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CASE REPORT

Trismus-pseudocamptodactyly syndrome (Hecht–Beals' syndrome): case report and literature review

R Carlos[1], E Contreras[1], J Cabrera[2]

[1]*Centro de Medicina Oral de Guatemala;* [2]*Hospital General San Juan de Dios, Guatemala City, Guatemala*

The trismus-pseudocamptodactyly syndrome (TPS) is a rare condition inherited as an autosomal dominant trait with variable expressivity. Clinically the syndrome is characterized by decreased ability to open the mouth and curvature of the fingers at the level of interphalangic joints while attempting dorsiflexion of the wrist (pseudocamptodactyly). A 6-year-old male patient from Guatemala with this syndrome is presented. The surgical treatment consisted of bilateral coronoid amputation to resolve the pseudotrismus. The procedure was successful and the patient achieved normal function.
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Keywords: trismus; pseudocamptodactyly; coronoid hyperplasia

Introduction

Trismus-pseudocamptodactyly syndrome (OMIM# 158300, TPS, Dutch–Kentucky or Hecht–Beal's syndrome) is a rare hereditary condition with autosomal dominant inheritance with variable expressivity, presenting partial inhability to open the mouth and limited extension of the interphalangic joints while attempting dorsiflexion of the wrists (pseudocamptodactyly).

Hecht and Beals (1969) first described the syndrome in 1969. A review of the literature revealed at least 11 families and approximately 314 affected individuals since 1969 (Wilson *et al*, 1969; De Jong, 1971; Mabry *et al*, 1974; Tsukahara *et al*, 1985; Rombouts and Verellen-Dumoulin, 1992; Teng *et al*, 1994). Most cases described to date are from of North America, Europe, Japan and Taiwan. The term 'Dutch–Kentucky syndrome' was introduced by Mabry *et al* based on the fact that a Dutch girl who migrated to the United States soon after the American Revolution and most cases reported to date in the US are probably related.

Trismus-pseudocamptodactyly syndrome is part of a group of related congenital conditions known as distal arthrogryposes, all of which present with camptodactyly and other defects. Other syndromes within this group are Freeman–Sheldon syndrome, congenital contractural arachnodactyly and familial camptodactyly (Hall *et al*, 1982).

Responsible genes for this condition have not been yet identified and analysis using blood group markers in one family failed to show linkage to any chromosome (Robertson *et al*, 1982).

Case report

In February 2001 a 6-year-old Guatemalan male presented with trismus of unknown etiology with a maximum opening of 23 mm (Figure 1); the trismus was apparently congenital. Because of this limitation the patient had difficulty with dental hygiene procedures and had developed extensive carious lesions. Restorative procedures were not possible at that time for the same reason. A panoramic radiograph revealed severe bilateral enlargement of the mandibular coronoids (Figure 2). A 3-D CT scan was ordered after the cause of trismus was discovered in the panoramic radiograph (Figure 3). The child also had difficulty with feeding. He used to push small amounts of food into the mouth between the limited opening. During physical examination a limitation in performing dorsal flexion of both hands was discovered, making the classic clawshaped hand. This restriction was because of the short finger-flexor-tendons (Figure 4). During volarflexion a full digital extension was observed with no finger stretch or movement limitation. The patient had no talipes equinovarus (Figure 4). The family history was not complete, as the mother was single and they had no contact with the father or his family. The mother and her family had no history of similar anomalies. The condition was finally diagnosed as trismus-pseudocamptodactyly syndrome (Hecht–Beal's syndrome).

Correspondence: Dr E Contreras, Centro de Medicina Oral de Guatemala, 16 calle 4-53 zona 10, Edificio Marbella, Guatemala City, Guatemala 01010. Tel: (502) 363 0769, Fax: (502) 337 0705, E-mail: roman@guate.net.gt
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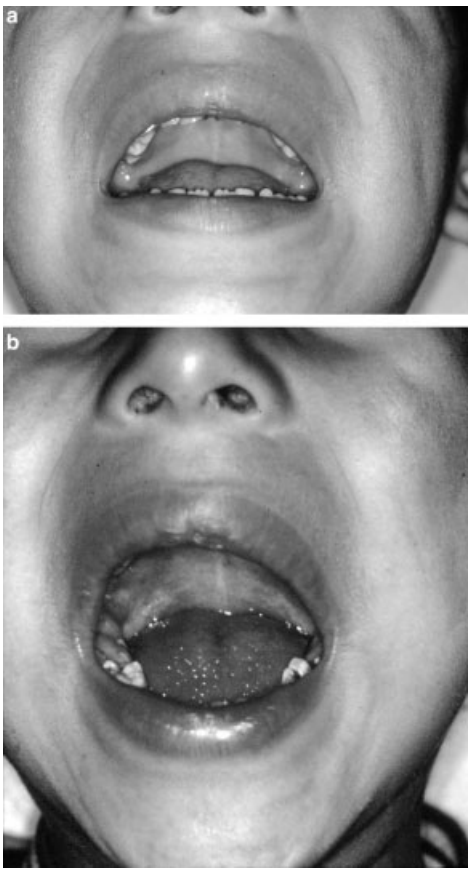

Figure 1 (**a**) Preoperative: Reduced opening at initial presentation. (**b**) Preoperative: Increased opening after coronoid amputation, at eighth month follow-up (lower)

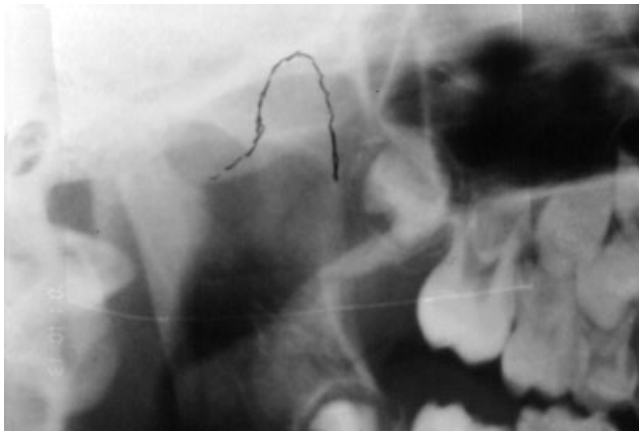

Figure 2 Panoramic X-ray showing one of the enlarged coronoids

The patient underwent intraoral bilateral coronoidectomy under general anesthesia using nasotracheal intubation. After the surgical procedure a mouth-opening device was placed which was only removed for eating and physical therapy sessions in order to prevent a new opening problem secondary to fibrosis or scarring. Immediately after the surgical procedure a normal opening was obtained (Figure 1b). No complications were noted after surgery. Restorative dentistry was completed successfully and the child is currently on orthopedic rehabilitation.

Discussion

Trismus-pseudocamptodactyly syndrome (Hecht–Beals' syndrome) is a rare autosomal dominant inherited disorder with affected families reported in the USA, the Netherlands, Canada, Japan and Taiwan (Wilson *et al*, 1969; De Jong, 1971; Tsukahara *et al*, 1985; Rombouts and Verellen-Dumoulin, 1992; Teng *et al*, 1994). To the best of our knowledge no cases have been reported in Latin America to date. The autosomal dominant inherited trait was not confirmed in the present case because of the loss of contact with the father. However, this possibility cannot be excluded.

The syndrome is characterized by trismus secondary to bilateral hyperplasia of the mandibular coronoid processes. Few of the published reports mention the cause of the trismus. The limited mouth opening is secondary to a prominent overgrowth of the coronoid processes that causes mechanical interference with the maxillae. The temporomandibular joints are normal. Therefore, this condition does not represent a true trismus but rather a pseudotrismus. Patients experience a functional deficit and dental hygiene is compromised due to this opening limitation. The limited opening hampers prophylactic and restorative dental treatment. They have an overall normal development but their stature is below average.

In our case this problem was successfully resolved by intraoral amputation of both coronoid processes. The patient has been followed up for 31 months without complications or side effects. There is a recent case published of a 4-year-old Italian girl affected by this unusual syndrome treated also with bilateral resection of the coronoid processes and postoperative ambulatory treatment with physiokinetic therapy (Pelo *et al*, 2003). Another case with attempted surgical correction via endoscopic release in a 28-month-old boy resulted in early postoperative failure. The same patient subsequently underwent bilateral coronoidectomy, the latter being successful after 12 months of follow-up (Lefaivre and Aitchison, 2003).

The other component of this syndrome is the inability of dorsalflexion of the hands because of short finger-flexor-tendons that in turn forces the patients to flex the fingers into a claw-shape. During volarflexion affected individuals are able to achieve full digital extension. Because involvement of the hands does not interfere with normal function and is completely asymptomatic, treatment is usually not necessary.

Leg or foot problems are found in at least 10%. Short leg muscles result in a variety of mild foot deformities: talipes equinovarus or calcaneovalgus, which our patient did not have (Gorlin *et al*, 2001).

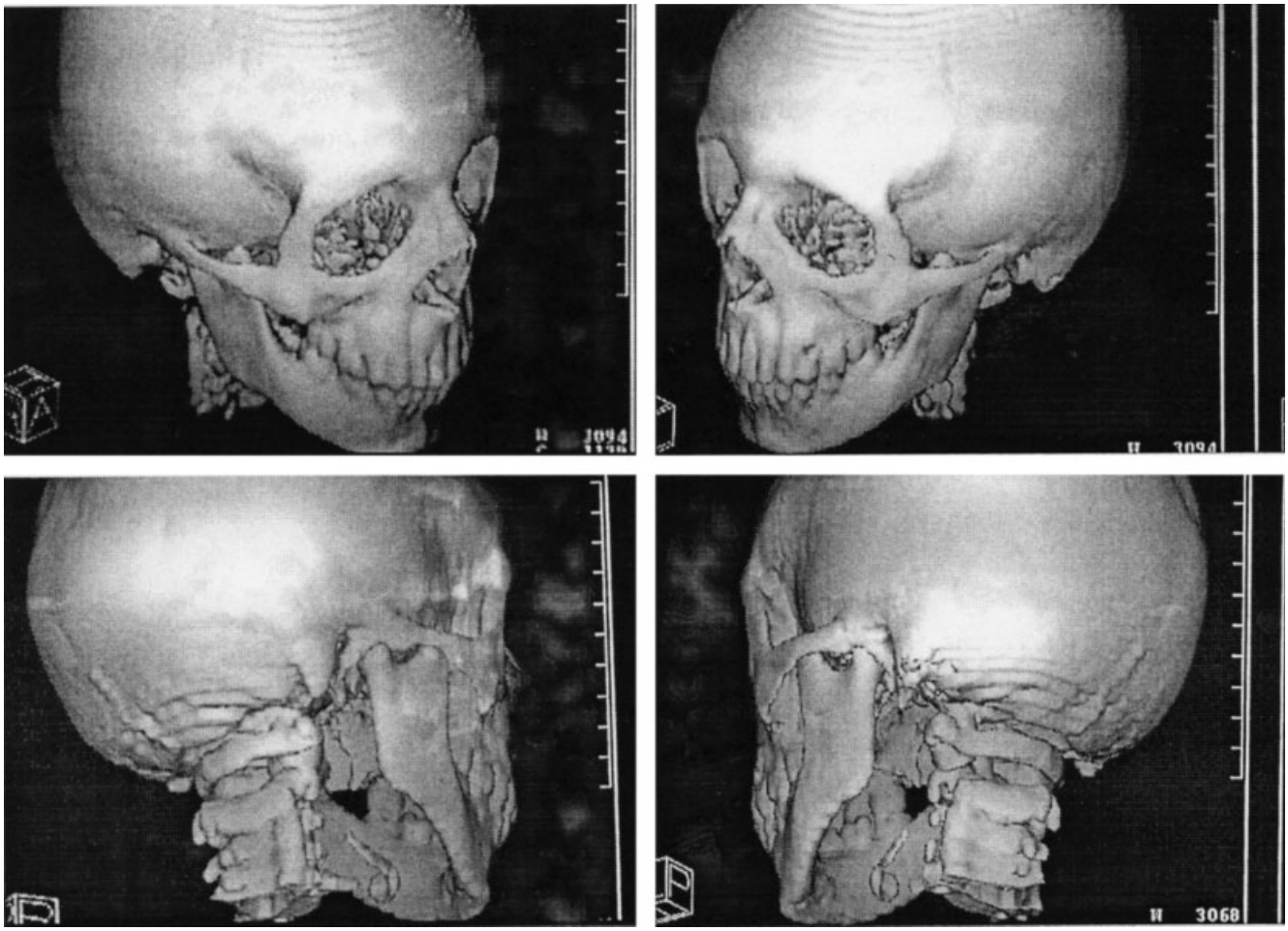

Figure 3 3D-reconstruction showing the bilateral coronoid enlargement, before surgery

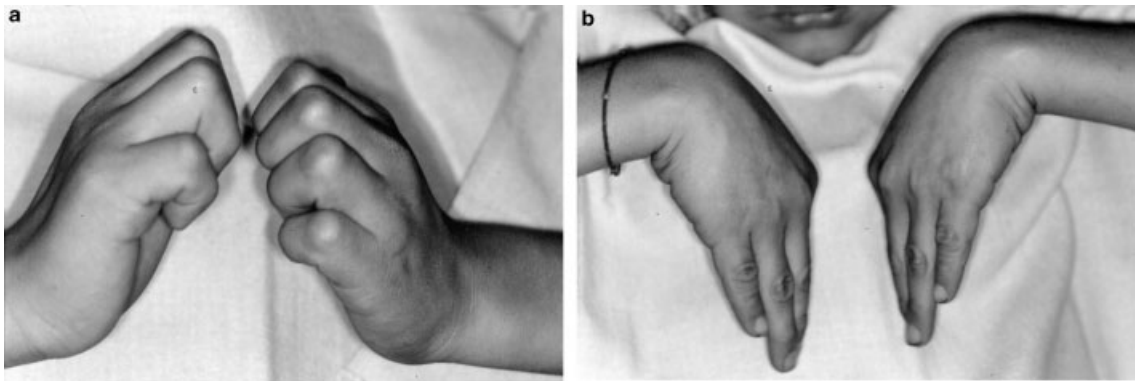

Figure 4 (**a**) Claw-shaped hands during dorsiflexion. (**b**) Normal hand movement during volarflexion

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