Trismus-pseudocamptodactyly syndrome: a case report


ABSTRACT: Background Hecht and Beals in 1969 described an autosomal dominant syndrome characterised by severe restriction of mouth opening, camptodactyly, shortness of leg muscles and, as a direct consequence, foot deformities. Case report A case of a 4 year old girl affected by this unusual syndrome is described. The patient underwent bilateral resection of coronoid processes by intraoral approach. An intraoral device was used in the immediate postoperative period in order to maintain mouth opening. Once at home, the patient has had, for six months, phisiokinesic therapy by means of a modified Darcissac device.

KEYWORDS: Trismus-pseudocamptodactyly syndrome

Introduction

Trismus-pseudocamptodactyly syndrome (TPS) was primarily described by Hecht and Beals [1969] as an autosomal dominant syndrome and those authors presented a family in which a father and his four sons were affected.

The main features of the syndrome are:
- limited excursion of the mandible;
- shortened muscle tendon units of the hands (consequently, flexion deformity of the fingers that occurs with wrist extension, camptodactyly);
- foot deformities related to shortened muscle legs, preventing a normal growth and development.

In the same year Wilson et al. [1969] identified the syndrome in nine members of a family spanning four generations. In 1974, Mabry et al. described the syndrome in nine members of an American family that had a Dutch ancestor (descending from a Dutch girl who emigrated in the USA soon after the American revolution), affected by trismus-pseudocamptodactyly anomalies. It was of note that the Dutch ancestry could be proved for four out of the eight families presented in the literature, so that the syndrome is also defined as Dutch-Kentucky syndrome. Nevertheless, in 1985 Tsukahara et al. reported on a Japanese family in which five individuals through three generations were affected with TPS. This syndrome is basically a rare neuromuscular disorder that manifests as a severe restriction of oral opening accompanied by various muscular and skeletal alterations without involving the intelligence’s development. Other rarer stigmata are:
- blefarocalasis;
- webbed neck;
- micrognathia;
- hardened cord behind the prominent naso-labial fold;
- occasional incidence of papula on these cords;
- chronic fibrosis myositis of the masseter muscles [Karras and Wolford, 1995].

The limitation of mandibular movements might be related to a fibrous abnormal ligament extending from the mandible to the maxilla, bilaterally [Horowitz et al., 1973; Mercuri, 1981].

Case report

The patient reported on here, along with the other anomalies, displayed hyperplastic enlargement of the coronoid process, probably ascribed to shortening of the muscle-tendon temporal units [Van Hoof, 1973]. This caused excessive strength during the oral opening attempts, affecting the growth of the coronoid processes.
Diagnosis. A 4 year old girl was referred to the George Eastman Dental Hospital Maxillo-Facial department for evaluation of limited oral opening (maximum incisal opening, MIO = 9 mm) (Fig. 1) by another medical department. After a physical examination confirming the TPS diagnosis, the following findings were revealed:
- camptodactyly (Fig. 2);
- bilateral pes equinus deformities (Fig. 2);
- brachicephalya, hipotrophic thick masseter muscles and bilateral hyperplasia of the coronoid processes, as shown by the radiographic examination (Fig. 3).

As a surgical correction of the valgismus, with lengthening of the Achilles tendon, was necessary, an early surgical resolution of the trismus was mandatory. This was in order to facilitate safe airway management during orthopaedic treatment under general anaesthesia [Browder et al., 1986; Vaghadia and Blackstock, 1988].

Treatment. The patient underwent intubation general anaesthesia using fibre optics. Using an intraoral approach, bilateral resection of the coronoid processes was performed, carefully forcing open the mandible intraoperatively, to achieve an interincisal opening of 42 mm (Fig. 4). A mouth prop was placed to maintain a good opening for the first five days after surgery. Beginning from the fifth day an intensive physical therapy regimen was instituted, using an intraoral device (Darcissac modified device) to continue after hospital discharge. The physical therapy regimen was continued at home for not less than six months.

Follow up. The case was followed up at weekly intervals for two years. A partial relapse of trismus was
observed and after four years the MIO was 17 mm and stable (Fig. 5). At the age of six years the patient underwent further surgery, under general anaesthesia, to correct the valgismus and the mouth opening at that time was sufficiently large to allow a safe airway management.

At the age of 10 years, unfortunately, a significant reduction of oral opening (MIO = 7 mm) was observed (Figs. 6, 7). Late relapse has been reported by several authors [Yamashita and Arnet, 1980; Mercuri, 1981; Karras and Wolford, 1995], so that further surgery is necessary.

It is of interest to note that, despite a severe reduction of mandibular function, affected patients show normal development of the maxillo-mandibular complex. In the present case loss of function seems to have played a minor influence on mandibular growth centres (Fig. 6).

**Discussion**

The TPS is a rare autosomal dominant syndrome with variable expressivity. It is characterized by limited oral opening, camptodactyly and foot deformities. The inability to open the mouth is related to an enlarged coronoid process and/or to a fibrous ligament, anterior to the masseter muscle, extending, bilaterally, from the mandible to the maxilla. This abnormal ligament was not present in the present case.

All the other reports present in literature [Yamashita and Arnet, 1980; Mercuri, 1981; Karras and Wolford, 1995] showed a surgical approach to the problem with bilateral coronoidectomy and removal of the enlarged coronoid process. Mercuri also made a resection of the fibrous bands anterior to the masseter muscle. According to the findings present in their case, Karras and Wolford [1995] resected the inferior one centimetre of the hypertrophic lateral pterigoid plates. An early relapse with limitation of oral opening was observed. Their patient was operated on after 4 weeks and a large amount of amorphous white tissue, occupying the superior two thirds of the masseter muscle, was resected. Bilaterally, the excessive mediolateral width of the mandibular ramus was reduced and adhesions that had formed between mandibular ramus and maxilla were resected. In all these previous reports intensive physical therapy, after hospital discharge, was immediately started for not less than six months.

As a delayed surgical treatment for relapse may often be necessary, the main goal in this present case was to obtain a mouth opening sufficiently large to allow a safe airway management during further surgery. Resection without removal of the coronoid process, performed bilaterally using an intraoral approach, makes surgery safer and less complicated, considering the reduced operative field due to the
patient’s age. An intraoral approach allowed us to avoid anaesthetic problems and to interfere, as little as possible, with the patient’s residual growth, avoiding growth deficiencies caused by scar tissue.

A normal growth of the mandible of the child reported on here was evident during the follow up period despite an obvious reduction of mandibular function due to relapse. This observation can be compared with cases of high condylectomy and loss of articular cartilage, in which a normal function allows growth of the mandible.

Conclusion

According to the literature, partial relapse of the trismus in cases of trismus-pseudocamptodactyly syndrome is frequently observed, requiring delayed surgical treatment. Nevertheless, early treatment is necessary to allow a safe treatment of orthopaedic deformities under general anaesthesia. Considering the necessity of secondary surgery it is suggested that a non-aggressive early surgery should be attempted. Delay is easier if mandibular growth is normal. Accordingly, a long-term postoperative physical therapy, to maintain mouth opening, is mandatory.

References


