmatory rectal suction biopsy was ordered.

Of note, anorectal manometry could also be performed; however, it is not a test routinely done in this facility

3. When is the optimal time for surgery for this particular patient?

We needed to take into account that this patient also has the underlying disease of Tetralogy of Fallot; therefore, when planning for surgery, the cardiology and anesthesiology teams were consulted.

The surgical correction can be either one- or two- stage procedure. For this patient, we could also think about performing a two- stage procedure, with the second stage in conjunction with the time he receives his cardiac repair. After evaluation, however, a one-stage approach is more beneficial to him. His cardiac problem is not as urgent for he has a pink Tet instead of the more severe cases, and he did not have a long-segment or complicated Hirschsprung. Therefore, an early, one-stage correction will place him on a healthier and normal road to development.

4. Which type of procedure is most suitable for this patient?

There are three commonly used techniques—Swenson, Duhamel, and Soave procedures. There are no strong evidences to which one is the best option, so the choice lies with the surgeon; he/she will often opt for the method he/she is trained and performs most frequently. For our case, the Soave procedure was used. The attending believed that this method would also provide him with lower risks of injury to other pelvic structures and less need for post-operative dilations.

Minimal access approaches are the current the mainstream; these procedures can be done transanally or with laparoscope assistance. Laparoscopic pull-through is associated with shorter hospital time and fewer complications. Moreover, we would be able to mark the transition zone and obtain intra-operative specimens for pathology confirmation. However, we needed to take into consideration whether our patient could tolerate abdominal insufflations. The increased pressure could affect his cardiovascular or pulmonary functions. Therefore, a lower intra-abdominal pressure was set for his laparoscope procedure.

5. How should we care for the patient while waiting for the operation?

Initially, the patient was kept NPO and NGT was used for gastric decompression. Fluid supply and other supportive care were given. As the clinical condition stabilized and abdominal distension subsided, feeding was initiated. During the days leading up to the surgery, rectal irrigation was diligently performed. The patient was able to stool normally and could sometime pass stool without irrigation. The frequency of irrigation was determined more subjectively; care providers could decide on the number of times based on the patient's clinical response.

Other preparations included consultations with the Cardiology and Anesthesiology Departments. Most importantly, we needed to be alert for any signs of infection or complications.

Other findings not discussed here

- TOF
- Physiologic jaundice
- Transient anemia
- Diaper dermatitis
- Poor feeding

References

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2. UptoDate:
   - Approach to the infant or child with nausea and vomiting
   - Constipation in infants and children: Evaluation
   - Functional constipation in infants and children: clinical features and differential diagnosis
   - Congenital aganglionic megacolon (Hirschsprung disease)
3. Prem Puri. *Hirschsprung's Disease* (Chapter 26). Pediatric Surgery: Atlas Series. Springer 2006

This is a preliminary report on a patient encountered during this rotation. More detailed description and discussion are beyond the scope of this report.