



POSTER PRESENTATION

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Oral manifestation of Goltz-Gorlin syndrome in a young girl

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Introduction

Focal dermal hypoplasia (Goltz-Gorlin syndrome) is a multi-system disorder characterized by involvement of skin, skeletal system, eyes and face. It is caused by loss-of-function mutations in the *PORCN* gene. We report the case of a young female, focusing on the dental features.

Aim

To describe the oral manifestation of a rare disorder that resembles ectodermal dysplasia (ED).

Case report

Clinical, radiological and genetic findings revealed common features of Goltz-Gorlin syndrome and pure ED. Oro-dental characteristics of the patient mostly corresponded to those described in the literature. However, previously unreported oro-dental findings such as taurodontism, peg-shaped teeth and microdontia are considered unusual for Goltz-Gorlin syndrome, but similar to the dental features of hypohidrotic ED. Clinical characterization of the patient by a multidisciplinary approach is described and a comprehensive review of the literature is presented.

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