

Pseudohypoparathyroidism: case report

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Abstract

A patient with pseudohypoparathyroidism is presented. The phenotypic appearance, known as Albright heredity osteodystrophy includes: short stature, round face, brachydactylia, and ectopic calcifications in the soft tissues. Dental manifestations reported in the literature are enamel hypoplasia, hypodontia, malformed roots, enlarged pulp chambers, microdontia, and pulp calcifications. Additional findings in this case are ankylosis and an enlarged frontal sinus. The delayed diagnosis of pseudohypoparathyroidism with the early presentation of multiple dental anomalies is discussed.

Introduction

Pseudohypoparathyroidism (PHP) was first described by Albright et al. (1942). The typical phenotype, known as Albright hereditary osteodystrophy, includes: short stature, round face, brachydactylia, and ectopic calcifications in the soft tissues (Nyhan and Sakati 1987). Additional characteristics include obesity, thickening of the calvarium, mental retardation, cataracts, and dental irregularities (Gorlin et al. 1976). This clinical picture is the first described example of a disease resulting from an end-organ resistance to a hormone, rather than a failure to produce or release that hormone.

In PHP the parathyroid glands are present and functional, whereas in primary hypoparathyroidism the glands either are not present or function abnormally. Although adequate levels of parathyroid hormone (PTH) are produced, in PHP the kidneys fail to respond to the PTH, resulting in hypocalcemia and hyperphosphatemia. Recent studies indicate a deficient receptor cyclase coupling (N) protein as the etiologic factor. The defective N protein adversely affects the binding of PTH

to its receptor site in the renal tubule cell membrane, resulting in a lack of conversion of ATP to cyclic adenylic acid (cAMP). The failure of a patient to respond to exogenous parathyroid extract with an increase in urinary excretion of cAMP, as occurs in normal individuals or those with primary hypoparathyroidism, leads to a diagnosis of PHP (Nelson 1979; Nyhan and Sakati 1987).

PHP is rare; however, it is of concern to the dental professional. Despite differing etiologies, the clinical pictures of PHP and hypoparathyroidism are similar. Hypocalcemia often presents initially as tetany or seizures leading to the misdiagnosis of epilepsy. The striking dental anomalies associated with this condition provide an opportunity to aid in early diagnosis.

The purpose of this paper is to review the dental manifestations of PHP and present a case report.

Literature Review

The dental manifestations of PHP are rarely mentioned in the medical literature. Delayed eruption and enamel hypoplasia are most commonly noted (Gorlin et al. 1976; Devogelaer et al. 1984; Nyhan and Sakati 1987; Jones 1988). Dental aplasia is cited as a frequent occurrence (Nyhan and Sakati 1987; Jones 1988). Other incidentally presented findings include dental malocclusion and a high arched palate (Nyhan and Sakati 1987), widened root canals (Gorlin et al. 1976; Assif 1977), and melanodontia (Devogelaer et al. 1984).

A paucity of information concerning PHP exists in the recent dental literature. Ritchie (1965) reviewed the incidental dental manifestations in the literature and presented his findings in four cases. He summarized the dental findings as small crowns, thin hypoplastic enamel, short roots with blunt apices, and large pulp chambers with calcified deposits. He also noted dental aplasia and delayed eruption as common occurrences.

Croft et al. (1965) reviewed the literature and presented a case report. Their findings were similar to those of Ritchie. They reported delayed eruption, enamel hyp-

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oplasia, large pulp chambers and "dagger-shaped" pulp stones in the incisal areas. Additionally, detailed histologic and microradiographic examination revealed normal mineralization of the enamel. The lines of Retzius remained tangential to the enamel "pitting," indicating that the defects represented true hypoplasia of the enamel, rather than resorption of a fully formed crown.

Jensen et al. (1981) reviewed the dental manifestations in idiopathic hypoparathyroidism and PHP in the Scandinavian literature and reported on nine cases of PHP. Their findings were delayed eruption (100%), enamel hypoplasia (67%), apical blunting (67%), hypodontia (67%), pulp calcifications (17%), thickened lamina dura (17%), and excessive caries (17%). Dental disturbances were reported in all cases of PHP, in contrast to earlier reports of dental anomalies in less than half of the PHP patients (Assif 1977).

Case Report

A 14-year, 7-month-old white male with PHP was referred to the Pediatric Dental Residency Program, Ft. Lewis, WA, for a dental evaluation. He presented with the typical Albright hereditary osteodystrophy phenotype of short stature (5th percentile), round face, and brachydactyly (Fig 1), consistent with PHP (Rowe 1987). A comprehensive dental examination was performed.

The patient's weight was at the 75th percentile, his head circumference was greater than the 98th percentile (Rowe 1987), and he had a positive Albright's sign (Fig 2, failure to form epiphyseal protuberances on making a fist, Nakano et al. 1987). His medical history revealed mild mental retardation, basal ganglia calcifications, and "seizures" at age 4. The diagnosis was made at age 10. The hypocalcemia was treated with vitamin D and calcium supplements. Additionally, his medications included levothyroxine sodium USP to treat hypothyroidism, often associated with this condition (Nyhan and Sakati 1987). He had attended special edu-



Fig 1. Brachydactyly is a characteristic of the phenotype.

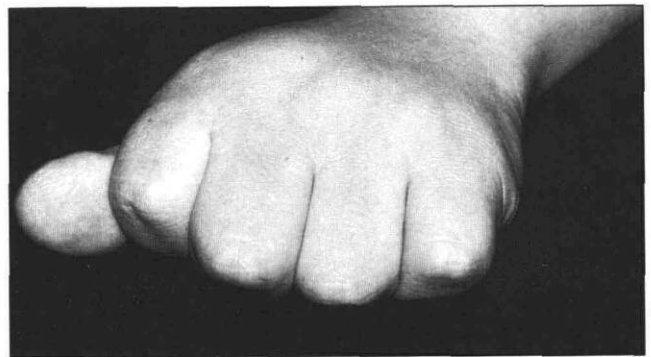


Fig 2. Albright's sign—failure to form epiphyseal protuberances on making a fist.



Fig 3. Generalized enamel hypoplasia is characteristic of PHP.

cation classes since kindergarten.

Intraoral examination revealed generalized enamel hypoplasia of all permanent teeth (Fig 3), with a pronounced hypoplastic band at the middle third of the incisors and the occlusal third of the first molars. Additional findings included microdontia, particularly of the mandibular incisors (below the 10th percentile); ankylosed primary right mandibular second molar; and a Class I malocclusion with a deep bite.

Radiographic examination revealed several dental anomalies. Enlarged pulp chambers were present in



Fig 4. Enlarged pulp chambers and "dagger-shaped" pulp calcifications.

multiple teeth and pulp calcifications, often "dagger-shaped" (Fig 4, previous page), were found in 26 of 27 teeth. The patient exhibited hypodontia with absence of the maxillary and mandibular right second bicuspid and all third molars (Fig 5). Delayed eruption of the mandibular left second bicuspid at age 14 was determined from serial panoramic radiographs. The cephalometric radiograph revealed an exceptionally large frontal sinus (Fig 6).

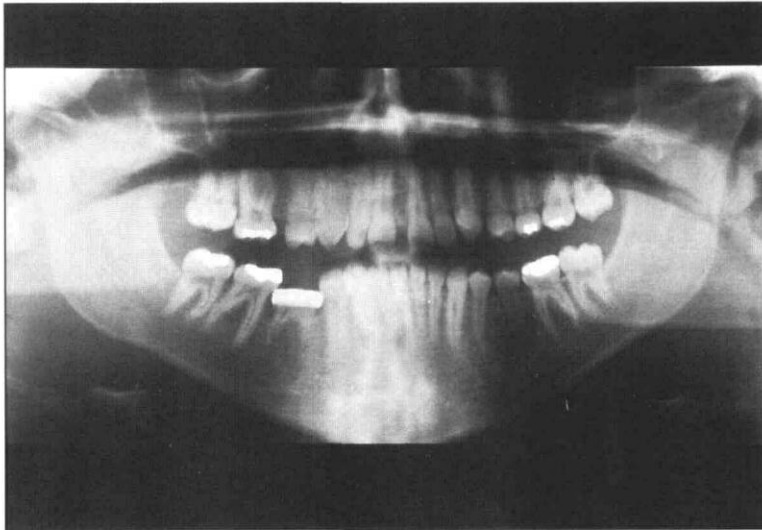


Fig 5. Ankylosis and hypodontia.

Discussion

The literature was reviewed for the dental manifestations of PHP and a case presented. The dental anomalies in this case were consistent with those reported earlier. Additional findings in this case included ankylosis and a large frontal sinus. The latter finding is interesting in that frontal bossing (Ritchie 1965), thick calvaria (Gorlin et al. 1976; Tsang et al. 1984; Jones 1988), and "hyperostosis frontalis interna" (Nyhan and Sakati 1987) have been reported as incidental occurrences associated with PHP. In this case, the clinical presentation appears due to an abnormally large sinus.

Enamel hypoplasia is associated with multiple pediatric disorders. Studies of children with chronic disorders of calcium and phosphate homeostasis indicate that the metabolic determinant of generalized enamel hypoplasia is hypocalcemia. The location of enamel lesions on specific areas of the dentition, coupled with a knowledge of the chronology of tooth development, allow for estimation of disease onset (Nikiforuk and Fraser 1981). The appearance in this patient of hypoplastic enamel in the incisal one third of the permanent first molars and in the middle one third of the incisors indicates hypocalcemia was present from birth through two years.

Unfortunately, this case parallels previously reported cases in which the correct diagnosis was not made until several years after the occurrence of initial manifestations. The presenting symptom is often tetany or seizures (Gorlin et al. 1976; Siejka et al. 1988) resulting in numerous reports of PHP misdiagnosed as epilepsy (Croft et al. 1965; Jensen et al. 1981) or other neuropsychiatric disorders (Nyhan and Sakati 1987). Though the dental manifestations of PHP are frequently apparent at younger ages, the diagnosis is often made at approximately 10 years of age (Croft et al. 1965; Ritchie 1965; Jensen et al. 1981; Nakano et al. 1987; Siejka et al. 1988). The dentist must investigate the etiology of enamel hypoplasia, hypodontia, and delayed eruption; particularly in patients with seizure disorders.

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Fig 6. Exceptionally large frontal sinus.

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