Cloacal Exstrophy

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

Overview - "What is it?"

Cloacal exstrophy is an abnormality where the muscles, skin and bones of the lower belly do not form at all. The bones of the pelvis do not meet in the middle as they do normally. The bladder and part of the intestines are exposed to the outside. The exposed bladder is split in two halves and the intestine is in the middle of the two halves of the bladder (Figure 1). In boys, the penis is deformed—small, split in half or not present. In girls, the clitoris is usually split in two, and the vagina is also usually split in two small channels instead of a single vagina or may not be present at all.

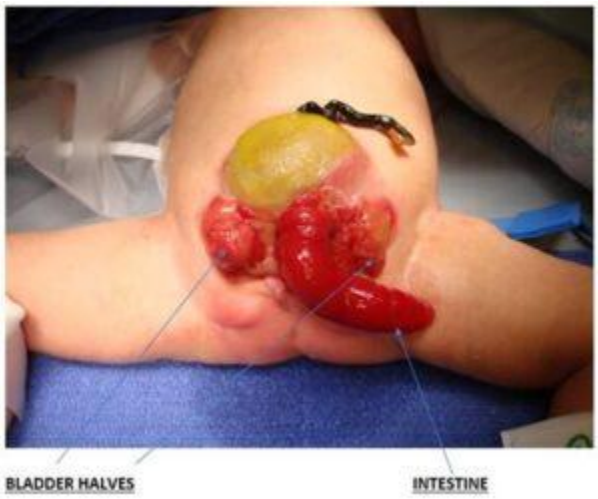

Figure 1: Cloacal extrophy
Photo courtesy of MJ Arca 11/2016

Cloacal exstrophy occurs in 1:200,000-400,000 among live births, boys are more commonly affected than girls, and almost 50% are premature. Other problems associated with this anomaly include problems with the intestine (shorter than normal, duplicated segments, no anal opening), spine, kidney, and genitals. Sometimes, there is a larger defect in the mid-abdomen with a thin covering through which one can see the inside organs. This covered defect is called omphalocele.

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

Often, cloacal exstrophy is diagnosed on ultrasound while the baby is still developing inside the mother. If not, the problem is readily apparent at birth.

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

- A **physical examination** by an experienced health care provider is essential.
- Cloacal exstrophy is considered part of **OEIS** association, named after the four commonly seen abnormalities: **O**mphalocele, **E**xstrophy, **I**mperforate Anus (no anal opening), and **S**pina Bifida
  - Omphalocele (opening of muscle and skin in center of abdomen, covered by a sac)
  - Exstrophy (see above discussion)
  - Imperforate anus (no normal anus present)
  - Spina Bifida (defect in the skin and spine overlying the main nerves of the middle of the back)
- **Labs and tests** include an Echocardiogram (heart ultrasound) to look for cardiac abnormalities, an ultrasound of kidneys, and blood tests to determine genetic makeup of the child, check hormone levels of the blood, as well as mineral levels.

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

Cloacal exstrophy requires several operations over months and years. The timing, type of surgery, and results will depend on your child's situation. Given possible severe deformities of the genitalia (vagina or penis), even gender assignment needs to be discussed thoughtfully and carefully.

Medicine: No medicine can fix cloacal exstrophy, only surgery

Special Consideration in cloacal exstrophy is gender assignment. A chromosome analysis determines the genetic makeup of the baby (genetically male or female). Decision-making should be in a multi-disciplinary fashion with the parents, an endocrinologist, a psychiatrist, a urologist, and your surgeon.

Surgery is performed in several stages:

First surgery (as newborn): Several surgeons are involved with the baby's care including general surgeons (intestine and genitals), orthopedic surgeons (bone), urology surgeons (kidney and bladder, genitals), neurosurgeons (brain and spine), and cardiac surgeons (heart).

Preoperative preparation includes a complete examination and studies to look for all possible abnormalities
Goals of the first surgery include separating the intestinal system from the urinary system as well as closing the belly. The omphalocele (abdominal wall defect) is closed and the bladder is separated from the intestine. The two halves of the bladder are sewed together and placed inside the belly. The intestine is brought out through the muscle and skin of the belly as a colostomy so the baby can eliminate stool. A bag will be placed over the colostomy to collect stool. Other procedures may be done at the same time. If there is an opening in the spine (spina bifida), closure of the defect may need to be done at the same time.

Postoperative care includes advancing to a regular diet once the colostomy starts to have stool come out. Parents are taught how to take care of stomas and may need dressing changes.

Risks of the first surgery include infection, bleeding, and risks of anesthesia and breathing tube.

Benefits of the first surgery include closure of the abdomen, placement of the organs inside the belly, and separation of the pathways for urine and stool.

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.

Second Surgery. Pelvis procedure. The goal of this second surgery is to close the ring of the pelvis. The pelvis is an open ring in these babies and the hip bones will need to be cut and adjusted. The surgery is called "osteotomies" and this operation is performed by an Orthopedic Surgeon. The baby will need to be casted for weeks after surgery. This operation is usually done several weeks or even months after the initial operation. The bladder may need to be repaired during this time as well. It is important to do this operation so that the pelvis can support the baby as the baby grows.

Third Surgery. Pull through procedure. The goal of this third surgery is to establish stooling out of the bottom. This is another separate procedure that may be necessary as your child gets

bigger. In this procedure, the colon is brought out into the area of the buttocks so eventually, stool can come out of the region of the buttocks. Sometimes, this is not possible if the muscles of the rectum or the nerves controlling elimination of stool are damaged. The situation is different for each child. If stooling through the buttock area is a possibility, your child's surgeon will discuss it with you.

Additional Surgery. The child may need multiple reconstructive surgeries for bladder and genital system (penis, vagina/uterus)

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

Diet: Usually normal for age. In cases where there is a shorter length of intestine, a special formula may be necessary for digestion.

Activity: Ask doctor before allowing tummy time, but otherwise, activity is normal for age.

Wound care: Surgical incisions should be kept clean and dry for a few days after surgery. Most of the time, the stitches used in children are absorbable and do not require removal. Your surgeon will give you specific guidance regarding wound care, including when your child can shower or bathe.

Medicines: Medicines for pain such as acetaminophen (Tylenol) or ibuprofen (Motrin or Advil) or something stronger like a narcotic may be needed to help with pain for a few days after surgery. Stool softeners and laxatives are needed to help regular stooling after surgery.

What to call the doctor for: Call your doctor for worsening belly pain, fever, vomiting, problems with urination, decreased stool output or if the wounds are red or draining fluid.

Follow-up care: Your child should follow-up with his or her surgeon 2-3 weeks after surgery to ensure proper post-operative healing. IT IS IMPORTANT TO KEEP ALL APPOINTMENTS WITH THE SURGEON, AS HEALING IS CRITICAL IN THE FIRST FEW MONTHS AFTER SURGERY.

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

The survival rate for patients with cloacal exstrophy approaches 100% but the quality-of-life outcomes are poor. These babies often have problems with potty training both urine and stool. Kids may not be able to feel when they have to pee or poop. For problems with controlling stool, they may need to be on medicines for bowel management or require other surgeries to try to keep the child clean. Ideally, these children need several specialists as the baby grows up to achieve the best outcome possible. There also may be a need for multiple surgeries for genital reconstruction.

Gender assignment in these children is a complicated issue. Many factors go into the decision of gender for the baby. This will be discussed and resolved carefully with the involvement of all the specialists to have the best possible long-term result for the baby.

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