Oral Lesion Diagnosis for Patient with Dyskeratosis Congenita

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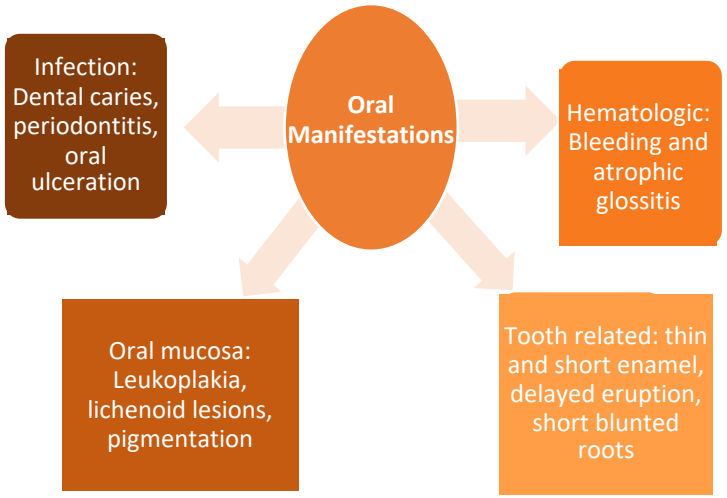
INTRODUCTION

- Dyskeratosis Congenita is a rare genetic syndrome with an inherited X linked recessive pattern caused by mutation in the DKC1 gene, resulting in dysfunction of the telomerase.
- It leads to impaired maintenance of telomeres and reduced telomere length which is crucial in cell division.
- Cells that divide rapidly are especially vulnerable to the effects of shortened telomeres like cells of the nail beds, hair follicles, oral mucosa, and bone marrow. It also causes premature aging.
- Clinically, the syndrome can manifest as cutaneous hyperpigmentation, nail dystrophy, pancytopenia, aplastic anemia, leukemia, and solid tumors (usually squamous cell carcinoma of the head/neck or anogenital cancer).



IMAGING

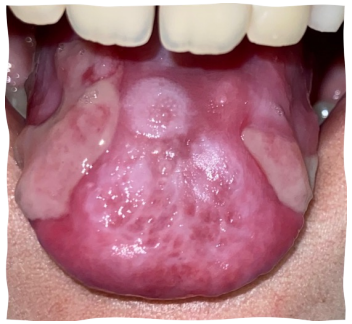


Figure 1: Patient dorsal and lateral tongue surfaces lesions

Figure 2: Patient nails dystrophy

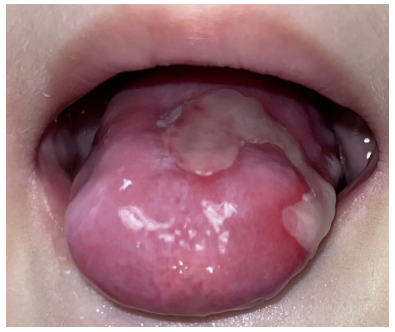


Figure 3: Patient brother similar tongue lesions

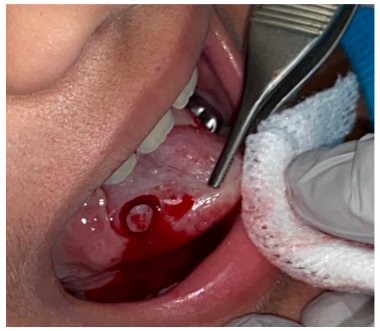


Figure 4: Punch Bopsy Procedure

CASE PRESENTATION

- 9-year-old male patient, with PMHx remarkable for Dyskeratosis Congenita (DCK1 linked) and anxiety, who was referred to our Pediatric Dentistry Clinic, with a chief complaint of excessive bleeding due to leukoplakia since patient started school, worsen by his anxiety. For which mother has been using magic mouth wash without relief.
- Dental History: every 6 months, brushes teeth twice a day, do not floss, and frequently snacks. With history of multiple dental restorations.
- Radiographic findings: late mixed dentition, and tooth #32 bud present.
- Punch biopsy: mild epithelial reactive change and subepithelial granulation tissue and no fungal organisms identified on periodic acid Schiff-stained sections, favoring a resolving inflammatory process.
- Differential diagnosis: Initially where erosive lichen planus, pemphigus, and pemphigoid. Nevertheless, they where rule out with patient's tongue biopsy.
- Treatment: continue the use of magic mouthwash without nystatin, a topical corticosteroid called lidex 0.05% gel BID.

DISCUSSION

Patients presenting with Dyskeratosis Congenita disease are at the high risk of developing oral manifestations including inflammatory processes.
Therefore, it is of outmost importance to follow up with pediatric dentist and oral pathologist to stipulate further management like lesion biopsy and then treatment.

CONCLUSION

Practitioners should suspect Dyskeratosis Congenita disease in patients that present with unresolved oral lesions and nail dystrophy as an associated findings, as well as performed a biopsy to discard any other malignant lesions.

REFERENCES

- Wright, J. T., Fete, M., Schneider, H., Zinser, M., Koster, M. I., Clarke, A. J. & Morasso, M. I. (2019). Ectodermal dysplasias: Classification and organization by phenotype, genotype and molecular pathway. *American Journal of Medical Genetics Part A*, *179*(3), 442-447.
- Thongprasom, K. (2017). A review of the effectiveness and side-effects of fluocinolone acetonide 0.1% in the treatment of oral mucosal diseases. *Acta Stomatologica Croatica*, *51*(3), 240.
- *Lidex Topical: Uses, Side Effects, Interactions, Pictures, Warnings & Dosing - WebMD*. (n.d.). WebMD. Retrieved December 1, 2021, from https://www.webmd.com/drugs/2/drug-3786/lidex-topical/details