Cleft lip and palate malformations: essential knowledge for the general practitioner

Giaquinto-Cilliers MGC, Plastic Surgeon and Head of Department; **Potgieter MD,** MBChB
Links DA, MBChB; **Van Schalkwyk GI,** MBChB
Department of Plastic Surgery and Burns Unit, Kimberley Hospital Complex, Northern Cape
Correspondence to: Magnus Potgieter, e-mail: mdpotgieter.article@gmail.com
Keywords: cleft lip and palate, congenital malformations

Abstract

Cleft lip and palate malformations are a common group of congenital abnormalities, and are therefore frequently encountered by the general practitioner, who is often the primary coordinator in the management of these patients. This is especially true in a South African setting, where specialist treatment is not always readily available. An incomplete understanding of the multiple components and complexities of the condition often results in unsatisfactory patient outcomes. This article aims to improve understanding of the condition and demonstrates how a multidisciplinary team approach is essential to successfully manage these patients. The roles of the different team members are described, and include the dentist, orthodontist, paediatrician, geneticist, speech therapist, dietitian, otorhinolaryngologist, as well as plastic and reconstructive and/or maxillofacial surgeon. A flow diagram with the various role players and the timing of their interventions has been designed to simplify the referral pathway. The authors aim to assist the general practitioner with the appropriate referral of patients with cleft lip and palate.

Ⓟ **Peer reviewed. (Submitted: 2013-03-31. Accepted: 2013-06-14) © SAAFP** **S Afr Fam Pract 2013;55(6):533-537**

Introduction

Cleft lip and palate malformations are a common group of congenital abnormalities, and are therefore likely to be encountered in general practice.[1] The incidence varies according to geographical location, ethnicity and socio-economic status, but is estimated to be around one per 1 000 births.[1] The incidence of the various deformities differs significantly in different population groups in South Africa. Butow et al found that combined cleft palate, alveolus and lip was the most common variant of the cleft spectrum, and that it was more common in the Caucasian patients (43.4%) in the study group vs. 29.6% (the African patients). The most common cleft defect in African patients was the cleft palate at 43.5%, which was only recorded in 35% of the Caucasian patients. Isolated cleft lip was found in 5.9% of the study group for Caucasians, and 4.5% for Africans. The remainder of the statistics consisted of combinations with other defects. Caucasian males were affected more often (58.2%) than their female counterparts, while clefts in African females were more common (54.9%) than in males.[2] A study in the Western Cape found that a high incidence was noted in coloured patients, while incidences in Caucasian and African patients were similar to figures reported from other countries.[3]

The condition is often viewed by parents and healthcare workers as a purely cosmetic affliction. The multiple components and complexities of the condition are sometimes not fully understood, and therefore neglected to the detriment of the child. Different cultural views of the malformation may also lead to inappropriate and/or delayed management. Early diagnosis and referral can help to ensure an optimum functional and cosmetic result for the patient. A multidisciplinary team is essential since no single discipline can fully manage these patients.

A basic understanding of the holistic management of these patients is important for the general practitioner (GP), whose role can range from simply informing the patient and family of what to expect, to acting as the primary coordinator within the multidisciplinary team. The determining factors in this regard include the knowledge of the practitioner and the availability of local resources. In this article, we aim to equip the GP with the essential knowledge required to play an effective role in this process, whatever his or her degree of involvement.

Aetiology

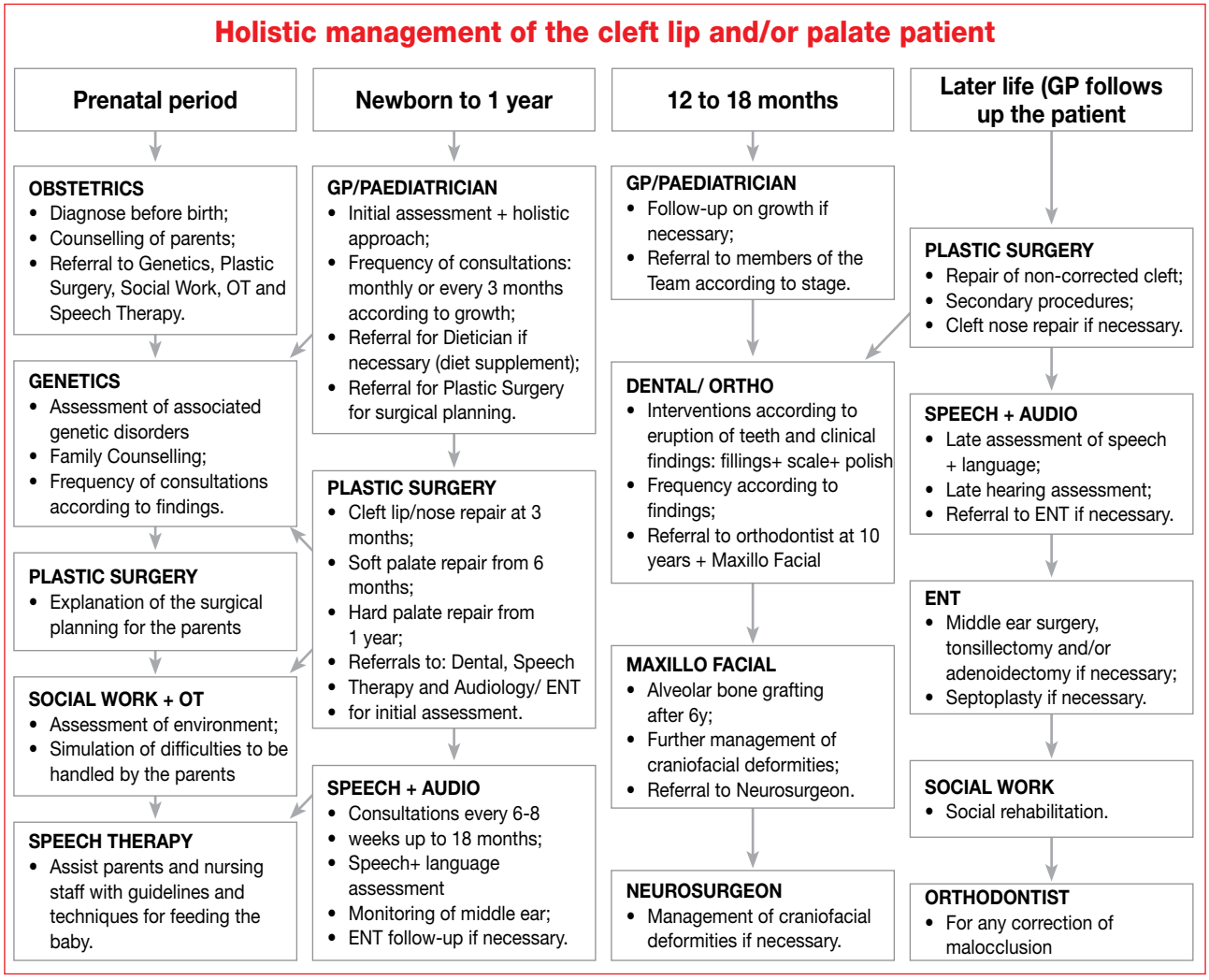
Over 300 genetic syndromes have been identified in association with cleft lip and palate malformations.[1] Aside from purely genetic factors, many environmental risk factors and gene-environment interactions have been found to play a role in the aetiology of clefts. Maternal smoking, even in the preconception period, has been found to be a potential

risk factor. Maternal alcohol consumption has also been suggested as a risk factor, but the evidence has been less consistent. (Data suggest that "binge" drinking patterns increase the risk more than more regular intake). It has been suggested that nutritional factors, such as folate deficiency, increase the risk, based on both observational studies and interventional trials that used folate supplementation to prevent recurrences in families. However, data on the efficacy of folate supplementation in preventing cleft lip and palate malformations remain inconclusive. Finally, some specific teratogens, such as valproic acid, have been shown to be associated with cleft lip and palate malformations.[4]

Overview of management

The condition requires multiple surgical procedures from birth to adulthood, as well as frequent outpatient visits. These are usually handled by plastic and maxillofacial surgeons. Many patients suffer from dental abnormalities and often need to be seen by a dentist and orthodontist.[5] The otorhinolaryngologist and audiologist are an essential part of the team as these children often display hearing problems and recurrent middle-ear effusions because of velopharyngeal insufficiency.[6] A speech therapist is frequently involved in the feeding and rehabilitation of these children. Referral to a psychologist, social worker and even a psychiatrist needs to be considered.[7] The geneticist and paediatrician also play an important role in the management of these patients owing to a high incidence of associated abnormalities and underlying genetic syndromes.[1]

Figure 1 is a suggested strategy for the management and referral of these patients at various stages. However, it is important to recognise that details may vary depending on available resources and the structure of local health services. Therefore, the rest of this paper is dedicated to providing an overview of the role played by each member of the multidisciplinary team. With this knowledge, the GP should have no difficulty in devising a contextually optimal approach.



ENT: ear, nose and throat, GP: general practitioner, OT: occupational therapy

Figure 1: Holistic management of a patient with a cleft lip and/or palate

The role of the obstetrician

The obstetrician is usually the first doctor to have contact with a baby with cleft lip and palate malformations, whether it is prenatally or at birth. His or her role is to identify patients at risk, e.g. a history of syndromic children in the family, and poor attendance of antenatal visits. The antenatal follow-up is often poor in the South African context because rural towns do not have access to medical care. The diagnosis of the intrauterine baby with cleft lip and palate malformations rests with the obstetrician, or often the GP. It is then his or her responsibility to initiate counselling, and to start the necessary referrals to other team members, as will be discussed in the rest of the paper.

The role of the speech therapist and dietitian

The role of the speech therapist and dietitian overlap to an extent, and will therefore be discussed under one heading. The initial referral can take place preoperatively, when feeding is the most important issue. The focus should be on establishing adequate nutrition as soon as possible. This is more important than the feeding method used, and will vary depending on the preferences of the mother and the nature of the deformity. The choice of feeding method may also change after the initial repair is carried out. It is important to remember that children with cleft lip and palate malformations may suffer from associated abnormalities, as well as developmental delays. Therefore, feeding problems may persist, even after repair of the defect, and follow-up with the speech therapist and dietitian is indicated.[8]

Regarding speech, data suggest that approximately 75% of patients with repaired cleft lip and palate malformations will require long-term intervention to achieve adequate speech.[6] Even in the context of a dedicated family and intervention by a speech therapist from an early age, some patients will have residual speech abnormalities.[9] The GP's role is to routinely screen the patient for any signs of delayed speech development and abnormal quality of speech, such as nasal speech. Prompt referral is indicated if this is detected at any stage.

The role of the paediatrician and geneticist

Data regarding the incidence of associated anomalies in patients with cleft lip and palate malformations are inconsistent, with centres reporting rates of between 3% and 63%.[10] Data further indicate that associated anomalies are more common in patients with both cleft lips and palates than in those with cleft lips alone.[10] A large prospective study conducted in Sweden found that abnormalities of the limbs and vertebra were the most common association, accounting for 33% of all associated abnormalities.[11] Cardiac defects were the second most common. It was found in at least one sample that an underlying genetic syndrome or association was present in up to 50% of patients with cleft lips and/or palates and associated anomalies.[10] More than 300 different syndromes have been described.[1]

Therefore, holistic care of the patient with a cleft lip or palate will require extensive screening, either by the family physician, or, if possible, a paediatrician. The goal of such screening is to identify any associated physical anomaly that requires further clinical attention. In addition, referral to a geneticist is important, if not as a matter of course, then certainly in all instances when an associated anomaly has been detected. Referral to a geneticist is important for a number of reasons. Firstly, many of the syndromes associated with cleft lips and palates are autosomal dominant in inheritance, e.g. Apert syndrome, thus necessitating genetic counselling for the parents of the patient, and later in life, the patient too. Secondly, if a syndrome is identified, this can help to focus the clinical investigation of anomalies associated with this syndrome.

An additional area of involvement for either the paediatrician or experienced family physician is to monitor the child for signs of developmental delay. Children with cleft lip and palate malformations are at higher risk of delayed development in the domains of gross motor, fine motor and expressive language.[12] The early identification of such difficulties can allow for timeous referral to speech and language therapists, as well as physio- and occupational therapists. A more detailed discussion of the role of these practitioners can be found in the next section.

The role of the orthodontist and dentist

The orthodontist and dentist can potentially be involved with both the preoperative and postoperative phase of management, depending on the needs of the patient. Therefore, good communication between these and other members of the surgical team, and specifically the plastic and maxillofacial surgeons, is of great importance. A referral to an orthodontist may be indicated in the preoperative phase to assess the potential for orthodontic devices to help "mould" the deformity into a position that is better suited to surgical repair.[13]

An assessment will be needed after repair of the cleft lip, as this procedure often inadvertently influences the condition of the maxilla. A final decision will then be made as to the need for either maxillofacial surgery or early orthodontic treatment. It is also important that the patient is followed-up when his or her permanent dentition begins to develop to assess the need for orthodontics[13] once more, as pure orthodontic problems are more common in this population. Children with cleft lip and palate malformations have an

increased risk of developing cavities and other dental caries, and should be routinely assessed by a dentist.[5]

The nature and timings of these interventions are highly individualised, and it is beyond the scope of most GPs to make such judgements. However, it is important that the GP is aware of the need for early referral in order for a specialised assessment to be made, and that communication between the orthodontist, dentist, maxillofacial surgeon and plastic surgeon is facilitated.

The role of the plastic surgeon, otorhinolaryngologist and maxillafacial surgeon

Considerable progress has been made in the field of the surgical management of cleft lip and palate malformations, and numerous techniques have been proposed. It has become a specialised field. Some surgeons dedicate their practice to this condition, and surgeons performing occasional surgery should arguably rather refer to larger centres, where more expertise is available. The different surgical techniques are beyond the scope of this article, but the basic principles and timing for referral will be discussed. The timing of surgery is not absolute and often depends on available surgical and anaesthetic expertise. However, is important to recognise that patients may require further surgeries much later in life after completing initial follow-up with their original surgeons. Therefore, the GP's role is to be alert to the development of any new indications for surgical referral.

Initially, only the cleft lip is repaired, usually at the age of three months. The majority of cleft surgeons do not favour neonatal repair, arguing that the presumed increase in anaesthetic risk is not justifiable, that it leads to compromises in surgical technique, and that it makes preoperative orthodontics impossible.[14] Primary rhinoplasty is often also required during primary cleft lip repair in order to correct nasal deformity.[15] The repair of the abnormally directed orbicularis oris muscle is the most important component of the repair. This will facilitate proper closure of the mouth, essential for feeding and normal speech.[16]

The surgical management of the soft palate usually occurs at six or 12 months of age. Essentially, this entails cleft palate repair, with or without bilateral myringotomy, the need for which must be assessed by the otorhinolaryngologist. There is significant debate as to the best timing of cleft palate repair. The potential benefits of early cleft palate repair from the standpoint of speech and hearing must be weighed against the increased technical difficulty of the procedure at a younger age and the possible adverse effect of maxillary growth. The crux of early palate repair is not to obtain complete closure, but the functional repair of the muscular sling,[16] which is necessary for normal functioning of the Eustachian tubes, and thus for normal speech and hearing.

By contrast, the hard palate should be repaired later, often at 12 months of age, to allow for a smaller adverse effect on maxillary growth than early repair.[17] For the same reason, early bone grafting has been largely abandoned in favour of secondary bone grafting by the time of mixed dentition. Secondary bone grafting can stabilise the dentomaxillary arch, improving the conditions for prosthodontic treatment, such as crowns, bridges and implants. It will also facilitate eruption of the teeth and increase the amount of bony tissue on the alveolar crest, allowing for orthodontic treatment.[17]

From this stage onwards, the need for further surgery needs to be assessed on an individual basis. Secondary palate lengthening or pharyngoplasty may be required at around 3-4 years of age. The need for this procedure can be established by referring a child with nasal speech to a speech therapist for a formal assessment. Complications of initial repair, such as oronasal fistulae, can also be repaired. Treatment for secondary lip and nasal deformities can be undertaken during the school years. Definitive open rhinoplasty may be performed at puberty. Definitive maxillary orthognatic surgery, if required, is performed by the plastic or maxilla-facial surgeon at skeletal maturity.[16]

Conclusion

Cleft lip and palate management represents a technical and logistical challenge to the medical profession. However, this challenge is also a potential opportunity for the dedicated GP, who is well positioned to coordinate and streamline this process. This is particularly true in the context of South Africa, where continuity of care between patients and multiple subspecialists is difficult to establish. In this article, we have provided essential information needed to help the GP take on a greater role in the management of these patients.

References

1. Cobourne, MT. The complex genetics of cleft lip and palate. Eur J Orthod. 2004;26(1):7-16.
2. Butow KW, vanWyLEI, Zwahlen RA. Differences in the clinical appearances of white versus black patients with facial cleft deformities: a retrospective study of a South African clinic. SADJ. 2007;62(7):298, 300-304.
3. Morrison G, Cronje AS, van Vuuren I, Op't Hof J. The incidence of cleft lip and palate in the Western Cape. S Afr Med J. 1985;68(8):576-577.
4. Dixon MJ, Marazita ML, Beaty TH, Murray JC. Cleft lip and palate: synthesizing genetic and environmental influences. Nat Rev Genet. 2011;12(3):167-178.
5. Trusler M, Bauer TB, Tondra JM, et al. Dental problems in cleft-lip cleft palate patients. Plast Reconstr Surg (1946).1957;19(5):409-419.
6. Habel A, Sell D, Mars M. Management of cleft lip and palate. Arch Dis Child. 1996;74(4):360-366.
7. Turner SR, Rumsey N, Sandy JR. Psychological aspects of cleft lip and palate. Eur J Orthod. 1998;20(2):407-415.
8. Clarren SK, Anderson B, Wolf LS. Feeding infants with cleft lip, cleft palate, or

cleft lip and palate. Cleft Palate J. 1987;24(3):244-249.

9. Peterson-Falzone SJ. Speech outcomes in adolescents with cleft lip and palate. Cleft Palate Craniofac J. 1995;32(2):125-128.
10. Shprintzen RJ, Siegel-Sadewitz VL, Amato J, et al. Anomalies associated with cleft lip, cleft palate, or both. Am J Med Genet.1985;20(4):585-595.
11. 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):180-186.
12. Neiman GS, Savage HE. Development of infants and toddlers with clefts from birth to three years of age. Cleft Palate Craniofac J. 1997;34(3):218-225.
13. Vig KW, Turvey TA. Orthodontic-surgical interaction in the management of cleft lip and palate. Clin Plast Surg. 1985;12(4):735-748.
14. Sommerlad BC. Surgical management of cleft palate: a review. J R Soc Med. 1989;82(11):677-678.
15. Thorne CH, Beasley RW, Aston SJ, et al. Grabb and Smith's plastic surgery. 6th ed. Philadelphia: Lippincott Williams & Wilkins, 2007; p. 201-225.
16. Lilja J. Cleft lip and palate surgery. Scand J Surg. 2003;92(4):269-273.
17. Liao Y, Cole TJ, Mars M. Hard palate repair timing and facial growth in unilateral cleft lip and palate: a longitudinal study. Cleft Palate Craniofac J. 2006;43(5):547-556.