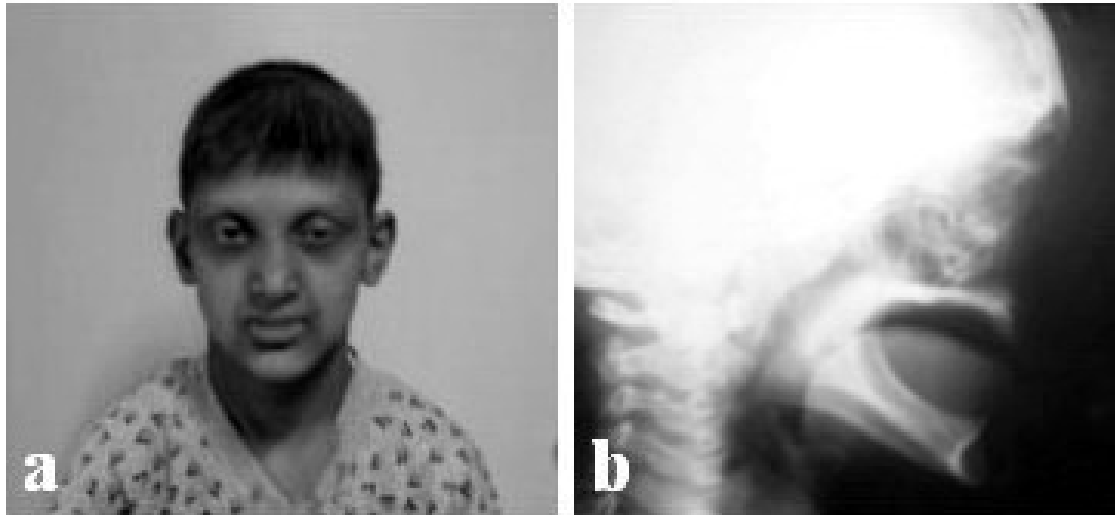


IMAGES IN PEDIATRIC ENDOCRINOLOGY

Congenital Anodontia in Ectodermal Dysplasia

An 11 year-old boy was referred to our department owing to abnormal appearance. He was the first child of consanguineous parents. There was no family history associated with his condition. He was born after 40 weeks of gestation with a birth weight of 3,000 g. No problems had been observed during the neonatal period. The patient weighed 25 kg (<3rd percentile), measured 130 cm in height (<3rd percentile), and had a head circumference of 48 cm (10th percentile). The patient had no family history of similar clinical features and unexplained pyrexia during infancy or childhood. His hair was fine, sparse, lusterless, and fair and he had normal nails (**Fig. a**). He had total anodontia denoted by the complete developmental absence of teeth in both primary and secondary dentitions. The patient appeared mentally normal. His behavior is normal for age. Routine investigation, including complete blood count, liver function, serum biochemistry, renal function, sedimentation rate, and detailed endocrinological tests demonstrated no abnormalities. The usual metabolic screening tests (e.g., serum amino acids, urine organic acids, urine metabolic screens) were normal. His bone age was reported to be within normal limits. Radiographs showed no signs of formation of tooth buds (**Fig. b**). Chromosome analysis in cultured peripheral blood lymphocytes was also normal. Electroencephalogram and magnetic resonance imaging of the brain were normal, as were abdominal ultrasound, pelvic ultrasound, and echocardiographic examinations. A diagnosis of ectodermal dysplasia in association with congenital anodontia was made and follow up was offered to the patient.

The causes of anodontia, whether total or partial, are attributed to hereditary ectodermal dysplasia, environmental factors, Sotos syndrome, Goltz Gorlin syndrome, etc. X-linked hypohidrotic ectodermal dysplasia is a rare disease characterized by the hypoplasia or absence of eccrine glands, dry skin, scant hair, and dental abnormalities¹. Common manifestations include defective hair follicles and eyebrows, frontal bossing with prominent supraorbital ridges, nasal bridge depression, and protuberant lips. Intra-orally, common findings are anodontia or hypodontia, conical teeth, and, consequently, generalized spacing^{2,3}.

Patients with severe dental abnormalities may develop feeding difficulties, which may result in malnutrition and failure to thrive. Therefore, pediatric endocrinologists can be the first to diagnose ectodermal dysplasia.

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