Generalized aggressive periodontitis in a child with 92, XXYY / 46,XY mosaicism: report of a second case

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SUMMARY: Olgun-Erdemir E, Yıldırım MS, Karşıyaka M. Generalized aggressive periodontitis in a child with 92,XXYY / 46,XY mosaicism: report of a second case. Turk J Pediatr 2010; 52: 94-96.

The present case report describes the oral features of tetraploid/diploid mosaicism. An 11-year-old boy with severe periodontal destruction is presented in this report. He was examined clinically, radiologically, immunologically, and genetically. Significant edema of the gingiva, severe sulcular bleeding on probing and mobility of many teeth were detected on intraoral examination. There was severe generalized maxillary and mandibular bone loss as determined by radiological examination. He was diagnosed as generalized aggressive periodontitis. The cytogenetic examination revealed 92,XXYY (4%) / 46,XY (96%) karyotype indicating tetraploid/diploid mosaicism. Non-surgical periodontal therapy was applied and he is currently under a routine follow-up period. In this report, the oral characteristics of tetraploid/diploid mosaicism are described. Dental practitioners should see these patients in some distinct periods, because tetraploid/diploid mosaicism subjects may have aggressive periodontitis.

Key words: mosaicism, tetraploid/diploid, therapy, aggressive periodontitis.

Tetraploidy is rare in human pregnancies. It is a relatively common finding in spontaneously aborted fetuses during the first trimester[1-3]. The mechanism leading to tetraploidy consists of inhibition of the movement of the two asters and aberration of spindle formation. The mosaic form develops from eggs in which only one of the nuclei became tetraploid while the other one divided normally[4].

Some clinical features of tetraploidy are severely delayed growth and craniofacial abnormalities including micrognathia, cleft palate, small mouth, malformed low-set ears, and prominent forehead[5-9]. The oral manifestations of this genetic abnormality have been described in only one article in the literature[10]. The aim of this case report is to present the oral and radiological manifestations of a male with tetraploid/diploid mosaicism.

Case Report

An 11-year-old boy (Fig. 1) was referred to the Periodontology Clinic in Kırıkkale University Faculty of Dentistry due to his severe periodontal symptoms. He is the first child of a healthy and non-consanguineous family. Associated findings were sparse hair, long eyelashes and retrognathia. Intraoral examination revealed significant edema of the gingiva, severe sulcular bleeding on probing and mobility of many teeth (Fig. 2). He had stopped brushing his teeth due to bleeding and sore gums. Slight calculus formation, moderate plaque and materia alba accumulation were present. The periodontal probing depths ranged from 2 to 10 mm. Class I-II and III mobilities were determined in both incisors and molars.

Significant alveolar bone loss affecting both maxillary and mandibular alveolar bones could be easily seen by radiographic examination (Fig. 3). There was severe bone destruction around the mandibular and maxillary anterior and posterior teeth and Class IV furcation defects in the mandibular right and left molars. Class II and III furcation involvements were

also detected in the maxillary left and right first molars, respectively. According to the clinical and radiological examinations, he was diagnosed as having generalized aggressive periodontitis[11]. The patient was referred for clinical genetic consultation.

Complete blood count, immunologic assessments and thyroid hormone levels were normal. His growth parameters and motor/mental developmental skills were appropriate for his age. However, karyotype analysis from peripheral blood lymphocytes revealed 92,XXYY (4%) / 46,XY (96%) karyotype, indicating tetraploid/diploid mosaicism.

After baseline measurements and extraction of mandibular and maxillary first molars and mandibular central and lateral incisors due to severe mobility and pus formation, the patient received a primary phase of periodontal treatment including oral hygiene instruction, scaling and root planing. Full-mouth supragingival professional tooth cleaning (scaling and polishing) was performed in two sessions and oral hygiene instructions were given at that time. He was instructed in the use of a soft nylon brush and dental floss. One week later, the subsequent non-surgical treatment consisted of subgingival debridement applied using Gracey curettes (Hu Friedy Instruments, Chicago, IL, USA) under local anesthesia in two sessions. Mouth rinse and antibiotics were prescribed. All extracted teeth were restored by a flipper. Re-evaluation procedures were conducted one, three and six months later. He was given non-surgical periodontal treatment and advised to improve his oral hygiene measures. He is still attending the routine follow-up visits (Fig. 4).

Discussion

Although tetraploidy mosaicism appears to be very rare[12], it may occur with a wide variation of congenital anomalies and different degrees of growth and mental retardation[13]. The present case had some signs also found in other cases of tetraploid/diploid mosaicism: long eyelashes, mandibular retrognathia and highly arched palate. He did not have growth delay, mental retardation or congenital anomalies except for the minor dysmorphic features mentioned earlier.

Fig. 1. Clinical photograph of the patient with micrognathia.

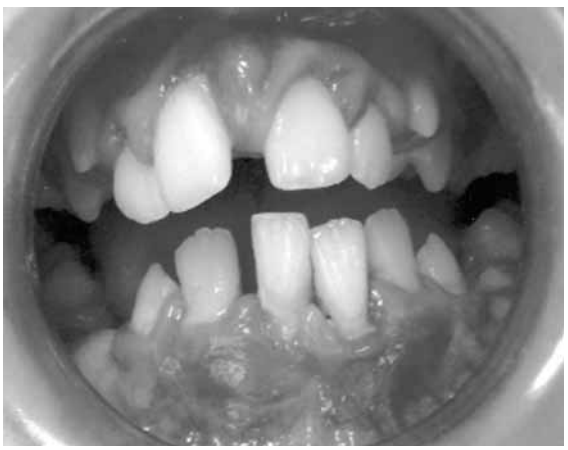

Fig. 2. Intraoral photograph of the patient with generalized aggressive periodontitis.

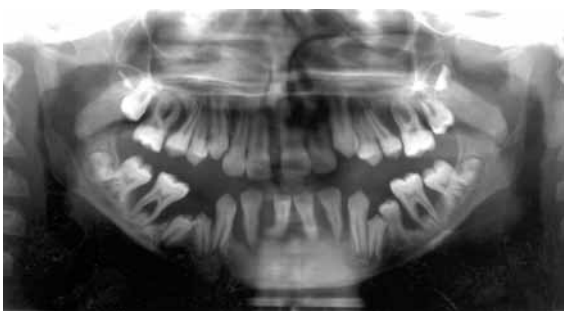

Figure 3. Panoramic radiograph showing generalized alveolar bone loss.

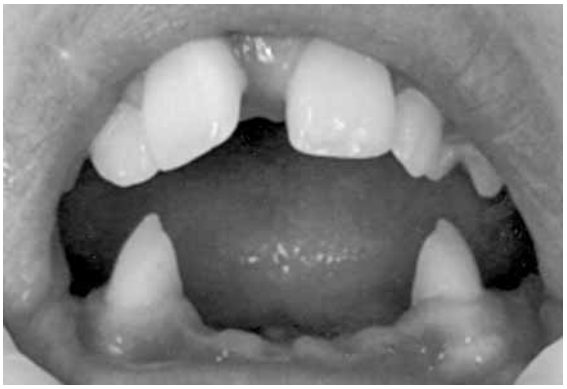

Fig. 4. Clinical photograph of the patient, six months after the non-surgical periodontal therapy.

This is the second case of tetraploid/diploid mosaicism with generalized aggressive periodontitis with severe periodontal destruction[10]. The clinical and radiological oral manifestations are very similar in these two reports. Significant general inflammation, edema of gingiva, severe sulcular bleeding on probing, slight calculus formation, deep periodontal pocket depths, and significant alveolar bone loss affecting maxillary and mandibular alveolar bones were noted in both cases. In the case by Tözüm et al.[10], 92,XXYY (25%) / 46,XY (75%) karyotype, indicating a higher percentage of tetraploid/diploid mosaicism, was reported. Sousa et al.[13] suggested that clinical signs were present in multiple organ systems and that there was no relationship between the percentage of tetraploid cells and the severity of malformations.

The present case was treated only with non-surgical periodontal treatment because there was indication of extraction of many teeth and the remaining teeth did not necessitate surgical intervention. In the follow-up periods, the patient was also advised to increase his oral hygiene measures.

In conclusion, this is the second patient with mosaic tetrasomy reported to have aggressive periodontitis, supporting the previous observation in the literature. Periodontal problems should be kept in mind in individuals with tetrasomy, and conversely, cytogenetic analysis should be performed in individuals with severe periodontal and minor dysmorphic findings.

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