

Swallowing and Dysphagia

Swallowing is the action that moves food or drinks from the mouth via the esophagus into stomach.

Dysphagia means difficulty swallowing. This difficulty is common in individuals with 22q11.2 deletion syndrome (22q11.2DS) and has also been reported in children with 22q11.2 duplication (22q11.2DupS). Among those with 22q11.2DS, there is no significant difference between late-pre-term and full-term babies with regards to the extent of dysphagia.

Normal Swallowing

Please watch this [video](#) about normal swallowing.

1. Oral Preparatory Stage

- This is the only voluntary stage out of the 4 stages.
- The **teeth** grind the food into smaller pieces.
- The **saliva** moistens and softens the food.
- The food becomes a small round **bolus**.

2. Oral Transit Stage

- The front of the **tongue** rises up towards the roof of the mouth.
- The back of the tongue lowers, and the bolus moves to the back of the mouth.
- The **soft palate** moves upward and prevents food and drinks from going to the nose.

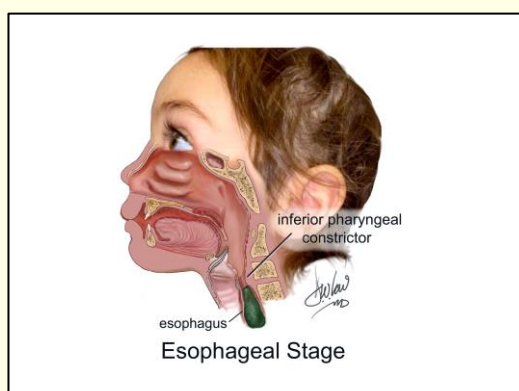
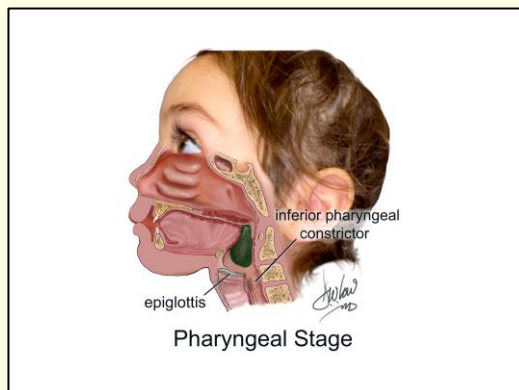
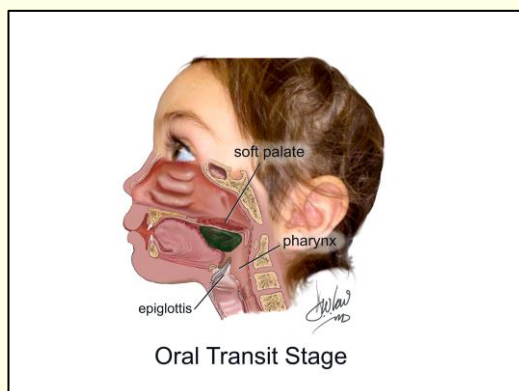
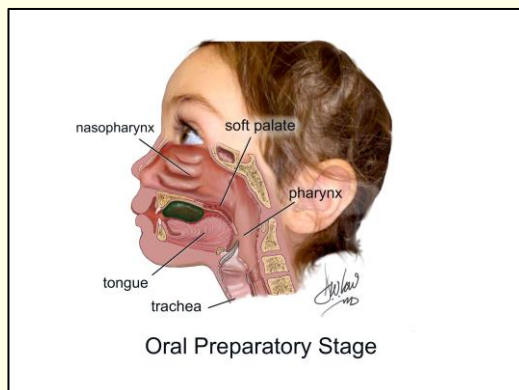
3. Pharyngeal Stage

- In this stage, a few things happen quickly to propel the bolus into the esophagus.
- The **larynx** (voice box, which is above the windpipe) lifts up, and the **epiglottis** flips down to cover the airway.
- Muscles in the throat squeeze the bolus down towards the throat.
- The **inferior pharyngeal constrictor** (also called cricopharyngeus muscle) at the top of the esophagus relaxes. This lets food and drinks go into the **esophagus**.

4. Esophageal Stage

- The inferior pharyngeal constrictor closes to prevent food and drinks from going back up.
- The esophageal muscles contract and relax in a wave motion called **peristalsis**. This moves the bolus down the esophagus towards the stomach.
- The mouth and throat goes back to the original state to get ready for the next swallowing event.

Four stages of swallowing



Swallowing and Dysphagia (continued)

Dysphagia

Swallowing is a quick yet complex motion that requires multiple nerves and muscles to work together the right way at the right time. If any part of the swallowing machinery fails, or if the timing is not precise, swallowing will become difficult or may not happen at all. It is important to talk to the doctor to find out the exact cause of the **dysphagia** (difficulty swallowing) so that the correct treatment can be prescribed.

Some of the Causes of Dysphagia in 22q11.2DS

Dysphagia usually happens due to problems with the **structure or the nerves** involved in swallowing. For example:

- The **timing** of the suck-swallow-breathe pattern is not right.
- Some babies suck in a way that makes food and drinks **collect** at the back of the mouth. They may be messy eaters and have trouble taking in non-slippery food that requires chewing.
- The tongue does not **retract** correctly for swallowing.
- Poor muscle control result in having bits of food **stay** in the mouth without going down.
- **Nasopharyngeal reflux** – food and drinks go up the nose because the soft palate fails to close the area between the mouth and the nose completely. Sometimes the back wall in the pharynx bulges and splashes the bolus up to the nose.
- The inferior pharyngeal constrictor muscle (cricopharyngeus muscle) **projects too much**. It **opens too late and closes too early**, so the bolus gets pushed back up into the pharynx.

Making Swallowing Easier

- Have a **thorough medical check** regarding **heart and palate structures** as well as **swallowing and GI function**.
- **Resolve acid reflux and constipation problems**.
- Feed food and liquids in **small bites**. Larger volumes increase the risk of choking.
- **Thicken liquids** using a thickener approved by the doctor to slow down swallowing and reduce the chances of the liquid from going down the airway.
- **Stay upright** during and after eating
- **Oral motor or feeding therapy** may be a part of the solution to improve swallowing.
- Some medications may help the food move faster through the digestive tract, but be aware of side effects.
- Ask a pharmacist for options for individuals (usually adults) who have problems swallowing pills.
- In severe cases, a **gastric tube (G-tube)** is inserted to deliver nutrients directly to the stomach or the intestines. This feeding bypasses the mouth and esophagus entirely.

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
- [Patterns of Dysphagia and Airway Protection in Infants with 22q11.2-Deletion Syndrome](#) – 2019
- [Dysphagia in children with a 22q11.2 deletion: Unusual pattern found in modified barium swallow](#) – 2000

Websites of medical institutions and professionals

- [Trouble Swallowing](#) (contains helpful videos) – Fauquier ENT