

Gastroesophageal Reflux Disease (GERD)

Acid in the stomach is an important tool to digest food. Usually, the **lower esophageal sphincter** stays shut to keep the acid and any partially-digested food in the stomach.

If the lower esophageal sphincter is weak or is open at the wrong time, stomach acid escapes up into the esophagus, leading to **acid reflux** (also called “heartburn”). If stomach acid leaks back up into the esophagus very frequently, potentially causing injury, the individual has a more serious form of acid reflux called **gastroesophageal reflux disease (GERD)**. Gastroesophageal reflux is a common problem in the general population, whether they have 22q differences or not.

Symptoms of GERD

- “**Heartburn**” – Stomach acid can irritate the lining of the esophagus, causing a burning feeling in the chest.
- Liquid or food rushing back up to the throat
- Sore throat or hoarseness of the voice
- Sour taste in the back of the throat
- Difficulty swallowing or lump in back of throat
- Dry cough
- Hiccoughs that last a long time
- Unexplained irritability in infants and young children
- Decreased appetite
- Pain in the chest or abdomen

Risk factors of GERD

- **Obesity** – The body has too much fat
- **Hiatal hernia** – The upper part of the stomach bulges and pushes upward through an opening in the diaphragm that should normally fit the esophagus
- **Pregnancy** – The growing uterus pushes on the stomach. Also, digestion is slower during pregnancy.
- **Gastroparesis** (also called gastric stasis) – The food contents in the stomach take longer than normal to move onto intestines
- **Asthma** – The inflammation and narrowing of the airway
- **Scleroderma** (systemic sclerosis) – This condition usually leads to the hardening of the skin but can also affect internal organs.

Factors that can make GERD worse

- Smoking
- Having big meals or eating late at night
- Eating fatty or fried foods
- Drinking alcohol or coffee
- Taking certain medications, such as aspirin

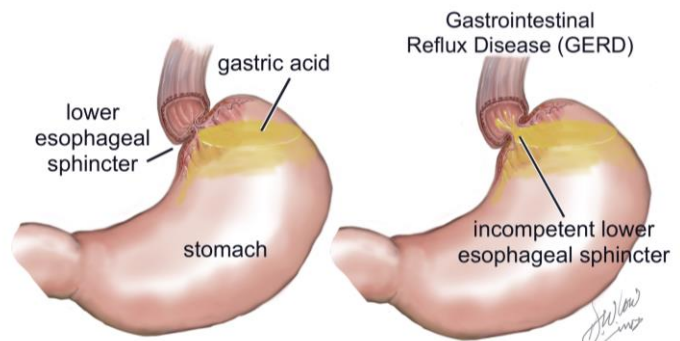
GERD and 22q11.2DS

Among individuals with 22q11.2DS, GERD tends to improve over time, primarily from childhood to teenage.

- < 13 years: ~58% have GERD
- 13 to 17: ~25%
- > 17 years: ~24%

Among adults with 22q11.2DS, there is a correlation between GERD and each of the following conditions:

- Depressive disorder
- Anxiety disorder
- Nausea / vomiting
- Abdominal pain
- Constipation



Left: The lower esophageal sphincter prevents acid from escaping upward to the esophagus.

Right: The sphincter is too weak to stop the acid from escaping upward to the esophagus.

If GERD is untreated...

- **Esophagitis** – Stomach acid damages the esophagus, leading to inflammation, bleeding, and open sores, making swallowing difficult.
- **Esophageal stricture** – Once the esophagus is damaged, scar tissues can form, which narrows the esophagus and makes swallowing difficult.
- **Barrett’s esophagus** – Damages to the esophagus can lead to changes to the cells that line it. The changes can lead to an increased risk of esophageal cancer.

Gastroesophageal Reflux Disease (GERD) (continued)

Diagnosing GERD

If your symptoms point to GERD quite certainly, the doctor may choose to proceed directly to treatment without doing further tests. However, if your symptoms are unusual or if there may already be damage to your esophagus, the doctor may choose to do some of these tests:

- **Barium Upper GI study** – You drink a liquid containing barium, which lights up under x-ray. This liquid will coat the esophagus so that the doctor can check for any narrowing on x-ray. The barium will be followed into the stomach and the upper part of the small bowel to make sure the intestines are placed correctly.
- **Upper Endoscopy** – A tiny camera is inserted into your mouth down the esophagus, stomach, and intestines to check the lining and get biopsies. The images and biopsies reveal if there are any damages to the esophagus.
- **Reflux Testing** – A tiny tube with an acid-sensitive tip or a wireless chip is inserted into the esophagus to check for acid reflux for 24 hours.
- **Esophageal Manometry** – A pressure-sensitive tube is placed into the esophagus to check whether the muscles contract and relax properly. This test does not check for GERD but can rule out other conditions that may look like GERD. It is a necessary step before anti-reflux surgery.

Treating GERD

➤ Medications

- The medications may reduce the amount of acid, heal the esophagus, or strengthen the lower esophageal sphincter.
- Some of the medications are available over the counter while others require prescriptions.
- Please discuss your situation with your medical provider before using any medications for GERD

• Lifestyle change

- Adopting an active lifestyle may reduce obesity, which is one of the major risk factors for GERD.

• Nissen Fundoplication

- This surgery involves the folding of the top part of the stomach around the lower esophageal sphincter to strengthen it.
- It is an option if medications and lifestyle changes cannot control GERD
- Watch the [Fundoplication video](#) from Mayo Clinic.

References / Resources

Clinical practice recommendations and research reports

- [Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome](#) – 2023
- [Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome](#) – 2023
- [Gastrointestinal Features of 22q11.2 Deletion Syndrome Include Chronic Motility Problems From Childhood to Adulthood](#) – 2022

Websites of medical institutions and societies

- [Gastroesophageal Reflux Disease \(GERD\)](#) – Johns Hopkins Medicine (contains helpful videos)
- [Gastroesophageal Reflux Disease \(GERD\)](#) – Mayo Clinic
- [Gastroesophageal Reflux Disease \(GERD\)](#) – GI Society / Canadian Society of Intestinal Research
- [Children with Reflux \(GERD\)](#) – GI Society / Canadian Society of Intestinal Research
- [GERD & Reflux](#) – GI Kids by the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (NASPGHAN)

When to treat GERD

Treatment is recommended for GERD if children have:

- Pain
- Irritability
- Excessive vomiting/spitting up that leads to poor weight gain
- Food refusal
- Swallowing problems
- Recurrent choking or pneumonias
- Interrupted breathing or apnea

In a study of 206 adults with 22q11.2DS, 4% had a history of fundoplication.

Helping babies with reflux

- Avoid overfeeding. Provide smaller feeds more often
- Burp frequently
- Avoid tight diapers or waistbands
- Keep baby upright for at least 30 minutes after feeding
- Avoid using the car seat while at home
- Keep baby away from secondhand smoke

For more information, please visit <https://gikids.org/gerd/>.