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Oral manifestation of Goltz-Gorlin syndrome in a young girl

M Callea^{1*}, I Yavuz², L Deroma³, M Montanari⁴, G Clarich¹, M Maglione⁵, E Albertini⁶, L Garavelli⁷

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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.

Author details

¹Department of Maxillo-Facial Surgery and Paediatric Dentistry, Institute for Maternal and Child Health, Trieste, Italy. ²Dicle University, Diyarbakır, Turkey. ³University Hospital Santa Maria della Misericordia, Udine, Italy. ⁴University of Bologna, Italy. ⁵University of Trieste, Italy. ⁶University of Ferrara, Italy. ⁷S. Maria Nuova Hospital, Reggio Emilia, Italy.

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¹Department of Maxillo-Facial Surgery and Paediatric Dentistry, Institute for Maternal and Child Health, Trieste, Italy

Full list of author information is available at the end of the article



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