Trismus-pseudocamptodactyly syndrome: a 20 year follow-up

ABSTRACT

Background Trismus-Pseudocamptodactyly Syndrome (TPS) is a rare autosomal syndrome characterised by the inability to open the mouth fully, pseudocamptodactyly, short stature and foot deformities. The maxillofacial feature entails hyperplasia of the coronoid processes which mechanically interfere with the zygomatic processes during mouth opening.

Case Report A 22-year-old girl affected by a severe form of TPS was followed from the age of three years. Bone reossification was observed after two coronoidotomies of both hyperplasic coronoid processes. After the decision to perform a coronoidectomy, the four-year follow-up showed a favourable outcome. Meanwhile the patient developed an anterior open bite which was treated with a fourth orthognatic surgery. The follow-up underscores how the correction of malformation leads to the generation of EMG activity of the masticatory muscles after many years of passiveness.

Introduction

Trismus-Pseudocamptodactyly Syndrome (TPS) is a rare condition also known as Hetch and Beals syndrome as the authors who first described it in 1969. TPS is a rare dominant autosomal syndrome with variable expression [Lano and Wekhaven, 1997] caused by a mutation of MYH8 codifying perinatal myosin heavy chain [Bonapace et al., 2010]. The main features of the syndrome are short muscle-tendon units, limited extension of interphalangic joints, foot deformities and limited excursion of the mandible. The inability to open the mouth fully seems to be caused by bilateral hyperplasia of the coronoid processes which mechanically interferes with the zygomatic processes during opening. Abnormal growth of coronoid processes develops from shortened temporal muscle-tendon units [Markus, 1986], and the resulting trismus is caused by shortened masseter, temporalis and pterygoid muscle-tendon units with healthy TM joints [Lafaivre, 2003]. The craniofacial malformation associated to this syndrome [Carlos, 2005], based on restriction of mouth opening, also creates problems in airway management for the anaesthesiologist.

This study, in which we report the case of a patient followed since 3 up to 22 years of age, sets out with a literature review about this rare syndrome The absence of vertical muscular control caused post-rotation of the mandible with an anterior open bite: the correction of the maxillo-mandibular malformation led to the generation of electromyographic activity after many years of passiveness.

Case report

A 22 year-old Italian girl affected by a severe form of TPS was followed at our operatory unit from the age of three years. Her main problem was recurring hyperplasia of the coronoid processes of the mandible.

The patient first underwent three surgeries at our department. At the age of four (1996) and at the age of fourteen (2006) the patient underwent intraoral bilateral coronoidotomy, both resulting in reossification of the bone gaps previously created with a relapse to a mouth opening of 10 mm. In June 2010 (at the age of 18), and after a new CT which confirmed a recurring hyperplasia of both coronoid processes, we performed a third surgery.

After endoscopically guided nasal tracheal intubation, a bilateral coronoidectomy was performed, obtaining an interincisal opening of 30 mm. In the early post-operative period the patient underwent a long course of physiotherapy. Two years later, at the age of 20, a CT scan confirmed the good outcome of the coronoidectomy without any signs of recurring hyperplasia; however, in the meantime the patient had developed an anterior open-bite. A conventional cranial X-ray showed the presence of a posterior dental premature contact that made impossible any type of masticatory function. The electromyography test showed no activity of temporalis and masseter muscles bilaterally. So we decided to submit the patient to a fourth surgery after one year of presurgical orthodontic treatment. The orthognatic surgery consisted in impaction and advancement of the maxillary bone and sagittal mandibular osteotomy. This surgical therapy made it possible to obtain a
correct intercuspation. After the surgery, the patient entered a long physiotherapy programme with monthly checkups for the first year post-surgery. At one month postoperatively, a first examination showed improvement in the mandibular excursion in protrusion and lateral movements; the electromyography showed signs of muscular activity. The last follow-up at 12 months, January 2014, confirmed the positive results with a further increase of the muscular activity and mandibular movements. The Relaxation Test showed an average increase of 276% of the temporalis anterior activity and an average increase of 357% of the masseter activity. The bite test showed an average increase of 176% of the temporalis anterior activity and an average of increase of 638% of the masseter activity.

Discussion

Trismus Pseudocamptodactyly Syndrome (TPS) or Hecht and Beals syndrome (BHS) is a rare autosomal disorder with sporadic incidence. The disease has a variable expressivity, and the clinical features vary greatly. This condition was first reported by Hecht and Beals in 1969 and is most frequently characterised by a decreased ability to open the mouth and curvature of the fingers at the level of interphalangic joints while attempting at dorsiflexion of the wrist [Wilson et al., 1969; Tsukahara et al., 1985; Teng et al., 1994]. To date only a single mutation, c.2021G>A (p.Arg674Gln) in MYH8, the gene encoding the perinatal myosin heavy chain, has been reported to cause TPS [Bonapace et al., 2010]. Alteration of this gene results in a phenotype of shortened muscle-tendon units. This mutation might also be associated to an increased risk of cardiac myxomas but it rarely, if ever, causes Carney complex [Toydemir et al., 2006].

The maxillofacial disorder concerns the inability to open the mouth fully, caused by bilateral hyperplasia of the coronoid processes [Carlos et al., 2005] and/or by the presence of fibrous ligaments extending bilaterally and anteriorly to the masseter muscle from the maxilla to the
mandible [Adams and Rees, 1999]. The inability to fully open the mouth is one of the most serious threats to these individuals. Intubation can be extremely difficult and hazardous [Seavello et al., 1999]. Adequate caloric intake, speech development, and dental care may be compromised [O’Brien et al., 1984].

The condition is treated by surgery. In the literature all reports about treatment of TPS show bilateral coronoidectomy as the surgical approach of choice [Karas and Wolford, 1995; LaFaire and Aitchinson, 2003; Carlos et al., 2005]. According to McLoughlin’s systematic review of the literature on the treatment of coronoid process hyperplasia, the most common surgical method is intraoral coronoidectomy, followed by extraoral coronoidectomy [McLoughlin et al., 1995]. The intraoral coronidotomy is seldomly applied [Chen et al., 2011; Gerbino et al., 1997]. In both surgeries, the most important complication consists in the reformation or rebuilding of the hyperplastic coronoid process [Yoshida et al., 2013; Smyth and Wake, 1994]. In our case, bone re-ossification was observed after the first two coronidotoanies [Pelo et al., 2003]; after the coronidotomy of both hyperplastic coronoid processes, the 4-year follow-up showed a good outcome.

In literature there is no reported case of development of an anterior open-bite after correction of bilateral hyperplasia of the coronoid processes as it occurred in our patient. However, due to the lack of function we expected a reduced maxillofacial growth (Moss functional matrix theory), but this did not occur in our case. The aetiology of skeletal anterior open-bite in such a case may possibly be attributed to a muscle imbalance between the jaw-closing and jaw-opening muscles during mandible development [Cai et al., 2012]. In this case report reduced EMG readings were recorded for temporals anterior and masseter muscles on both cranial sides. Therefore, after the third surgical correction, thanks to which the coronoid processes did not mechanically interfere anymore with the zygomatic processes, two causes may have contributed to the relapse of the anterior open bite: the muscular imbalance and the release of the joint. A fourth surgery was then required to obtain a correct skeletal and dental relationship.

The activation of muscular activity during the physiotherapy programme was linked to improvement of the mandibular excursion. In fact, the modifications of the craniofacial morphology by means of orthognathic surgery reflect not only on aesthetics, but also on the masticatory muscles, specifically the masseter and anterior temporals [Frongia et al., 2013]. Raustia and Oikarinen [1997] noted an increase in EMG activity of the masseter and anterior temporals during chewing and maximal bite force in the intercuspal position 12 months after surgery. A balanced EMG activity during clenching is considered a sign of good adaptation of the neuromuscular system to the new occlusal condition [Piancino et al., 2012].

Conclusion

According to the literature, coronoidectomy is the surgery that yields the best outcomes in the treatment of coronoid processes hyperplasia. Nevertheless, the development of the anterior open-bite which occurred after surgeries underlines the importance of muscular control in the vertical skeletal growth. Finally, the beginning of the muscular activity obtained after the last orthognathic surgery suggests that masticatory muscles and generally the whole masticatory system maintain adaptive capability, and that the surgical correction of skeletal disorder improves occlusion and masticatory muscles balance.

References