Congenital partial hypoplasia of the lower lip: a rare form of 28-29 Tessier cleft?

Olina RIOS[1], Virginie Lafontaine[2], Cyril Debortoli[1], Charles Savoldelli[1], and Beatrice Morand[2]

[1]University Hospital Centre Nice
[2]Grenoble Alpes University Hospital

August 29, 2024

Congenital partial hypoplasia of the lower lip: a rare form of 28-29 Tessier cleft?

Olina Rios (a), Virginie Lafontaine (b), Cyril Debortoli (a), Charles Savoldelli (a), Beatrice Morand (b)

Corresponding author: rios.o@chu-nice.fr

(a) Department of Oral and Maxillo-facial surgery, Head and Neck Institute, University Hospital Center of Nice, 06100 Nice, France

(b) Department of Maxillo-facial and Plastic surgery, University Hospital Center of Grenoble-Alpes, 38000 Grenoble, France

Key Clinical Message:

We aim to describe a case of congenital partial hypoplasia of the lower lip in a 2-year-old child. As this anomaly was associated with other oral anomalies, we explored the possibility of a modified 28-29 Tessier cleft. However, other hypotheses were proposed as we may be dealing with an unknown mechanism.

Keywords: labial hypoplasia, lower cleft lip, craniofacial abnormalities, Tessier clefts.

Introduction:

First described by Couronne in 1819, cleft mandible and/or lower lip is one of the rarest craniofacial clefts with less than 100 cases reported in the literature [1,2]. In almost all cases, the midline location of the lip and/or mandible has been noted. According to Tessier's classification of 1976, cleft lip and/or mandible midline is defined as no. 30.

The paramedian lower cleft lip is even rarer, with only 5 cases described. However, in these cases the defect occurs in a very specific part of the lower lip and/or mandible. In the case described below, there was a defect of the entire lateral third of the lower lip. Therefore, it could not be formally associated with a 28-29 Tessier cleft and may not have the same origin.

Case history:

We aim to report the case of a 2-year-old boy who was born with hypotrophy of the lateral third of the lower lip. To the best of our knowledge, this type of anomaly has not been previously reported in the literature. Written informed consent was obtained from the parents of the child. This case report was submitted with their consent and with the approval of the Ethics Committee of the Face and Neck Institute.

The patient was first referred to the craniofacial cleft team at the Grenoble University Hospital Centre at the age of 5 months. He was the first child of healthy parents with no consanguinity. The pregnancy was

spontaneous, and the mother had a previous miscarriage. There was a family history of congenital dental anomalies: the mother's aunt had only milk teeth and the mother of our patient was missing two permanent teeth (34 and 44). There was no exposure to teratogens or x-rays during pregnancy. The mother was 30 years old at the time of delivery. The boy was born at term by normal vaginal delivery. Birth weight was 3215 g with a cranial circumference of 35 mm. The Apgar score was 10/10. He didn't have any other anomalies.

Investigations:

Cardiological examination and echocardiography were normal. Feeding was normal, although the parents noted a tendency to drool due to difficulties in achieving complete lip closure. Growth was normal. Examination revealed hypoplasia of the right lateral third of the lower lip **(Figure 1A).** There was no facial paralysis and the smile appeared symmetrical. Intra-oral examination revealed a gingival notch, a mucosal bridle and the median labial frenulum was deviated on the right side **(Figure 1B).** Lip eversion was possible. There were no abnormal findings on the tongue, oral mucosa, pharynx or tonsils, but a kystic lesion was visible on the anterior part of the uvula. No abnormality was seen on the rest of the body. Circumscriptions and amputations of digits or limbs weren't noticed. As the development was normal, a simple follow-up with an annual visit was chosen.

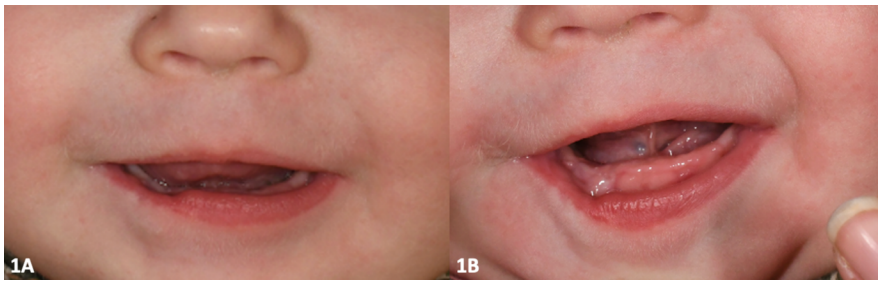

Figure 1. View of the lower lip at the age of 5 months (1A). Mucosal bridle (1B)

At 27 months, tooth 83 wasn't in place and tooth 82 was abnormal (while 72 and 73 were on the arch). The orthopantomogram made for the consultation showed good mandibular continuity but the absence of teeth nos. 42, 43 (whereas 32 and 33 were visible). This could be related to the known history of missing teeth, but as this anomaly is unilateral, it could also be related to the lip deformity. **(Figure 2)**

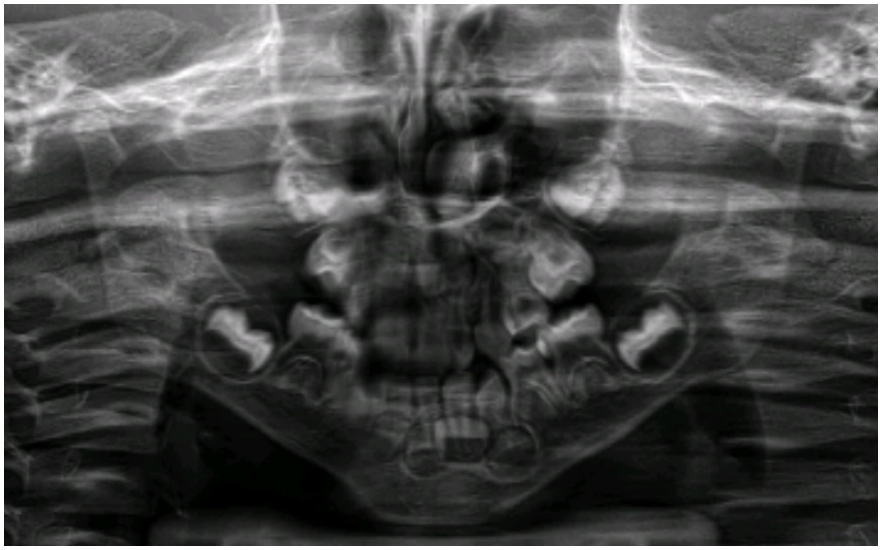

Figure 2 : Orthopantomogram at 27 months.

Treatment:

The management of this labial hypoplasia can be discussed, but the less invasive technique remains lipofilling with autologous fat graft or hyaluronic acid. In the meantime, follow-up is required to monitor speech development and eruption of the teeth.

Discussion:

The presence of a gingival notch, a mucosal bridle, and difficulty erupting teeth led us to believe that this may be an incomplete Tessier cleft 28-29. There are only five articles in the literature reporting a paramedian cleft of the lower lip [3–7] from localised lower lip hypoplasia associated with bilateral upper lip and palate clefts (Oka et al. 1983), to complete lower lip clefts without mandibular clefts (Hassanpour et al. 2018, Chauvel-Picard et al. 2018, Ghorpade et al. 2023), to incomplete lower lip cleft with mandibular defect (Morritt et al. 2007). In all these cases, the lip defect is localised on a thin part of the lower lip without extension to the commissure. Regarding the anatomical differences between a lower cleft lip defect and this total external third lower lip hypoplasia, we believe that this may be the first congenital partial lower lip hypoplasia reported in the literature.

For Chisholm et al [8], atrophic skin conditions are caused by abnormalities in the dermis and/or subcutaneous tissue. Although the epidermis may be thin and atrophic, there must be a loss of substance in the subepidermal tissue for the skin to look and feel atrophic. In this case, we can assume that both the orbicularis oris and the depressor labii inferioris are hypotrophic but remain functional

(Figure 3A and 3B).

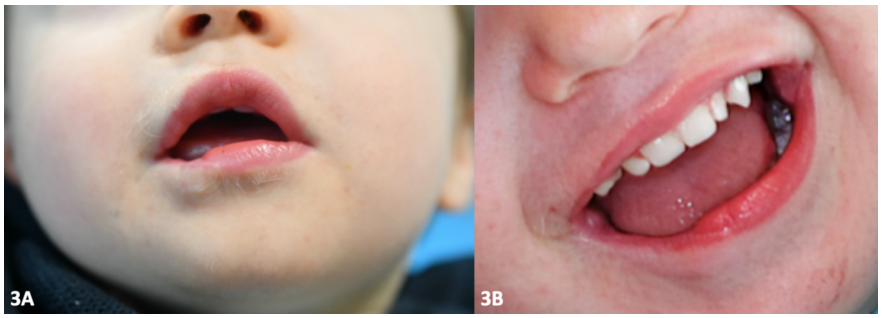

Figure 3. Evolution of the hypotrophy of the lower lip at 24 months (3A). Smile at 27 months showing no asymmetry (3B).

While much research has been done to understand and explain the embryological mechanism of upper cleft lip, the aetiology of paramedian lower cleft lip is still unclear and there is no consensus on its embryogenesis.

There are several hypotheses. The first is a fusion defect between the mandibular prominences of the first branchial arches. According to Warbrick [9], it is due to three depressions in the mandibular process during the foetal period. He suggested that a disturbance in the embryological formation of the mandibular process could lead to a disturbance in one of these depressions, resulting in paramedian defects.

Secondly, clefts may result from failure of mesodermal migration and penetrance. In addition, growth centres within the developing mandible may be required for formation. Partial or complete failure of growth centre differentiation may contribute to mandibular defects, rather than a simple failure of mandibular protrusions to fuse at the midline[10].

Thirdly, there is a belief that intrauterine trauma may be responsible for cleft lip and palate [5,11]. In 2018, Chauvel-Picard et al observed a paramedian cleft of the lower lip in a child whose mother had undergone fetal

reduction for a multiple pregnancy. They believed that the introduction of a transabdominal or transvaginal needle could cause craniofacial abnormalities. Burton et al studied 394 fetuses and infants whose mothers had undergone chorionic villus sampling. Thirteen (3.3%) had major congenital anomalies, including four with missing limbs or parts of limbs and three with cleft lip, with or without cleft palate [12]. In our case, the mother didn't experience any trauma or fetal reduction. However, it is important to note that our patient was sucking his thumb on the first and second fetal scan. Prolonged intrauterine lip pressure could explain this anomaly, but it would probably have been seen in many other babies.

Fourthly, two main syndromes could explain this anomaly. Amniotic band syndrome and Van der Woude syndrome. The amniotic band syndrome is known to be responsible for many forms of clefting. It wasn't considered here because there were no ringstrings and no amputations of digits or limbs. Van der Woude syndrome involves paramedian pits or clefts. This syndrome is highly variable and has been reported as lip pits in combination with a cleft. However, the present case has neither the characteristic lip pits nor the cleft. It has therefore been excluded.

A fifth hypothesis could be a vascular origin. Ischaemia of the stapedial arterial system, which arises from the second pharyngeal arch and is responsible for the vascularisation of the face during embryogenesis, could eventually lead to facial malformation. In 1973, Poswillo attempted to demonstrate this vascular theory in an animal model by injecting triazene into the carotid artery, causing a haematoma in the distribution area of the stapedial artery and eventually leading to the formation of macrostomia and hemifacial microsomia. This theory remained controversial and was criticised in 1995 by Louryan et al.[13]

Our case has many similarities with cases of paramedian inferior cleft lip, but the anatomical analysis of the lip deformity led us to believe that it could not be formally classified as a 28-29 Tessier cleft. Thus, the mechanism may be completely novel.

Conclusion:

This may be the only report of a congenital partial hypoplasia of the lower lip that does not quite fit the Tessier classification, as it does not have the shape of a common cleft. However, the associated anomalies (mucosal bridle, gingival notch, difficulty in eruption of teeth) can be seen in usual cleft lips and suggest a longer follow-up of this young patient to evaluate speech and dentition development. Surgical lip correction will certainly be performed by minimally invasive lipofilling with autologous fat graft or hyaluronic acid.

Author contributions

Olina Rios: Conceptualization; data curation; methodology; writing – original draft; writing – review and editing.**Virginie Lafontaine:** Supervision; visualization; methodology.**Cyril Debortoli:** Visualization; methodology. **Charles Savoldelli:** Supervision; visualization; methodology. **Beatrice Morand:** Project administration; data curation; methodology; writing – review and editing; validation.

Acknowledgments

None.

Funding information

The study had no financial support.

Conflict of interest statement

None.

Data availability statement

The data supporting the findings of the present study are available from the corresponding author upon request.

Ethics statement

This case report was submitted with the approval of the Ethics Committee of the Head and Neck Institute.

Consent

Written informed consent was obtained from the patient's parents to publish this report in accordance with the journal's patient consent policy.

ORCID

Olina Rios https://orcid.org/0000-0002-7537-6112

References:

1. Lou Q, Wang G, Cai M, Chen Y, Wu Y. Median cleft of the lower lip and mandible: Clinical experience and surgical treatment. *Journal of Craniofacial Surgery* . 2019;30(3):e238-e241. doi:10.1097/SCS.0000000000051912. Liu W, Ma L, Zhang S, Zhao T. Clinical correction of complete median cleft of the mandible and lower lip: A 17-Year Follow-Up of a Case Report With Literature Review.*The Cleft Palate-Craniofacial Journal* . 2021;58(12):1577-1584. doi:10.1177/10556656219901703. Oka M, Shinohara M, Kubo K, Torigoe Y, Koguchi H. A bilateral total cleft of lip and hard and soft palate and transverse cleft face with an atypical paramedian defect of the lower lip and lower jaw on the right side. A case report of a new syndrome?*Journal of Maxillofacial Surgery* . 1983;11(5):232-235. doi:10.1016/s0301-0503(83)80054-x4. Morritt DG, Goodacre TE, Wall SA. A paramedian cleft of the lower Lip. *Journal of Craniofacial Surgery* . 2007;18(3):704-706. doi:10.1097/scs.0b013e318052e2ce5. Hassanpour SE, Shirazi FH, Karim F. Lateral Lower Lip Cleft: A report of a rare craniofacial cleft in a child. *Journal of Craniofacial Surgery* . 2018;29(7):e656-e658. doi:10.1097/SCS.0000000000046696. Chauvel-Picard J, Gleizal A. Para commissural cleft of the lower lip: A first case described in the literature. *Journal of Stomatology, Oral and Maxillofacial Surgery* . 2018;119(4):288-290. doi:10.1016/j.jormas.2018.04.0037. Ghorpade SS, Shetty PN, Bonanthaya K, Rao D. Isolated paramedian cleft of lower lip: A rare entity. *The Cleft Palate Craniofacial Journal* . Published online July 13, 2023:10556656231185971. doi:10.1177/105566562311859718. Chisholm C, Miedler J, Etufugh CN, Ghali F, Cockerell CJ. Unusual and recently described cutaneous atrophic disorders: Recently described cutaneous atrophic disorders. *International Journal of Dermatology* . 2011;50(12):1506-1517. doi:10.1111/j.1365-4632.2011.04938.x9. Warbrick, J. G. Remarks on the aetiology of congenital bilateral fistulae of lower lip. *British Journal of Plastic Surgery. 4* . 1952.10. Tanna N, Wan DC, Perry AD, Kawamoto HK, Bradley JP. Paramedian mandibular cleft: revisiting the Tessier classification. *Journal of Craniofacial Surgery* . 2012;23(1):e38-40. doi:10.1097/SCS.0b013e318241db5811. Chauvel-Picard J, Massardier J, Gleizal A. Could fetal reduction induce facial cleft? Report of a case. *British Journal of Oral and Maxillofacial Surgery* . 2018;56(6):543-545. doi:10.1016/j.bjoms.2018.05.00912. Burton BK, Schulz CJ, Burd LI. Limb anomalies associated with chorionic villus sampling. *Obstetrics & Gynecology* . 1992;79(5 (Pt 1)):726-730.13. Louryan S. Contribution and limitations of experimental models to the understanding of certain abnormalities of the face and ear. *Bulletin et Memoires de l'Academie Royale de Medecine de Belgique* . 1995;150(7-9):320-326; discussion 327.