Advanced periodontitis associated with Larsen's syndrome: case report

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Abstract

Larsen's syndrome is a genetic disorder of connective tissue characterized by multiple large-joint dislocations, abnormalities of hands and feet, and anomalous facial features. Although findings involving multiple systems have been added to its clinical and pathological entity, oral pathology besides cleft palate has not been reported. We report here advanced periodontitis with severe gingival inflammation in a 29-year-old Larsen's syndrome patient. Bacterial culture of subgingival plaque identified the periodontal pathogens that were present in numbers above the normal range. We speculate that the underlying defect in connective tissue may predispose Larsen's syndrome patients to periodontal destruction provoked by microbiological factors. (Pediatr Dent 17:62–64, 1995)

Larsen's syndrome is a genetic disorder of connective tissue characterized by multiple large-joint dislocations, abnormalities of hands and feet, and anomalous facial features. The transmission of Larsen's syndrome is unclear with both autosomal dominant and autosomal recessive forms having been reported.[1] Since the original description by Larsen et al. in 1950,[2] a few Larsen's syndrome cases have been reported in the literature and new findings have added to our understanding of the clinical and pathological nature of the syndrome.[3-7]

The typical clinical findings in Larsen's syndrome are multiple joint dislocations that involve the knees, hips, and elbows, talipes equinovarus or equinovalgus deformities of the feet, cylindrical-shaped fingers and spatulate thumbs.[1-6] The patients usually have a flattened face with depressed nasal bridge, frontal bossing, and widely spaced eyes.[1-6]

Several reports describe malformations of the larynx, trachea, and bronchi, which subsequently cause serious respiratory distress.[3, 6, 7] Cardiovascular disorders including dilatation of aortic roots, tortuosity, and dilatation of cranial and abdominal arteries also are associated with Larsen's syndrome.[8-9] Another recently reported finding was mixed hearing loss.[10] Also, conductive and sensorineural components are thought to be abnormal in patients with Larsen's syndrome.[10] Intelligence is normal.[1, 2, 10] The only oral manifestation reported is cleft palate or bifid uvula.[2, 4-5] There have been no reports describing the periodontal condition in patients with this syndrome. We report here a case of advanced periodontitis in a 29-year-old female with Larsen's syndrome.

Case report

Medical and dental history

RB, a 29-year-old African-American female was referred to the University of Illinois pediatric dentistry clinic for general dental care. Her medical and dental histories were provided by her mother and taken from her medical records. Diagnosis of Larsen's syndrome was based on the presence of bilateral equinovarus, subluxation of the knees, dislocation of the radial heads, and thoracolumbar scoliosis. From age 2 months to age 19 years, a series of orthopedic surgeries and castings were performed to correct the deformities. At age 17 years, RB suffered from cor pulmonale, which was thought to be secondary to restrictive lung disease resulting from severe scoliosis. She was placed on digoxin and had a tracheostomy. During the next 10 years, she remained stable with no medication.

Medical history revealed myopia corrected by glasses, no hearing impairment, and no other medical problems. Family history revealed no musculoskeletal or periodontal problems similar to those of the patient.

RB received restoration of a fractured maxillary left central incisor several times up until 5 years prior to this visit. She reported that she brushed her teeth once a day, but never used dental floss. The patient and her mother said they did not notice premature exfoliation of primary teeth. The patient said that her teeth had become loose during her second decade of life and two lower central incisors had been lost. She had fallen and fractured the mesio-incisal aspect of the maxillary left central incisor when she was a child. RB had not received health care until she was referred for consultation at the University of Illinois Hospital.

Clinical and radiographic evaluation

RB presented with dwarfism (109 cm in height and 26.3 kg in weight). Her head was symmetrical and normocephalic. Her face was flattened with a depressed nasal bridge, exophthalmos and widely spaced eyes

(Fig 1). She had difficulty opening her mouth for more than a few seconds. Macroglossia and difficulty in controlling tongue movement also was observed. There was no pain upon palpation of the temporomandibular joints.

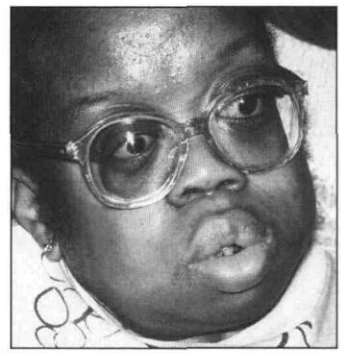

Fig 1. Characteristic face of Larsen's syndrome: flattened face, depressed nasal bridge, exophthalmos and widely spaced eyes.

Intraoral examination revealed a permanent dentition with two mandibular central incisors missing. Caries was present only on the maxillary right third molar. Oral hygiene was poor with an O'Leary's Plaque Index[11] of 100%. Gingival inflammation was severe throughout the mouth with Gingival Index[12] of 3 (Fig 2). All the teeth demonstrated mobility varying from degree I (combined facial-lingual movement totaling 1 mm) to degree III (combined facial-lingual movement totaling 3 mm or more and/or tooth depressable into socket)[13] except three maxillary right molars and the mandibular right second molar. Full mouth probing demonstrated extensive loss of periodontal attachment in all the teeth. The probing depth ranged from 3 mm to 11 mm with an average of 6 mm. The occlusal examination revealed an Angle Class I malocclusion with posterior cross bite.

Full mouth radiographic examination revealed 40 to 95% alveolar bone loss associated with all the teeth, especially maxillary and mandibular incisors (Fig 3). Supra- and subgingival calculus was evident throughout the mouth. There appeared to be no pulpal pathology.

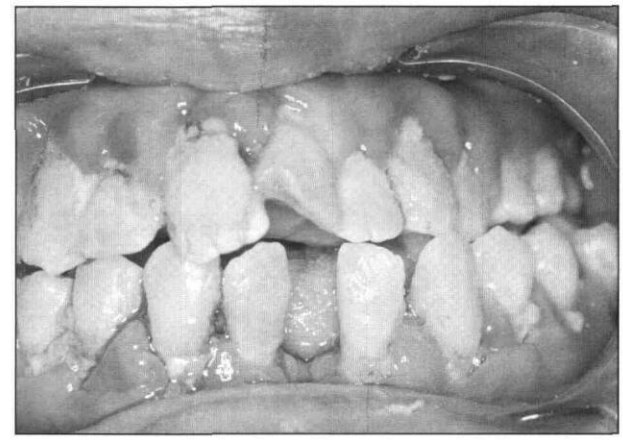

Fig 2. Poor oral hygiene with severe gingival inflammation is evident.

Laboratory evaluation

Bacterial culture of subgingival plaque identified the following pathogens that were above normal ranges: *Bacteroides forsythus* (1.8%), *Porphyromonas gingivalis* (3.90%), and *Peptostreptococcus micros* (3.33%). Hematological tests were performed with the following results: RBC $4.99 \times 10^6/\mu l$, hematocrit 39.7%, hemoglobin 12.7g/dl, WBC $3.7 \times 10^3/\mu l$ with differential showing polymorphonuclear leukocytes 54.0%, lymphocytes 38.8%, monocytes 6.3%, eosinophils 0.2%, basophils 0.7%, and platelets $275 \times 10^3/\mu l$. Serum alkaline phosphatase was 71U/l, and fasting glucose 79.0 mg/dl. All the above test results appeared to be within normal limits except the WBC, which was slightly low.

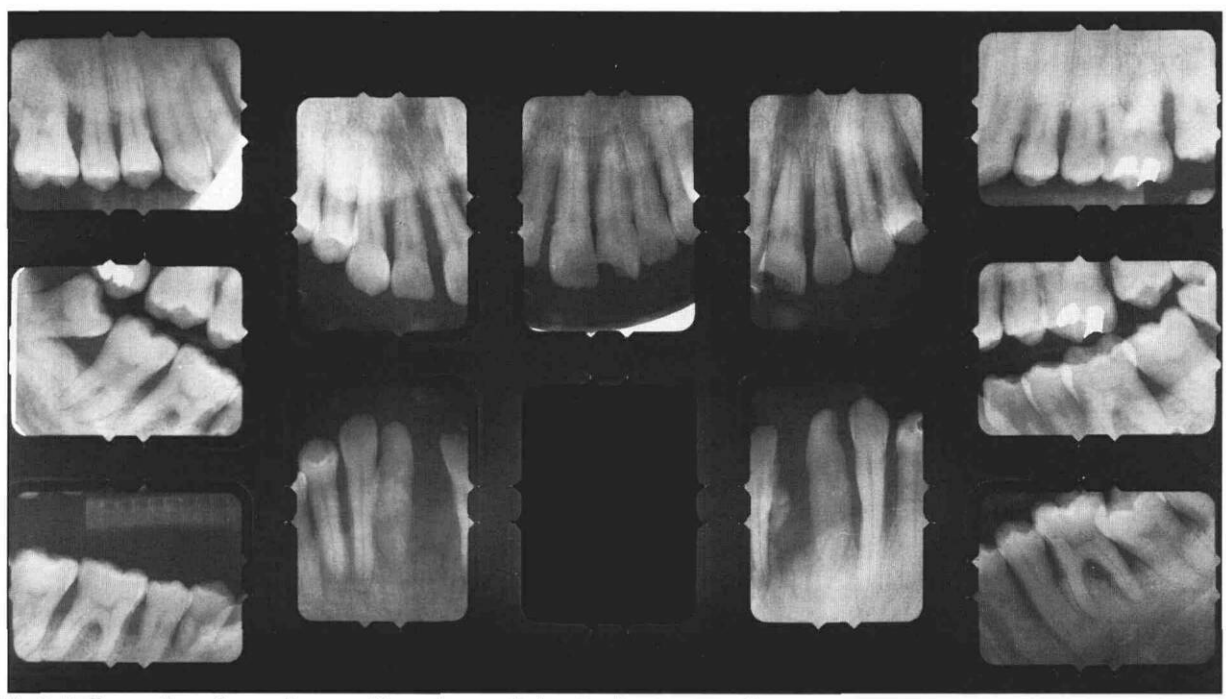

Fig 3. Full-mouth radiographs revealing advanced alveolar bone resorption.

Discussion

This report presents a case of Larsen's syndrome with advanced periodontitis. Oral or dental pathology besides cleft palate or cleft uvula in Larsen's syndrome patients has not been reported previously. Larsen's syndrome was reported initially as an orthopedic abnormality and has been associated with problems of many other systems that reflect a generalized mesenchymal disorder. Microscopic observation of malformed larynx, trachea, and bronchi in a Larsen's syndrome patient by Ronningen and Bjerkreim[3] revealed a marked reduction of elastic fibers. Lutter[1] described wound healing problems in some patients undergoing orthopedic surgery, suggesting a defect in collagen production. In studies at the molecular level, analysis has demonstrated a mutation of a gene involved in collagen production.[14]

The etiology of periodontitis is bacteria and their byproducts. However, it is believed that the periodontitis is more advanced if a subject has a defect in the host defense system[15–17] or defects in certain cell types present in periodontium.[18–20] Such examples are Chédiak-Higashi syndrome,[15] leukocyte adhesion deficiency syndrome,[16] neutropenia,[17] Papillon-Lefèvre syndrome,[18] hypophosphatasia[19] and Ehlers-Danlos syndrome.[20]

Neutrophils are believed to be the first line of defense against bacterial plaque in the gingival crevice. In cases of Chédiak-Higashi syndrome, leukocyte adhesion deficiency syndrome, and neutropenia, neutrophils either have a defect in killing mechanisms, a defect in migration, or are present in low numbers, respectively. In cases of Papillon-Lefèvre syndrome, Ehlers-Danlos syndrome, and hypophosphatasia, the suggested etiologies are defects in epithelia, collagen, or cementum, respectively. These defects in different cell types seem to accelerate the periodontal breakdown when it is superimposed with bacterial infection.

We suspect that a collagen defect is an underlying contributing factor for the advanced periodontitis seen in Larsen's syndrome. As with other types of periodontal disease that affect children and young adults (Chédiak-Higashi syndrome, leukocyte adhesion deficiency syndrome, neutropenia, and Papillon-Lefèvre syndrome) early detection is important in providing the opportunity to prevent or retard further tissue destruction.[21] The periodontal condition of Larsen's syndrome patients should be closely evaluated from a young age. Given the fact that patients with Larsen's syndrome may have other systemic involvement including cardiovascular, respiratory, and spinal problems, appropriate medical consultation is important before any dental treatment is rendered. Since wound healing has been a concern in Larsen's syndrome patients, any procedures that provoke bleeding such as probing, scaling and root planning, or tooth extraction should be performed with caution.

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