

Case Report

Bilateral microform cleft lip

David Pace, Simon Attard-Montalto, Victor Grech

Abstract

Microform cleft lip (MCL), also called congenital healed cleft lip or cleft lip “frustré”, is a rare congenital anomaly. MCL has been described as having the characteristic appearance of a typical cleft lip which has been corrected *in utero*. We present a girl with bilateral microform cleft lip associated with a preauricular sinus and bilateral camptodactyly.

Introduction

Microform cleft lip (MCL) is an uncommon congenital anomaly. Bilateral microform cleft lip is even more unusual. MCL has been previously associated with several anomalies and we here present a girl with bilateral MCL, a right preauricular sinus and camptodactyly of both fifth fingers.

Case report

A female neonate was born by normal vaginal delivery at 38 weeks' gestation weighing 1925 g with Apgar scores of 9 at 1 minute and 9 at 5 minutes. She was the first child born to healthy unrelated Maltese parents. Neonatal examination revealed bilateral paramedian scars above the upper lip indicative of MCL (Figure 1). The scars extended from the nostrils into the vermilion border. No deformity of the nostrils was present and the palate was not cleft. She had a right preauricular sinus, bilateral camptodactyly of the little fingers and a single crease on the left fifth digit. An echocardiogram and abdominal ultrasound did not reveal any abnormality and the karyotype was normal. Her father has had bilateral cleft lip repair and also has a preauricular sinus. She is now 5 years old and is growing and developing normally.

Keywords

Cleft lip/classification, case report, cutaneous fistula, ear, external/abnormalities

David Pace* MRCPCH

Department of Paediatrics
St Luke's Hospital, Guardamangia, Malta
Email: dpace@mail.global.net.mt

Simon Attard-Montalto MD, FRCPCH

Department of Paediatrics
St Luke's Hospital, Guardamangia, Malta

Victor Grech PhD, MRCPCH

Department of Paediatrics
St Luke's Hospital, Guardamangia, Malta

*corresponding author



Figure 1: Bilateral paramedian scars can be seen extending from the nostrils to the upper lip (arrows)

Discussion

The basic morphology of the face is created between the fourth and tenth week *in utero* by the development and fusion of the frontonasal process plus the two maxillary swellings and two mandibular swellings of the first pharyngeal arches.¹ It is thought that MCL may result from either partial failure in the mechanism of fusion of the frontonasal and maxillary processes before week seven of embryonic life, or otherwise from spontaneous late foetal repair of an open cleft lip. MCL is frequently associated with an ipsilateral notch in the vermilion border and a collapsed nostril. Our patient had bilateral vermilion border notches but lacked nostril abnormalities.

Family members of individuals with MCL may not only have MCL but also cleft lip or palate, as in our case.² Furthermore, parents of individuals with open clefts may have subtle nasal deformities and/or microform cleft lip.³ The features described in this case report partially satisfy those found in the Branchiooculofacial Syndrome and Van der Woude Syndrome, both autosomal dominant conditions that are associated with clefting.^{4,5}

MCL is a variant of cleft lip and is likewise more common in males.² The most frequent orofacial anomalies are cleft lip (CL) and cleft palate (CP), with cleft lip with or without cleft palate (CL/P) being a distinct entity from isolated CP.⁶ CL/P and CP may occur in isolation or as part of various syndromes. Both CP and CL/P are caused by a complex interplay between genetic and environmental factors with a significant familial contribution which, however, does not follow a clear Mendelian pattern of inheritance.⁶ The recurrence risk of CL/P in subsequently born children has been estimated to be 4% if one parent or one child is affected, 17% if one parent and one child have it, and 9% if two children have it.⁷ The recurrence risk of cleft palate is 6% if one parent has it, 2% if only one child is affected, and 15% if one parent and a child are affected.⁷

MCL appears to be very rare, since the only reported cohort in the literature gives a rate of 0.06/10,000 live births (95% CL: 0.04-0.09/10,000 live births; n=25). Bilateral MCL is even rarer, and in this only and largest series reported to date, only one male had bilateral MCL.² A possible explanation for the very low incidence of MCL could be due to under-reporting since isolated MCL represents a minimal type of cleft lip which in most cases might be clinically insignificant, similar to spina bifida occulta.

However MCL might be associated with other major congenital anomalies similar to children with open clefts of both lip and/or palate who have a higher incidence of anomalies of the skeleton, the central nervous system and the heart.⁸⁻¹⁰ The

only reported series of MCL documented five other congenital anomalies which included two cases with hydrocephalus, two cases of the VACTERL (vertebral anomalies, anal atresia, cardiac malformations, tracheo-oesophageal fistula, renal anomalies and limb deformities) association and one infant with a cleft following the junction of the frontonasal and maxillary processes and associated with a coloboma of the lower eyelid (corresponding to an atypical oblique facial cleft type 3 of Tessier) and a single umbilical artery. The majority of MCL were on the left side and involved males.² An association between unilateral MCL and coarctation of the aorta has also been described in another Maltese infant.¹¹

Conclusion

Infants born with microform cleft lip should be screened for other associated congenital anomalies. This is the first report of a girl with bilateral MCL who, however, did not have any other associated major abnormalities. It may be that the father and daughter suffer from the same condition with a variable degree of expression.

References

1. Larsen WJ. Development of the Head and Neck. In: Schmitt WR, Otway M, Bowman-Schulman E, editors. Human Embryology. New York: Churchill Livingstone; 1993. p. 309-40.
2. Castilla EE, Martinez-Frias ML. Congenital healed cleft lip. Am J Med Genet. 1995; 58:106-12.
3. Sigler A, Ontiveros DS. Nasal deformity and microform cleft lip in parents of patients with cleft lip. Cleft Palate Craniofac J. 1999;36(2):139-43.
4. Hall BD, deLorimier A, Foster LH. A new syndrome of hemangiomas of the branchial clefts, lip pseudoclefts, and unusual facial appearance. Am J Med Genet. 1983;14:135-8.
5. Ranta R, Rintala AE. Correlations between microforms of the Van der Woude syndrome and cleft palate. Cleft Palate J. 1983; 20:158-62.
6. Weinberg SM, Neiswanger K, Martin RA, Mooney MP, Kane AA, Wenger SL, et al. The Pittsburgh Oral-Facial Cleft study: expanding the cleft phenotype. Background and justification. Cleft Palate Craniofac J. 2006;43(1):7-20.
7. Curtis EJ, Fraser FC, Warburton D. Congenital cleft lip and palate. Am J Dis Child. 1961;102: 853-7.
8. Milerad J, Larson O, Hagberg C, Ideberg M. Associated malformations in infants with cleft lip and palate: a prospective, population-based study. Pediatrics. 1997; 100(2 Pt 1):180-6.
9. Stoll C, Alembik Y, Dott B, Roth MP. Associated malformations in cases with oral clefts. Cleft Palate Craniofac J. 2000; 37(1):41-7.
10. Natsume N, Niimi T, Furukawa H, Kawai T, Ogi N, Suzuki Y, et al. Survey of congenital anomalies associated with cleft lip and/or palate in 701,181 Japanese people. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2001; 91(2):157-61.
11. Grech V, Lia A, Mifsud A. Congenital heart disease in a patient with microform cleft lip. Cleft Palate Craniofac J. 2000; 37(6): 596-7.