



The newsletter of



**Providing support, information, and connections  
to families of children with undiagnosed medical conditions**

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## **What is behind the feelings?**

A few messages on our e-mail group got me thinking about how we process our thoughts and feelings and make sense of it all. It is often hard to understand the feelings that we experience when we are on a diagnostic journey for our children, especially when we have been in the undiagnosed world for an extended period of time. As you can imagine it is even harder for our families and friends to understand what we are facing.

I'm gradually learning different stages that one may face when they have an undiagnosed child. These stages are not scientific and mostly come from experience and talking with other families.

**Denial** - It is easy to have a denial stage when there is an undiagnosed child in the picture. The symptoms often aren't all presented to us at once and the doctors don't really know what is going on with the child. This stage can even come and go; things may be going well for a period of time so you may feel everything is ok. There may even be a sense of denial about the child's future since there is so much unknown. There is also a sense of denial of how affected our children are, since we tend to compare to others who are more affected. This is where time and experience may help us; we will naturally prepare ourselves for what might be in the future.

**Blame** – Since we are unsure of what our children have we may blame ourselves, our partners, doctors or even a higher power. This blame might last longer than children who receive a diagnosis, because we have no evidence to change our thoughts. If you can learn about the

diagnosis and understand it the blame stage may be easier to overcome.

**Searching for answers** – We seem to go in stages of searching and wanting to know the diagnosis. Sometimes we are at a type of peace with not know, often because no issues are presenting a sense of urgency to understanding. With every new symptom, sign or issue we tend to feel that urgency to have a better understanding for what it is that is causing all of our child's symptoms. If we knew we would at least have a better understanding of what is happening. In some situations there may be a plan of action to take that may improve the situation. Some of us may also have fear standing in our way of searching for answer. The fear of what it is that we don't know, would it be better not to know?

**Our extended family and friends** – Many of us have different experiences with family and friends. Sometimes we receive great compassion and understanding from those around us. It's not always like that, others don't really understand. They may see our children as a fear of what could be for them, and they just can't handle those thoughts so they stay away. They may also be unsure of how to approach us and fear saying the wrong things. Then some are very insensitive and again likely because their lack of understanding our situations. This is a reminder that we are all different. It is best to try not to let these situations get the best of you. Take small steps to open the lines of communication and try to understand others ignorance of our situations.

Your Friend,  
Amy Clugston



## Living an Undiagnosable Life

By Jennifer McKay

You start out with the best of intentions. Their nursery is going to be the best ever. It's going to have puppies everywhere and a train going around overhead. They have every possible childhood toy and gadget. And the clothes and shoes are just so much fun.

They have the most beautiful smiles. All they want is to be loved and protected. Why don't we get the option to protect them? We do everything humanly possible and it's not nearly enough. There are no answers and there's a good chance there never will be. The best minds in the world are baffled. All you can do is tell them every day that you love them, are proud of them, and will be with them every step of the way. You also have to tell them if it's too much you understand and it's okay because you don't want them to be scared.

Their spirit amazes you every day. They have such strength. Each day they teach you that you have so much more strength than you ever knew you had. They teach you things about the human spirit and experience that you would never have known existed if everything was okay. They teach you to sweat the small stuff because each one of those little things is so priceless.

No matter how bad it is you still hold out hope for a miracle. Maybe one day they will get to spend some time in their nursery playing with all of their toys, watching their train, and naming all of the puppies. Sometimes these miracles do happen, but it's very rare. You take them to regular doctors, witch doctors, and anything else in between just for a chance to change the inevitable outcome. You try hippie products, every vitamin known to human kind, healing meditations, and even crystal tones.

Then you have to say goodbye. How do you ever say goodbye? You've had your heart and soul torn out over and over again. How is that ever repaired? They look so beautiful - being sent off to play. You keep expecting them to just wake up and say something, anything or give you that look - and we all know that look. It never happens.

Now you have to rebuild your life. Do you go back to the way it was before like nothing ever happened? How can you because you will never be the same person again? Where is that strength to put one foot in front of the other and conquer all obstacles that was here not that long ago? How do you tap back into that? How do you respond to others? There are just so many questions and still now answers.

### Jimmy McKay Story

I had a type 1 mitochondrial disorder. My first symptom was Nystagmus. My case looked like Cookie Monster Eyes that rolled all over the place. Later I developed cataracts (the Nystagmus was actually my first sign) and had them removed. Basically I had rapid body movements when I got upset or agitated. Due to these movements I began aspirating my food and saliva into my lungs which was just compounded since I also had acid reflux. These problems caused me to have a trach put in so I could breathe and a g-tube in my tummy so I could eat. As time wore on I needed oxygen boosts because breathing became so difficult. So here are my overall things: type 1 mitochondrial disorder, undiagnosed neurological movements, cataracts, developmental delays, hypotonia, g-button with nissen due to reflux, trach due to laryngomalasia, hiatal hernia, brain atrophy, and seizures. And many think learning how to spell your name is hard! I passed away after a long and hard fight to live on December 27th, 2007 at 9:59 AM since I had an appointment at 10 AM. I will be missed by many and hope I taught people a lot while I was here for my little mission from God.



# Resources

This section is meant to offer you information on a variety of opportunities and resources. We are not endorsing or suggesting that these are best for you. Use your own judgment on any of the Resources.

## Genetic Information Nondiscrimination Act

Through the hard work of many organizations involved in the Coalition of Genetic Fairness, GINA was passed by President Bush on May 21<sup>st</sup>. With so many advancements in gene testing it is sometimes found that we carry gene defects that we weren't aware of. Since we are in the mists of all this testing this could very well affect the families of SWAN USA. We no longer need to worry that if a genetic difference is found in our children or even ourselves, it may affect how we are treated. We can expect not to be discriminated against in the United States. You will have to stay tuned on just how this may affect us. You can learn more about GINA at the link below.

<http://www.geneticfairness.org/>

## United Health Care Children's Foundation

It is always difficult for families to have to pay for all the medical needs of our children; This is one way to help with that problem. The United Health Care Children's Foundation is offering grants to families in need of financial help with their children's medical needs. The grants will be for medical needs that are not covered or not fully covered by their commercial health benefits. You must have a commercial health plan, which excludes Medicare and Medicaid. If you are interested and meet the criteria you can submit an online grant application. The application would then be reviewed by their Regional Board of Directors. If your grant is approved your commercial health plan would be billed first, then the remaining balance to you. You would be responsible for sending bills to UHCCF, who would then pay the portion of the bill to the service provider. To find out more information use the information below.

<http://www.uhccf.org>  
1-800-328-5979 ex.24456

## NIH Undiagnosed disease program

NIH released information in May of 2008, about their new Undiagnosed Disease Program. The program will be held at the NIH Clinical Center in Bethesda, Maryland. The program will review every case that comes to them, but they



will only be able to see about 100 patients a year. There will be 25 specialists to review the cases. A referral is needed from a physician who will provide all medical records and test results that is needed by NIH.

1-866-444-8806

<http://rarediseases.info.nih.gov/Undiagnosed>

## ZAC Browser

Zone for Autistic Children was created for the developer's grandson who has autism. It is easy for children to use and free to download. Like many things that are created to benefit children with autism, this can be a great tool for many of our children. Many of our children have some type of similarity with children who have autism such as; communication difficulties, social needs, sensory and behavioral issues. Don't shy away from products that are intended for children with a specific diagnosis, they often can be used to benefit all children.



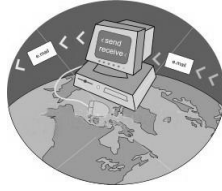
<http://www.zacbrowser.com>

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## Connecting with other families

For those who are new to SWAN USA, you can connect with other families through our e-mail group. The group is now up to 260 members.

[http://groups.yahoo.com/group/undiagnosed\\_syndromes](http://groups.yahoo.com/group/undiagnosed_syndromes)



You can also connect with other families by telling your story in this newsletter or on our website. If you leave contact information families can contact you if they see similarities.

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## Calendar of Our Kids

SWAN USA will be creating a calendar full of our kids. This will be a great way to connect with other families and see the cute faces of our kids. Siblings are also welcome to show their faces to us all. The calendar will be for 2009 and will be very colorful. It will also be a great awareness tool. We are hoping for many faces to be included, and will only show first names in the. If you would like to include your child send a picture, name and birthday to the e-mail address or postal address in the contact section.



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## SWAN USA Logo

You can purchase items from cafepress with SWAN USA's logo on it and a percentage will come back to the organization. Here are a few of the items you can purchase.



[www.cafepress.com/swanusa](http://www.cafepress.com/swanusa)

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## Thank You for your Donations!

From Scott and Lynn Myers  
In Honor of Lorna, Jadon & Cloey Clugston

From Gary Strickland  
In Memory of Jimmy Mckay

From Bert and Gwen  
In Memory of Jimmy Mckay

Tup Strickland  
In Memory of Jimmy Mckay

Little Tresoros Therapy Services  
In Memory of Jimmy Mckay

Marcia Muse  
In Memory of Jimmy Mckay

JLC Underground  
In honor of the Clugston Family

Temple Inland Foundation  
Match on behalf of Marcia Muse

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## SWAN USA Contact Information

Amy Clugston  
1745 Lorna Lane  
Otsego, MI 49078

**New Phone Numbers**  
Phone: 269-692-2090  
Family Line: 888-880-SWAN

[amyclugston@undiagnosed-usa.org](mailto:amyclugston@undiagnosed-usa.org)  
<http://www.undiagnosed-usa.org>

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## Board Members

**Amy Clugston** – President/Treasurer  
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**Michelle Maleski**  
**Heather Long**  
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**Jennifer McKay**

## Treating GERD By Lesley Bennett

In many cases, your child's symptoms can be treated by simple lifestyle or dietary changes. However, GERD can be difficult to treat in some medically complex children and your specialist may recommend a medical approach using prescription or over the counter (OTC) medications to control GERD, or in rare cases, a surgical approach to prevent the severe complications of GERD. Neurologically impaired children are at high risk for developing GERD complications and physicians know that these children may need preventive **medical** management (medications) that focus on controlling stomach acid or avoiding motility problems and **surgical** interventions that provide nutrition (tube feedings) or that prevent complications from acid reflux (fundoplication). Many children born with congenital malformations are also more likely to develop GERD due to their problems with gastric emptying, strictures, or dysmotility; and these pediatric patients may also need more aggressive GERD management. Treatment of GERD will depend on your child's age, overall health, underlying condition, medical history, physical exam, your child's ability to tolerate certain therapies or medications, your preferences, and the severity of your child's GERD or GERD complications. Your child's physician will discuss treatment options with you, and these options may include: **diet or lifestyle changes, medications, or surgical interventions.**

### Lifestyle changes

Based on your child's physical exam findings, the medical history, and your answers to GERD questions, your GERD specialists can recommend strategies to avoid reflux such as dietary, feeding, and positioning changes to help your child. These changes may affect your entire family. You and your family may have to consider changes in the home or way your family eats and cooks to help your child lose weight, avoid tobacco smoke, or eliminate foods that provoke GERD symptoms. Lifestyle and dietary changes a specialist or GERD team often recommends include:

- **For Infants:** If your baby is being breast-fed, you may be asked to avoid eating any foods that provoke acid reflux such as milk products, soy, wheat, nuts, chocolate, peppermint, caffeine, carbonated beverages, spicy foods, or acidic foods such as tomatoes or oranges. If your infant is taking formula, you may be asked to try thickening the formula with rice (helps reduce vomiting) or provide your infant with smaller, more frequent, amounts of formula throughout the day. GERD specialists may also recommend feeding or positioning strategies to help your infant such as burping your baby several times during a feeding, keeping your infant in an upright position for at least 30 minutes after a meal, propping your child up in bed at night, and not feeding your baby just before bedtime.
- **For Children and Teens:** The nutritionist on a GERD team may recommend that your child avoid some or all foods that can provoke acid reflux such as beverages containing caffeine, chocolate, spicy foods, fried or fatty foods, peppermint, garlic, onion, citrus fruits, and tomato-based foods including childhood favorites like spaghetti or pizza.

The nutritionist or feeding experts may also recommend limiting the size of your child's meals to avoid overeating, giving your child small meals frequently throughout the day, switching prescription medications to more stomach friendly drugs, elevating the head of your child's bed 6 inches to prevent nighttime bouts of acid reflux, and NOT eating 2 or 3 hours before bedtime. Feeding specialists on the team may show your child the proper way to chew food, positioning (ways to sit) techniques to improve swallowing, or even recommend that your child wear loose clothing while eating to prevent reflux. If none of these strategies help your child's acid reflux symptoms, then it is time to consider ways to medically manage your child's condition.

### Medical Intervention

There are a number of prescription and over-the-counter medications used to treat acid reflux. The medication or combination of medications your child's specialist recommends will depend on your child's symptoms, GERD evaluation, underlying condition and your preferences. Prescription medications are usually stronger than over-the-counter medications and can help reduce the severe symptoms or complications of GERD. Many of these medications can reduce your child's symptoms with 2 weeks. Your child will probably need a prescription medication if the esophagus is inflamed or your child has severe acid reflux. Currently there are 5 classes of medications used to treat GERD: acid neutralizers, cryoprotectives, acid reducers, proton pump inhibitors, and prokinetic agents.

#### Antacids

Antacids are over-the-counter drugs that can temporarily neutralize stomach acid and they are often used in combination with other GERD medications. These drugs can give your child quick relief from acid reflux symptoms and they are often one of the first drugs used to treat acid reflux. Most antacids are liquids that come in a number of flavors and will coat the esophagus--which can help relieve the pain or burning sensation from stomach acid. Antacids are fast acting but they are not recommended for long term treatment (only occasional use) since they do not have any long lasting effects and cannot not heal an inflamed esophagus. Antacids with magnesium can sometimes cause diarrhea while antacids with aluminum can cause constipation. Aluminum containing antacids are not recommended for infants due to possible neurologic and bone development side effects. Some common antacids are:

- Aluminum and Magnesium Hydroxide: **Maalox®**, **Mylanta®**, or **Gaviscon®**
- Calcium Carbonate: **Tums®** or **Roloids®**
- Magnesium Hydroxide: **Philips' Milk of Magnesia®**
- Sodium Bicarbonate

#### Cryoprotective Agents

These drugs work by coating (protecting) the lining of the digestive tract, and often they can help heal any tissue damage or erosions, especially in the esophagus. The most commonly used cryoprotective agents are:

- **Carafate®** (Sucralfate) is a sucrose sulfate-aluminum compound that reacts with stomach acid to form a paste. The sucralfate paste can attach to the surface of an ulcer (erosion) and form a barrier that allows the ulcer to heal by preventing further tissue damage. Sucralfate is capable of coating an ulcer for 6 to 8 hours after a single dose. It is a prescription medication that comes in an oral suspension, and is used by many children to heal painful erosions caused by acid reflux.
- **Cytotec®** (Misoprostol)-- is a synthetic prostaglandin (hormone-like) drug which stimulates mucus production to protect the tissues lining the GI tract.

### **Histamine 2 Receptor Antagonists (H2RA)**

These drugs are also called H2 (acid) blockers or acid reducers. H2 blockers prevent histamine (a substance found in the body) from stimulating stomach acid production. This class of drugs works by blocking the histamine-2 receptors on stomach cells that produce gastric acids and this reduces the amount of acid released into the stomach and GI tract. These drugs come in prescription and over-the-counter (OTC) forms and many can be prescribed for children as a flavored syrup, suspension, or drink. These medications do not act as quickly as antacids (they take about 30 minutes to work), but they provide relief for a longer period of time than antacids, greatly reduce acid production, and are very effective in reducing nighttime GERD symptoms (when taken before bed). H2 blockers have few side effects and are one of the first drugs used by gastroenterologists to treat acid reflux since there are a number of studies showing the effectiveness of the drugs in children from ages 1 month to 18 years. H2 Blockers commonly used to treat pediatric GERD are:

- **Zantac®** (Ranitidine) comes in prescription and OTC forms. The prescription medication comes in pill, syrup or liquid, effervescent drink, IV, and injection forms. Zantac can be prescribed for children as a flavored syrup (mint) or as Efferdose® (an effervescent citrus drink that contains no lactose fillers).
- **Tagamet®** (Cimetidine) also comes in OTC and prescription pill, syrup, IV, and injection forms. For children it is often prescribed as a flavored (fruit/pineapple) syrup.
- **Pepcid®** (Famotidine) comes in prescription and OTC forms. As a prescription medication it comes in gels, oral suspension, IV and injection forms. For children it comes in a flavored oral suspension (cherry, banana, mint) and tablets (cherry).
- **Axid®** (Nizatidine) comes in a prescription oral solution that is currently recommended for patients 12 years and older.

### **Proton Pump Inhibitors (PPI)**

The “proton pump” is a system in the acid producing (gastric) cells in the lining of your stomach that is responsible for making the acids necessary for digestion. PPIs are a class of drugs that enter these gastric cells and block or suppress the “proton pump” which greatly reduces the amount of acid released into the stomach over a LONG period of time. The

great benefit of PPIs is their healing power. Since these long lasting, powerful drugs can reduce stomach acid production by as much as 90%, PPIs help the erosions in your child’s esophagus heal. The main disadvantage of PPIs is their cost. PPIs are usually tried after H2 blockers have failed to reduce your child’s symptoms since prescription PPIs are very expensive (~\$8/day or more). Although some PPIs are not yet FDA approved for pediatric patients, PPIs are frequently prescribed for children with recurrent acid reflux symptoms that do not resolve with H2 blockers such as children with asthma, erosive esophagitis, or recurrent pneumonia; and in infants with recurrent vomiting, failure to thrive, or life-threatening events such as apnea. The only PPI currently available as an OTC medication is Prilosec (Prevacid is scheduled to be released in an OTC form in 2009/2010). You should not use the OTC form of a PPI unless your child’s specialist recommends it. These drugs are safe but very powerful and your physician will probably recommend the lowest dose possible to prevent side effects such as stomach cramping or diarrhea (sometimes long term use of PPIs can cause bacterial infections such as C. diff). Since most of these prescription medications come in time-release or time-delayed capsules, PPIs should not be chewed or crushed. PPIs commonly used by children are:

- **Prilosec®** (Omeprazole) is available in both prescription and OTC (tablet) forms. The prescription form is a delayed-release tablet or capsule. For pediatric patients unable to swallow (including those fed by tube), the capsules can be opened and the granules of Prilosec may be mixed with applesauce, water, or an acidic juice (orange, cranberry, pineapple, tomato).
- **Prevacid®** (Lansoprazole) is a prescription (an OTC version should be available in 2009) medication available in delayed-release capsule, solutab (chewable), injection, IV, and an oral suspension or powder packet. For pediatric patients who have trouble swallowing the time released capsules may be opened and then mixed with a tablespoon of a soft food such as applesauce, yogurt, water, or 2 ounces of juice (such as apple, grape, or orange) and the drug information sheet gives very detailed instructions for mixing the capsules with food or water. In addition, there is an oral suspension that is available as 15 or 30 mg strawberry-flavored (powder) packets which can be mixed with 2 tbsp of water right before ingestion (packets should not be mixed with juice).
- **Nexium®** (Esomeprazole) often called the purple pill is available in prescription capsule, injection, and IV forms. For pediatric patients who cannot swallow, or have difficulty swallowing medications, the capsules can be opened and the granules added to an acidic liquid such as orange, pineapple, tomato, or cranberry juice—or mixed with water for G tube administration.
- **Zegerid®** (Omeprazole® and Sodium Bicarbonate) is a prescription combination PPI and antacid that is available in capsule, chewable tablet, and oral suspension (powder packet) forms. The oral suspension (powder pack) is recommended for children who are unable (or unwilling) to swallow a capsule—do not sprinkle capsule granules on juice or soft foods, since Omeprazole® in this formulation does not have an enteric coating to protect it.

- **Protonix®** (Pantoprazole) is available in prescription capsule, injection, and IV forms. Currently it is only FDA approved for treating erosive esophagitis.
- **Aciphex®** (Rabeprazole) is rarely used for pediatric patients due to serious interactions with some antibiotics such as amoxicillin.

### Prokinetic (MOTILITY) Agents

Prokinetic agents are GI motility drugs. Some conditions such as delayed gastric emptying (gastroparesis) are associated with acid reflux or GERD. When food stays in your child's stomach for a long period, stomach contents along with stomach acid can reflux into the esophagus. The longer the food stays in your child's stomach, the greater the chance for reflux--the faster food moves out of your child's stomach into the intestines, the lower the chance of reflux. Motility drugs improve muscle tone or peristalsis (coordination of muscles) in the GI tract and they work to move food more quickly or efficiently through your child's digestive tract. Unfortunately, several of these drugs have caused severe adverse reactions in patients and are only used in children who are critically ill.

- **Reglan®** (Metoclopramide) stimulates and coordinates peristalsis (the rhythmic muscle contractions that move food through the digestive tract). It also works to increase muscle tone in the lower esophageal sphincter (LES) and stimulates stomach contractions. Metoclopramide crosses the blood-brain barrier and can cause muscle spasms, dystonia, tremors, restlessness, irritability and aggressive behavior. This drug may be tried in children over 2 years old who have recurrent vomiting that does not respond to H2 blockers or PPIs.
- **Urecholine® (Bethanchol)** is really a urinary tract drug that helps to empty (or contract) the bladder. One side effect of this drug is that it often increases gastric muscle tone and helps to restore peristalsis-- improving gastric emptying time.
- **Erythromycin** is an antibiotic with a beneficial side effect-- it can stimulate gastric motility. Erythromycin is often used in low doses to treat gastroparesis since it does not have the harmful side effects of Reglan.
- **Motilium®** - (Domperidone) Is currently not available in the USA, but it is available in Canada and other parts of the world.

### Surgical options

Due to the effectiveness of many of the newer GERD medications, today surgery is only recommended for a few children diagnosed with GERD who do not improve with aggressive medical treatment. Your child's specialists may recommend surgery if your child cannot tolerate GERD medications; has atypical symptoms especially respiratory symptoms such as recurrent pneumonia or bronchitis due to acid reflux; has complications of GERD such as erosive (bleeding) esophagitis or recurrent esophageal strictures; has an underlying medical **condition** that may contribute to GERD such as a GI motility disorder or large hiatal hernia; or chooses

surgery—many families are unable to afford the long-term use of some of the newer (expensive) PPI medications and opt for surgery. Surgery may be recommended for infants who have failed as little as 2 weeks of aggressive medical therapy or in older children after several medication trials

The most common surgical procedure for pediatric GERD is the **Nissen fundoplication** (or a variation). In this surgery, the upper portion of the stomach (called the fundus) is wrapped around the lower end of the esophagus to help strengthen, or tighten, the lower esophageal sphincter (LES). A fundoplication helps the LES close better and prevent acid reflux, but your child will still be able to eat since the LES will open enough to allow food to pass down the esophagus into stomach. Depending on your child's age, symptoms, overall health, underlying condition, and your child's specialists or medical center, the Nissen fundoplication can be done by open surgery, endoscopy, laparoscopic or even robotic techniques. Your child's specialist will explain the risks and benefits of the procedure to you. The Nissen fundoplication is not a cure for GERD but it is one of the most effective treatments and may help control your child's GERD symptoms for years. However, your child may still need to take medication as an extra protection or to help heal a damaged esophagus.

If your child has a severely damaged esophagus, trouble swallowing, reflux-associated respiratory condition, is at risk of aspiration, or is losing weight; your child's specialists may recommend a gastrostomy (G tube) or Jejunostomy (J tube). These are procedures to place a feeding (or drainage) tube through the abdominal wall into your child's stomach (gastrostomy) or intestines (Jejunostomy). Depending on your child's specialist or center, G or J tubes can be placed by open surgery, laparoscopy, endoscopy, or interventional radiology. There are a number of reasons for placing a G or J tube but the primary objective is to improve your child's nutrition and overall health. G and J tubes can be placed temporarily or permanently. Specialists will often recommend feeding tube placement for babies with anatomic anomalies such as tracheal fistula or esophageal atresia or for older children who have had a fundoplication and are at risk of aspiration.

If GI motility seems to be contributing to your child's GERD symptoms, another surgical procedure that may be recommended is a pyloroplasty. This surgery is done to widen the opening at the lower end of the stomach called the pylorus, or pyloric sphincter. In this procedure a surgeon will usually cut the muscles around the pylorus so that food can move from the stomach to the intestines more easily and efficiently.

Although this surgery greatly improves GI motility, it can cause complications such as dumping syndrome—the stomach contents empty too quickly causing metabolic problems.

