Sotos syndrome with enamel hypoplasia: a case report

Mikiko Inokuchi* / Jouji Nomura** / Yoshihiko Mtsumura*** / Motoko Sekida**** / Toshiro Tagawa*****

A rare case of Sotos syndrome with enamel hypoplasia is described. Dental abnormalities include enamel hypoplasia, expansion of the pulp cavity, high arched palate, and absence of the bilateral premolar teeth of the mandible.
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INTRODUCTION

Sotos syndrome was first reported in 1964 by Sotos et al.[1] and was characterized by facilitated prenatal growth and bone age acceleration, abnormal craniofacial morphology, and psychomotor developmental retardation. Although a possibility of autosomal dominant inheritance was indicated, the pathogenesis and incidence have yet to be elucidated.[2]

This article reports a case of Sotos syndrome with enamel hypoplasia.

CASE REPORT

The patient was a 2-year and 2-month old male infant, who was first brought to our hospital for precise oral examinations on December, 1998. The family history showed that the parents were not related by blood, and the brother and other blood relatives were healthy. The medical history revealed that the patient was born by spontaneous delivery without respiratory disorders or jaundice after 40 week of pregnancy, and weighed 4,024 kg, and was 54 cm long. Both the values of body weight and stature were above the 90th percentile values in the st stature-growth percentile curves (physical growth values in infants, (The trend of national health, 1990). Apgar score was 10 points.

In the postnatal second month, the patient was found to have micrognathia, umbilical hernia, and light undescended testis. In the postnatal third month, because the patient showed developmental retardation, CT examination of the head, electroencephalography, and thyroid function test were performed, and slight encephalotrophy by CT and electroencephalogram of S and W were observed (Figure 1). Chromosome examination by G band staining showed that the chromosomes were composed of 46, MY chromosome with no abnormal nuclear types. Simultaneously, the patient was diagnosed as having atrial septal defect (ASD) by the presence of cardiac murmur, pulmonary emphysema-like chest radiographic image, and increase in pulmonary bloodstream. In the postnatal fifth month, the patient was diagnosed as having epilepsy with apnea, and has taken anti epileptic medication since then.

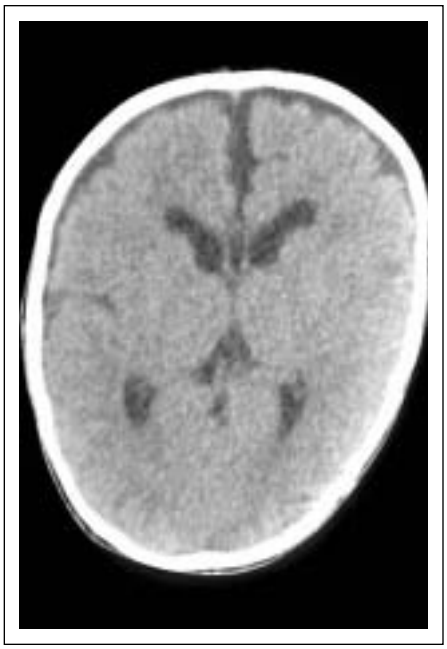

Figure 1. Cerebral CT findings with slight encephalatrophy.

* Mikiko Inokuchi, DDS, Senior Resident
** Jouji Nomura, DDS, PhD, Lecture
*** Yoshihiko Mtsumura, DDS, Assistant
**** Motoko Sekida, DDS, Assistant
***** Toshiro Tagawa, DDS, PhD, Professor and Chief of Department of Oral and Maxillofacial Surgery, Faculty of Medicine, Mie University, Japan.

Send all correspondence to Dr. Mikiko Inokuchi, Department of Oral and Maxillofacial Surgery, Faculty of Medicine, Mie University, 2-174 Edobashi, Tsu, Mie 514-8507, Japan.

TEL: +81-59-232-1111
FAX: +81-59-231-5207

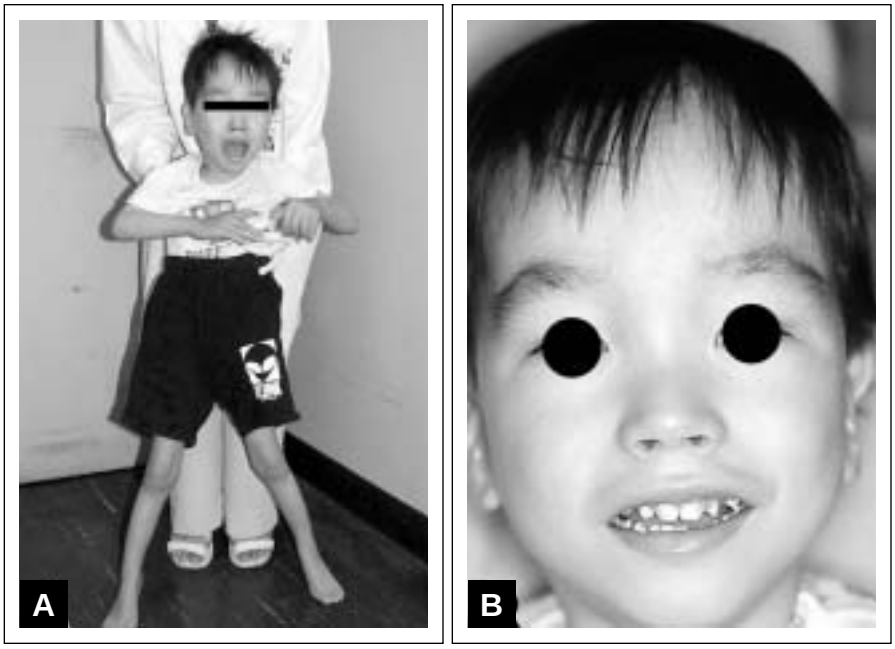

Figure 2. Extraoral findings. The large circumference (A), and the ocular hypertelorism (B) was revealed, with no abnormalites such a micrognathia.

At the age of one year and six months, the patient was diagnosed as having Sotos syndrome with ASD and psychomotor developmental retardation. At the age of one year and eleven months, the patient underwent an ASD direct closure operation, and was admitted to the department of thoracic surgery because of mediastinitis, and was referred to our department for precise oral examinations.

The general status showed, the height was 87.5cm, weight was 9.3kg, and head circumference was 50.1cm in the first examination. The height was average, and the weight was below the 10 percentile; however, the head circumference was above the 90 percentile (physical growth values in infants). (The trend of national health, 1990).

The intelligence of the patient was less than that of one-year old infants, and although the patient could hold his head up, he could not walk with tonicity of the muscles in the upper and lower limbs (Figure 2 A).

Although ocular hypertelorism was observed, the face was symmetrical without other abnormal findings (Figure 2 B).

Noting the oral findings, all deciduous teeth had erupted, and enamel hypoplasia with partial exposure of dentin was observed in all teeth except BAAB. However, no dental caries were observed, and the hardness of the enamel was confirmed by the examination with an explorer. A high arched palate was observed (Figure 3).

The head A-P radiographic findings showed absence of the 5445 tooth germs with no abnormal findings of the morphology of germs of the following permanent teeth and the jaw bones. Cephalometric radiography showed no abnormal findings such as micrognathia; however, regarding Facial Diagram (Japanese type), the cephalometric radiogram corresponded to that between six-year and two-month old and eight-year and eleven-month old children. Dental radiographic images showed expansion of the pulp cavity in all deciduous teeth (Figure 4).

The radiographic image of the left carpal bone showed four ossification nuclei and abnormality in the order of ossification. The inferior extremity of the radius was absent, and although capitate and hamate bones were normal, ossification of the triangular bone was facilitated and ossification of the scaphoid bone was retarded. Hypertrophy in the ends of the bones of digits were confirmed (Figure 5).

MODEL ANALYSIS

The maxillary C-C dental arch width was 27.5mm, which was more than the mean value ±1SD, and dental arch length was 34.0mm, which was than the mean value ±2SD. the mandibular C-C dental arch width was 20.0mm, which was less than the mean value, and dental arch length was 29.1mm, which was more than the mean value ±2SD. The mesiodistal width of the teeth, the width of all maxillary teeth except BB were less than the mean value, and in the mandibular, EBAABCE was less than the mean value, and showed a tendency in which teeth were small compared with the size of the jaw bone (Japanese, measurement of deciduous teeth).

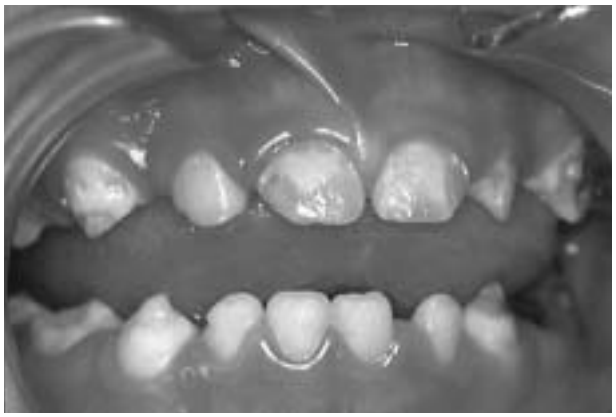

Figure 3. Oral findings. Enamel hypoplasia with partial exposure of dentin was observed in all deciduous teeth except BAAB.

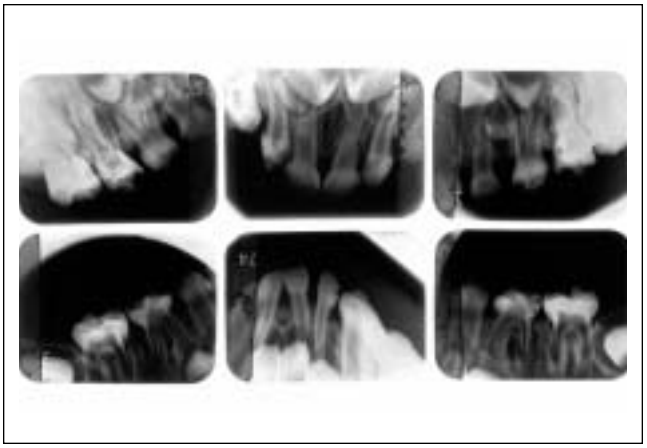

Figure 4. Dental radiographic findings showed expansion of the pulp cavity in all deciduous teeth.

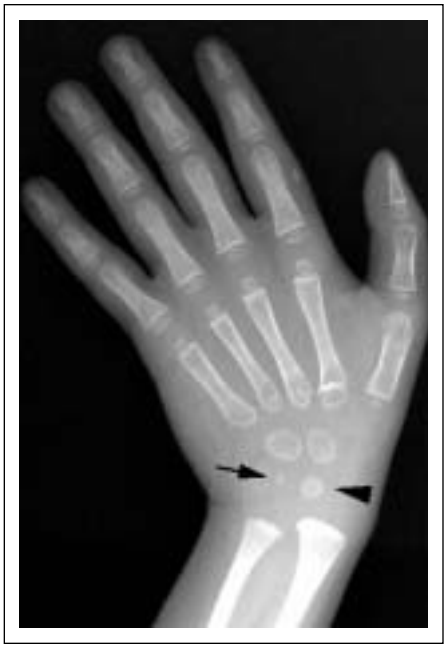

Figure 5. Radiographic findings of the left carpal bone. The scaphoid bone was retarded (arrow), whereas the triangular bone was facilitated (arrow head).

DISCUSSION

The Sotos syndrome has been speculated to be autosomal dominant. Schrander-Stumpel *et al.*[3] inferred that the genetic locus of this syndrome was in 3p21 or 6p21; however, relationships with autosomal recessive inheritance[4] or fragile X chromosome syndrome were indicated,[5] and no certain results were obtained regarding heredity.

There were other reports in which retardation of cerebrospinal fluid circulation from the embryonal period and intermittent intracranial hypertension during sleeping were related to the occurrence.[6] Others believe that oversecretion of growth hormone from the embryonal period to infancy was related to growth acceleration;[7] however, these reports were uncertain and showed no certain notions regarding the male-to-female ratio or incidence.

The other diseases associated with overgrowth are gigantism and fragile X syndrome. Gigantism is differentiated from Sotos syndrome by the abnormally great arm span, a special physiognomy, advanced bone age, and a variable degree of psychomotor retardation.[8] Fragile X syndrome is differentiated by the presence of a fragile site in the terminal site (Xq27.3) of the long arm of X chromosome, and X-linked mental retardation.[9] Sotos syndrome is differentiated from these two disease by the correction of overgrowth with aging.[10]

Sotos syndrome is characterized by facilitated prenatal growth, bone age acceleration, craniofacial abnormality similar to that of acromegaly, and nonprogressive supraneural symptoms.[11,12] Cole *et al.*[10] indicated that diagnostic criteria of Sotos syndrome are specific appearance like ocular hypertelorism and four other signs: tall height, macrocranium, bone age acceleration, and developmental retardation.

Complications of Sotos syndrome are epilepsy,[2] heart disease, genitourinary diseases,[3] and incidence of hepatoma, Wilms tumor, vaginal tumor,[2] and fibroblastoma[14] is high. Cohen *et al.*[14] reported complications such as small cell carcinoma, multiple hemangioma, osteochondrosis, and giant cell granuloma.

Oral findings included, prognathism,[1,2,10] premature eruption of the teeth and high arched palate.[1,7,10,11] These findings were reported without other detailed reports. In the present case, absence of tooth germs of the permanent teeth, high arched palate, and as interesting findings, enamel hypoplasia of all deciduous teeth and expansion of the pulp cavity were found.

Generally, enamel hypoplasia by autosomal dominant inheritance is classified into hypoplastic type, hypocalcification type, and hypomaturation type.[1] Hypoplastic type shows primary derangement of

enamel formation with no substantial changes in dentin, dental pulp, or periodontal ligament. Although the enamel is thin, it is hard and shows pits or a granule-like concavo-convex surface.[2]

Hypocalcification type is the most commonly observed type, in which formation of normal thickness of the enamel accompanied by hypocalcification is observed.[3]

Hypomaturation type is related to maturation of the enamel or derangement of secondary calcification. Thickness of the formed enamel is nearly normal, and the enamel is harder than that in hypocalcification type.

Hypomaturation type is classified into four types: autosomal recessive pigmentation type, X-linked recessive type, snow-capped tooth type, and autosomal dominant type with hypoplasia and taurodont.[10,13] Among hereditary diseases regarding formation of the tooth morphology, abnormality in the enamel matrix is observed in Aarskog syndrome (X-chromosome-linked), Goltz syndrome (X chromosome-linked), and Trichodento osseous syndrome (autosomal dominant). As previously described, Sotos syndrome was indicated to be possibly associated with autosomal dominant inheritance, and the present case of with that possibility was regarding as hypoplastic type, because hardness of the enamel as preserved.

On the other hand, dentinogenesis imperfecta related to heredity is classified into three types.[1,15,16] Type 1, which is accompanied by osteogenesis imperfecta;[2] Type 2, which is not accompanied by osteogenesis imperfecta; and[3] Type 3, Brandywine type.

Most cases of type 1 show autosomal dominant inheritance, and some show recessive inheritance.

Type 2 shows autosomal dominant inheritance, and the pulp cavity and root canal are rarely observed, whereas, the expansion the pulp cavity has been occasionally reported.[12]

Type 3 shows autosomal dominant inheritance, and some of which were reported to have shell teeth.

The radiographic images showed marked expansion of the pulpal cavity. We did not analyze the components of the teeth in the present case, because none of the teeth required extraction; however, the expansion of the pulp cavity in this case is considered to be a result of dentinogenesis imperfecta. Considering that together enamel hypoplasia and dentinogenesis imperfecta can genetically occur, this patients symptoms should be added to those of Sotos syndrome.

Further study is necessary to obtain a certain prognosis of Sotos syndrome; however, symptoms of the central nervous system such as psychomotor developmental retardation improve with aging[10] and the terminal height usually is within the normal range, with some cases exceeding ±2SD.[10]

On the other hand, because the incidence of malignancies as complications is high, sufficient care must be exercised for the general conditions. Regarding the changes in discrepancy between the jaw bones and teeth, long-term observation of the changes with growth after succedaneous replacement of the permanent teeth.

This paper is the first detailed report to our knowledge regarding the oral symptoms of Sotos syndrome.

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