

Swallowing Issues in Infants and Young Children with Prader-Willi Syndrome (PWS)

Children and adults have experienced the concerning feeling that “something going down the wrong pipe” triggering coughing to clear the material. Aspiration is the medical term for this type of choking - food or liquid enters the windpipe (trachea) instead of going down the throat (esophagus) to the stomach which can result in inflammation, infection and long term injury to the airways if the material is not cleared from the airway. A study published by Dr. Salehi and colleagues in 2017¹ reviewed medical records of 10 infants and children under 3 years of age followed at Seattle Children’s Hospital who underwent a total of 23 swallow studies (videofluoroscopic swallow study (VFSS)) to evaluate feeding, choking and respiratory issues. About ½ of the children received only tube feedings for nutritional support with the remainder receiving full or partial oral feeding. 87% of the swallow studies showed evidence of aspiration of liquid into the lungs without outward signs of concern “silent aspiration”. Half of the children were on growth hormone at the time of the testing which did not impact upon the swallow study results. In this small study, there was no difference in the results in the children who were on GH or differences in the genetic subtype of PWS. Swallowing concerns among young children were discussed in a recent publication by Roy and colleagues², including 18 infants/young children with PWS followed at Cook Children’s with swallowing issues noted in all the initial swallow studies and silent aspiration seen in 1/3 of the swallow studies. Several of the young children followed by the Cook Children’s team had a history of lung issues; less than ½ of the children received growth hormone treatment.

A recent study published by Scheimann and colleagues³ suggests that laryngeal clefts may cause some of the swallowing issues seen in PWS. A laryngeal cleft is a birth defect which causes a connection between the larynx or trachea and esophagus and allows food to enter the airway. They reported 3 children with PWS and history of silent aspiration on swallow studies who had a laryngeal cleft found by pediatric otolaryngology (ENT) during airway exam under anesthesia with repair completed followed by improvement in symptoms.

¹Salehi, P., H. J. Stafford, R. P. Glass, et al. 2017. “Silent Aspiration in Infants with Prader-Willi Syndrome Identified by Videofluoroscopic Swallow Study.” *Medicine* **96**, no. 50: e9256.

²Roy, S. M., A. Trejo, J. McReynolds, et al. 2025. “High Rates of Dysphagia and Silent Aspiration in Infants With Prader-Willi Syndrome.” *American Journal of Medical Genetics, Part A* **197**, no. 10: e64121

³Rodrigo, M. L., C. Heubi, E. Chiou, and A. Scheimann. 2024. “Laryngeal Clefts in Prader-Willi Syndrome: Feeding Difficulties and Aspiration Not Always Caused to Hypotonia.” *American Journal of Medical Genetics, Part A* **194**: e63634.