Trismus-pseudocamptodactyly syndrome. Case report ten years after

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ABSTRACT. Background In 1969, Hecht and Beals described for the first time a rare dominant autosomal syndrome characterised by reduced mouth opening, pseudocamptodactyly, short stature, and foot deformities. Recent studies have confirmed that TPS is caused by a mutation of MYH8 that is common to another disease called Carney syndrome. Case report The authors describe the long term follow-up of a case presented in 2003, ten years after the first surgical procedure: a 14-year-old girl, affected by this rare syndrome, had undergone an early (at 4 years) surgical treatment of bilateral coronoidotomies to ensure safe airway management to allow subsequent surgical treatment to correct foot deformities. After six years, a complete relapse of the trismus occurred. Three years later, the patient underwent a second surgery of bilateral coronoidotomies to definitely solve trismus. At the 18 months follow-up, the mouth opening was stable.

KEYWORDS: Pseudocamptodactyly; Carney syndrome; Coronoidotomies; Temporal muscles trismus; Dutch–Kentucky syndrome.

Introduction

The TPS or Hecht and Beals syndrome, named after the authors who first described it in 1969, is a rare dominant autosomal syndrome with variable expression [Lano and Wekhaven, 1997].

Recent studies have discovered that TPS is caused by a mutation of MYH8 codifying for perinatal myosin heavy chain [Toydemir et al., 2006]. Patients affected by TPS with recurrent mutation of MYH8 also had Carney syndrome, which is characterised by a high incidence of mixomas, skin pigmentation disorders, endocrine tumors, or overactivity in shwannomas that manifest with cardiac mixomas and spotty skin pigmentation [Veugelers et al., 2004]. TPS is also called the Dutch-Kentucky syndrome since Mabry et al. in 1974 discovered it in nine members of a Kentucky family that descended from Dutch ancestors, more precisely, from a Dutch woman who immigrated to the United States right after the American revolution [Mabry et al., 1974].

Recent studies have proved that the mutation of MYH8 took place independently in North American and European TPS pedigrees [Toydemir et al., 2006]. During the last two decades, TPS has been discovered not only in North America and Holland but also in Japan, Taiwan, Central America (Guatemala), and Europe (Germany, Italy, Belgium); thus, TPS cannot be considered exclusively Dutch–Kentucky [Tsukahara et al., 1985; Robertson et al., 1982]. TPS is also classified as distal arthrogryposis [Toydemir et al., 2006]. Males are more frequently affected than females [Lafavire and Aitchinson, 2003; Robertson et al., 1982]. TPS is characterised by shortened muscle-tendon units, inability to open the mouth fully, limited extension of interphalangic joints while attempting dorsiflection of the wrists (pseudocamptodactyly), and foot deformities. Limited mouth opening seems to be caused by bilateral hyperplasia of the coronoid processes that mechanically interfere with the zygomatic process during opening. Abnormal growth of coronoid processes should be mechanically stimulated by shortened temporal muscle-tendon units [Markus, 1986; Mabry et al., 1974].

Moreover trismus is favoured by shortened masseter, temporalis, and pterygoid muscle–tendon units [Lafavire and Aitchinson, 2003]. The temporomandibular joints are normal. For these reasons it does not represent a trismus but it is rather a pseudotrismus [Carlos et al., 2005; O’Brien et al., 1984].
Literature reports that fibrous ligaments bilaterally pull the masseter muscle forward, which could contribute to the trismus onset [Horowitz et al., 1973; Mercuri, 1981; Adams and Rees, 1999]. Thus, the presence of the fibrous band represents a variant of the Hecht and Beals syndrome [Lano and Wekhaven, 1997]. The patients under consideration do not exhibit any evidence of dominant autosomic inheritance and pseudocamptodactyly [Adams and Rees, 1999]. This finding is not always present in TPS cases (as also in the case described here). Beside making the normal functions like mastication and swallowing difficult, the limited mouth opening also creates problems in airway management for the anesthesiologists. Problems related to lower limbs are documented in at least 10% of the cases with TPS. The shortened leg muscles determine the foot deformity, whether equine, valgus, or varus foot [O’Brien et al., 1984].

Case Report

A 14-year-old Italian girl affected by TPS, whose father is also affected by TPS, has been followed by our operative unit, since the age of four (Fig. 1). At that time, the patient underwent bilateral coronoidotomy, which led to increased mouth opening from 9 to 42 mm. This allowed her to undergo, under general anesthesia, a subsequent surgery to correct the problem of valgus foot without difficulty, as it involved intubation. The patient subsequently followed a rigid protocol of kinesis physiotherapy.

Despite this, at four years from intervention, a partial relapse of the trismus occurred (mouth opening = 17 mm). In the following two years, a further reduction of mouth opening to 6 mm [Pelo et al., 2003] was observed, and the CT examination showed reossification of the bone gaps (Fig. 2). Since no specific problems were present, and no caries were detected [Giannoni et al., 2005] it was decided to

Fig. 1 - The patient at age 4. Mouth opening was 9 mm.

Fig. 2 - Patient in 2005. Mouth opening was 6 mm.

Fig. 3 - CT confirmed the interference of the coronoid processes with the zygomatic processes of the maxillary bone.

Fig. 4 - Surgical view: a bilateral coronoidotomy was performed, achieving a bone gap of 1.5 cm.
postpone further surgeries until the child reached puberty.

In September 2005, the patient underwent a new CT (Fig. 3) which confirmed reossification of the bone gaps observed in the previous CT (in 2001) and underlined the interference of the coronoid processes with the zygomatic processes of the maxillary bone.

The patient also underwent electromyography, which showed complete absence of muscular activity in the jaw elevators. The parents refused our proposal to carry out bilateral amputation of the coronoid processes with an extraoral surgical approach. Therefore, after endoscopically guided nasal tracheal intubation, we performed a bilateral coronoidotomy creating bone gaps of 1.5 cm (Fig. 4), and placing Bichat’s bulla bilaterally between the gaps (Fig. 5). Intraoperatively was obtained a mouth opening of 45 mm (Fig. 6). A bite block was placed for 24 hours to stabilise the mouth opening. Antibiotic, analgesics and myorelaxants were administered. The bite block was removed only for feeding and mouth cleaning.

Five days after surgery an intense physiotherapy programme had to be started because, while jaw opening was possible, jaw closing movements were completely absent; after 7 days the patient succeeded in having muscular control.

During the postoperative period the oral opening varied between 25 and 30 mm.

At six months postoperatively, a second electromiography was performed, which confirmed the development of muscular activity in the jaw elevators as well as in the lower part of the jaw. A CT control at 12 months confirmed the positive result of surgery (Fig. 7). At present, 18 months after surgery, mouth opening remains stable at 28 mm with good exertion of the mandible in the protrusive and lateral movements (Fig. 8). The patient has been instructed to use the bite block weekly and clinical examinations every three months are scheduled.

**Discussion**

TPS is a rare syndrome of autosomal dominant inheritance with variable expression. Recent studies have shown that it is caused by a recurrent mutation of the MYH8 [Toydemir et al., 2006; Veugelers et al., 2004], the gene codifying for perinatal myosin heavy chain [Veugelers et al., 2004]. Mutation of this gene results in a phenotype of shortened muscle-tendon units. It has been shown that mutation of the MYH8 is common to another disease called Carney complex.

The patients affected by Carney complex have a high incidence of mixomas, skin pigmentary disorders,
The fibrous ligament described by Mercuri in 1981, interposition of Bichat’s bulla between the bone gaps. In our opinion, if clinical conditions are not life-threatening for the patient and he/she has proper nutrition, it is always better to wait until puberty for greater patient compliance. We are positive that postoperative therapy requires a bite block along with physiotherapy to avoid scar bridile and reossification of bone gaps. In our experience, the use of bite block is a useful aid for mouth opening stabilisation and long-term control of the results.

Conclusion

We conclude that the age of intervention is dictated by the case-by-case needs: ensure safe airway management in orthopaedic surgical correction of valgus foot, or oral pathologies that cannot be treated due to limited mouth opening [Carlos et al., 2005; Pelo et al., 2003].

In our opinion, if clinical conditions are not life-threatening for the patient and he/she has proper nutrition, it is always better to wait until puberty for greater patient compliance. We are positive that postoperative therapy requires a bite block along with physiotherapy to avoid scar bridile and reossification of bone gaps. In our experience, the use of bite block is a useful aid for mouth opening stabilisation and long-term control of the results.

References


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