PedsCases Podcast Scripts

This is a text version of a podcast from Pedscases.com on "Approach to Hirschsprung Disease." These podcasts are designed to give medical students an overview of key topics in pediatrics. The audio versions are accessible on iTunes or at www.pedcases.com/podcasts.

Approach to Hirschsprung Disease

This podcast was developed by Kieran Purich and Dr. Ioana Bratu for PedsCases.com. October 18, 2016.

Introduction

Hi, my name is Kieran Purich, 3rd year medical student at the University of Alberta. This podcast was developed in conjunction with Dr. Ioana Bratu. Dr. Bratu is a pediatric surgeon and Associate Professor at the University of Alberta, and practices out of the Stollery Children's Hospital in Edmonton. Today we will be discussing an approach to Hirschsprung disease.

Case Presentation

You are a first year family medicine resident in an urban setting. A concerned G1P1 mother comes in with her full-term one-month-old baby boy who has had difficulty gaining weight. She describes feeding problems as the child often spews green vomit when she tries to breastfeed. The mother also mentions that the child has irregular, yet explosive bowel movements (<1 every 24 hours) and that often her son's "belly feels full." On further questioning, mom reveals that baby passed meconium late on day 3 of life.

Objectives:

1) Recognize the presenting signs and symptoms of Hirschsprung disease
2) Develop an organized approach to a child with suspected Hirschsprung disease including differential diagnosis, history, physical exam and investigations
3) Review the treatment and prognosis of Hirschsprung disease

What is Hirschsprung disease?

Hirschsprung disease is a congenital defect, affecting approximately 1 child in every 5000 births. This condition is attributed to the absence of nerve cells in the intestine, specifically the absence of ganglion cells in all or part of the myenteric and submucosal plexuses of the distal colon. This is thought to be due to the failure of neural crest cell migration during development. The result is abnormal peristalsis in the aganglionic area, leading to severe constipation and megacolon. Approximately 85% of the time this aganglionic segment is limited to the rectosigmoid colon, yet there are variants where the aganglionic segment extends further upwards along the GI tract, harboring worse prognoses. Failure to identify Hirschsprung disease can lead to malnutrition or bacterial overgrowth and inflammation in the gut known as enterocolitis, which can lead to sepsis and may become quickly fatal, making it critical to not overlook.

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Risk Factors

Risk factors for Hirschsprung disease include male sex (4:1 M: F), the presence of Down syndrome (0.8% risk) & family history of Hirschsprung disease (4% risk if affected sibling). Inheritance is complex, as it appears to be a multigenic disorder. Children with Hirschsprung disease also have a 30% risk of other serious congenital abnormalities including: gastrointestinal abnormalities, congenital heart disease, limb abnormalities, cleft lip/palate, hearing loss, urogenital disease and ophthalmologic abnormalities.

Clinical Presentation

More often than not Hirschsprung disease is detected shortly after birth. As seen in the case presented earlier, classic symptoms include: failure to pass meconium within 24 hours of birth, failure to thrive, constipation, abdominal distention and signs of bowel obstruction with bilious vomiting. If enterocolitis is present the child also may have explosive diarrhea, fever, lethargy, and dehydration.

It is important to remember that Hirschsprung disease occasionally presents as severe chronic constipation with failure to thrive in an older child.

Differential Diagnosis

Hirschsprung disease is imperative to consider when a neonate presents with intestinal obstruction, chronic constipation or signs of enterocolitis. The gold standard for diagnosis is confirmation by rectal suction biopsy – a relatively invasive procedure. To ensure this procedure is not done in vain physicians must distinguish Hirschsprung disease from other causes of constipation. Important GI causes of constipation include meconium ileus, intussusception, Meckel's diverticulum, intestinal atresia, malrotation, necrotizing enterocolitis, anorectal malformations, functional constipation, lactose intolerance, pyloric stenosis and celiac disease. Important non-GI causes of constipation include inadequate feeding, lead poisoning, sepsis, neuromuscular disorders and metabolic disorders. The differential diagnosis is largely driven by how unwell the child is, along with other features on history and physical exam.

History

In this podcast we will focus primarily on the presentation of Hirschsprung disease, but a thorough pediatric history and physical should be completed in all newborns.

Specific symptoms to ask about on history are feeding problems, and any associated failure to thrive. Ask about abdominal distention, any vomiting but in particular bilious vomiting, diarrhea and any explosive infrequent bowel movements requiring laxatives/enemas. If concerned about enterocolitis you should also ask about fever, diarrhea, vomiting, progressive abdominal distention, lethargy and bloody stools. It is important to distinguish how many hours after birth meconium was passed; as if it is over 24 hours then it is worth considering a surgical consultation.

Physical Exam

As with all patients the physical exam should begin with inspection to see whether or not the child is acutely ill. Obtain vital signs to ensure hemodynamic stability and to look for signs of

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sepsis. A child with enterocolitis from Hirschsprung disease will appear ill, lethargic, malnourished and have a distended, tender abdomen. Interestingly, children with enterocolitis may have diarrhea, which is opposite of most children with Hirschsprung disease. Look for abdominal distention and discomfort. A digital rectal exam is important in a child with suspected Hirschsprung disease as they may have a tight anal sphincter and there may be an explosion of watery stool and gas on the removal of the finger. But, be sensitive and mindful to the appropriate setting. In all children, complete a thorough physical exam including looking for other associated congenital abnormalities. Long-term, children should have their development monitored.

Investigations

If you suspect Hirschsprung disease then a referral to pediatric surgery is necessary. An urgent referral will expedite access to the appropriate diagnostic tests. A standard workup, which should be completed at the time of referral, includes a complete blood count, thyroid hormone and electrolytes to rule out constipation from hypercalcemia, hypokalemia and diabetes insipidus. The primary care provider should order 3 views abdominal x-rays, which if the patient has Hirschsprung disease, will show dilated loops of bowel with the absence of stool and gas in the rectum. The presence of an obstruction or perforation should warrant immediate referral to the emergency department.

The preferred initial test is a water-soluble contrast enema, ideally preformed prior to rectal irrigations. The contrast enema will classically show a funnel shaped “transition zone” indicating the approximate plane at which the normal bowel becomes aganglionic. This abnormal recto-sigmoid ratio is pathognomonic of Hirschsprung disease. Normally, the rectum should have a larger diameter with capacity to hold stool when compared to a smaller diameter normal sigmoid. In Hirschsprung’ the rectum is “tonically” contracted and thus the rectal diameter is “narrow” while the proximal bowel, usually the sigmoid, diameter is very dilated because it cannot overcome the pressure of the contracted rectum. Be aware that contrast enemas are contraindicated if you suspect enterocolitis due to a risk of perforation, so only order this investigation if the patient is clinically well. In older patients over the age of 12 months, anal manometry can be useful to look for the presence of a rectoanal inhibitory reflex.

Diagnosis of Hirschsprung disease is confirmed by rectal suction biopsy, which takes tissue from above the dentate line. This procedure can be performed at the bedside, and inspects for aganglionosis and hypertrophic nerves. In non-neonates a more invasive full-thickness open rectal biopsy may be necessary.

Treatment

While the diagnosis is being confirmed, children that present unwell with suspected enterocolitis will need rehydration, broad spectrum antibiotics, nasogastric decompression and rectal irrigations which usually means urgent hospital admission. If the child does not respond to empiric therapy, and remains unwell, the child may require a laparotomy and placement of a decompressive stoma to relieve the obstruction.

Treatment for Hirschsprung disease is surgical excision of the affected bowel segment. Surgery should be completed shortly after presentation in infants, with the time frame depending on the child’s overall health. The surgery is known as a “pull through” procedure in which the diseased aganglionic portion of colon is resected and an anastomosis is made

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between the healthy colon and the rectal cuff proximal to the dentate line to preserve normal sphincter function. There are three accepted “pull through” methods known as the Duhamel, Swenson and Soave techniques. Techniques can be completed by open abdominal incision, laparoscopic assisted or transanally which does not require an abdominal incision. The technique used will differ from surgeon to surgeon, as there is no strong evidence suggesting that one technique is superior to another. In all cases, the child must be monitored for postoperative enterocolitis and constipation. Patient’s courses will vary depending on the severity and extent of aganglionic bowel, and some may require a temporary or permanent ostomy bag.

Post-operatively, up to 60% of children have diarrhea and fecal soiling, however this should normalize with time. Perianal excoriation is also common, which can be treated with barrier creams including zinc oxide. The child should then be followed regularly for a minimum of 5 years by a pediatric general surgeon.

Prognosis

The overall reported mortality from Hirschsprung disease is less than 1%, and is usually related to enterocolitis and delayed diagnosis. Fortunately, the long-term outcomes for most patients are good, and the majority report good or fair quality of life (5). However, patients will not be morbidity free. Current evidence suggests that only about 50% of children will develop optimum bowel function. Long-term complications range from constipation to anastomotic strictures, fecal soiling, achalasia of the anal sphincter or residual aganglionosis, which may require special high fiber diets or further treatment for bacterial overgrowth with probiotics and antibiotic cycling. In addition, patients can be plagued with fecal incontinence, obstructive symptoms including abdominal distention, constipation and bloating or recurrent enterocolitis. Long-term urinary and sexual dysfunction may also occur due to dissection near the pelvic plexi at the time of surgery. All of these complications must be identified and therapeutic interventions can occur.

Case Resolution

Now that we have discussed the details of Hirschsprung disease, lets return to the case presented at the beginning of the podcast.

Following a thorough physical exam, including a DRE, the baby is referred to a pediatric surgeon. The pediatric surgeon completes a rectal suction biopsy, and the patient is scheduled for a “pull through” surgery. The surgery harbors no complications, and the patient is discharged a few days afterwards.

The patient follows up with their pediatric surgeon for the next 6 years, and suffers from occasional constipation, which is treated by his diet, as well as laxatives when necessary."

Conclusion

This brings us to the end of the podcast. Remember, if you suspect a child has Hirschsprung disease ensure to:

- Rule out enterocolitis and sepsis
- Ask how many hours after birth the child passed meconium

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- Complete a through physical exam including a digital rectal exam or DRE. The rectal exam is perhaps diagnostic but more importantly therapeutic as it will help evacuate an explosion of built up pressure from bacteria in the colon overgrowing. But, check with your supervisor before doing a DRE on a pediatric patient.
- Refer to a pediatric surgeon immediately for assessment.
- Be aware that diagnosis is made by rectal biopsy and treatment is surgical
- Remember, this disease **can be fatal,** making it critical to not overlook.

Thanks for listening.

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