Esophageal Dysmotility

Normally, in a motion called **peristalsis**, the muscles in the **esophagus** (the pipe connecting the throat to the stomach) tighten and relax to bring liquid and food towards the stomach. At the bottom of the esophagus, the **lower esophageal sphincter** relaxes and opens to let incoming food enter the stomach.

Esophageal dysmotility (or **esophageal motility disorders**) is a group of serious swallowing abnormalities in which the esophagus and the lower esophageal sphincter do not work well, so that liquid and food get stuck in the pipe without arriving at the stomach. Treatments depend on the type of disorder, and may include dilating the sphincter using a balloon, surgery to widen the sphincter, and changes in diet and eating habits. It is often difficult to distinguish the various types of esophageal motility disorders, so please discuss your situation with your healthcare provider.

Esophageal dysmotility and 22q differences

Esophageal dysmotility was seen in 9% of adults with 22q11.2DS who had undergone GI imaging studies.

Classification of Esophageal Motility Disorders

Lower esophageal sphincter – Normal Peristalsis – Failed

Major disorders of peristalsis

All 3 are rare, and treatments are not very effective.

- Absent contractility
  - No peristalsis at all
  - Common in patients with systemic sclerosis (or scleroderma, which is the hardening of the skin and internal organs)
  - Patient often has reflux but not dysphagia or chest pain
- Distal esophageal spasm
  - Contractions at bottom end of esophagus happening too soon
  - Poor peristalsis
- Hypercontractile (jackhammer) esophagus
  - Esophageal muscles contract with too much force

Minor disorders of peristalsis

- Fragmented peristalsis
  - >50% Muscle contractions too short
- Ineffective esophageal motility
  - >50% Swallowing unsuccessful

Lower esophageal sphincter – Failed Peristalsis – Failed

Achalasia (3 types)

Peristalsis & lower esophageal sphincter fail in all 3 types

- Type I (Classic)
  - 100% failed peristalsis
  - May respond to myotomy (surgery that widens the sphincter) but less so to dilation (inflating a balloon to widen the sphincter)
- Type II (with esophageal compression)
  - Some pressurization but no peristalsis
  - All treatments are usually effective
- Type III (spastic)
  - No peristalsis but have uncontrolled muscle contractions instead in >20% swallows
  - Poor response to treatment

Esophagogastric junction outflow obstruction

- Blockage at the lower esophageal sphincter
- Lower esophageal sphincter fails to relax
- Some peristalsis happening

Adapted from Table 1 of Esophageal Motility Disorders

References / Resources

- 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
- Esophageal Motility Disorders – 2020

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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