

HEREDITARY HYPOHIDROTIC ECTODERMAL DYSPLASIA WITH ANODONTIA: A CASE REPORT

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إن مرض احتباس العرق الناتج عن سوء تكون الوريقة الخارجية . هو مرض وراثي يتميز بتشوه واحدة أو أكثر في البنية الخارجية، مثل الشعر، الأسنان، الأظافر، والغدد العرقية. ويظهر بوضوح وكثرة عند الذكور أكثر من الإناث. والمظاهر السريرية تكون نقص في الشعر ويكون الشعر أشقر ودقيق مع ملمس غير طبيعي للجمجمة والحواجب وريف العين وجفاف البشرة، وتشوه في الأظافر، بروز في الجبهة، انخفاض في الأنف وتشقق في الشفاه. وإن الغياب الجزئي أو الكامل، للغدد العرقية يمكن أن يسبب أكتريا أو حرقة في الجلد. من النادر حدوث غياب كامل للأسنان المؤقتة أو الدائمة، ولكن غياب جزئي لها كثير الشبوع مع ظهور فراغات بين الأسنان السنية الشكل. ولكن شامو، ويرودي وصفا حالات لغياب كامل للأسنان المؤقتة والدائمة. وفي هذه الحالات يكون هناك نقص في تطور عظم السنخ مما يؤدي إلى نقص في البعد العمودي. الغرض من هذه الدراسة هو تسجيل حالة وراثية لسوء تكون الوريقة الخارجية عند مريض سعودي عمره أربع سنوات.

راجع العيادة الخارجية بجامعة الملك عبدالعزيز لغياب أسنانه. السيرة الطبية أظهرت أن الطفل يشتكي من جفاف في العيون وكان يتحسس من رائحة البطاطا، واللحم وهو يعالج من قبل الطبيب بسبب الربو. وهو يشعر بعطش مستمر مما يجبره على شرب الماء البارد بشكل مستمر. وذكر والده أن الطفل يعاني من صعوبة في التعلق والأكل، ويأكل الأطعمة الطرية فقط.

وبين الفحص السريري غياب في الأسنان المؤقتة العلوية والسفلية وضيق في التوء السنخي، جفاف متوسط في الفم، التهاب الغشاء المخاطي، عدم انسياب في اللعاب. وأظهرت الصورة البانورامية تاجين مخروطيين دون جذور ضمن العظم ويعتقد أنها نائين، ولا يوجد دليل على وجود أسنان أخرى، ولم يكن هناك سيرة سريرية تدل على وجود هذا التشوه لدى الأب والأم أو العائلة.

A four-year-old Saudi boy presented, for the first time, with the characteristic clinical features of hypohidrotic ectodermal dysplasia. Intraoral examination revealed total anodontia of the deciduous teeth. Roentgenographic examination showed four cone-shaped crowns with incomplete roots in bony crypts consistent with permanent canines. No other calcification of the permanent successors was noted. The child was the only member of his family who suffered from hypohidrotic ectodermal dysplasia.

Introduction

Hereditary hypohidrotic ectodermal dysplasia is a hereditary disease characterized by deformity of at least two or more of the ectodermal structures, hair, teeth, nails and sweat glands.^{1,2} It is typically inherited as a cross-linked recessive trait so that the frequency and severity of the condition is more pronounced in males than in females.^{3,6} The

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clinical features include sparse, fine blond hair with abnormal texture of the scalp, eyebrows and eyelashes, dry skin, nail defects, prominent forehead, depressed nasal bridge and protuberant dry and cracked lips.^{2,4,6,7} Beckerman⁸ reported the case of a female patient with anhidrotic ectodermal dysplasia and lacrimal gland anomalies. Complete, as well as partial, absence of sweat glands can cause dry skin and eczema and there may be heat intolerance or hyperthermia in warm condition.^{4,7,9} Xerostomia due to hypoplastic accessory salivary glands may cause drying and cracking of the lips.¹⁰ A complete anodontia of both deciduous and permanent dentition is rare. A partial anodontia is more common with the patient showing a few widely spaced malformed teeth.^{4,5,7,11,12} Shaw⁶, Brodie and Sarnat¹³ described cases of complete anodontia of both deciduous and permanent dentitions. In these reports,^{6,13} the alveolar process failed to develop normally with a reduction in vertical dimension resulting in protuberant lips.^{4,7}

The objective of this study is to report a case of hereditary hypohidrotic ectodermal dysplasia with anodontia in a four-year-old Saudi boy.

Case History

A four-year-old Saudi boy was brought to the Out Patient Dental Clinic at King Abdulaziz University because of the absence of teeth. Medical history revealed that the child complained of dry eyes and was allergic to the smell of potatoes, meat and freshly washed clothes. He had suffered under the care of a physician for an asthmatic condition. He also had repeated episodes of unexplained hyperpyrexia and thirst which necessitated consistent drinking of cold water. His parents stated that he had difficulty in speaking and eating, and has been on soft diet.

The oral examination revealed absence of maxillary and mandibular deciduous teeth and narrow alveolar ridges. Moderate dryness of the mouth, inflamed mucosa and diminished salivary flow were evident (Figs. 1, 2). A panoramic radiograph showed two maxillary and two mandibular cone-shaped crowns (without roots) within bony crypts suggestive of permanent canines, no other evidence of tooth formation was seen [Figs. 3, 4]. There was no history of this anomaly in the child's parents or grandparents.

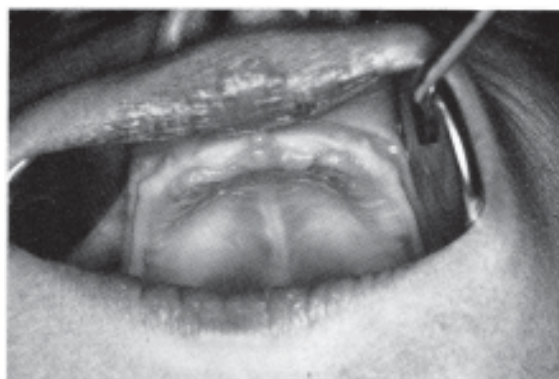


Figure 1. Intraoral photograph showing thin edentulous maxillary ridge and dry and pale mucosa.



Figure 2. Intraoral photograph demonstrating complete anodontia of the mandibular deciduous teeth, sharp alveolar ridge and inflammation of the oral mucosa.

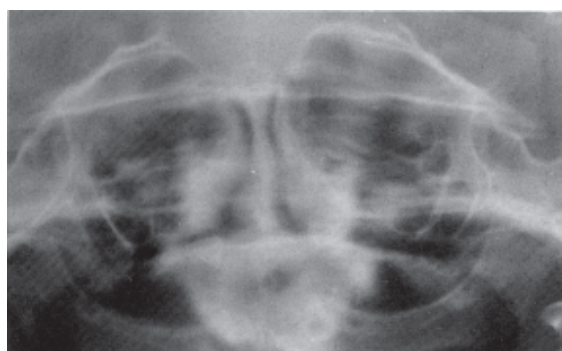


Figure 3. A panoramic radiograph showing 2 maxillary and 2 mandibular cone-shaped crowns, probably canines, in bony crypts. The other teeth are congenitally missing.

Physical examination revealed a boy with atypical features which *included fine, sparse blond scalp hair, scanty eyebrow and eye lashes, mild*

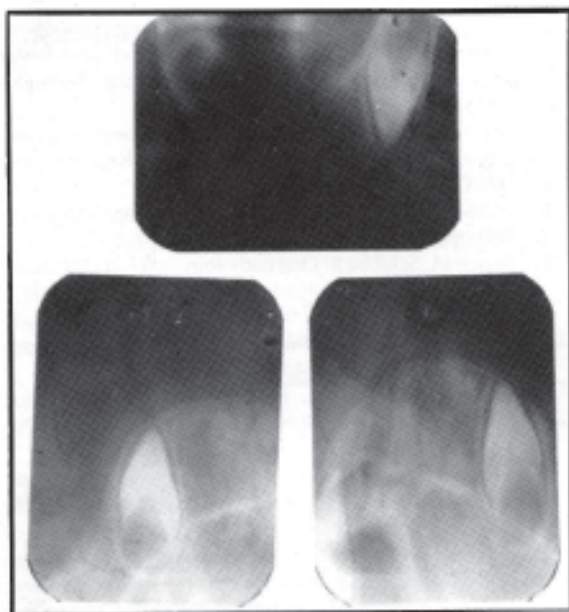


Figure 4, Periapical dental radiographs showing 4 cone-shaped crowns.

frontal prominence, depressed nasal bridge, hypoplastic auricular bobs, dry and wrinkled skin with eczematous patches, and dry, cracked protuberant lips [Figs. 5, 6].

The form of his finger and toenails were normal. The oral and skin findings were consistent with hypohidrotic ectodermal dysplasia.

Discussion

The presentation of facial deformity, dry skin, sparse hair of the scalp, eyebrows and eyelashes and anodontia observed in the case is similar to previous reports.^{3,5,6,7} Xeroderma and eczema are due to anomalies of the skin appendages which may include partial or total absence of the hair follicles, sweat glands and sebaceous glands.^{4,7} Dry eyes, a sequela of diminished tears, observed in the present case report, could be due to partial or total absence of the lacrimal glands or deformity of the gland ducts as suggested in an earlier study.⁸ The observation of normal form and shape finger and toenails in this case agrees with similar observation made by Shaw.⁶

The repeated episodes of hyperthermia observed in this patient has been attributed to defective skin appendages in previous reports.^{2,6,7,9,14} Xerostomia, cracked lips and



Figure 5. Photograph of a facial features of four-year-old boy showing fine, scanty blond hair on the scalp, eyebrows and eyelashes, dry chin and protuberant lips.

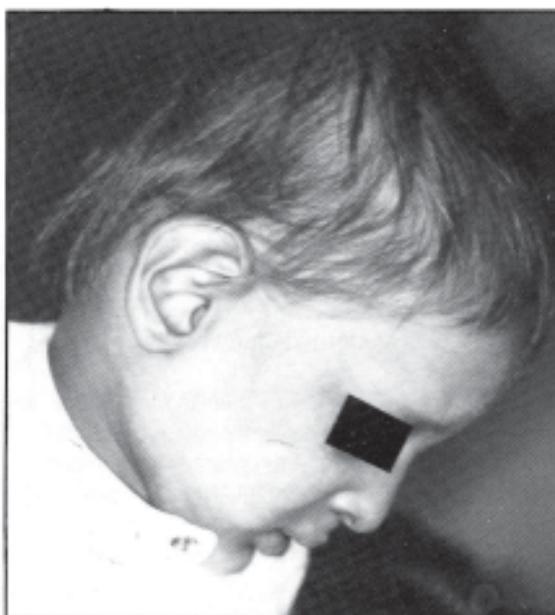


Figure 6. A profile view revealed frontal prominence, depressed nasal bridge, auricular malformation and wrinkle skin of the lower eyelid.

inflamed oral mucosa observed in the present case could be due to partial or complete absence of the intraoral accessory salivary glands, as suggested in the reports by Shaw⁶ and Basserman Nielsen¹⁰.

Freire-Maia and Pinheiro¹ and Giansanti et al.³ gave a useful classification of ectodermal dysplasia,

and have reviewed extensively, the associated syndromes. In their studies, hereditary hypohidrotic ectodermal dysplasia was the most common.

In studies that utilized serial cephalometric measurements, Sarnat *et al*¹⁵, Tuchine *et al*¹⁶, and Borg and Midtgaard¹⁷ have reported that in general facial growth proportion, and pattern of jaw growth appear to be normal in these children despite the absence of tooth development. Therefore, the protuberant lips observed in these children may be attributed to the reduction in the height of the alveolar process.

Congenitally, missing maxillary lateral incisors, maxillary and mandibular third molars, are more common than total anodontia of deciduous and/or permanent teeth.^{4,6,9} In the present case, there was complete anodontia of the deciduous teeth and partial anodontia of the permanent successors.

It is important to emphasize that information in respect of absence of anhidrotic ectodermal dysplasia in the patient's family was given by the child's father. This case is being documented for the rarity of its occurrence. It is recommended that the non-dental problems of such patients be managed by physicians.¹⁸ It is further recommended that partial or complete dentures should be constructed for these patients and readjusted from time to time to allow for normal growth of the orofacial musculature.

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