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Alstrom syndrome: a case report

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Abstract: Alstrom syndrome is a rare disorder characterized by early obesity, loss of central vision, diabetes mellitus, hearing loss and short stature. Previous studies, have reported no information regarding oral findings. This article describes oral findings in two cases of Alstrom syndrome. In both cases, gingivitis was present and also light yellow-brown discolored enamel bands were observed on the anterior teeth. This staining may have resulted from discoloration of the preexisting slight band-like enamel hypoplasia. The gingiva was examined histologically by light and transmission electron microscopy. Irregular thickness of the basal lamina and delamination of the myelin sheath were detected by transmission electron microscopy. There is no information about pathological odontogenesis in Alstrom syndrome in previous reports. Oral present findings may contribute further information about the clinical manifestations of Alstrom syndrome. (J. Oral Sci. 43, 221-224, 2001)

Key words: Alstrom syndrome; diabetes mellitus; discolored enamel band.

Introduction

Alstrom syndrome was first described in a Swedish family in 1959 (1,2). This syndrome affects the ocular system during the 2nd decade (3-6), the auditory system between the 2nd and 3rd decades (3-5) and the genitourinary (1,3,5,6) and integumentary systems (1,3,4,6) during the 4th decade. It is characterized by early obesity (1,4), loss of central vision due to atypical retinal degeneration, diabetes mellitus (3-6), sensorineural hearing loss (3-5) and short stature (3,7,8). Radiologic examination of affected patients has also revealed scoliosis and hyperostosis frontalis interna (5,9). Laboratory findings include carbohydrate intolerance (1,8), renal impairment manifested by albuminuria and elevated blood urea nitrogen, hypertriglyceridemia (1), increased urinary gonadotropin levels and low plasma testesterone levels in some cases (8,9). In all cases reported, the parents of affected children were normal. This syndrome occurs in both sexes, most probably by autosomal recessive inheritance.

Alstrom syndrome is a rare disorder, with fewer than 30 cases reported (3-10). In recent studies, patients with Alstrom syndrome were examined for endocrine insufficiency, and it was reported that growth hormone deficiency may account for the short stature. The advanced bone age and normal early growth were thought to be due to hyperinsulinism resulting from a specific defect in the signal transduction of insulin action and possibly hormone spillover on another receptor (7).

Although extensive morphological and laboratory examinations were carried out on these subjects, previous reports gave no information regarding oral findings. This article describes oral findings in two cases of Alstrom syndrome.

Case Reports

The patients were siblings aged 14 and 20 years who visited our clinic for dental examinations and treatment. In their family, not only these siblings but also their 7-year-old sister were affected by early-onset atypical retinitis pigmentosa, mild sensorineural hearing loss, short stature, truncal obesity, hyperinsulinism and hypertriglyceridemia (Fig. 1). The parents, who were first cousins, had eight

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children altogether. The parents and other family members were completely healthy, except for the third youngest child of the family who developed Behcet's disease at 13 years of age (A in Fig. 1). A paternal aunt had died at 23 years of age. All that was known about her was that she was relatively short and obese, and was suffering from visual loss.

Medical Examination

Case 1

The patient was 14 years old and the fourth child of the family (B in Fig. 1). Physical examination showed that she had short stature and truncal obesity (BMI 23.2 kg/m^2). She had been totally blind for the last four years. Furthermore, she had livedo reticularis on the extremities, butterfly facial rashes and bilateral palmar erythema. Arterial blood pressure (BP) was 150/95 mmHg and pulse rate was 100/min and regular. Urinalysis revealed proteinuria (0.5 g per 24 h), marked glycosuria and mild acetonuria. Her bone age was estimated to be around 17-18 years. A renal biopsy revealed mesangial proliferative-type glomerulopathy with hyaline arteriosclerosis, mild interstitial fibrosis and no amyloidosis. An electromyographic investigation showed that motor and sensorial nerve conduction velocities were within the normal ranges.

Fig. 1 Three brothers and 5 sisters together (A suffered from Behcet's disease and B (Case 1), C (Case 2) and D suffered from Alstrom syndrome).

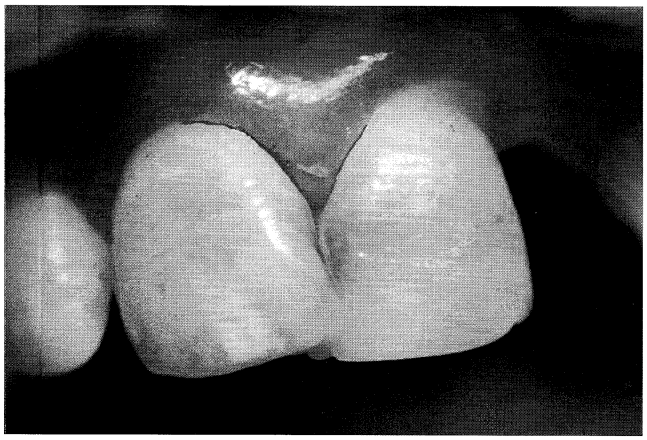

Fig. 2 Localization of the discolored bands on the enamel in Case 1.

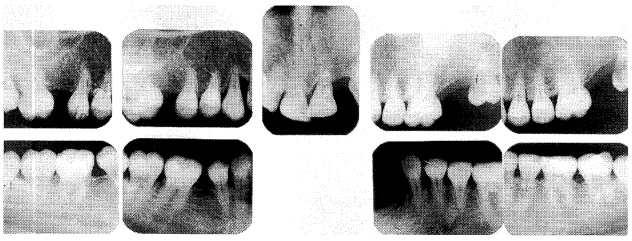

Fig. 3 Radiographs of Case 1.

Case 2

The patient was 20 years old and the eldest child in the family (C in Fig. 1). She suffered from blindness and also had short stature and truncal obesity. She showed bilateral sensorineural hearing loss and mildly retarded psychomotor development.

The clinical findings for both patients are shown in Table 1.

Oral Findings

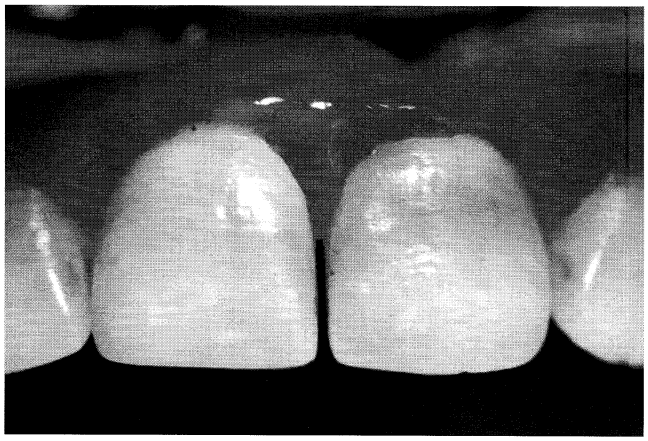

Fig. 4 Localization of the discolored band on the enamel in Case 2 at the corresponding areas of the anterior teeth of Case 1.

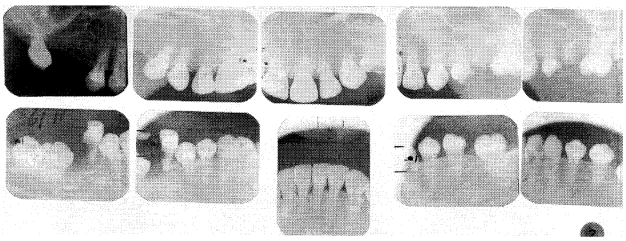

Fig. 5 Radiographs of Case 2.

Table 1 Physical characteristics and clinical findings relevant to Alstrom syndrome in both cases

PARAMETERS	CASE I	CASE II
Age	14	20
Sex	F	F
BMI (kg / m^2)	29.1	29.7
Obesity	truncal	truncal
Height (m)*	1.40	1.36
Retinitis pigmentosa	present	present
Visual loss	total blindness	total blindness
Hearing loss	sensorineural	sensorineural
Psychomotor development	normal	normal
Diabetes Mellitus	insulin-treated diabetes	OGTT overt diabetes
Nephropathy (nondiabetic)	present	absent
Hypertriglyceridemia	present	present

*Growth development was found to be less than the third percentile of normal children according to height charts.

During oral examination, it was observed that the oral hygiene of both siblings was poor and they had gingivitis which scored 3 according to the SBI index.

Case 1

Oral examination of the patient revealed eight missing teeth, two decayed teeth and two fillings. The incisal half of the anterior teeth featured moderate white patches and on the cervical half a light yellow-brown discolored enamel band was observed. This enamel band also exhibited a pronounced accentuation of perikymata (Fig. 2). Radiographs exhibited vertical and horizontal alveolar bone loss (Fig. 3).

Case 2

Oral examination of the patient revealed six missing teeth and three decayed teeth. Moderate white patches and light yellow-brown discolored enamel bands with an accentuation of perikymata were observed at the corresponding areas of the anterior teeth of Case 1 (Fig. 4). Horizontal alveolar bone loss was observed radiographically (Fig. 5).

Discussion

Clinical manifestations of both siblings seemed to be consistent with the features of Alstrom syndrome (1,3-6). Enamel opacities which scored between 2 and 3 (11), and discolored enamel bands on the central and lateral teeth of both sisters are characteristics of a moderate form of systemic band-like enamel hypoplasia. Band-like enamel hypoplasia is an acquired hypoplasia caused by systemic disease at a certain stage during tooth formation. Systemic diseases such as fever, allergy, immunization, infections, nutritional deficiencies, intoxication, cerebral palsy, congenital heart disease, gastro-intestinal disease and endocrinopathies might cause such disturbance of tooth formation (12). In both cases, the band-like enamel hypoplasia was located within the same limited area of the crowns of permanent teeth which contained a growth line indicating that this part of the teeth developed between the ages of 2 and 3 years. Enamel hypoplasia was characterized by involvement of contralateral teeth which exhibited a pattern of interference corresponding to the chronology of tooth formation (13). According to the medical history of the two siblings, there were no other etiological factors that could cause band-like enamel hypoplasia or enamel opacities. This interesting finding of a similar growth line on the teeth of both sisters leads us to propose that a systemic disorder in connection with Alstrom syndrome may have been the etiological factor which affected both siblings at the same age and influenced tooth formation for a limited time. This proposition is supported by the fact that the enamel hypoplasia located within specific and limited areas of the tooth crowns coincides with the tooth formation of the same chronology. Presumably, the sisters suffered from an inherited systemic disorder associated with the syndrome which disturbed the normal process of amelogenesis (14). Staining on the hypoplastic enamel band may have been due to pigment absorption from extrinsic materials after eruption (15). Since the discolored enamel band occurred in both sisters within the same area of the contralateral teeth, it may be considered a complication of Alstrom syndrome.

There is no information about pathological odontogenesis in previous reports of Alstrom syndrome. The results of this first attempt to characterize the oral-dental pathology in Alstrom syndrome may contribute to existing knowledge about the clinical manifestations of the syndrome.

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