Albright’s Hereditary Osteodystrophy: A Constellation of Clinical Features

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ABSTRACT
Pseudohypoparathyroidism (PHP) is an inherited metabolic disorder characterized by end-organ resistance to the action of PTH (Parathyroid Hormone). There are four types of PHPs namely Ia, Ib, Ic and II. PHP Ia is associated with a constellation of clinical features referred to as Albright’s Hereditary Osteodystrophy (AHO). The oral manifestation of AHO found in the literature includes aplasia and or enamel hypoplasia, late tooth eruption, and enlarged radicular channels susceptible to caries. Here, we are reporting a rare case of a 14-year-old girl with Albright’s hereditary osteodystrophy with distinctive oral manifestations.

Keywords: Albright’s hereditary osteodystrophy, Pseudohypoparathyroidism, Oral manifestation.

INTRODUCTION
Albright’s hereditary osteodystrophy (AHO) was first described by Fuller Albright in 1942. It is a rare inherited metabolic disorder characterized by a typical phenotype. It may be associated with or without resistance to parathyroid hormone. It occurs as a consequence of reduced erythrocyte membrane coupled with Gs alpha activity. The clinical features of these patients include stocky built, short neck, round face, brachydactyly, short metacarpals, short metatarsals, low nasal bridge, osteoporosis and mild mental retardation. Various clinicians have reported the oral manifestations of AHO, which could be imperative in diagnosing the case. Here, we present a case of AHO with additional oral findings.

CASE REPORT
A 14-year-old girl was referred to the Department of Oral Medicine and Radiology by a team of neurophysician, pediatrician and psychiatrist for oral examination. Patient’s complaints were halitosis and gum bleeding. Patient was a known epileptic since 3 years for which she was under regular medication. There was no significant family history, except her mother had hypertension during her first trimester. There was no history of consanguineous marriage of her parents. She was the only child with normal natal and perinatal histories. She had normal developmental milestones. General physical examination revealed her normal stature and weight. There were no abnormalities on systemic examination. Extraoral examination revealed short neck, round face, hypertelorism, low-set ears, low hair-lines, depressed nasal bridge, synophrys (Fig. 1), short ring finger (Fig. 2) and short fourth toe. Her intraoral examination illustrated crowded teeth, anterior deep bite, malposed maxillary left canine, missing mandibular second premolars, deep caries in the mandibular molars, coated and fissured tongue and generalized gingival inflammation. Then patient was subjected for laboratory investigations. Patient’s hematological investigations were within normal limits and biochemical investigations revealed low serum calcium–5.2 mg/dl (7-12 mg/dl) and alkaline phosphatase–277 U/L (50-440 U/L) levels and raised inorganic phosphorus–9.1 mg/dl (2.5-4.8 mg/dl), parathyroid hormone–236.2 pg/ml (10-55 pg/ml) and thyroid stimulating hormone–10.1 (0.5 to 5.0) level. Patient’s orthopantomogram revealed impacted mandibular premolars and enlarged pulp chambers in all the teeth (Fig. 3). Hand wrist radiograph and skull radiograph showed short metacarpals (Fig. 4) and thickened calveria in parieto-occipital region, respectively. Computed Tomogram suggested calcification in bilateral basal ganglia and frontal lobe (Fig. 5). Based on history, patient was subjected for laboratory investigations. Patient’s hematological investigations were within normal limits and biochemical investigations revealed low serum calcium–5.2 mg/dl (7-12 mg/dl) and alkaline phosphatase–277 U/L (50-440 U/L) levels and raised inorganic phosphorus–9.1 mg/dl (2.5-4.8 mg/dl), parathyroid hormone–236.2 pg/ml (10-55 pg/ml) and thyroid stimulating hormone–10.1 (0.5 to 5.0) level. Patient’s orthopantomogram revealed impacted mandibular premolars and enlarged pulp chambers in all the teeth (Fig. 3). Hand wrist radiograph and skull radiograph showed short metacarpals (Fig. 4) and thickened calveria in parieto-occipital region, respectively. Computed Tomogram suggested calcification in bilateral basal ganglia and frontal lobe (Fig. 5). Based on history,

Fig. 1: Showing dysmorphic facial features
Fig. 2: Shortened fourth finger

Fig. 3: Illustrating impacted mandibular premolars and enlarged pulp chambers in all the teeth

Fig. 4: Radiograph showing shortened 4th metacarpal

Fig. 5: CT scan showing calcification in the basal ganglia

DISCUSSION

Albright’s hereditary osteodystrophy (AHO) is also known as Acrodysostosis and pseudohypoparathyroidism. Patients with AHO present inactivating mutations in the GNAS1 gene, which codify the subunit $\alpha$ of protein Gs, mapped in chromosome 20q13.2-20q13.3. The feature consists of a rounded face with a short, stocky built. There is brachydactyly with dimpling of the dorsum of hand. Shortening of the digits only rarely involves the second digit (both upper and lower limb), so that the ring finger is shorter than the index finger. The abnormalities of the fourth and fifth metacarpals and metatarsals are characteristic. Thickened calvaria, short and wide phalanges with metastatic calcification, and exostoses may also be present. Mild mental retardation, basal ganglia calcification and lenticular cataracts are found later in life.

The oral findings include dull white and hypoplastic pitted teeth, small crowns and blunt roots, thin enamel and large pulp chambers, unerupted teeth, early loss of teeth, short and wide jaws. Different clinicians have quoted distinctive oral features in the previous literature, such as ankylosis of TMJ, severe periodontal disease with gingivitis, labial hypotonicity, deep hard palate, xerostomia, multiple caries teeth, Angle class II malocclusion, and anterior open bite.

But our patient had additional oral features, such as anterior deep bite, crowded teeth, malposed maxillary canine, gingival recession, carious teeth, mild coating and fissuring of the tongue and Angle class I malocclusion. The consistent oral findings with the previous literatures were short and wide jaw, impacted mandibular second premolars, multiple carious teeth, gingivitis, and enlarged radicular canals in all the teeth.

In the current case, it was found that extraoral features and laboratory findings were consistent enough in diagnosing the case. But in order to get in-depth knowledge of the same, probing intraorally can be of great asset to both oral as well as general physician. So, after all the examination and observation, we can finally conclude that clinical as well as oral features can pave the way for further diagnosis.
REFERENCES