Oral Care in a Patient with Long Arm Deletion Syndrome of Chromosome 18: A Narrative Review and Case Presentation

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Patient: Male, 7-year-old
Final Diagnosis: Tooth decay and ankyloglossia
Symptoms: Dental pain • feeding problems
Medication: —
Clinical Procedure: —
Specialty: Dentistry • Genetics

Objective: Congenital defects/diseases
Background: Long arm (q) deletion syndrome of chromosome 18 is a congenital chromosomal disorder. The specialist dental management of patients with 18q deletion is a challenge, as these individuals fall into the category of patients with special needs. The aim of this work was to describe the surgical and dental management in hospital of a patient with long arm deletion syndrome of chromosome 18 (18q).

Case Report: An 8-year-old patient with deletion syndrome of chromosome 18 (18q) was referred to the Department of Dentistry and Oral Surgery. The patient presented dental pain and difficult feeding. The examination of the oral cavity revealed a destructive carious lesion of the lower right second deciduous molar and the need to perform a frenectomy due to the short lingual frenulum, which limited the movements of the tongue. Given the complex management of the patient, it was necessary to carry out surgical procedures in the operating room. Frenectomies of the lower labial and lingual frenulum were carried out with the aid of an electric scalpel with an ultra-sharp microdissection needle.

At 2-month follow-up, the patient presented with good extraction site healing and satisfactory lingual mobility, along with improvements of speech and feeding.

Conclusions: Dental involvement in patients with deletion syndrome of the long arm of chromosome 18 is poorly documented in the literature. The hospital regimen appears to be the criterion standard for the management of the patient with long arm deletion syndrome of chromosome 18.

Keywords: Chromosome 18 Deletion Syndrome • Labial Frenum • Lingual Frenum • Pediatric Dentistry

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Background

Long arm (q) deletion syndrome of chromosome 18, also called De Grouchy syndrome, is a congenital chromosomal disorder. It was first studied by De Grouchy in 1964, who described a patient who exhibited a wide range of abnormalities, including short stature, characteristic facial features, ear malformations suggestive of congenital auricular atresia, foot edema, and severe disability [1]. Deletion occurs “de novo” in 94% of cases and in the remaining 6% it is inherited from a parent carrying a chromosome-balanced translocation. [2] The incidence of the deletion of the long arm of chromosome 18 is approximately 1 in 40 000 live births [3] and usually affects the terminal portion [4]. The extent of the deletion varies among patients and has extremely variable phenotypic manifestations [5], even if no association was observed between the size of the deletion and the severity of the manifestations [6]. Phenotypic manifestations affect different organs and systems and are extremely variable. The clinical developmental profile is characterized by mild to moderate mental retardation, developmental delay [7] and a known relationship with growth hormone deficiency [8]. There may also be multiorgan involvement with genitourinary malformations [9], cardiac malformations with an incidence of 24-36% [3], and IgA deficiency [10]. The limbs are also affected and present malformations [9] with foot anomalies and hypotonia [3]. Craniofacial dysmorphisms can also be found [3], cleft lip and palate [14], midface hypoplasia, depression of the corners of the mouth [15], proptosis, congenital heart disease, brain anomalies [11,12], and orofacial malformations. In particular, cleft palate was observed with or without cleft lip, upper palate [14], midline hypoplasia, depression of the corners of the mouth [15], proptosis, nasal saddle flat, palpebral fissures projected downwards or upwards [16], malformations of the external auditory canal which will be narrow or with atresia, and anomalies of the entire auditory organ [12,14].

The specialist dental management of patients with 18q deletion is a formidable challenge, as these individuals fall into the category of patients with special needs. “Disablement pertinent to dentistry” has been defined as a physical or mental condition that does not allow the patient to undergo proper oral hygiene and that does not foster patient compliance during dental treatments [17]. Poor cleaning ability and the difficulty of carrying out adequate dental care have repercussions on oral health, and these individuals tend to have a high incidence of caries and inflammatory processes affecting gingival tissue [18]. Dental patients with special needs may have anxiety, lack of collaboration, and defensive attitude, making dental treatment extremely complex and treating such patients is often is not feasible in private practice. In these cases, it is necessary to carry out interventions in a protected environment, such as a hospital, and for optimal treatment, general anesthesia is required [19]. In patients with special needs, several variables come into play that can affect treatment, since most of them have a complex clinical profile, including congenital diseases, with physical and psychological limitations [20].

The aim of this work was to describe the in-hospital surgical and dental management of a patient with long arm deletion syndrome of chromosome 18 (18q).

Case Report

An 8-year-old pediatric patient with deletion syndrome 18q came to the Pediatric Department of the A.O.U San Giovanni di Dio and Ruggi D’Aragona for a specialist evaluation due dental pain that made feeding difficult.

For this reason, was referred the patient for dental consultation at the Department of Dentistry and Oral Surgery of the same hospital. The patient, who presented mental retardation, was uncooperative. Examination of the oral cavity revealed the presence of inflammation of the gingival tissues due to the accumulation of bacterial plaque; therefore, the parents were instructed on how to help the patient maintain proper oral hygiene.

The mucous membranes were healthy and none of the teeth showed mobility. A destructive carious lesion of tooth 8.5 was detected, along with the need to perform a frenectomy due to the short lingual frenulum, which limited tongue movements. Given the complex management of the patient, it was necessary to carry out surgical procedures in the operating room; therefore, it was decided to perform 2 frenectomies (lingual and lower labial) and the extraction during a single surgery to avoid subjecting the patient to subsequent general anesthesia.

The patient was taken to the operating room and general anesthesia was carried out. The extraction of tooth 8.5 was performed first. Local anesthetic with vasoconstrictor was injected into the buccal region to reduce intraoperative bleeding. Subsequently, atraumatic extraction of the tooth was carried out and, after alveolar revision, the wound was sutured with a resorbable 5/0 Vicryl suture to avoid need for removal of the suture (Figure 1). Subsequently, the 2 frenectomies were performed (lingual and lower labial). For the control of intraoperative bleeding, the same procedure of local anesthetic infiltration was used (Figure 2). Frenectomies of the lower labial and lingual frenulum were performed using an electric scalpel with an ultra-sharp microdissection needle (Colorado) to facilitate cauterization and hemostasis (Figures 3, 4) The suturing used resorbable Vicryl 5/0 (Figure 5).
After 2-month follow-up, the patient presented with good extraction site healing and satisfactory lingual mobility, along with improvements of speech and feeding.

Discussion

After a complete review of the literature, few documents emerged regarding the dental phenotypic manifestations of the syndrome of deletion of the long arm of chromosome 18, as there are no indications for the dental management of these patients. Syndromic orofacial cleft represents the most frequent alteration of orofacial dysmorphisms in patients with deletion of the long arm of chromosome 18, with a prevalence of 17-38%. If anomalies such as high and arched palate are added to the cleft, the prevalence rises to 33-55% [21]. Mental retardation, developmental delay [8,9], and growth hormone deficiency [10,11] and among the oral pathological conditions...
that are dental manifestations secondary to metabolic conditions are associated with orofacial cleft. Among these, it was observed that in patients whose neurological development is impaired there is a greater risk of poor oral health, as well as delays in acquiring personal care skills, which are essential to sustain and maintain good oral health [22].

Understanding the importance of and role that plaque removal plays in maintaining oral health and developing the skills to perform such maneuvers is much more difficult for patients with special needs, and for these reasons external support is required [23]. In addition to these factors, other elements must also be added, such as inadequate dental health care, difficulty in accessing dental services, an incorrect diet, and intake of drugs that induce xerostomia. These elements make patients with special needs more susceptible to dental caries, and the development of gingivitis leading to periodontal disease [24]. The syndrome of deletion of the long arm of chromosome 18 can manifest itself with a growth hormone deficiency [10], which, as widely reported in the literature, is associated with dental and developmental anomalies of the bone structures of the jaws. It has been observed in several studies that bone structures in patients with GH deficiency are less developed [25], in particular mandibular retrusion, open gonion with an increased angle, and increased length of the lower third of the face have been observed [26]. The maxilla seems to be less affected, although cases of retrusion of the maxilla have also been reported [27].

The development and anomalous rotation of the jaws during growth can influence and alter the eruptive path of the teeth. This involves, in many cases, the onset of malocclusions and dento-basal disharmony with crowding due to incorrect spacing of the teeth caused by the reduced development of bone bases [28]. These conditions represent an element that make these patients more susceptible to development of various processes, as malocclusions are considered a risk factor for incorrect dento-basal ratio and the consequent dental crowding favor the formation of bacterial plaque, and make plaque removal difficult with home hygiene measures [29].

All these findings must be strongly taken into account when approaching dental treatment in patients with long arm deletion syndrome of chromosome 18, and the management of the patient must be personalized and individual. In our case, due to the patient’s poor ability to perform effective home oral hygiene maneuvers, due to the delay in mental and motor development, and given the presence of dental crowding which therefore guided our therapeutic choice of tooth 8.5 towards the extraction, rather than a conservative treatment that did not guarantee the possibility of success in the medium- and long-term. For this reason, we opted for the operation to be carried out in a protected environment, such as that of an operating room with the aid of general anesthesia. Several reviews have highlighted the lack of patient cooperation due to anxiety, intellectual disability, or some other deficit as the main indication for treatment under general anesthesia [30]. Our patient showed little ability to cooperate due to intellectual deficit and delayed cognitive development, and this was the main indication for treatment under general anesthesia.

General anesthesia, when performed by an expert team, appears to be a safe procedure [20,31]. However, particularly in patients with a complex clinical picture, there remains a certain risk of peri- or post-operative complications [32]. The mortality rate of patients under general anesthesia is 1/40 000 [33]; 37% of the deaths are drug-related, with halothane being the most commonly implicated agent, 38% being cardiac complications, and 20% are due to respiratory complications [32]. There is little scientific evidence on the suitability or the safety of the repeated use of general anesthesia over time in patients with special needs. We decided to perform 2 frenectomies (lingual and inferior vestibular) and an extraction in a single surgery to avoid subjecting the patient to repeated general anesthesia.

Another element to consider is the follow-up (short- and long-term) and the difficult management of post-operative control sessions. To improve the immediate post-operative period, instruments that could potentially lower the percentage of complications were used during the surgical procedure; in particular, the electric scalpel was used for frenectomies. By frenectomy we mean the complete removal of the frenulum. It is a more invasive procedure than frenulectomy and more difficult to perform but has more predictable results, with a lower recurrence rate [34]. Lingual frenectomy was performed after a clinical evaluation requested by the speech therapist who highlighted the presence of ankyloglossia; speech articulation problems are the most common indications for lingual frenulum surgery in preschool patients. [35]. The possible consequences of ankyloglossia range from limiting lingual movement, which can lead to the development of atypical swallowing, to impaired mandibular growth [36].

Our patient was referred again to the specialist to carry out functional rehabilitation sessions of the lingual organ; in fact, when the frenectomy procedure is performed in older children, they should be referred to a speech therapist to restore normal language functions [34].

The use of the electrosurgical unit was aimed at obtaining the same adequate hemostasis as with conventional scalpels, because despite the advantages such as ease of use, precision, low cost, and minimal damage to the surrounding tissues, they are unable to provide adequate control of bleeding, which is a priority in highly vascularized tissues such as those of the oral cavity [37,38]. Clinical evidence and studies in the literature
report the better performance of electrosurgery compared to others methods, including those with traditional scalpels, regarding hemostasis [39].

Alternatively, the use of the dental laser has also been proposed in the literature, given the advantages offered by the low invasiveness of the treatment and better post-operative management [40]. Lasers are already widely used in pediatric patients for surgical excision of lesions of the oral mucosa or for removal of caries [41,42].

The choice of the type of suture is also fundamental for a correct dental management of the patient 18q. We chose use of 5/0 sutures in resorbable material as this is the ideal choice for patient comfort and for the management of subsequent follow-up sessions. Non-resorbable sutures are not associated with a worse cosmetic outcome or increased wound complications, but the use of resorbable sutures would be preferred as there is no need for an additional visit for suture removal and anxiety (particularly among children) associated with the removal of the suture [43].

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Conclusions

The long arm deletion syndrome of chromosome 18, in its dental meaning, is poorly documented in the literature and the management of these patients remains a field still to be explored and delineated with scientific evidence that guides the clinician toward correct management of the syndromic patient. In present case report discusses the hospital regimen under general anesthesia, which seems to be the criterion standard for management of patients with deletion syndrome of the long arm of chromosome 18. This allows performance of all dental treatments in a single operating session.

Declaration of Figures’ Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.
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