CASE REPORT

Unilateral macrostomia in the newborn: a rare congenital anomaly of the oral commissure

Eva Simonse,¹ Bianca Panis,¹ Jamiu O Busari^{2,3}

¹Zuyderland Medisch Centrum, Heerlen, The Netherlands

²Department of Pediatrics, Zuyderland Medical Center, Heerlen, The Netherlands

³Department of Educational Development and Research, Maastricht University, Maastricht, The Netherlands

Correspondence to

Dr Jamiu O Busari,
ja.busari@zuyderland.nl

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SUMMARY

Macrostomia is a rare medical condition, defined as an enlargement of the mouth at the oral commissure. The incidence varies between 1 in 60 000 to 1 in 300 000 live births. Macrostomia is a form of a facial cleft. Macrostomia can present as a unilateral or bilateral anomaly with a partial or complete cleft. Associated anomalies of the surrounding bone, muscle and soft tissue can also be present with or without the presence of a syndrome. Macrostomia results in aesthetic disharmony and also in functional problems. In both cases surgery is the treatment of choice. In cases of macrostomia, additional investigations should be performed to rule out accompanying cardiac and renal anomalies and associated syndromes. A multidisciplinary approach and good collaboration between healthcare providers is essential for optimal care of these patients.

BACKGROUND

Macrostomia is a form of a facial cleft that occurs as an enlargement at one (or both) of the oral commissures of the mouth.¹ Clefts are maxillofacial deformities which develop from disturbances from the union of the embryonic facial processes.² The recognition of macrostomia as a cleft is therefore important for the proper treatment and support of children with these conditions. There are several forms of facial clefts. A primary or true cleft always develops at an early stage of facial development and in four specific areas namely: (1) between the lateronasal and maxillary process (or oro-naso-ocular cleft), (2) between the medionasal and maxillary process (or cleft of the lip), (3) between the maxillary processes (or cleft of the palate) and (4) between the maxillary and mandibular process (or macrostomia). Secondary clefts emerge at a later stage, usually after the embryonic facial processes have been fused. The facial abnormalities associated with secondary clefts arise due to a lack of the normal synergy and harmonious development of the different ossification centres. This can give rise to a break in skeletal, muscular and cutaneous continuity.¹

In this article we present the case of a newborn infant with a unilateral macrostomia. Other names for macrostomia are Tessier cleft number 7 and transverse facial cleft.

CASE PRESENTATION

A newborn male infant was presented to the paediatrician after birth with a unilateral macrostomia, preauricular pit and preauricular skin tags. The pregnancy was complicated with a vanishing twin.

Folic acid was used in prophylactic dose during pregnancy. The child's delivery was uneventful besides a mild fetal distress that required assisted fundal expression. The infant recovered swiftly post partum, and his birth weight was 2905 g between the 5th and 10th centile. On physical examination, several facial anomalies were seen. The most prominent was a lateral cleft lip at the left side of the mouth (figure 1). Also, palpation of the left cheek suggested a possible abnormality of the mandibular joint. The zygomatic muscle was intact. In front of the right ear, four preauricular skin tags were seen and on the left side only one with a pit in front of it (figures 2 and 3). Further physical examination did not yield any additional abnormalities. During the first days of life, feeding difficulties arose and following two episodes of hypoglycaemia (serum glucose 1.5 and 1.9 mmol/L) feeding by a nasogastric tube was initiated. Enteral feeding was facilitated by a special needs feeder and nasogastric tube feeding was discontinued on the fourth day post partum.

The family history was negative for macrostomia, although the mother had a brother and an aunt with spinal dysraphism.

INVESTIGATIONS

Ocular investigation did not reveal any congenital anomalies of the eyes. Renal ultrasound scan was performed which showed a normal anatomy of the renal system. Cardiac ultrasound scan did not reveal any structural abnormalities. Genetic screening was performed which showed a normal copy number variation profile and normal whole exome sequencing.

DIFFERENTIAL DIAGNOSIS

- Hemifacial microsomia
- Treacher Collins syndrome

TREATMENT

The boy was referred to the department of oral and maxillofacial surgery for surgical correction. The literature on macrostomia describes various methods of surgical correction. The ideal outcome, however, is dependent on the correct placement of scars between different aesthetic subunits of the face. For example, closure of the commissure can be achieved using triangular mucosal flaps. This method transposes the skin into the corner of the mouth, giving it an unnatural appearance. Vermilion square flap is another method that tends to leave a scar at the vermilion-cutaneous junction of the commissure. Overlapping myoplasty, another surgical method, provides a natural overlap of the upper lip over the lower lip. Finally, simple straight

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Figure 1 A lateral cleft lip at the left side of the mouth.

line closures or improper approximation of muscle bundles might result in fish mouth deformity.²

OUTCOME AND FOLLOW-UP

Evaluation of the child's growth and drinking will be necessary. Surgical repair of the mouth was scheduled at the age of 1 year.

DISCUSSION

Development of the face occurs through differentiation and migration of the first and second branchial arches during week 5–8 of embryonic development. Macrostomia occurs when there is an abnormal fusion of the orotragal line, which is an embryonic fusion line extending from the lateral commissure to the tragus of the ear.³ Macrostomia is rare facial abnormality with a varying incidence of 1 in 60 000 to 1 in 300 000 live births.⁴ In contrast, the incidence of the more familiar isolated cleft lip and cleft palate is 1 in 1000 and 0.5 in 1000 live births, respectively.³ Macrostomia is more common unilaterally, with only 10–20% bilaterally. In the unilateral cases, the right side is most commonly affected. The cleft may be partial or complete extending up to the tragus.⁴ However, most clefts do not extend beyond the anterior border of the masseter.⁵ Men are more commonly affected than women.^{3 4}

Macrostomia can occur in isolation or be associated with other congenital abnormalities or syndromes, like Treacher Collins syndrome and hemifacial microsomia.⁴ Nearly 25% of unilateral macrostomia patients have hemifacial microsomia.³ Totally isolated transverse clefts are however rare.⁵ Macrostomia is often accompanied by defects of the surrounding bone, muscle and soft tissue derived from the first and second branchial arch. Bone anomalies usually include mandible hypoplasia, maxillary duplication and zygomatic bone anomalies. About 60–70% of patients have abnormalities of the external and

Figure 2 Right ear: four preauricular skin tags.

Figure 3 Left ear: one preauricular skin tag with a pit in front of it.

middle ear and preauricular skin and cartilaginous tags are the most common. Macrostomia is rarely associated with anomalies of the parotid gland and ducts, the trigeminal nerve and facial nerve.³ Meanwhile, congenital anomalies such as polydactyly, cardiac and renal anomalies have been reported.⁶

Macrostomia causes both aesthetic as well as functional problems of the oral cavity. These include feeding difficulties, drooling, speech incoherence and difficulty in blowing and the aim of surgical treatment is to correct both problems. In 2010, Bütow and Botha provided a classification for the Tessier 7 clefts and the differences in their severity. In order to achieve the best aesthetic results, these subclassifications need to be considered prudently and the surgery tailored appropriately.² Early operative closure has been associated with normal speech development. Timing of the operation, however, is usually determined by the presence (or absence) of severe orofacial deformities.³

In this report, we present a case of a newborn male infant with unilateral macrostomia, accompanied with preauricular skin tags and a preauricular pit. Feeding difficulties were the most prominent problems during the first days of life, for which the use of a special needs feeder was required. Genetic testing was performed to rule out hemifacial microsomia or Treacher Collins syndrome. Hemifacial macrosomia is a rare disorder involving abnormalities of the ears, facial soft tissue deficiency, nerve palsies and maxillary and mandibular hypoplasia. In total 25% of unilateral macrostomia patients have hemifacial macrosomia. Bilateral macrostomia, however, is very rarely associated with hemifacial macrosomia.³ Treacher Collins syndrome is an

Learning points

- ▶ Macrostomia is a primary cleft, defined as an enlargement of the mouth at the oral commissure.
- ▶ It is a rare clinical condition presenting at birth, mostly unilateral and often associated with anomalies of surrounding bone, muscle and soft tissue.
- ▶ It can occur in isolation or in combination with a clinical syndrome.
- ▶ Aesthetic disharmony and functional problems caused by macrostomia can be corrected by surgery.
- ▶ A multidisciplinary approach and good collaboration among healthcare providers is mandatory for an optimal management and treatment of the condition.

autosomal dominant congenital disorder characterised by the absence or deficiency of the malar bone, flattening of the cheeks and eyelid coloboma. In this congenital condition the deviations are usually bilateral.¹ Surgical repair would be necessary to correct the aesthetic disharmony of the face and to improve any functional problems of the mouth this patient may have. In our patient surgical correction was planned at the age of 1 year. Treatment and coaching of macrostomia requires a multifactorial approach and good collaboration between local healthcare services and the specialist centres is mandatory.

Competing interests None declared.

Patient consent Obtained.

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