

Abstract

Kozlowski syndrome is the most common type of spondylometaphyseal dysplasia (SMD). It is characterized by short stature (130 to 150 cm), pectus carinatum, limited elbow and hip movement, mild bowleg deformity, and curvature of the spinal column. Children with Kozlowski dwarfism usually are not recognized at birth, since they have normal clinical features, weight, and size. This article reports the dental treatment and oral findings of a 14-year-old female patient with Kozlowski dwarfism. (Pediatr Dent 15:49-52, 1993)

Spondylometaphyseal dysplasia (Kozlowski type): case report

Cynthia M. Guzman, DMD Gerald R. Aaron, DDS, MS

Introduction

Spondylometaphyseal dysplasia (SMD) is a disorder comprising a group of diseases in which the spine and metaphyses of the tubular bones are affected. The most common type of spondylometaphyseal dysplasia is Kozlowski dwarfism; several forms of this syndrome have been defined by radiographic and genetic analysis.

Children with Kozlowski dwarfism usually are not recognized at birth, since they have normal clinical features, weight, and size. This disease is recognized in pre-school age as shortening of stature and kyphotic appearance.¹ Patients with Kozlowski dwarfism often were reported to have Morquio's disease, because of the similar characteristics. In Morquio's disease, patients have short stature, short neck, and "knock knees," but are differentiated by their corneal opacities, cardiac problems, specific dental findings, and urinary excretion of keratin sulfate.²

Also present in Kozlowski dwarfism are pectus carinatum, limited elbow and hip movement, mild bowleg deformity, and kyphosis (curvature of the spinal column). These associated problems often require surgical treatment. Patients reach 130 to 150 cm in height,² and usually have a normal life span.

This article reports the oral findings and dental treatment of a patient with Kozlowski dwarfism.

Literature Review

The term "metaphyseal dysostosis" first was used to designate a condition with extreme shortness and metaphyseal irregularities of the lower limbs and hands.³ A milder form of metaphyseal dysostosis was reported by

Schmidt.⁴ Sporadic cases of metaphyseal dysostosis associated with generalized platyspondyly and with radiographic alterations predominantly in the spine, pelvis, and the proximal femora, were termed spondylometaphyseal dysostosis.⁵

Spondylometaphyseal dysplasia (SMD) is a generic term for a heterogeneous group of skeletal dysplasias in which spinal involvement (platyspondyly) is associated with varying degrees of metaphyseal abnormality.⁶ The most thoroughly defined SMD (Kozlowski type, a form of short-trunk dwarfism) is diagnosed readily by characteristic radiographic findings which include: 1) generalized platyspondyly with anterior tapering of vertebrae (especially in the thoracic region); 2) generalized metaphyseal dysplasia, which often is most severe in the femoral necks (coxa vara is common) but variable in other sites; and 3) apparent retardation of bone age in the carpal and tarsal bones.^{5,7}

Kozlowski-type SMD should be differentiated from Morquio's disease. Morquio's disease is a rare skeletal dysplasia characterized by abnormal metabolism of the mucopolysaccharide keratin sulfate. Patients with Morquio's disease have disproportionate short stature, neck shortness, sternal protrusion, laxity of the ligaments, "knock knees," flat feet, and disproportionately long extremities.⁸ It is inherited as an autosomal recessive trait, and is estimated to occur in 1/40,000 births.⁹ The pattern of bone changes in Morquio's disease depends very much upon the patient's age. The platyspondyly in Morquio's disease, like Kozlowski's dwarfism, is characterized by a central anterior tongue-like protrusion, hypoplasia of the vertebral bodies at the thoracolumbar junction, and hypoplasia of the odontoid process.² Just as in Kozlowski dwarfism, associated thoracic kyphosis in Morquio's usually is prominent. Corneal opacities and dental anomalies are constant features in Morquio's disease. In Morquio's, the enamel layer of the teeth is very thin, and the teeth are smaller and more opaque than usual.⁸ The permanent

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posterior teeth have generalized pitting on the buccal surface, and the permanent maxillary incisors are spade shaped and spaced. An anterior open bite may be observed and has been associated with the presence of an enlarged, protruding tongue.¹⁰ Midface hypoplasia with a depressed nasal bridge and protrusion of the mandible also have been reported by Behrman.¹¹ In addition, children with this disorder are much shorter than those with Kozlowski dwarfism.

SMD (Kozlowski type) has been reported mainly in the European literature. It usually is described as autosomal dominant but probably also occurs as an X-linked recessive type with most cases representing fresh mutations¹² with a prevalence of one per million or fewer live births.

The most common clinical findings in SMD (Kozlowski type) are short neck and disproportionately short trunk with dorsal kyphosis (curvature of the thoracic spine). Diagnosis can be made by exposing radiographs of the spine and pelvis. Spinal changes are well-established by 2 years of age with platyspondylia with an anterior deformity in the form of a "tongue" present in the upper thoracic, cervical, and lumbar spine.¹³ The dorsolumbar vertebrae are more oval-shaped with a recessed twelfth thoracic or first lumbar vertebra. Generalized platyspondylia and femoral neck changes are the two main diagnostic features in a radiographic examination. Generalized platyspondylia is greatest in the high dorsal spine by 5 years of age. The femoral heads are well seated. The femoral necks are short, and the femoral neckshaft angle shows a mild coxa valga, which is maintained with growth.¹³ A unique radiographic feature in childhood is the thick metaphyseal collar around the femoral neck. Calcium and phosphorus metabolism has not been found to be abnormal, nor has abnormal mucopolysacchariduria been observed.¹⁴

In SMD (Kozlowski type), the thorax is described as pectus carinatum (pigeon breast) and the pelvis has short iliac wings with flat irregular acetabula.¹⁴ Coxa vara (deformity of the hip joint with decrease in the angle of inclination between the neck and shaft of the femur) usually is present. Genu valgum (knock knee) also has been observed. Limbs have irregular metaphyses but are normal in length. The elbows often are more disturbed than the knees. Epiphyseal development is relatively normal and bone age tends to be delayed. Short, stocky hands also are seen in this type of SMD due to hypoplastic carpal bones with late ossification. Waddling and halting gait with limitation of joint mobility are characteristic of Kozlowski dwarfism. This condition, plus early degenerative joint changes, usually lead to great discomfort and make surgery necessary.

In morphological and biochemical studies,¹⁵ a probable pathogenic mechanism of a particular biochemical defect was suggested as etiologic in several of the chondrodysplasias. Abnormalities, probably related to a degradative lysosomal process of proteoglycans in chondrocytes, were found in SMD of the Kozlowski type.¹⁵

Unaffected in this disease are the facial appearance, intelligence, sense organs, and viscera. Literature concerning the orofacial findings of Kozlowski dwarfism was not found. No description of dental abnormalities was reported in the literature.

Case Report

A 12-year-old Caucasian female diagnosed with Kozlowski dwarfism was referred to pediatric dentistry at Fort Lewis, Washington. The patient was an alert child with normal mental development.

She was the third child of healthy parents. The pregnancy was uncomplicated. Her birth-weight was 6 pounds 11 ounces and her length was 19 inches.

Motor development was normal but growth was delayed. At 3 years of age, the child was 20 inches tall, making her shorter than other children of her age. This difference became increasingly apparent when she first went to school. She sat at 7 months, walked at 14 months, and was completely toilet trained by 16 months. The first teeth were noted at 6 months. She reportedly developed "knock knees" after she began walking. Since birth, she has undergone cervical fusion, spinal fusion, and rebuilding of hip and knees. Additional surgery is planned.

Dental history included a Class III malocclusion and TMD musculature pain resulting from posterior disocclusion and prognathic position in centric relation. A night guard was fabricated, and the treatment plan was to evaluate eruption guidance and recall at every six months.

The most striking feature of this patient at 14 years of age was her height of 37 1/2 inches (Fig 1). (According to the National Center for Health Statistics, at 14 years of age the average height of a female adolescent is 63 inches).

Dental examination revealed a late mixed dentition with maxillary and mandibular primary first molars still present.

Other significant dental findings were small permanent teeth (microdontia), diastema, and an upper midline shift to the left of approximately 3 mm. The patient's maxillary and mandibular incisors were compared to Moyer's¹⁶ average mesiodistal diameters of permanent dentition in females. The maxillary centrals were on average 1.3 mm smaller than Moyer's standard. Other anterior teeth also exhibited decreased size.

In addition, the patient had a relatively large tongue (macroglossia), which overlaid the occlusal surfaces of her mandibular teeth. The patient had good oral hygiene and her soft tissues were healthy. The maxillary first molars had occlusal caries. The crown of the mandibular right permanent canine was fractured.

Steiner analysis was used for the cephalometric tracing: SNA-65°; SNB-67°; ANB- -2°; 1/NA-29°; I\NB-28°; Occl-SN-29°; GoGN-SN- 43°; FMA-32°; IMPA-88°; 1/1- 135°. These results describe a profile of anterior divergence with a skeletal Class III malocclusion (Figs 2, 3). The high angle represents a clockwise growth with dolichofacial growth trends.

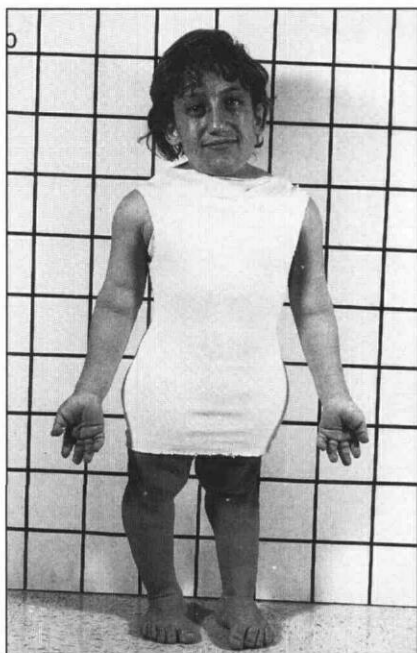


Fig 1. Full body view showing normal head and size and length of limbs, with a short body, knock knees and short and stocky hands.



Fig 2. Lateral view showing anterior divergent profile.

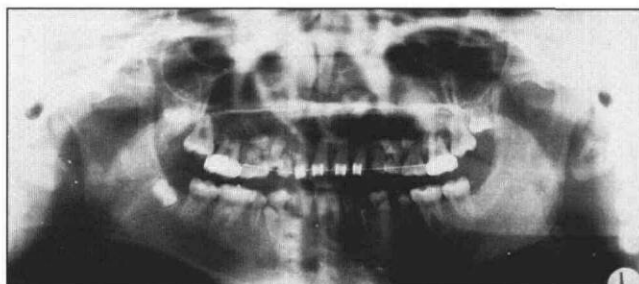


Fig 3. Panelipse radiograph showing delayed eruption of permanent teeth..

Discussion

Unique dental findings were observed in a patient with a disease that has been reported in the European literature but is not well described in the American literature. No information was found in the literature concerning the orofacial structures or dental treatment for these patients.

One feature of this child's dentition was microdontia. According to Shafer,¹⁷ in true generalized microdontia, all teeth are normally formed but smaller than normal and this occurs in some cases of pituitary dwarfism. This patient had a complete primary dentition (as evidenced by radiographs) and was developing a complete complement of permanent teeth. She also had a large tongue (macroglossia), which according to McDonald and Avery¹⁸ may cause both an abnormal growth pattern of the jaw and malocclusion. Flaring of lower anteriors and an Angle Class III occasionally are the result of macroglossia.

The patient was treated with fixed appliances for correction of the anterior crossbite and closure of the midline diastema. To encourage the permanent premolars into occlusion, the first primary molars were extracted. During her limited orthodontic treatment, she developed periapical radiolucencies on the mandibular left permanent canine, and the lower right mandibular central incisor which were asymptomatic. The teeth were treated with root canal therapy. After the crossbite and diastema closure were corrected, she developed a posterior crossbite on the right side which was corrected with an active transpalatal bar.

Morquio's disease is one of the diseases whose features are similar to SMD (Kozlowski type). One of the main dental findings in Morquio's reported by Kinirons⁸ was spade-shaped and spaced permanent maxillary incisors. In this patient with Kozlowski dwarfism, the maxillary incisors were spaced but not shaped abnormally. Langer¹⁹ reported that eight of 10 patients with Morquio's

syndrome showed thin enamel with sharp pointed cusps in both primary and permanent teeth. This patient had thin enamel, with no pitting and no pointed cusps in her primary or permanent dentition.

Major Guzman is currently a pediatric dentist in the U.S. Army, Fort Clayton, Republic of Panama. Dr. Aaron, U.S. Army retired, is in private practice in Vancouver, WA.

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