HEAD & FACE MEDICINE

POSTER PRESENTATION Open Access

Oral manifestation of Goltz-Gorlin syndrome in a young girl

M Callea[1*], I Yavuz[2], L Deroma[3], M Montanari[4], G Clarich[1], M Maglione[5], E Albertini[6], L Garavelli[7]

From 5th International Conference on Ectodermal Dysplasia (ED2012)
Erlangen, Germany. 1-3 June 2012

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.

doi:10.1186/1746-160X-8-S1-P8
Cite this article as: Callea *et al.*: Oral manifestation of Goltz-Gorlin syndrome in a young girl. *Head & Face Medicine* 2012 **8**(Suppl 1):P8.

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

Published: 25 May 2012

[1]Department 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