

BECKWITH-WIEDEMANN SYNDROME (BWS)

MACROGLOSSIA AND OBSTRUCTIVE SLEEP APNEA

Beckwith-Wiedemann syndrome (BWS) is a rare disorder involving changes on a region of chromosome 11p15 that influence pre- and postnatal growth. These changes disrupt the normal balance of growth gene expression and lead to the overgrowth seen in patients with BWS. Depending on which parts of the body are affected, children with BWS can have different features. These include **macroglossia**, or an abnormally large tongue. Macroglossia is more common in some genetic/epigenetic types of BWS, but it can affect any patient with BWS, even in those with negative genetic testing.

What is macroglossia?

Macroglossia, or an enlarged tongue, is a common feature in patients with BWS. Macroglossia can result in a longer and/or thicker tongue than other children. In some children with BWS, the tongue may also be larger on one side than the other.

How can macroglossia affect my child?

Macroglossia can cause a variety of issues depending on its severity and presentation. Children with macroglossia may have issues with eating, speaking, or breathing. Some children can have **obstructive sleep apnea (OSA)**, a condition where the tongue blocks the airway when they sleep. This causes pauses in breathing throughout the night that can wake the child up or cause their oxygen levels to drop. If left untreated, macroglossia can also cause issues with jaw development and tooth formation.

What specialists should evaluate my child for macroglossia?

There are a variety of specialists that your child may need to see, depending on the extent of their macroglossia and the clinical manifestations. These may include:

- Pulmonologist/sleep specialist – sleep apnea evaluation
- Plastic surgeon – consideration of jaw formation and possible surgical intervention
- Otolaryngologist – evaluation of the airway, tonsils, and adenoids
- Speech therapist – speech therapy
- Feeding specialist – feeding evaluation and optimization
- Orthodontist – evaluation of tooth and jaw development

How do we evaluate macroglossia in children with BWS?

There are ways to measure the extent of macroglossia and test children for OSA.

- Jaw radiographs can demonstrate the effects of macroglossia on jaw development
- Bronchoscopy is used to visualize the airways and can help determine if the tongue blocks the airway.
- Polysomnography, or sleep study, is used to evaluate for OSA.
- Swallow studies can demonstrate feeding difficulties

Even among children diagnosed with OSA, the actual disruption of breathing can vary significantly. As a result, treatment will depend on the needs of the child. In some cases continuous positive airway pressure (CPAP), which is used to treat OSA in patients with other conditions can be used in BWS. Further research is needed to determine which children with BWS are most likely to benefit from CPAP. In other cases, a tongue reduction may be helpful.

Will my child need a tongue reduction?

Currently, there are no standard criteria to determine if and when a child will need a tongue reduction. Careful evaluation by a multi-disciplinary team of specialists can help determine if a child needs a tongue reduction. Although there are no set criteria, the following factors may be considered:

- If the child is having issues with feeding or speech
- If the child is having difficulty breathing – this can include noisy breathing, snoring, and/or obstructive sleep apnea (OSA)
- If the child is experiencing issues with jaw development or tooth formation
- If the physical appearance of the tongue is distressing to child or parents

Summary

- BWS can cause macroglossia, or abnormally large tongues, in children.
- Macroglossia can cause OSA, disrupting sleep by obstructing the airway.
- Although the exact relationship between macroglossia and OSA in children with BWS is not yet known, we do have reliable techniques for determining the extent of macroglossia and diagnosing OSA.
- There are multiple treatment options to explore for BWS patients with OSA, though further research is needed to fully appreciate the efficacy of each option.

Patient family education materials provide educational information to help individuals and families. You should not rely on this information as professional medical advice or to replace any relationship with your physician or healthcare provider.