Anorectal Malformation (Imperforate Anus)

Patient and family information, brought to you by the Education Committee of APSA

Overview - "What is it?"

This condition occurs when the rectum of the baby does not come all the way through the tissue of the bottom, leaving no opening for the stool to be passed from the body. Depending on the severity of the condition, it is often classified as a low, intermediate, or a high anorectal malformation. It is also known as "imperforate anus".

Anorectal malformations (ARM) occur in 1 baby in every 5,000 live births. It is equally seen in boys and girls.

Figure 1: Picture of a baby's bottom without an anal opening. Picture courtesy of MJArca 11/2016.

Signs and Symptoms - "What symptoms will my child have?"

Early signs: After the baby is born, an ARM is diagnosed as no opening for stool to be evacuated from the body or there is an abnormal position of the anus. Sometimes, ARM is noted when a baby does not stool within 24 hours or if a rectal thermometer cannot be

inserted. If the anus is abnormally positioned, stool may come out of a different location—seen coming out of the vaginal area or mixed in the urine.

Later signs/symptoms: Most of the signs/symptoms occur after an attempt at surgical repair of the condition. One of the most common symptoms is constipation.

Diagnosis - "What tests are done to find out what my child has?"

Labs and tests: Babies with ARM have higher risk of certain types of other abnormalities. It is important to find out if the baby has these abnormalities so nothing is missed that can impact the baby's health moving forward. The most common abnormalities are grouped into the "**VACTERL**" association (named after the first letter of the most common anomalies).

- **V** (spine): Spine X-ray to look for abnormalities of the bones and ribs. Spine ultrasound or MRI may also be obtained.
- **A**nus: Baby has high imperforate anus/cloaca
- **C** (heart): An ultrasound of the heart (echocardiogram) is needed to check for problems such as abnormal holes, problems with valves, etc.
- **T**racheo-**E**sophageal Fistula (TEF): Abnormal connection between airway (trachea) and esophagus (tube that connects mouth to stomach) and a blind-ending esophagus in neck. This may be seen on X-ray, or earlier if the baby has problems breathing or swallowing.
- **R**enal (kidney): Ultrasound of kidneys to look for abnormalities
- **L**imbs (arms and legs): Examine arms and legs for deformity. Arms are the most common place for abnormal bones.

An abdominal film/X-ray, abdominal ultrasound, and pelvic magnetic resonance (MRI) scan are often done to understand the extent of the imperforate anus.

Conditions that mimic this condition: Babies can have variations in their rectal, genital, and urinary anatomy. In girls, the rectum may join with the vagina and urethra to form what is known as "cloaca."

Treatment - "What will be done to make my child better?"

Medicine: Medications are not helpful before surgical repair. Since air and stool in the intestine are not being emptied properly because of the anal problem, the baby's belly can get swollen. The baby is not fed and occasionally, a tube is passed from the mouth into the stomach to remove air and fluid to keep the abdomen from becoming distended before surgery. Some babies are kept on antibiotics by mouth until the actual anus is repaired to prevent urinary tract infections.

Surgery: The first decision by your surgeon will be whether to perform a colostomy on your baby (bringing the intestine to the abdominal wall to empty into a bag). This will depend on many factors including the size of the baby and other conditions that they may have. The designation of "low" "intermediate" or "high" ARM is given depending on the location of the abnormal opening in relation to the muscle that controls stooling or the sphincter muscle.

- Babies who have a low ARM and who are otherwise healthy may undergo repair of the anus soon after birth. In this repair, the end of the anus is matched with the center location of the sphincter muscle. In these patients, a colostomy is not usually needed.
- In babies with an intermediate or high imperforate ARM, or if the baby has conditions that would make it unsafe to undergo an anus repair (such as complex heart condition, very small in size, etc), a colostomy may be performed first. This will allow for growth of the baby, which helps increase the success of the anus repair procedure. The baby's stool comes out of this colostomy (Figure 2). The colostomy also allows the surgeon to perform special x-ray tests to get accurate information on the type of abnormality. This information will help the surgeon plan the repair of the anus.

Figure 2: Photo of colostomy and mucus fistula (encircled in purple ink).
Photo courtesy of MJArca 11/2016.

Preoperative preparation: Your baby will be kept from eating until the surgery is performed. They will be given nutrition through an IV (intravenous catheter) until then.

Informed consent: A consent form is a legal document that states the tests, treatments or procedures that your child may need and the doctor or practitioner that will perform them. Before surgery, your doctor should tell you what the operation is, the goal of the surgery and other possible treatment options that are available. Your doctor should explain the risks and

benefits of the surgery. You give your permission when you sign the consent form. You can have someone sign this form for you if you are not able to sign it. You have the right to understand your child's medical care in words you know. Before you sign the consent form, make sure all of your questions are answered. It is important to know that during surgery, there are things that can happen that your doctor may have not predicted before going in. He or she will explain these to you after the surgery.

Postoperative care: Many times, babies will require a period of anal dilation to decrease the risk of developing a stricture or narrowing where the anal opening is relocated. This can be done at home for a number of months until healing has occurred, and the risk of post-operative stricture has decreased.

Risks/Benefits:

- **Risks:** Infection or breakdown of the wound, bleeding, damage to surrounding structures may occur during any surgery.
- **Benefits:** The infant requires a way to evacuate stool effectively, whether relocation of the anal canal in ARM or a colostomy.

Home Care - "What do I need to do once my child goes home?"

Diet: Normal

Activity: Normal

Wound care: Cleaning the surgical incision after going home is important, as long periods of stool on the skin and incision can lead to wound breakdown in the immediate post-operative period. Use of barrier creams such as zinc oxide are helpful to prevent this skin breakdown.

Medicines: Laxatives are not typically used immediately after colostomy or definitive repair. Your surgeon will decide when and if your child needs any medicines to have a bowel movement, depending on their long-term bowel habits.

What to call the doctor for: Any fevers or redness around the incision. Also call your doctor if your child goes more than two days without having a bowel movement, is vomiting, or has abdominal pain.

Follow-up care: You should plan to see your surgeon within two weeks of having the surgery performed and will likely have several visits a year for the first few years of life to make sure your child is able to pass stool without difficulties.

Long Term Outcomes - “Are there future conditions to worry about?”

Patients with low ARM have very good results in terms of continence (able to control bowel movements), but they also have a very high risk of constipation. Children with intermediate and high ARM have lower success rates, as the severity of the anatomic and neurologic abnormalities are increased in these patients. These children may have more problems with soiling of stool and poor sphincter control. It may take several years to determine the final outcome of the surgical procedure.

Specialized diets and medications may be needed to help your child have regular bowel movements and to reduce the chance of incontinence or soiling. In intermediate to high ARM, further studies and operations may be required in the future to create the best situation in terms of stool control.

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