Oro-dental manifestations of the Schwartz-Jampel syndrome

L.X.G. Stephen* / P.H. Beighton**

A boy with the Schwartz-Jampel syndrome (chondrodystrophic myotonia) had a number of oro-dental complications. These included difficulty in tooth extraction and orthodontic care due to a small oral aperture and rigidity of the temporo-mandibular joints. General anaesthesia was hazardous because of a propensity to malignant hyperthermia, and endotracheal intubation was difficult because of shortness and rigidity of the neck and the small size of the laryngeal structures. Awareness of these potential problems is crucial for anaesthesia and comprehensive dental management. The radiological demonstration of dentigerous cysts is a hitherto unreported observation in this disorder.
J Clin Pediatr Dent 27(1): 67-70, 2002

INTRODUCTION

The Schwartz-Jampel (SJS) [MIM 255800] syndrome is an unusual autosomal recessive genetic disorder in which skeletal abnormalities are associated with myotonia. The main clinical manifestations are short stature, stiff joints, and spinal malalignment. The facial appearance is characteristic, with blepharophimosis and muscle rigidity producing a mask-like appearance, drooling and a high pitched voice.[1] Mentality is usually normal and there are no systemic ramifications, but the skeletal and muscular abnormalities cause considerable disability and the spine malalignment may lead to potentially lethal complications.[2]

The combination of muscular and skeletal abnormalities predisposes to a number of primary and secondary oro-dental manifestations and complications. These problems, which are of practical importance in dental management, are reviewed in this article and illustrated by a report of an affected boy. The radiographical demonstration of dentigerous cysts represents a previously undocumented observation.

* L.X.G. Stephen, BChD, PhD, Dental Genetic Unit, Department of Oral Medicine and Periodontology, Faculty of Dentistry, University of the Western Cape, Cape Town, South Africa.
** P.H. Beighton, MD, PhD, FRCP, Department of Human Genetics, Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa.

Send all correspondence to Dr. L.X.G. Stephen, Department of Oral Medicine and Periodontology, Faculty of Dentistry and WHO Oral Health Collaborating Centre, University of the Western Cape, Private Bag X08, Mitchell's Plain 7785, Cape Town, South Africa.

Voice: +27-21-328116
Fax: +27-21-323250

CASE REPORT

MM, a boy of indigenous African stock was born in 1988. At birth he was noted to be small, with clubfeet (talipes equinovarus) and an immobile face. His parents were young and non-consanguineous and neither his younger brother nor any other family members were similarly affected. Ambulation was delayed and when walking commenced, his gait was disturbed due to the foot deformities, limb malalignment and muscle rigidity. At the age of five years, the hip joints were examined under anaesthesia and at 9 years valgus osteotomy and adductor tentotomy of the left hip were performed. Rigidity of the temporo-mandibular joint, the constricted buccal aperture, mandibular hypoplasia and the small size of the larynx made endotracheal intubation difficult. The risk of malignant hyperthermia posed a significant hazard at operation and necessitated an appropriate anaesthetic approach. The postoperative course was complicated by airway obstruction due to secretions blocking the constricted larynx. Essential relief was obtained by suction.

In later childhood, the intellect was normal and general health was good. Physical handicap was considerable and he had a waddling gait with flexion contractures of the hips, knees feet and elbows. He received his education at a special residential facility in Cape Town.

In, October 2000 at the age of 13 years, he was seen at the Dental Genetic Unit, University of the Western Cape for the purpose of dental assessment and formulation of an appropriate plan for dental management. At this time his stature was very small (height 125 cm), and he had an immobile facies, with blepharophimosis, ptosis and micrognathia (Figure 1). Extra-oral examination revealed limited opening of the mouth; the temporo-mandibular joints only displayed hinged movement with very little translocation on opening. The midline of the maxillary dentition was deviated 3 mm to the

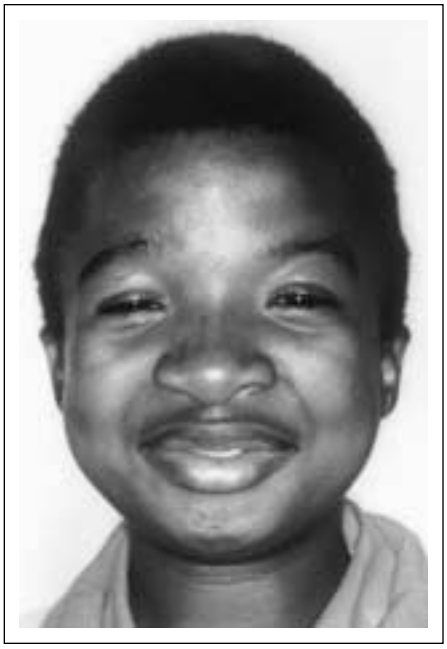

Figure 1. MM at the age of 13 years. The characteristic mask-like facies results from muscular rigidity.

left. The facial muscles were rigid and the lips were pursed.

Intra-oral examination revealed partial ankyloglossia that limited movement and function of the tongue. The midline frena of the upper and lower lip were very prominent (Figure 2). Maxillary teeth that were present included 11,12,13,14,15: 21,22,23,24,26. (Teeth number 13,23 were buccally displaced). The mandibular teeth that were present included teeth number 31,32,33,34,35: 41,42,43. There was no dental caries, but moderate inflammation of the gingival tissues was evident.

Lateral skull radiographs showed that the condylar heads of the mandible were small and underdeveloped, possibly as a consequence of the impaired function of the masticatory muscles.

A pantograph revealed several impacted teeth in the maxillary arch (Figure 3), [teeth number 14,17,18,24,27,28]. The crowding and impaction was severely complicated in the mandibular arch and included teeth number 36,37,38,44,45,46,47,48 (Figure 4). Radiolucent cystic lesions were observed in association with teeth number 37,38,46,47,48.

Consultation with various specialists including a maxillo-facial surgeon, oral pathologist and a radiologist resulted in the decision that these radiolucent lesions should be observed bi-annually. If there was any change or growth of these lesions, then surgery would be necessary to remove the impacted teeth and cysts simultaneously.

At follow-up, six months later, his mal-occlusion and other oral complications were unchanged, but he complained of intermittent discomfort in the distal region of third and fourth quadrant.

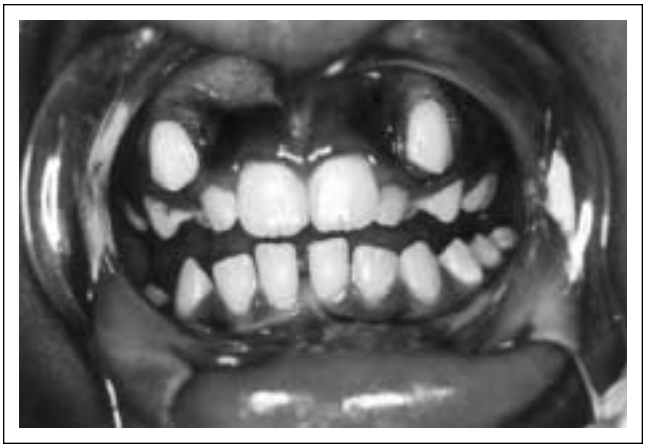

Figure 2. The midline frena of the upper and lower lips are very prominent. Midline deviation and severe crowding of the permanent dentition is evident.

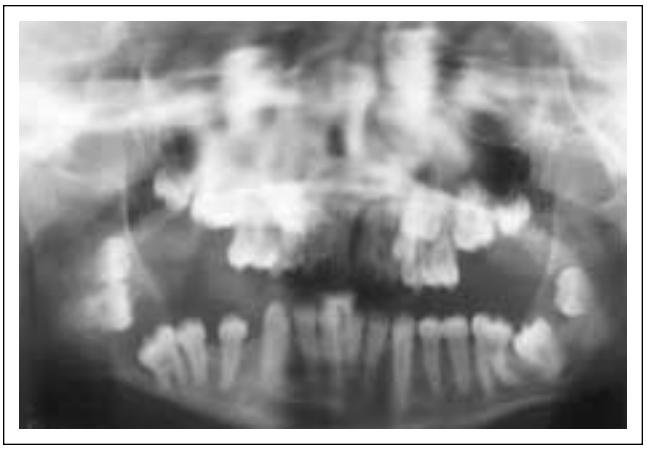

Figure 3. Pantograph showing several impacted teeth in the maxillary and mandibular arches. A large dentigerous cyst is associated with tooth number 47. A cystic lesion is also associated with tooth 38.

A pantograph demonstrated a substantial increase in the cystic areas associated with the lower second and third molars. In view of the unusual presentation and rapid increase in size of the lesion associated with tooth 47, MR and CT scans of the brain, deep face and mandible were obtained. The differential diagnosis of the mandibular lesions included dentigerous or keratocysts and there was evidence that the cyst associated with tooth 47 was infected.

From a surgical and dental viewpoint, the impacted teeth and dentigenous cyst will have to be removed before complications arise. The maxillary, mandibular and lingual frenulae need to be resected since they impair the function of the lips and tongue. The comprehensive management plan for this patient involved speech therapy to rectify problems of articulation that had arisen as a result of ankyloglossia.

This extensive regime of surgical management will be difficult because of the microstomia and limitation of temporo-mandibular movement. In addition, as the

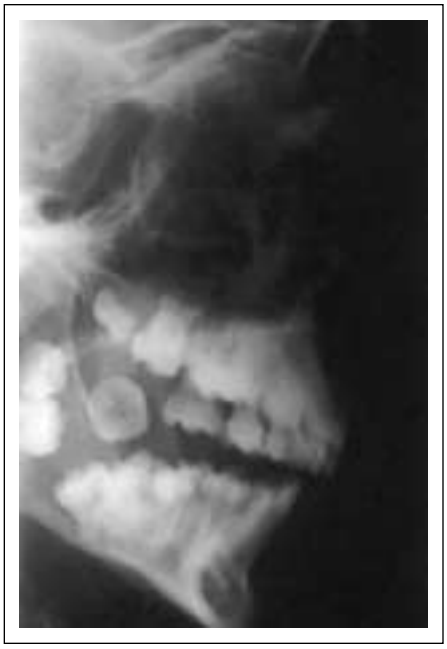

Figure 4. Lateral view of the jaws. Severe orthodontic malocclusion and the presence of impacted molar teeth are evident.

Table 1. Oro-dental manifestations and complications of the Schwartz-Jampel syndrome.

1. Temporo-mandibular joint rigidity.
2. Facial muscular rigidity
Drooling.
Indistinct speech.
Small oral aperture.
3. Laryngeal hypoplasia
High pitched voice.
Dyspnoea.
Difficult endotracheal intubation.
Post-operative airway obstruction by secretions.
4. Mandibular hypoplasia
5. Propensity to hyperthermia
6. Dentigerous cysts.

planned procedures will have to be performed under general anaesthesia, there may be problems with endotracheal intubation due to the short neck and small larynx. Equally, the propensity to malignant hyperthermia poses a major anaesthetic hazard. Post-operatively, obstruction due to secretions blocking the constricted airways, represent a major risk. Suction may well be necessary for relief. For these reasons, a comprehensive pre-operative anaesthesiological approach is warranted. The manifestations and complications of the condition, which are inherent in dentistry and anaesthesia are listed in Table I.

COMMENT

After the initial description of an affected brother and sister by Schwartz and Jampel,[3] other reports followed from several parts of the world. Various combinations of the eponyms "Catel", "Schwartz", "Jampel" and "Aberfeld" have been employed, as has the descriptive designation "chondrodystrophia myotonica". The term "Schwartz-Jampel syndrome" (SJS) is now in general use.

By 1973, reports of 8 affected persons had appeared in the medical literature and it became evident that SJS was inherited as an autosomal recessive trait.[4] Since that time, numerous persons with SJS have been recognised.[5] It is of interest that our patient, MM was one of the individuals with SJS in whom the determinant gene was initially localised to chromosome 1 by research collaborators at the Saltpêtriére Hospital, Paris.[6] Thereafter, the gene was characterised[7] and the gene product identified.[8] The pathogenetic mechanisms are currently being explored.

The stunted stature, muscle rigidity and skeletal abnormalities cause to significant disturbance of gait, and corrective orthopaedic operations have been performed on a number of affected persons.[9] Problems with anaesthesia, notably facial rigidity, mandibular hypoplasia, small buccal aperture, temporomandibular joint rigidity are well recognised in orthopaedic practice. In particular, the short neck and small larynx make endotracheal intubation difficult, while the anaesthetic risk of malignant hyperthermia is an additional hazard.[10]

The anaesthetic complications that have been encountered during orthopaedic surgery are equally applicable to dentistry, although thus far, to the best of our knowledge these have not been reported, reviewed or discussed in this context. During the past three decades, 15 affected persons, including 2 sets of siblings have been documented by PB in South Africa. Many of these patients have attended special facilities for the physically handicapped and it is to be expected that they will be encountered by dentists and orthodontists, who provide dental services to these institutions. Awareness of the clinical manifestations and potential dental and anaesthetic complications are essential for effective dental management.

REFERENCES

1. Gorlin RJ, Cohen MM, Levin LS. Chondrodystrophic myotonia. In: Syndromes of the Head and neck. 3rd ed. New York, Oxford University Press, pp. 631-633, 1990.
2. Viljoen D, Beighton P. Schwartz-Jampel syndrome (chondrodystrophia myotonica). J Med Genet 29: 58-62, 1992.
3. Schwartz O, Jampel RS. Congenital blepharophimosis associated with a unique generalized myopathy. Arch Ophthal 68: 52-57, 1962.
4. Beighton P. The Schwartz syndrome in Southern Africa. Clin Genet 4: 548-555, 1973.
5. Giedion A, Boltshauser E, Briner et al. Heterogeneity in Schwartz-Jampel chondrodystrophic myotonia. Europ J Pediat 156: 214-223, 1997.
6. Nicole S, Ben Hamida C, Beighton P et al. Localization of the Schwartz-Jampel syndrome (SJS) locus to chromosome 1p34-p36.1 by homozygosity mapping. Hum Mol Genet 4: 1633-1636, 1995.
7. Fontaine B, Nicole S, Topaloglu H, et al. Recessive Schwartz-Jampel syndrome (SJS): confirmation of linkage to chromosome 1p, evidence of genetic homogeneity and reduction of the SJS locus to a 3-cM interval. Hum Genet 98: 380-385, 1996.

8. Nicole S, Davoine C-S, Topaloglu H, et al. Perlecan, the major proteoglycan of basement membranes, is altered in Schwartz-Jampel syndrome (chondrodystrophic myotonia). Nature Genetics 26: 480-483, 2000.
9. Horan F, Beighton P. Orthopaedic aspects of the Schwartz syndrome. J Bone Joint Surg 57-A: 542-544, 1975.
10. Seay AR, Ziter FA. Malignant hyperpyrexia in a patient with Schwartz-Jampel syndrome. J Pediatr 93: 83-84, 1983.

ACKNOWLEDGEMENTS

We are grateful to Professor M.H. Moola of UWC for his encouragement and to Mrs Greta Beighton for preparing the manuscript with enthusiasm and efficiency. PB acknowledges with gratitude partial support from the South African Orthopaedic Association.