

GLOSSARY of some NEWBORN CONDITIONS for which the NICU is highly likely to be NECESSARY

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Down Syndrome (Trisomy 21)

Definition:

Babies born with Down syndrome have an extra copy of chromosome number 21. There are three ways this can happen:

- Babies can have 3 copies of chromosome 21 in each of their cells. They have a total of 47 chromosomes in each cell instead of the normal 46. This is called a trisomy, the most common chromosome makeup in Down syndrome
- Uncommonly, babies can have 46 chromosomes in some of their cells, but have 47 in others. In other words only some of the cells have the extra copy of chromosome 21. This is called mosaic trisomy. Sometimes just one particular type of cell, such as the skin cells, will have the extra chromosome. In other cases, a percentage of all of the cells will have the extra chromosome
- Also uncommonly, babies can have chromosome 21 or part of chromosome 21 stuck to another chromosome

These babies are born with a variety of specific physical features and some degree of developmental delay and mental retardation. They usually have round heads that may be smaller than normal. The back of the head is often flat. They usually have a flat nasal bridge and their eyes are slanted up at the corners. They usually have a small mouth and chin and their tongue may look large. It may stick out of their mouth a little. The lines or creases across the palms of their hands are different. The little finger may curve in towards their other fingers.

Signs and Symptoms: (In addition to the ones listed above)

- These babies tend to be floppy and have poor muscle tone. This is called hypotonia
- Small ears that are often low placed
- Extra skin may be present on the back of the neck

- A blockage in the esophagus. The esophagus is the tube that goes from the mouth to the stomach. This is called esophageal atresia
- A blockage in a part of the intestines. This is called duodenal atresia
- The doctor may hear a heart murmur and there may be heart defects, which can cause very serious problems.
- Retarded growth and development are usually seen.
- Delayed mental and social skills are always present. They may be mild or severe.

Tests:

- Amniocentesis can be done before the baby is born. Fluid from the water around the baby is removed. The cells in it are tested for abnormal chromosome numbers.
- A doctor can usually tell if Down syndrome is present from a careful physical exam.
- The baby's blood will be tested to confirm that there is an abnormal number of chromosomes. High-resolution chromosome tests will show whether the baby has a full trisomy, a mosaic or a translocation.
- An ultrasound exam of the heart can find any abnormalities. This is called an echocardiography.

Treatment:

There is no cure for Down syndrome. All treatment is aimed at supporting the parents and babies.

- Physical therapy. Parents can be taught to do exercises and positioning techniques with the baby, to help prevent physical problems.
- Occupational Therapy (OT). An OT will be available to help parents with feedings issues. Some babies need more help with feedings because of poor tone and a larger than normal tongue size.
- Surgery. If there is a heart problem found, surgery may or may not be necessary to correct the problem.

Recovery Process:

Children with Down syndrome are can be very affectionate, kind and cheerful; however, they are at a higher risk for developmental disabilities. The disabilities will affect behavior and learning. A genetic counselor can provide counseling for families, so that they can understand what to expect as the child grows up. With appropriate care and social stimulation, these children can have the opportunity to reach their full potential.

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Duodenal Atresia

Definition:

The duodenum is the first part of the bowel, after the stomach. Food combines with acid in the stomach and then moves to the duodenum which mixes with bile and other juices that help digest the food. From there, the food mixture moves through the bowel. The next part of the bowel is called the small intestines. In the small intestine, the body removes the nourishment from the food mixture. When a baby has duodenal atresia (i.e. underdevelopment with blockage), the duodenum has not formed properly. Instead of being the first part of an open tube, it is not open. There is no hole for the stomach contents to move through. Doctors think, as one possibility, that sometimes when the baby is developing, the blood supply to the duodenum gets interrupted. This interruption causes the duodenum to get narrow and blocked. The result is duodenal atresia. No one knows why this interruption of the blood supply occurs.

Signs and Symptoms:

- During pregnancy the mother may have more than the usual amount of “water” (i.e. amniotic fluid) around the baby. This condition is called polyhydramnios
- After birth the baby will start vomiting large amount of green fluid, possible even if he/she is not being fed
- The baby may have Down syndrome. About 25% of babies with Down syndrome have duodenal atresia

Tests:

- X-rays of the bowel (called a KUB and an upper GI)
- Blood tests will be done to see if the salts in the baby’s blood have become abnormal from the vomiting

Treatment:

- Intravenous Therapy (IV). Because these babies are vomiting and cannot eat, they need to get fluid and nourishment through an IV. They will also receive antibiotics through the IV
- Orogastric tube (OG). A nurse will pass a tube through the baby’s mouth into the stomach to drain the green fluid
- Surgery. A baby born with duodenal atresia needs surgery to remove the blocked portion of the duodenum. The baby will go to the operating room. The surgeons will remove the abnormal part and reconnect the intestines.

Recovery Process:

It will take several days for the baby to recover from the surgery. The baby will be given pain medication to keep for comfort. He/she will be given fluids and nourishment through an IV. When the baby is ready, the doctors will start to feed the baby very slowly. At first, the feeding will be given into a tube that goes from either the nose or the mouth directly into the stomach. It might take several weeks before the baby can take a full feeding from a bottle. Mothers are encouraged to save their breast milk so that it can be fed to the baby.

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Congenital Diaphragmatic Hernia (CDH)

Definition:

Congenital diaphragmatic hernia is a condition in which an abnormal hole exists in the diaphragm. The diaphragm is a muscle located below the lungs. This is the main muscle used in breathing. It also serves as a barrier between the lungs and the organs of the abdomen, such as the stomach and liver. Sometimes during fetal development, this hole does not close causing CDH. The organs such as the stomach, intestines and liver can move into the space where the heart and lungs are growing, called the thorax. As a result, the lungs do not grow to their normal size. Both the heart and lungs can become severely compressed. The lungs remain small and poorly developed. They are called hypoplastic. A life-threatening condition called persistent pulmonary hypertension of the newborn (see PPHN) can result. However, not all babies with CDH are this sick, and in fact many do not even need the help of a respirator. But in all cases surgery is best for the babies with CDH

Signs and symptoms:

- Difficulty breathing
- Abdomen appears small. Much of the bowel and other organs may have moved into the chest

Tests:

- Ultrasound can often be used to detect hernia even before birth
- Chest x-rays show the abdominal contents in the chest (thorax) and the heart can be shifted to one side
- Blood tests will show low levels of oxygen and high levels of carbon dioxide in the sickest babies with CDH

Treatment:

- As mentioned, not all babies with CDH are so sick as to need a respirator, but will still need surgery to repair the problem

- Ventilator (breathing machine). The baby will be supported on a ventilator to help with breathing when needed
- Nitric oxide therapy. This is a special gas given through the ventilator, and it may be used to improve lung function
- Medicine that helps to support the heart and maintain blood pressure
- In extreme life-threatening cases, the baby may require ECMO support (please the information on ECMO on this web site)
- Surgery. The surgery involves moving the abdominal organs out of the chest and to repair the hole in the diaphragm

Recovery process:

It will take several days to weeks for the baby to recover from the effects of surgery. The baby will be on a ventilator. This will help the baby's breathing until the diaphragm and lungs heal from the surgery. Sometimes the doctors will need to give the baby medicine to keep him/her from moving. This might be needed for the first few days following the surgery. Pain medications will be given to keep the baby comfortable following surgery. Outcomes are difficult to predict. Many infants require prolonged hospital stays to treat problems after surgery such as infections, PPHN, decreased diaphragm function, and problems with the intestines after they have been pushed back into the abdomen. Also, the baby needs to learn to suck/swallow, meaning to learn to take a bottle like a normal baby. This takes time because the reflexes for feeding need to return after the period of being sick.

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Gastroschisis

Definition:

Gastroschisis is a disorder in which the baby is born with a defect in the wall of the abdomen (stomach). The contents of the abdomen (bowel, stomach, and liver) are pushed outside of the abdomen. There is no covering or sac over these contents. The absence of the sac will expose the intestines to the fluids in the womb. This can sometimes cause the contents to become swollen and inflamed. Babies with this problem can also have smaller than normal abdomens

Signs and symptoms:

- Visible bowel on the outside of the baby
- Fluid losses

Treatment:

- Surgery. This condition requires surgery to prevent infection and large fluid losses, performed in the first day after birth. Swelling of the bowel and lack of skin and muscles can make it difficult to put the contents back into the abdomen with one surgery. Surgeons specializing in this type of surgery will put as much bowel as possible back into the abdomen. A silo or pouch may be placed over the rest of the bowel. Every day or so as the bowel swelling lessens, the surgeon will place more of the intestines into the abdomen. A second surgery will need to be done to close the muscles and skin over the abdomen
- Intravenous Infusion (IV). An IV will be needed to give fluids and nourishment until the baby's intestines are ready to receive milk
- Antibiotics will be given through the IV to prevent infections
- Orogastric tube: a tube which passes from mouth or nose into the stomach. It helps to keep the bowel from filling up with swallowed air causing distention, aspiration and discomfort
- Assisted breathing with the use of a ventilator (breathing machine)

Recovery Process:

It will take several days/weeks for the baby to recover from the effects of surgery. The baby will be given pain medications to keep him/her comfortable. The baby will not eat by mouth until the intestines are healed, during which time he/she will receive nourishment through an intravenous infusion. When the baby is ready, the doctors will start to feed your baby very slowly through a tube that runs from either mouth or nose directly to the stomach. It will take several weeks before your baby can take a full feeding from a bottle. Mothers are encouraged to save their breast milk so that it can be used to feed her baby

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Hydrocephalus

Definition:

The brain and spinal cord are surrounded by a fluid called cerebrospinal fluid (CSF). This fluid is produced in the ventricles (channels in the brain) and circulates around the brain and spinal cord. CSF acts as a shock absorber to cushion the nervous system. Hydrocephalus (too much fluid on the brain) occurs when the circulation of the CSF is blocked or when the fluid is not absorbed properly. The CSF then backs up in the ventricles, causing them to expand and push on the brain tissues. This pressure causes the fontanel (soft spot on the head) to bulge and the head to get larger. The continued accumulation of fluid and pressure may cause damage to the nerve cells in the brain

Signs and symptoms:

- Soft spot on head is full and/or tense

- Enlarged head or increased measurement of the size of the baby's head
- High-pitched, shrill cry
- Widened space between the baby's bones of the head (keep in mind that it's normal for the flat bones of the baby's head not to be connected at birth)

Tests:

The following can be done to check the size of the ventricles or to reduce CSF pressure

- Ultrasound of the brain
- Computed tomography (CT) or magnetic resonance image (MRI) of the brain
- Surgery: placement of a ventriculo-peritoneal shunt (VP shunt)

Treatment:

In some cases, the swelling of the ventricles may stop on its own. Surgical treatment will be necessary when the ventricles continue to expand. This can reduce the pressure in the ventricles

- A temporary surgical procedure called a reservoir is placed in special circumstances, for preterm babies, to allow excess fluid to be withdrawn from the ventricles by a needle. A bump under the skin of the head can be felt where the reservoir is located
- Surgical placement of a VP shunt will be done at a later time, when the baby is larger. Surgery involves inserting one end of a thin tube into the baby's ventricle. The other end of the tube is passed underneath the skin and drains into the abdominal cavity. The body will reabsorb the extra spinal fluid

Recovery process:

It will take several days for the baby to recover from surgery. The baby will be given pain medication for comfort. Once the VP shunt is inserted it usually remains in place and must be lengthened every two to four years as the child grows. Infection and blockages are the two most common complications

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Imperforate Anus

Definition:

The bowel (intestines) normally ends in a pouch called the rectum. The rectum releases stool from the body through the anus. It does this by using specialized nerves and muscles. Babies born with an imperforate anus do not have a normal opening, rather a

blind ending to the pouch. Because of this they can't pass stool. There are several forms of imperforate anus:

- The anal opening can be in the wrong place
- It can be a small tube-like opening called a fistula. The fistula can go from the rectum to the skin, the vagina or the urethra (tube that allows urine to leave the body)
- There may be no opening at all

In either situation the rectum can be very close to the skin, nerves and muscles that allow the anus to function. This is called a low type of imperforate anus. Or the rectum may end further away from the skin, nerves and muscles. This is higher up in the abdomen. This is called a high type of imperforate anus

Signs and symptoms:

- There is no opening for stool to pass
- The opening is in the wrong place
- The baby does not pass stool in the first 24-48 hours
- Stool may come from the vagina, penis, or scrotum
- The baby's stomach may appear large or even swollen

Tests:

- X-ray of the stomach and bowels
- A careful physical examination

Treatments:

- Intravenous Therapy (IV). If these babies are fed, they may vomit and will not be able to continue taking milk. An IV will be started to give the baby fluid and nutrition
- Orogastric tube (OG). A tube will be passed through the baby's mouth into the stomach. This will prevent vomiting
- Surgery. A baby with an imperforate anus needs surgery fairly quickly. During the surgery, the baby will be put into a deep sleep to be pain-free. The kind of surgery that is done depends upon whether the baby has a low-type or high-type imperforate anus. If the baby has a low-type imperforate anus, the surgeon is usually able to repair the problem with one operation. If the baby has a high imperforate anus, the surgeons will need to do two or more surgeries. In the first surgery, the surgeon will create a temporary opening of the large intestines (colon) onto the abdomen. This is called a colostomy. This allows stool to leave the body. Once the baby has recovered from this surgery, he/she can be fed. The baby is allowed to grow for several months, before he/she undergoes the more complex repair of the imperforate anus. After the repair is completed, the surgeon is able to close the colostomy

Recovery Process:

After surgery, the baby will receive pain medication to keep for comfort. Babies usually recover quickly from these surgeries and can eat in a few days. If the baby has had a colostomy, the parents are taught to care for it before the baby goes home. We encourage mothers to save their breast milk so that it can be used to feed their baby

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Intraventricular Hemorrhage

Definition:

Intraventricular hemorrhage (IVH) means bleeding into or near the normal fluid channels (ventricles) within the brain. The area where IVH usually begins in preterm babies has a very fragile network of tiny blood vessels. Poor oxygen levels or fluctuations in blood pressure can damage these vessels. The bleeding occurs when these vessels are unable to hold the blood within the channels. The more premature and the sicker the baby is, the higher the chance the baby will have an IVH. The extent of the IVH is graded from I to IV:

- Grade I IVH is the least severe grade. It involves a small bleed and is contained within the tissue. Grade I IVH does not usually result in permanent neurological injury
- Grade II IVH results when a portion of the ventricle is filled with blood. Grade II IVH has a slightly worse prognosis than grade I but the likelihood of long-term neurological injury is also low
- Grade III IVH involves bleeding into the majority of the ventricular channels with enlargement of the ventricle. Here there can be problems with development as the child grows older
- Grade IV IVH involves bleeding into the brain tissue itself, in addition to the ventricles. Those infants with grade IV IVH are at a very high risk for permanent brain injury, including mental retardation, developmental delay and cerebral palsy

Signs and symptoms:

- Apnea (a pause or stoppage in breathing)
- Low heart rate
- Anemia
- Seizures
- Poor muscle tone and decreased activity called lethargy
- Bulging soft spot on the top of the baby's head
- A baby with an IVH may not show any symptoms

Tests:

- Ultrasounds of the head
- Measurements of the size of the head of the baby

Treatment:

- In many cases, repeat ultrasounds will be all that needs to be done. The doctor will follow the progress of the bleed or the reabsorption of the blood
- Babies with IVH may also develop hydrocephalus (increased cerebrospinal fluid). Surgery may be needed to fix this problem. See Hydrocephalus for more information

Recovery Process:

A number of babies under 1500 grams at birth will have IVH. The outcome for these babies varies with each case. Generally, grades I, II, and III IVH have a somewhat more optimistic prognosis, although these infants still have a risk for problems like hydrocephalus, mental retardation, cerebral palsy and vision loss. Babies with Grade IV IVH usually always have serious neurological problems

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Jaundice of the Newborn

Definition:

Jaundice is the yellow color of the skin and eyes that is often seen in newborn babies. This is caused by bilirubin in the blood. Bilirubin is a normal by-product of the breakdown of red blood cells. All red blood cells get old and break down. The liver processes the bilirubin and deposits it in the intestines. The bilirubin then leaves the body in the stool. All newborns have some jaundice and this doesn't present a problem. Their livers are still a little immature, and to some extent the bilirubin provides positive benefits to the baby's health. Low levels of bilirubin are not dangerous, but very high levels can cause permanent brain damage.

Sign and symptoms:

- Eyes and skin will have a yellow tinge
- The baby may be very sleepy and hard to wake, or even very irritable with a high pitched cry
- The baby may be eating poorly or refusing to eat at all

Tests:

- Physical exam

- Blood tests will show higher than normal bilirubin levels

Treatments:

- Phototherapy. If a baby's bilirubin levels are too high, the baby is put under special artificial lights. This is called phototherapy. Sometimes a light-producing blanket is placed under the baby. The baby's eyes are protected from the light by eyeshades. This therapy will help break down the bilirubin in the skin by a chemical photo-reaction. Babies usually need to be under the lights for only a few days. The baby's blood will be checked regularly to make sure that the bilirubin level is dropping
- Intravenous therapy (IV). If babies are too sick to eat well, they will have an IV so that they can get enough fluid
- Exchange transfusion. Occasionally, the baby's bilirubin will rise so high that the doctors need to do an exchange transfusion to avoid the danger of brain injury. The exchange transfusion removes the very high bilirubin levels directly from the blood. The procedure involves replacing baby's blood with blood from the blood bank

Recovery Process:

Some babies have more trouble getting rid of bilirubin. For example, premature babies have very immature livers. Babies that are sick and are not eating don't produce enough stools to get rid of the bilirubin. There are also a number of rare diseases which cause the red blood cells to die more quickly than usual, producing extra bilirubin. Most babies recover completely

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Meconium Aspiration Syndrome (MAS)

Definition:

Meconium is the sticky, odorless material (stool) that babies produce as fetuses during pregnancy. Sometimes it is released while the baby is still in the uterus. This is a sign of a problem like infection or not enough oxygen getting to the baby, or other kinds of stress. The release of meconium before birth can be a very serious problem if the baby breathes this material (i.e. aspirates the meconium) into the lungs. If the baby starts to breathe while in the uterus where the meconium is mixed with the mother's amniotic fluid, some of the meconium can end up in the lungs, making them very sick. A life-threatening chain of events can occur. Meconium can obstruct small airways. Because it is thick and sticky, it cannot be cleared easily from the lungs. It also decreases the effect of surfactant. Surfactant is a chemical material that keeps air sacs open in the lungs, and is thus very necessary for easy breathing. Meconium is highly irritating to the lungs. It may cause so much inflammation that the lungs do not work properly, causing great difficulty in getting oxygen into the blood so that the heart can pump it to all the organs and tissues of the body. Blood flow through the lungs can even become a problem (see PPHN)

Signs and symptoms:

- Difficulty in breathing.
- Presence of meconium-stained birth fluid or meconium in the trachea (windpipe or airway)

Tests:

- X-rays of the lungs
- Blood samples to check for low oxygen levels

Treatment:

At CHLA treatment is aimed at supporting the baby until their lungs have healed:

- Antibiotics are given to fight infections that are commonly seen in babies with MAS
- Orogastric tube. This is passed through the mouth or nose into the stomach. It will help remove any air that the baby might swallow
- A ventilator (breathing machine). It will help support breathing
- In severe cases, a gas called nitric oxide is given through the ventilator. This can help improve blood flow through the lungs
- A special medicine called surfactant (foamy liquid) is often placed in the lungs. Surfactant helps to keep the air sacs open
- In extreme cases, patients may require ECMO therapy. (See ECMO)

Recovery process:

With early treatment, even patients who require ECMO have a greater than 90% chance of survival. Although there is a risk of damage to the brain in cases where the fetus or infant was severely deprived of oxygen, the majority of babies will recover without long-term problems

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Myelomeningocele/Spina Bifida

Definition:

Spina bifida means a split or break in the spine. The spinal column, or backbone, is made up of 33 smaller bones called vertebrae. These bones protect the spinal cord by forming a bony ring around the cord. In spina bifida, one or more of the vertebrae is formed improperly. There is a gap in the protective ring. Where there is a gap, the spinal cord is not protected. The spinal cord is like a closed tube made up of nerves. The nerves send

messages to move muscles, and also take messages back from the body for processing in the brain, like sensing feeling on the skin

There are three types of spina bifida

- There is an opening in one or more vertebrae but the spinal cord does not stick out. A dimple or a bit of hair may mark the location of the defect. This usually does not cause any problems. Many people who have it do not know it
- Meningocele. This is a more serious form of spina bifida. In this case the protective covering of the spinal cord (the meninges) pushes out through the abnormal opening in the vertebrae. A kind of sac of the exposed meninges is formed. The sac is called a meningocele. The nerves in the spinal cord are not usually damaged in this case. The defect can be repaired with little or no damage to the spinal cord. This is the least common form
- Myelomeningocele, meaning cord (myelo-) out plus the meningocele, or meninges out as above. Thus, the covering with the spinal cord is exposed on the back of the baby. This is the most serious form of spina bifida. The nerves may not have developed properly or they may have been damaged when they get pushed through the gap in the abnormally developed vertebral column (bones). There may be weakness or paralysis of the bladder and bowel control, when nerves controlling these organs are involved. The amount of damage depends upon the location of the myelomeningocele. These problems are repaired quickly after birth, by specialized surgeons called neurosurgeons, to prevent infection and further injury to the nerves

Sign and symptoms:

- A big blister or sac on the baby's back
- Fluid may be leaking from the blister

Tests:

- A high level of alpha fetal protein in the mother's blood during pregnancy
- A special very accurate ultrasound during pregnancy can find the sac on the back

Treatment:

- Surgery. Babies with myelomeningocele will have surgery during their first few days after birth. They will be deeply asleep and pain-free during surgery and will be given medication for pain after surgery
- Intravenous Therapy (IV). These babies have a high-risk for infection and need to be on antibiotics. They will not be able to eat before their surgery and will need to get fluids and nutrition through their IV

Recovery Process:

After surgery, these babies will be given pain medicine in their IVs to keep them comfortable. They will get IV fluids until they can eat by mouth. They usually recover quickly from the surgery and are feeding normally in a few days. CHLA has a spina bifida team. This is a group of doctors, nurses and social workers that specializes in the care of babies and children born with spina bifida. This team arranges for physical and occupational therapy and any other specialized treatment babies may need after they go home. They help the family to make the best adjustment to any disabilities that are present

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Necrotizing Enterocolitis (NEC)

Definition:

NEC involves inflammation and infection occurring from injured surfaces of the bowel. It is one of the common surgical emergencies in newborns, when the problem is particularly severe. The exact cause is unknown. One theory is that the intestinal tissues of premature infants are weakened by too little oxygen or blood flow, but occasionally it also occurs in full term babies. For preterm babies when feedings are started, the added stress of food moving through the intestines allows bacteria that are normally found in the intestine to invade and damage the wall of the intestinal tissues. As a result of the bacteria, the body fights the infection and sometimes this causes inflammation that can actually injured the intestines even more

Signs and symptoms:

- NEC can occur within the first 2 weeks of life, usually after milk feedings have begun
- Baby may appear less active
- Increased respiratory problems
- Vomiting
- Increased size of the abdomen
- Redness or abnormal color to the abdomen
- Apnea (stoppage of breathing)

Tests:

- Blood in the stool
- Special X-ray of the bowel (KUB)

Treatment:

Treatment may include:

- Stopping feedings
- Orogastric (OG). The tube will be inserted through the mouth or nose into the stomach. This will help to keep the stomach from swelling up more from swallowed air and prevent aspiration.
- Intravenous (IV). Fluids for nourishment and fluid replacement will be given through the IV while the bowel heals
- Antibiotics to help fight the infection will be given through the IV
- Frequent X-rays of the bowel
- Severe cases may require surgery to remove the diseased bowel. An ostomy (surgical opening) may be needed to create an opening into the bowel. This allows passage of stool above the diseased bowel. The surgical area can then rest and heal

Recovery process:

It will take several days/weeks for the baby's bowel to heal. If surgery is required the recovery process will be longer. The baby will be given pain medication to keep him/her comfortable. When the bowel is healed, the baby will start feeding, but very slowly. At first the feeding will be given into a tube that goes from either the nose or the mouth directly into the stomach. It will usually take several weeks before the baby can take a full feeding from a bottle. Mothers are encouraged to save their breast milk so that it can be fed to the baby

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