

The Nance-Horan syndrome of dental anomalies, congenital cataracts, microphthalmia, and anteverted pinna: case report

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Abstract

A case of a male patient presenting with unusual dental morphology, anterior supernumerary teeth, and agenesis of premolars associated with congenital cataracts, microphthalmia and anteverted pinna is described. Although 3 similar cases are in the medical literature, this is the first case in which the dental features are described in detail.

Dental anomalies associated with many congenital and hereditary syndromes have been well documented. Hypodontia, the agenesis of one or more teeth, is observed in various ectodermal dysplasias¹ while hyperdontia or the presence of supernumerary teeth is a well-described feature of cleidocranial dysostosis² and Gardner's syndrome.³ Disorders of tooth form also are associated with many genetic diseases. Taurodontism, an anomaly in which the furcation of molars is displaced apically, resulting in elongation of the body and pulp chamber and in shortening of the roots, has been described in the trichonychondental (TOD) syndrome, the trichodento-osseous syndrome, the Mohr syndrome (oral-facial-digital syndrome II) as well as the Klinefelter's syndrome.⁴ Alteration in crown morphology also is seen in Ellis-van Creveld syndrome⁵ where conically shaped lateral incisors and canines and barrel-shaped central incisors are present.

In many of these inherited conditions, the dental findings are so consistent as to constitute important features of the syndrome. Thus, recognition of dental anomalies in patients with congenital medical conditions may play an important role in the diagnosis of the disease, especially in cases where dysmor-

phological features and metabolic changes are not well defined.

This report describes a patient who presented with peg-shaped central incisors, anterior supernumerary teeth, agenesis of premolars, and taurodont molars with unusual rhomboidal crown morphology and some prominent pulp horns. These multiple dental anomalies were associated with congenital cataracts, microphthalmia, and anteverted pinna.

Clinical Report

The patient, a Caucasian male was 8 years of age when first referred to the University Dental School by the School Dental Service for assessment and treatment of supernumerary teeth.

He was of normal intelligence, with height 132.4 cm (90th percentile) and weight (22.6 kg (25th percentile). He had almost total loss of vision which resulted from bilateral cataracts and attended a school for the blind. In addition, he also showed microphthalmia, convergent strabismus of the left eye, and nystagmus. The ears appeared large and anteverted, but no obvious abnormality of pinna morphology was noted (Fig 1). No other general dysmorphic features were found.

Dental Findings

At the time of the first dental examination, all primary teeth were present except for the mandibular incisors, the left maxillary second molar, and the maxillary right central incisor. The maxillary permanent first molars and mandibular incisors were fully erupted. The maxillary primary left central incisor was carious, nonvital and abscessed, and the root frag-

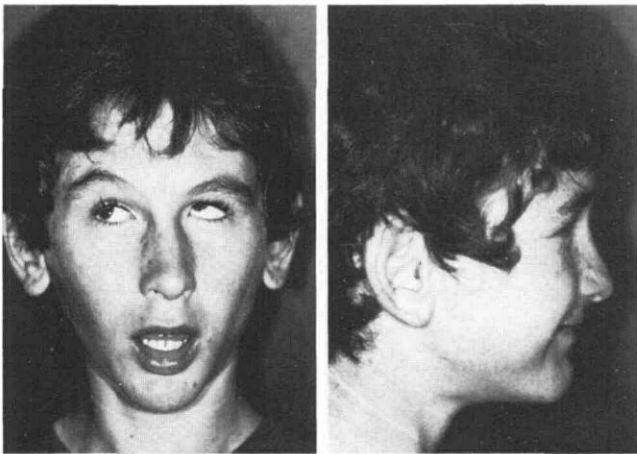


FIG 1. Photograph of the patient's face showing front and side views.

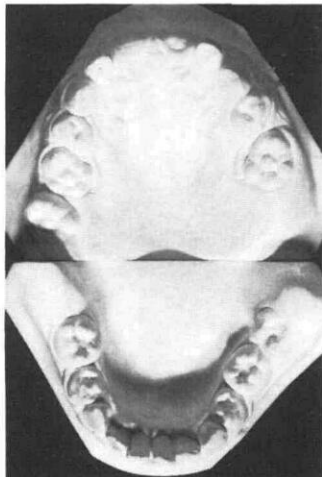


FIG 2. Models of the patient's dentition at age 8 years showing the abnormal morphology of the teeth.

ment of the maxillary left primary second molar present. The primary second molars appeared ankylosed.

The crowns of the primary molars were abnormal, with buccal, lingual, mesial, and distal surfaces converging toward the occlusal. The occlusal table thus appeared narrow. In addition, an extra cusp was present in the center of the occlusal surface of the maxillary right deciduous molar (Fig 2).

Examination of the occlusion revealed a deep overbite with the lower incisors contacting the upper palatal gingiva and an overjet of 3 mm. There was a bilateral posterior open bite of about 3 mm.

An orthopantomograph taken at age 7 years, 10 months revealed agenesis of both maxillary and left mandibular second premolars (Fig 3). In addition, 2 supernumerary teeth were present in the area of the maxillary central incisors. Further interesting findings included the taurodont first permanent molars with their large pulp chambers. Also, the mesial pulp

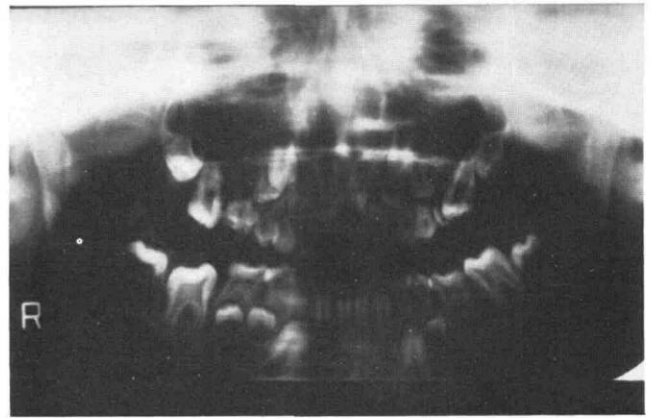


FIG 3. Orthopantomograph of the patient taken at age 7 years, 10 months, revealing agenesis of both maxillary and left mandibular second premolars, anterior supernumerary teeth, and taurodont first permanent molars.

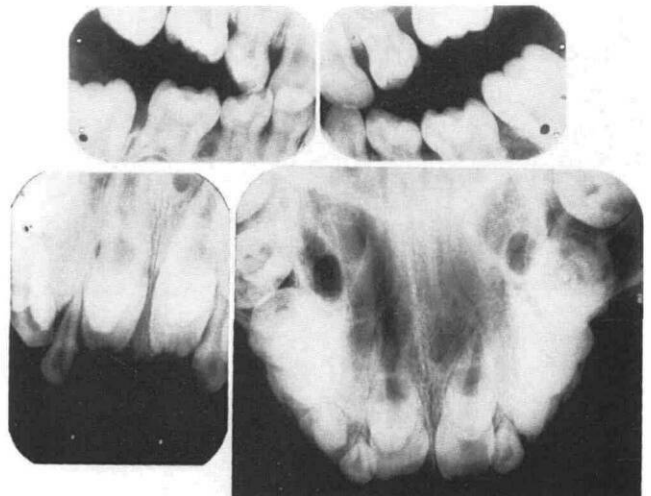


FIG 4. Intraoral radiographs of the patient.

horns of lower second primary molars appeared to extend very close to the occlusal surfaces. Intraoral radiographs confirmed the findings in the orthopantomograph (Fig 4). No proximal caries was noted from bite-wing radiographs.

The abscessed primary incisor, the root fragment of the maxillary second deciduous molar, and the supernumeraries were removed with the aid of local anesthesia. Healing was uneventful. A histological examination of the supernumeraries revealed no significant findings. The patient was followed up regularly for preventive care.

When the permanent teeth erupted, they were noted to be distinctly abnormal in crown morphology. The permanent incisors were peg-shaped with marked convergence of the proximal surfaces toward the incisal edges. This was more marked in the maxillary

incisors compared to the mandibular incisors. In addition, the maxillary central incisors were rotated and a large diastema was present between the teeth. The first permanent molars also had occlusally converging sides so that the occlusal surfaces were reduced. The cusps of these teeth were rounded and reduced in height. In addition, an extra cusp was present in the center of the occlusal surface of each of the maxillary first permanent molars. The dentition at age 12 years is shown in Figures 5, 6, and 7.

At age 12 years, another orthopantomograph revealed another supernumerary tooth developing in the area of the root of the right maxillary canine. In addition, the roots of the maxillary central incisors appeared dilacerated. Although the second permanent molars still were unerupted, it was evident from this radiograph that their crown morphology was also abnormal, with tapered proximal surfaces. These teeth were also taurodont, similar to the first permanent molars (Fig 8).

A hand-wrist radiograph revealed that the patient was about 6 months retarded in skeletal age compared to the reference standards of Greulich and Pyle.⁶ No morphological bone defects were detected. A mild skeletal II base was observed from a lateral skull radiograph.

The mandibular second primary molars were extracted to allow optimal eruption of the premolars. The dentition of the patient's mother and brother were examined clinically and radiographically but no significant features were noted.

Histological Findings

Microscopically, sections of the primary molar tooth showed a relatively thin layer of enamel with several sites of developmental disturbances characterized by disk-shaped depressions of the surface and accentuated within the body of enamel beneath these de-

fects. Highly irregular dentine with cellular cementum (or bone) was present in abundance on the floor of the pulp chamber, reducing its size considerably (Fig 9). This tooth also exhibited bony ankylosis.

Medical History

The patient was the older of 2 boys born of a non-consanguineous marriage. He was the product of a normal, full-term, uneventful pregnancy with birth weight of 2.78 kg. Two days after birth he developed hepatosplenomegaly and jaundice with bilirubin levels of 6.3 mg/dl. The jaundice persisted for several weeks and extensive investigations revealed no anatomical abnormalities of the biliary system. However, hepatosplenomegaly persisted and liver function tests remained abnormal, with elevated SGOT, LDH, and serum alkaline phosphatase levels. Because he was otherwise well, no further investigations were undertaken.

At 6 months of age it was discovered that the child had bilateral congenital cataracts with severe loss of vision, microphthalmia, and convergent strabismus of the left eye. These findings subsequently were confirmed by two ophthalmologists. Because of the presence of congenital cataracts and neonatal jaundice, the child was investigated for the possibility of congenital infection with TORCH (toxoplasmosis, other organisms including syphilis, rubella, cytomegalovirus, and herpes) organisms but laboratory tests were insignificant.

The child also had a history of asthma which started at a few months of age and was controlled by salbutamol aminophylline, and corticosteroid.

At 9 years of age, alpha₁-antitrypsin deficiency was diagnosed by measurements of the serum levels of this enzyme. Alpha₁-antitrypsin is an inhibitor of trypsin and other protease enzymes in serum. The normal phenotype of the Pi (Protease inhibitor) sys-

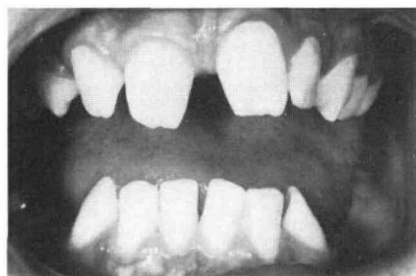


FIG 5. (left) Photograph showing abnormal permanent anterior teeth of the patient.

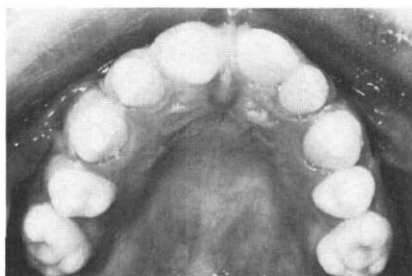


FIG 6. (center) Photograph of the maxillary teeth, depicting the unusual morphology of the maxillary permanent first molars.

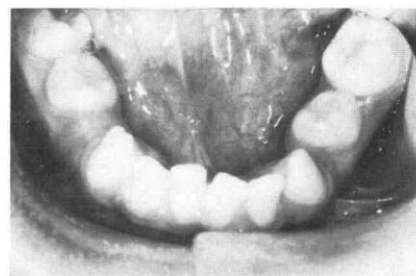


FIG 7. (right) Photograph of the mandibular permanent teeth showing their abnormal morphology.

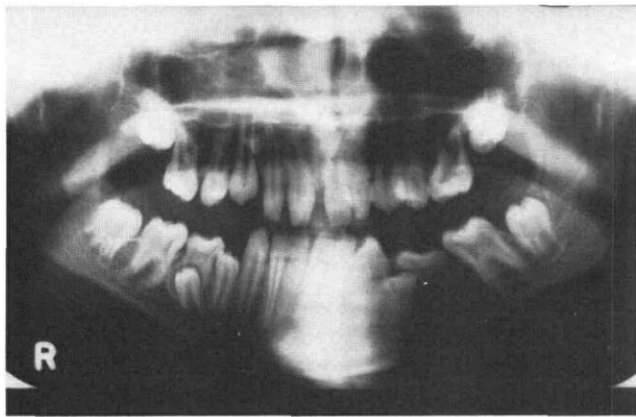


FIG 8. Orthopantomograph of the patient taken at 12 years of age. Note the dilacerated roots of the maxillary central incisors, the taurodont molars, and the supernumerary tooth present in the area of the root of the maxillary right canine.

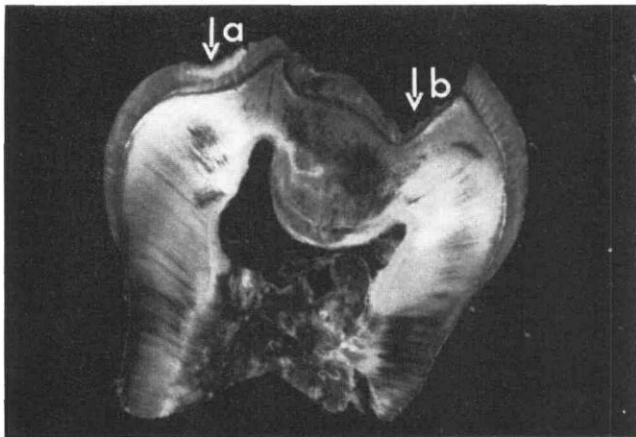


FIG 9. Photomicrograph of undecalcified section of the ankylosed second primary molar. Note highly irregular dentine with cellular cementum (or bone) at the floor of the pulp chamber. The arrows depict (a) zone of hypomineralization associated with enhanced striae of Retzius, (b) fractures of enamel during preparation of section.

tem is M; the type associated with liver disease is Z⁷. Phenotype analysis for the protease inhibitor was performed on the patient, his parents, and his younger brother. Both the patient and his brother were typed as homozygous (phenotype PiZ) for the codominant gene and both parents were heterozygous (phenotype PiMZ).

Other metabolic disorders such as congenital galactokinase deficiency were excluded by appropriate tests and chromosomal abnormalities were excluded by karyotyping.

Discussion

In this patient, it is most likely that the dental anomalies were associated with congenital cataracts,

microphthalmia, and the large anteverted pinna. A literature search revealed reports of a few patients with similar dental anomalies and ophthalmic features. Nance et al. first described a family with congenital X-linked cataracts, anteverted pinna, short metacarpals, and dental anomalies.⁸ These patients included anterior supernumerary teeth and abnormal dental morphology nearly identical to that seen in this patient. However, dental radiograph findings were not reported.

Van Dorp and Delleman⁹ also reported a family with X-linked congenital cataract, microphthalmia, and a peculiar form of the ear and dental anomalies similar to those seen in the present patient.

X-chromosomal cataract with microphthalmia, somatic anomalies, and mental retardation also have been described by Hoefnagel et al.¹⁰ and Goldberg and McKusick.¹¹ These authors reported that affected males also show a peculiar form of the ear, and the latter authors mentioned dental anomalies including a diastema between maxillary central incisors.

The patient described in this report appeared to be the first member of his family to show these clinical features. Apart from his fathers being color blind, no other family members had any ophthalmic conditions or unusual dental features. In particular, apart from myopia, no ophthalmic abnormalities were detected in the mother. The possible genetic basis of congenital cataracts in this case is thus unknown. It may be X-linked like the other reported cases; however, this is not definite because of the negative family history.

On the other hand, the etiology of the condition in this child may not have been a hereditary condition but one caused by an environmental agent. Many infections such as congenital syphilis and rubella can cause congenital cataracts, neonatal jaundice, and dental anomalies. However, in this patient, such congenital infections had been excluded by appropriate laboratory tests in the neonatal period.

Alpha₁-antitrypsin, a glycoprotein found in the serum in 24 phenotypic forms, is an inhibitor of trypsin and other proteolytic activities. Deficiency of this enzyme predisposes to chronic liver and pulmonary disease. The incidence of the Z phenotype which is associated with liver disease, is estimated at 1:2000–1:4000 as an autosomal codominant gene.⁷

Dental abnormalities have not been reported in alpha₁-antitrypsin deficiency before. It is unlikely that there is any relationship between this enzyme deficiency and the dental anomalies in this report. The patient's brother, a year younger in age, was identical in phenotype (PiZ) but had no dental anomalies. It is also of interest to note that the brother had little evidence of liver disease.

In conclusion, the syndrome of dental anomalies, congenital cataracts, microphthalmia, and anteverted

pinna appear to be a distinct clinical entity. To date only 3 similar reports are in the literature; in these reports, the dental features were described only briefly.

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Quotable Quote: man's best friend

If a dog is man's best friend, why is it there are an estimated 44,000 facial injuries yearly from dog bites? Children under age 10 seem to get the worst of it, accounting for nearly all of the 16,000 such injuries that are characterized as severe.

How can you protect your child from attack? The answer to the question is not an easy one. In most cases, the bite is not brought on by teasing or abuse; nor is the attacking dog a stranger to the child usually. The problem appears that biting around the head and mouth is a normal part of a dogs' aggressive play. In all likelihood, the dog is approaching the child in a playful spirit, without any intention of harming him.

The best solution probably would be not to keep large, aggressive dogs as pets, especially if you have young children. German shepherds, malamutes, and huskies seem to be the breeds most inclined to bite; hounds, the least. Young dogs pose a greater danger than older ones; males more than females. Whatever its age, breed, or sex, a dog should never be left alone with a child, even if the child is in a crib or playpen.

Dog. Parent's Magazine. August, 1985 p 18.