

Congenital hypothyroidism presenting as isolated macroglossia in a term neonate: A case report

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ABSTRACT

Macroglossia is a clinical condition in which the tongue protrudes beyond the teeth or alveolar ridge during resting posture, or there is an impression of a tooth on the lingual border with the mouth open. True macroglossia occurs in hypothyroidism, Beckwith–Wiedemann syndrome, and storage disorders. Apparent or pseudo macroglossia may result from neurological impairment of the tongue or a relatively small buccal cavity as in Pierre Robin syndrome and Down's syndrome. Rarely, macroglossia is inherited as an autosomal dominant condition. A wide variety of primary conditions of the tongue can cause tongue enlargement, e.g., lingual thyroid, cysts, tumors, hemangioma, lymphatic malformations, and myositis. Symptoms associated with macroglossia include noisy breathing, stridor, snoring, and feeding difficulties. Although macroglossia is described in hypothyroidism, there are few publications on macroglossia presenting in the neonatal period. We report a case of congenital hypothyroidism presenting as isolated macroglossia at birth in a term neonate.

Key words: *Congenital hypothyroidism, Macroglossia, Neonate*

Macroglossia is a clinical condition in which the tongue is not completely accommodated by the buccal cavity. Macroglossia is classified as true macroglossia, which is associated with definitive histopathological findings and pseudo macroglossia, which is a relative enlargement of the tongue secondary to a small mandible and without any histological abnormalities. True macroglossia may be primary, characterized by hypertrophy or hyperplasia of the tongue muscles or secondary to infiltration of normal tissue with anomalous elements [1,2].

Common causes of true macroglossia in children are hypothyroidism, lymphangioma, hemangioma, idiopathic hyperplasia, metabolic disorders, and chromosomal abnormalities [3,4]. Secondary macroglossia may also be a manifestation of both benign and malignant space-occupying lesions such as cystic hygroma, cysts in the lingual thyroglossal duct, dermoid cysts, neurofibromatosis, granular cell tumors, lymphoma, and rhabdomyosarcoma. Apparent enlargement of the tongue secondary to micrognathia is seen in Down's syndrome and Pierre-Robin syndrome and occasionally in conditions with underlying neuromuscular dysfunction such as cerebral palsy. The tongue may be displaced upward and forwards by lesions in the floor of the mouth including tumors, Ludwig's angina, ranula, and angioneurotic edema.

We report a case of congenital hypothyroidism presenting as isolated macroglossia at birth in a term neonate.

CASE REPORT

A full-term neonate was referred to our hospital in view of a large protruding tongue noticed at birth. The baby was born to a 27-year-old G2P1L1 mother at 39 weeks gestation by cesarean section (Ind: Cephalo pelvic disproportion and previous cesarean birth). Apgar scores were normal and the baby weighed 3320 g at birth. The baby had mild respiratory distress, and macroglossia was noticed at birth. The antenatal period was uneventful without any history of maternal fever, hypertension, or diabetes. Her thyroid profile was normal. Baby is a non-consanguineous product.

At admission, heart rate was 128/min, capillary refill time 3 s, respiratory rate 74/min, and SaO₂ 94% in room air. The tongue was protruding out, and the baby was unable to retract the tongue into the mouth (Fig. 1). There were no clinical features of Beckwith–Wiedemann syndrome (BWS). Except for macroglossia, there were no other clinical features of hypothyroidism (goiter, dry skin, or an umbilical hernia).

Laboratory investigations showed a hemoglobin of 15.8 g/dl, platelet count of 2,95,000/cu mm, and the leukocyte count of 9300/cu mm. The septic screen was negative (C-reactive protein: 2.1 mg/l). Chest X-ray showed streaky opacities suggestive of wet lungs. Blood culture was taken, and antibiotics (cefotaxime and amikacin) were started. Tachypnea subsided within 48 h. Thyroid profile sent after 48 h of life showed markedly elevated thyroid stimulating hormone (>150 μ IU/ml), low T3 (61 ng/dl) and T4 (4.3 μg/dl) suggestive of hypothyroidism.

X-ray of knee joints showed a small distal femoral epiphysis. The proximal tibial epiphysis was not visualized suggestive of delayed bone age (Fig. 2). Ultrasound of neck showed small echogenic thyroid gland. Isotope scan showed no uptake of radioisotope (Fig. 3). Ultrasound abdomen was normal (no visceromegaly). Bilirubin on day 4 of life was 16.8 mg/dl, with a direct fraction of 0.4 mg/dl. The baby received phototherapy for 2 days, had swallowing difficulty, and was on gavage feeds for the 1st week of life. Biopsy of the thyroid gland was not done as parent's consent could not be obtained.

Levothyroxine (15 µg/kg/day) was started on day 5 of life. The baby had difficulty at latching on the breast and was on gavage feeds. Baby accepted feeds from day 7 of life and was discharged on day 9 of life. Parents were counseled regarding daily thyroxine supplementation and regular follow-up. Hearing screening was normal. At 4 weeks of age, the tongue has reduced in size and thickness (Fig. 4). Thyroid profile was done every 2 weeks for initial 3 months of life and later every month up to 6 months of age. Growth and development were appropriate at 6 months of age.

DISCUSSION

The National Organization of Rare Disorders lists macroglossia as a rare disease, which has a prevalence of fewer than 200,000 affected individuals. The true incidence of macroglossia is unknown and difficult to assess because of its association with many genetic etiologies [4].

There are few publications on macroglossia presenting in the neonatal period. The majority of these publications are small case series or single case reports of macroglossia [3]. Autosomal dominant congenital macroglossia was first reported in 1986 in 2 unrelated families as an isolated finding [5]. Association of macroglossia with neonatal diabetes is also reported [6]. Prada *et al.* published the largest cohort of macroglossia in children and suggested a diagnostic approach [7].

The evaluation of a patient with macroglossia should begin with a thorough history and physical examination, which may allow the recognition of a syndrome of which the enlarged tongue is one component. Assessment of the tongue should include examination for masses and changes in color and consistency. Microscopic examination of tongue tissue is useful in secondary macroglossia and for localized lesions of the tongue [2].



Figure 1: Macroglossia at birth

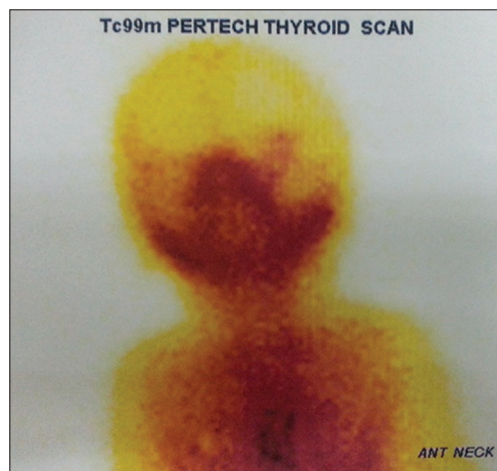


Figure 3: Isotope scan showing absent uptake

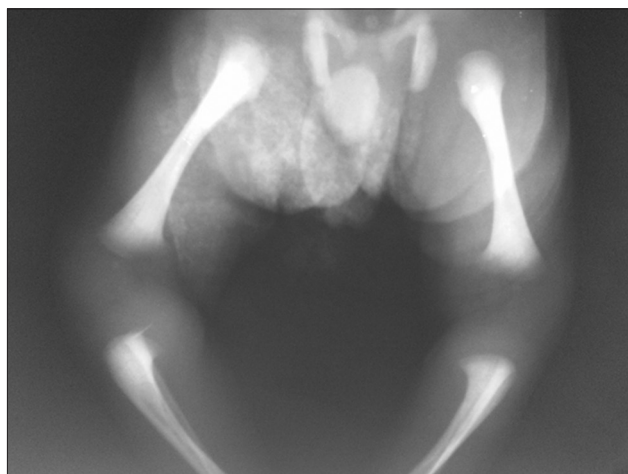


Figure 2: X-ray of knee joints showing absent proximal tibial epiphysis and small distal femoral epiphysis



Figure 4: At 4 weeks of age, the tongue has reduced in size and thickness

Laboratory investigations include thyroid function tests, isotopic imaging of the thyroid gland, abdominal ultrasound, chromosomal studies, molecular studies for BWS, metabolic profile, and urinary mucopolysaccharide assay [8].

Although the clinical course is usually benign for the majority of patients with isolated macroglossia, it causes medical problems independent of the underlying etiology, and potential interventions should be considered to improve the outcome of these patients [9,10]. Complications reported include speech disturbances, swallowing difficulties, drooling, recurrent upper respiratory tract infections, airway obstruction, and mandibular deformities. The most common problems encountered in the neonatal period are airway obstruction and feeding difficulty. Gavage feeding and oxygen supplementation for an average of 2 weeks were reported in neonates with isolated macroglossia and suspected BWS. Some patients, especially in the syndromic group may need prolonged assistance with nutrition and airway maintenance. Surgical correction of macroglossia is usually required when it constitutes a significant cosmetic concern, causes feeding difficulties, speech problems, airway obstruction, excessive tongue desiccation, and ulceration or risk of tongue trauma [9,10].

Our patient had feeding difficulty and required gavage feeds for initial 1 week of age. Baby responded well to thyroxine supplementation. Tongue size and thickness reduced in size by 4 weeks of age and it took 3 months for macroglossia to resolve completely.

CONCLUSION

Congenital hypothyroidism should be ruled out in all cases of macroglossia. A thorough history, physical examination, and

investigations help in identifying the etiology of macroglossia which is essential for prognostication and treatment. Airway obstruction and feeding difficulties need to be taken care which can be life-threatening. Follow-up of these patients to identify speech problems and dental malocclusion is important for timely intervention.

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