

Macroglossia is the abnormal enlargement of the tongue. In rare cases, macroglossia occurs as an isolated finding that is present at birth (congenital). In many cases, macroglossia may occur secondary to a primary disorder that may be either congenital (e.g., Down syndrome or Beckwith-Wiedemann syndrome) or acquired (e.g., as a result of trauma or malignancy). Symptoms and physical findings associated with macroglossia may include noisy, high-pitched breathing (stridor), snoring, and/or feeding difficulties. In some cases, the tongue may protrude from the mouth. When inherited, macroglossia is transmitted as an autosomal dominant genetic trait. Macroglossia is a disorder characterized by a tongue that is large in proportion to other structures in the mouth. In the congenital type of the disorder, protrusion of the tongue from the mouth may interfere with feeding of the infant. Later, talking may be affected. The large size of the tongue may also cause abnormal development of the jaw and teeth, resulting in misaligned or protruding teeth. Ulceration and dying tissue on the tip of the tongue may be other symptoms of the disorder. Isolated autosomal dominant macroglossia is very rare, with about 50 cases reported in the medical literature. The prevalence in other instances depends on the underlying disorder for which the macroglossia is secondary. For example, macroglossia occurs in most cases of Beckwith-Wiedemann syndrome, and the prevalence of that syndrome is estimated at 1 in 17,000 births. Infants born with isolated autosomal dominant macroglossia present with the obvious sign at birth. A family history and physical exam can confirm the diagnosis.