



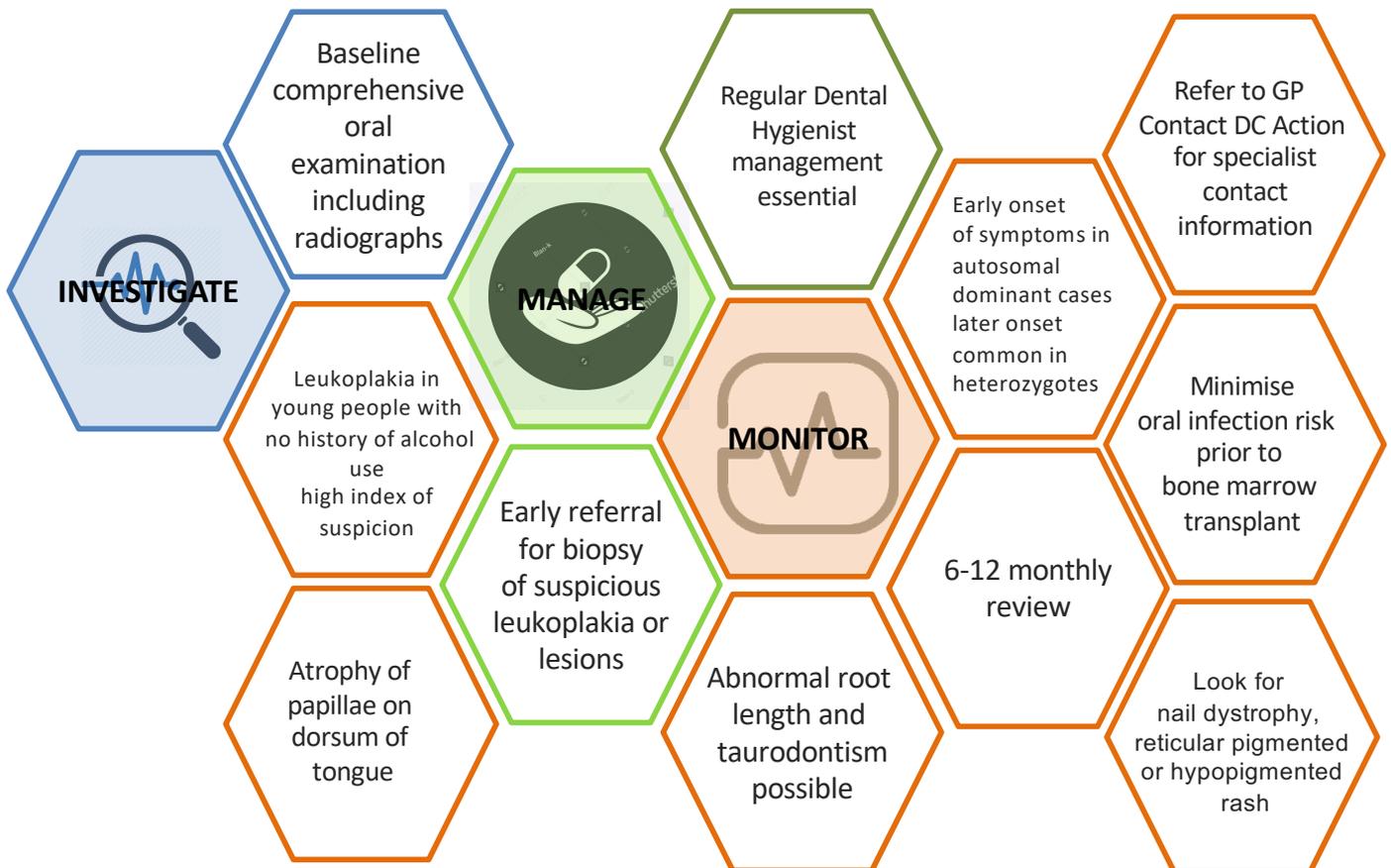
Telomere Biology Disorders (TBDs), including Dyskeratosis congenita (DC) are complex, genetic multi-system disorders that cause premature aging of cells and organs. The clinical symptoms of TBDs are varied. A person diagnosed with a TBD may not experience all of the symptoms below.

Oral / dental

Increased risk of developing oropharyngeal squamous cell carcinoma. Oral changes can include oral leukoplakia, increased dental caries, hypodontia, thin enamel structure, aggressive periodontitis, intraoral brown pigmentation, tooth loss, taurodontism and blunted roots.

Consider a diagnosis of DC or other inherited bone marrow failure syndromes / TBD in young persons with oral leukoplakia, particularly those with no history of smoking.

Multiple permanent teeth with decreased root/crown ratios warrant investigation and may suggest TBD. Age of onset of symptoms can be extremely variable depending on genetic alterations. Frequent monitoring and referral for biopsy of suspicious lesions for early diagnosis of potential malignant transformations recommended. Bone Marrow Transplant in TBD with bone marrow failure. Take steps to minimise oral infection prior to transplant.



Atkinson *et al.* Oral and Dental Phenotype of Dyskeratosis Congenita. *Oral Dis.* 2008 July; 14(5): 419–427.

Savage SA. Dyskeratosis congenita 2009 (updated 2019 Nov 21) in Adam MP, Ardinger HH, Pagon RA *et al.*, Eds. *Gene Reviews*® [internet] Seattle (WA): University of Washington Seattle; 1993-1019. Bookshelf URL <https://ncbi-nlm.nih.gov/books/>

HH = Høyeraal-Hreidarsson, CP = Coats Plus, RS = Revesz Syndrome

*No DC specific treatments currently indicated.

This information is based on reports from the medical literature. Please see your doctor if you have concerns about TBD.



ADVOCACY



EDUCATION



SUPPORT

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The US National Cancer Institute (NCI) monitors a large cohort of inherited bone marrow failure patients for cancer incidence. The latest 15 year follow up data suggests that those affected by Dyskeratosis Congenita (DC) / Telomere Biology Disorders (TBDs) developed head and neck and anogenital squamous cell carcinomas at rates that were hundreds-fold greater than those of the general population.

Alter BP *et al.* Cancer in the National Cancer Institute inherited bone marrow failure syndrome cohort after fifteen years of follow-up. *Haematologica* 2018 Volume 103(1):30-39.

CASE STUDIES

Condition	Oral / ENT features	Reference
DC (highly suggestive)	A three-month history of an ulcer on the left lateral border of the tongue.	Fatehi KS <i>et al.</i> Squamous cell carcinoma of the tongue in a patient with Dyskeratosis congenita: a rare entity. <i>British Journal of Oral and Maxillofacial Surgery</i> : 57(2019) 79-81
DC	Two case studies of Dyskeratosis Congenita (DC) and a brief review of the literature.	Karunakaran A <i>et al.</i> Dyskeratosis Congenita: A Report of Two Cases. <i>Case Reports in Dentistry</i> Volume 2013.
DC	A case study and recommendations for dental management of a child with Dyskeratosis Congenita undergoing bone marrow transplantation.	Koruyucu M <i>et al.</i> Oral and Dental Findings of Dyskeratosis Congenita <i>Case Reports in Dentistry</i> Volume 2014.
DC	A review of the spectrum of diseases encompassed by the term Dyskeratosis Congenita (DC). In its classic form it is usually characterized by the mucocutaneous triad of abnormal skin pigmentation, nail dystrophy, and leukoplakia.	Dokal I. Dyskeratosis congenita. <i>ASH Education Book</i> . December 10, 2011vol. 2011 no. 1 480-486.
DC	The prevalence of oral leukoplakia, increased dental caries, hypodontia, thin enamel structure, aggressive periodontitis, intraoral brown pigmentation, tooth loss, taurodontism and blunted roots was studied in a cohort of 17 patients with DC and 23 family members. The most common oral changes in DC patients were oral leukoplakia (65% of the entire DC population), decreased root/crown ratio and mild taurodontism.	Atkinson JC <i>et al.</i> Oral and Dental Phenotype of Dyskeratosis Congenita. <i>Oral Dis</i> . 2008 July; 14(5): 419–427.
DC	Oral and Dental Changes of Dyskeratosis Congenita: A Case Report with Literature Review.	Serindere G. <i>Journal of Advanced Oral Research</i> 2018 : 9(1-2) 20–23 DOI: 10.1177/2320206818789761
See also	White spongy nevus: a rare hereditary condition.	Surendran S and Wright G. P58 British Society of Paediatric Dentistry Conference Abstracts 2018 https://doi.org/10.1111/ipd.4_12407
See also	A decision tree for diagnoses predisposing to oral white lesions according to their clinical features.	Mortazavi H <i>et al.</i> Review. Oral White Lesions: An Updated Clinical Diagnostic Decision Tree. <i>Dentistry Journal</i> : 2019, 7, 15. doi:10.3390/dj7010015