

A constellation of dental anomalies in a chromosomal deletion syndrome (7q32): case report

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Abstract

A case is reported of a patient with deletion of the long arm of chromosome 7 at a highly specific locus (7q32). In addition to significant craniofacial stigmata and global developmental delay, the patient presented with numerous clinical and radiographic dental anomalies observed over a 10-year period. Hypodontia, accessory roots, dens invaginatus, hypoplastic enamel, and numerous pulpal anomalies all were noted. Some of these dental findings suggest trichodentoosseous syndrome (TDO), although the other stigmata do not. The wide variety of dental findings in this patient may help to define the role of chromosome 7q32 in dental development. (Pediatr Dent 16:306-9, 1994)

Introduction

A pattern of characteristic clinical manifestations such as nail dysplasia, patella hypoplasia, and iliac spurs seen in nail-patella syndrome, helps to define a specific syndrome. In turn, syndromes may be defined on the basis of chromosomal abnormalities. The process of meiosis gives rise to errors such as chromosomal maldistribution, duplication, or breakage resulting in deletion of a chromosome or a chromosomal portion.¹

Numerous chromosomal deletion syndromes have been described based upon whether the short arm (p) or long arm (q) is deleted. More precise identification is made on the basis of the locus or range of loci affected. Deletion of the long arm of chromosome 7 (7q-) is a rare phenomenon with less than 30 reported cases.² Deletions involving loci 21-32 of the long arm (7q21-q32) do not result in a recognizable clinical syndrome, but deletions from locus 32 to the terminal end of the long arm of chromosome 7 (7q32-qter) result in a constellation of significant stigmata.³ Commonly observed in 7q32-qter are low birth weight, postnatal growth deficiency, developmental delay, mental retardation, microcephaly, prominent forehead, eye anomalies, hypertelorism, bulbous nasal tip, broad nasal bridge, wide philtrum, large mouth, micrognathia, cleft lip/palate, ear anomalies, abnormal palmar creases, distal limb deformities, muscle tone disturbances, hernias, capillary hemangioma, and external genital anomalies.⁴

The simultaneous occurrence of multiple dental anomalies has been reported previously, particularly in cases of chromosomal abnormalities with multisystem involvement.^{5, 6} Additionally, multiple dental anomalies have been reported in individuals and within families, without evidence of other systemic manifestations.⁷⁻¹⁰ Thus, the study of chromosomal deletion syndromes can lead us to attempt to locate the affected chromosomal locus and to understand its molecular control. The purpose of this report is to outline the

numerous dental anomalies observed in an individual with a highly defined chromosomal abnormality.

Case report

Past medical history

The patient originally presented to the dental clinic at age 5. He was delivered prematurely by cesarean section with a birth weight of 3 lbs, 4 oz. Multiple dysmorphic features were noted at birth including microcephaly, short neck, symbrachydactyly, diastasis recti abdominis, hypospadias, and widely spaced nipples. Chromosome studies revealed a partial deletion of the long arm of chromosome 7 at q32.¹¹ The postnatal period was complicated by growth retardation and delayed motor and cognitive milestones.

Now aged 15, the patient is nonambulatory, nonverbal, and has profound hearing loss and profound mental retardation. He has been institutionalized since age 2 years. At that time, clinical examination revealed a small-statured male with microcephaly, prominent forehead, upward slanting palpebral fissures, hypertelorism, bulbous nose tip, wide philtrum, large mouth (Fig 1), symbrachydactyly, widely spaced nipples, diastasis recti abdominis, and hypospadias.

Past dental history

Most recently, the patient presented at age 15 years with congenital absence of two maxillary and two mandibular permanent incisors. The remaining maxillary incisors were microdontic. Also, the maxillary incisors and canines had invaginations on the lingual surfaces. All other dental hard tissues demonstrated normal morphology. There were no carious lesions. Although the soft tissue examination was otherwise within normal limits, there was a generalized marginal gingivitis.

The patient's initial dental examination at age 5 years revealed an early transitional dentition, with eruption of dysmorphic and divergent permanent maxillary incisors in the area normally occupied by the permanent

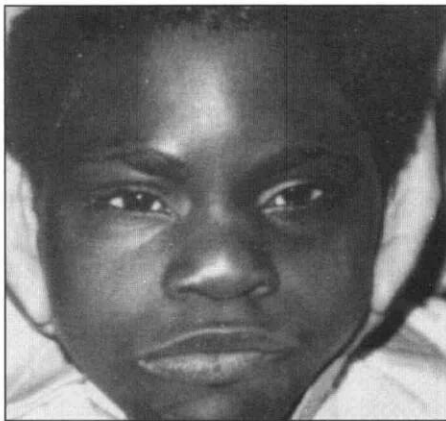


Fig 1. Facial features at age 10 years: microcephaly, upward slanting palpebral fissures, hypertelorism, and wide philtrum.

lateral incisors. The maxillary primary central incisors still were present. The mandibular arch permanent central incisors were erupting into the areas normally occupied by the permanent lateral incisors. Here too, the primary central incisors were present. There were no carious lesions. At age 8, the maxillary primary central incisors had exfoliated, as had the mandibular primary central incisors. At age 9, dental examination revealed a late transitional dentition with bilateral absence of maxillary and mandibular permanent incisors and rotation and convergence of the remaining maxillary incisors. The occlusal anatomy of the maxillary and mandibular first permanent molars was unremarkable. At age 10, clinical examination revealed an age-appropriate permanent dentition exhibiting bilateral congenitally missing maxillary and mandibular incisors.

Radiographic examination

At age 5 years, in the early transitional dentition, initial radiographic evaluation revealed resorption of the primary central incisors by anomalous teeth with crowns erupting divergently into the position of the lateral incisors (Fig 2). No primary lateral incisors or their successors were seen. At age 9 years, periapical radiographs revealed the presence of first and second premolars and first permanent molars. The mandibular left second primary molar had three roots (Fig 3).

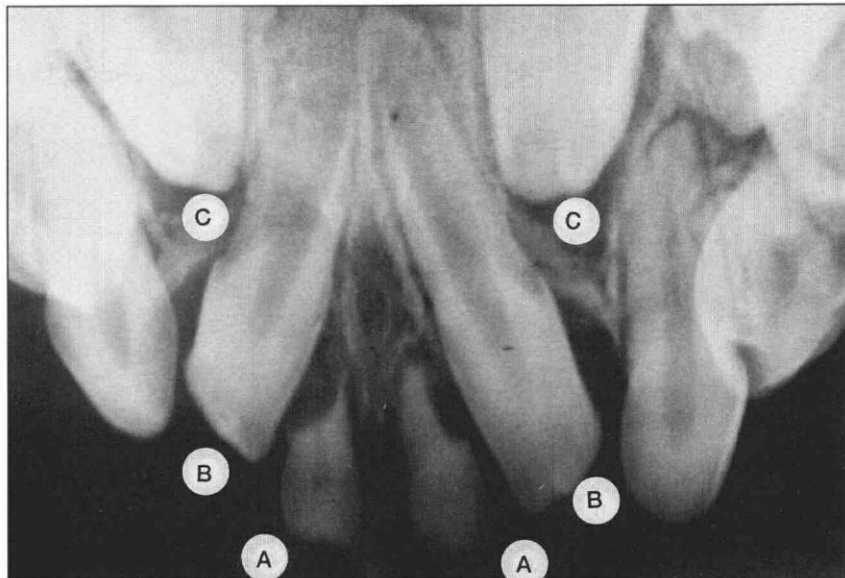


Fig 2. Maxillary occlusal radiograph at age 5 years: A. Primary maxillary incisors; B. Divergent eruption of dysmorphic permanent incisors; C. Permanent maxillary canines with suggestion of dens invaginatus.

The enamel appeared thin on all primary teeth, but exhibited a normal radiodensity gradient with dentin. Abnormal pulpal morphology was suggested on the radiographs including prominent pulp horns in the mandibular second primary molar and a suggestion of taurodontism of the maxillary first permanent molars (Fig 3). Additionally, radiographic evaluation revealed anomalous teeth with divergent roots and convergent crowns in the maxillary anterior region with a sugges-

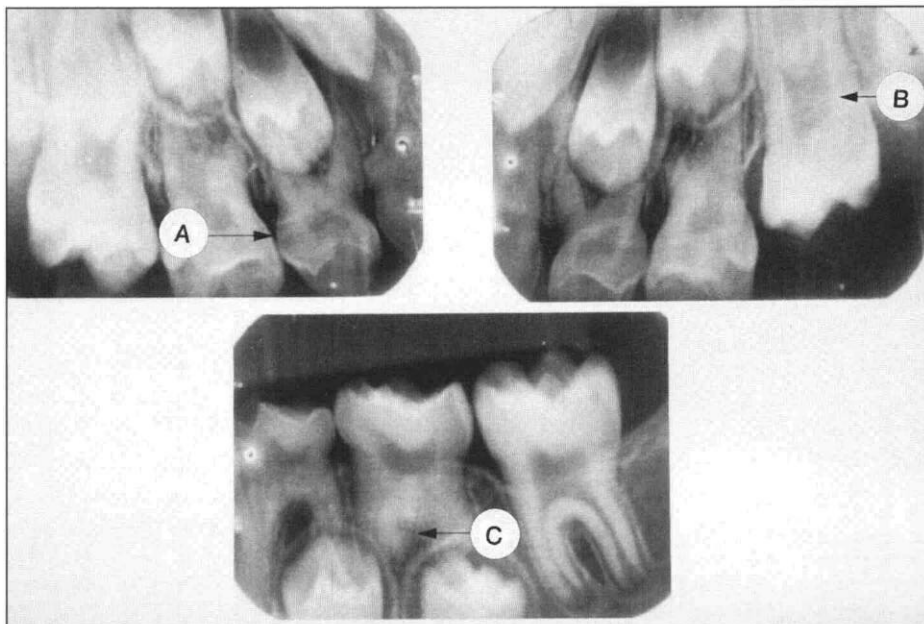


Fig 3. Periapical radiographs at age 9 years: A. Hypoplastic enamel on primary molars; B. Suggestion of taurodontism of maxillary first permanent molar; C. 3-rooted mandibular second primary molar with prominent pulpal horn.

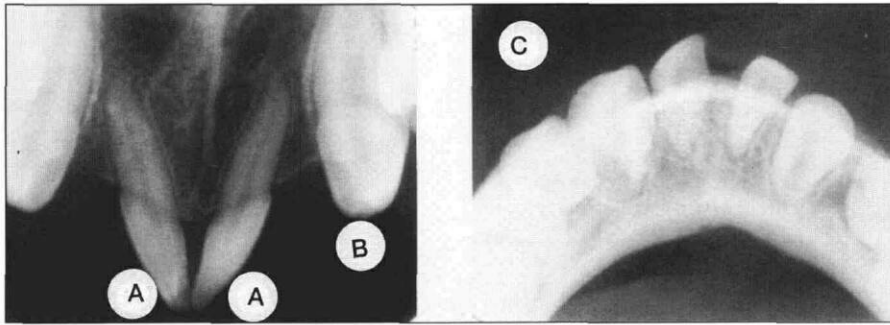


Fig 4. Maxillary and mandibular occlusal radiographs at age 10 years: A. Congenital absence of maxillary permanent incisors with remaining convergent anomalous maxillary incisors and invaginations; B. Dens invaginatus of permanent canine; C. Congenital absence of mandibular permanent incisors.

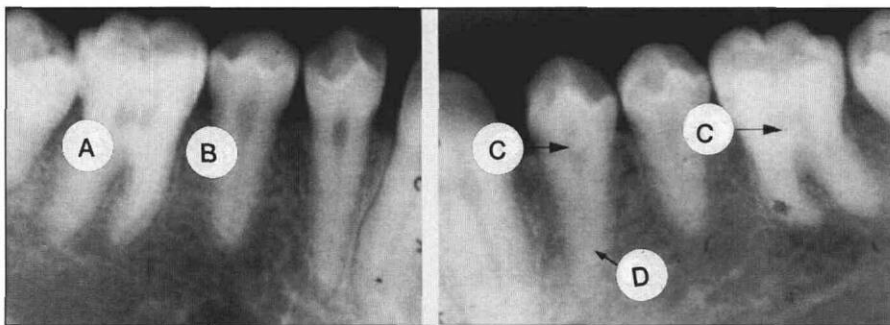


Fig 5. Mandibular periapical radiographs at age 15 years: A. Bow tie-shaped pulp outline; B. Shortened root with thistling of pulp chamber; C. Pulpal stones; D. Calcification of radicular canals.

tion of dens invaginatus on the incisors and canines. Congenital absence of two mandibular and two maxillary incisors was confirmed (Fig 4).

At age 15, the patient exhibited thistle-shaped pulp chambers of the mandibular premolars and bow-tie outlines of the mandibular permanent molar pulp chambers as well as calcification of the pulpal canals. The roots of the mandibular second premolars appeared shortened. These findings were not noted in the maxilla. The presence of pulpal stones was noted in both maxillary and mandibular teeth, as was decreased enamel thickness (Fig 5).

Discussion

Only one previous description of dental findings in patients with chromosome 7 deletion has been reported.¹² The dental findings in two unrelated patients with a terminal deletion were limited to absence of a single permanent maxillary central incisor. The dental findings in this case are associated with deletion of a highly defined locus, rather than a terminal segment deletion. The dental findings include hypodontia, thin

enamel, root dysplasia, pulpal stones, abnormal pulpal morphology, and abnormal coronal morphology. The findings associated with 7q32 deletion may indicate that the genetic material controlling these dental features is located on this locus. To what extent other factors such as systemic disturbances, gene expressivity, and penetrance influence clinical findings is unclear.¹³ Cytogenetic studies revealed that there was no translocation of the deleted locus, thus there is no insertion of genetic material at an inappropriate site to alter the expression of another locus by positional effect.

The dental anomalies observed in this patient each may be encountered as isolated findings with varying incidences. Agenesis of maxillary lateral incisors is not an uncommon finding. The frequency of absent and small lateral incisors among individuals of African descent is 2%.¹⁴ Agenesis of mandibular incisors is more rare. Hypodontia has been reported to occur in many syndromes, such as ectodermal dysplasia and Rieger's syndrome, but also may occur as an isolated event.¹⁴

The abnormal coronal morphology and divergent erupting pattern of the maxillary central incisors can be seen radiographically (Fig 2). In the maxillary occlusal radiograph at 9 years, the maxillary incisors are fully erupted and now are convergent (Fig 4). It is difficult to explain the final position of these teeth or to know which factors may have influenced them. The abnormal coronal morphology and convergent position of the maxillary central incisors suggest that these teeth may be supernumeraries with agenesis of the maxillary central incisors, but agenesis of maxillary central incisors is very rare, ranging from 0.66–3.6%.^{15, 16} Although the maxillary incisors may represent a microdontic form of permanent central incisors, congenital absence of maxillary central incisors has been reported in patients with 7q terminal deletions.

The invaginations observed clinically and radiographically on the maxillary incisors and canines were consistent with dens invaginatus, although there is no widely accepted definition.¹⁷ As is frequently reported, the patient exhibited bilateral occurrence.

The pulpal morphology of the permanent dentition suggests taurodontic maxillary first permanent mo-

lars. Although taurodontism may occur as an isolated finding, it has been reported to occur in Klinefelter's syndrome¹⁸ and as part of trichodontoosseous syndrome (TDO).¹⁹ Additionally, the enamel observed in the primary dentition is thin, but with a normal-appearing radiodensity. Thin enamel, high pulp horns, and taurodontism are part of the constellation of findings in TDO. The diagnosis of taurodontism, however, may be tenuous, affected by such factors as radiographic technique and angulation.

The abnormal pulpal morphology and pulp stones seen in the permanent dentition are consistent with dentinal dysplasia type II. However, the primary dentition exhibited neither the characteristically associated clinical nor radiographic findings. In fact, the prominence of the coronal pulp sharply contrasts with the pulpal obliteration commonly encountered.¹³ Although the shortened roots of the mandibular second premolars may be reminiscent of dentinal dysplasia type I, there are no other corroborating findings to support this diagnosis.

Although the incidence of three-rooted first permanent molars has been reported,²⁰ the occurrence of three-rooted primary mandibular molars has been reported only rarely.^{21,22} The occurrence of three-rooted primary mandibular molars has been reported in a variant of the Ekman-Westborg-Julin syndrome,⁸ as well as in a recently reported case of multiple dental anomalies in a patient with an otherwise noncontributory medical history.¹⁰

The individual dental anomalies observed in this patient each would be reasonably expected to occur in the absence of familial history or known chromosomal alteration. Each may be encountered as an isolated finding. However, the constellation of dental findings seen in this patient diagnosed with chromosome 7q32 deletion syndrome is certainly rare.

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