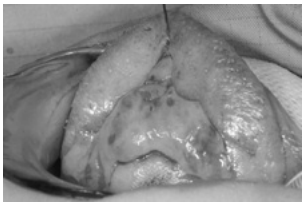
Case Report

Tongue ulcer in a case of X-linked diskeratosis congenita



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Introduction. Dyskeratosis Congenita (DKC) is characterized by immunodeficiency, bone marrow failure, anomalies of the skin, nails and mouth, and cancer predisposition. This syndrome affects predominantly males, with onset in childhood. All individuals with DKC have abnormally short telomeres for their age. The typical oral manifestation is leukoplakia, that may occur in any region of the mucosa.

Case details. A male child of 16 months, in apparent good health, was brought by his parents in the emergency room of the dental clinic of AO San Paolo in Milano, for a lesion of the back of the tongue.

The lesion had been present for about 4 months and its appearance was preceded by a condition that the pediatrician had identified as geographic tongue. The kid did not show any discomfort due to the lesion and he was eating regularly.

On examination, an ulcer of about 1 cm in diameter, at the center of the back of the tongue was detected. A cell sam-

ple was collected by brushing for cytology examination. Treatment with topical clobetasol and chlorhexidine gel was recommended, and complete blood count prescribed. The cytological diagnosis was: “sample consisting of granulocyte layer”. A biopsy was then performed under general anesthesia, but again the pathological diagnosis was inconclusive. Genetic tests were performed at the Ospedale Pediatrico Gaslini, in Genova, and a diagnosis of X-linked dyskeratosis congenita was eventually proposed. The child under wentallogenic stem cell transplantation.

Conclusions. The oral lesion of this young patient was not a typical manifestation of the condition (leukoplakia), but a putative sign of haematological abnormalities.

References

- Scully C, Langdon J, Evans J. Marathon of eponyms: 26 Zinsser-Engman-Colesyndrome (Dyskeratosis congenita). Oral Dis 2012; 18:522-3.