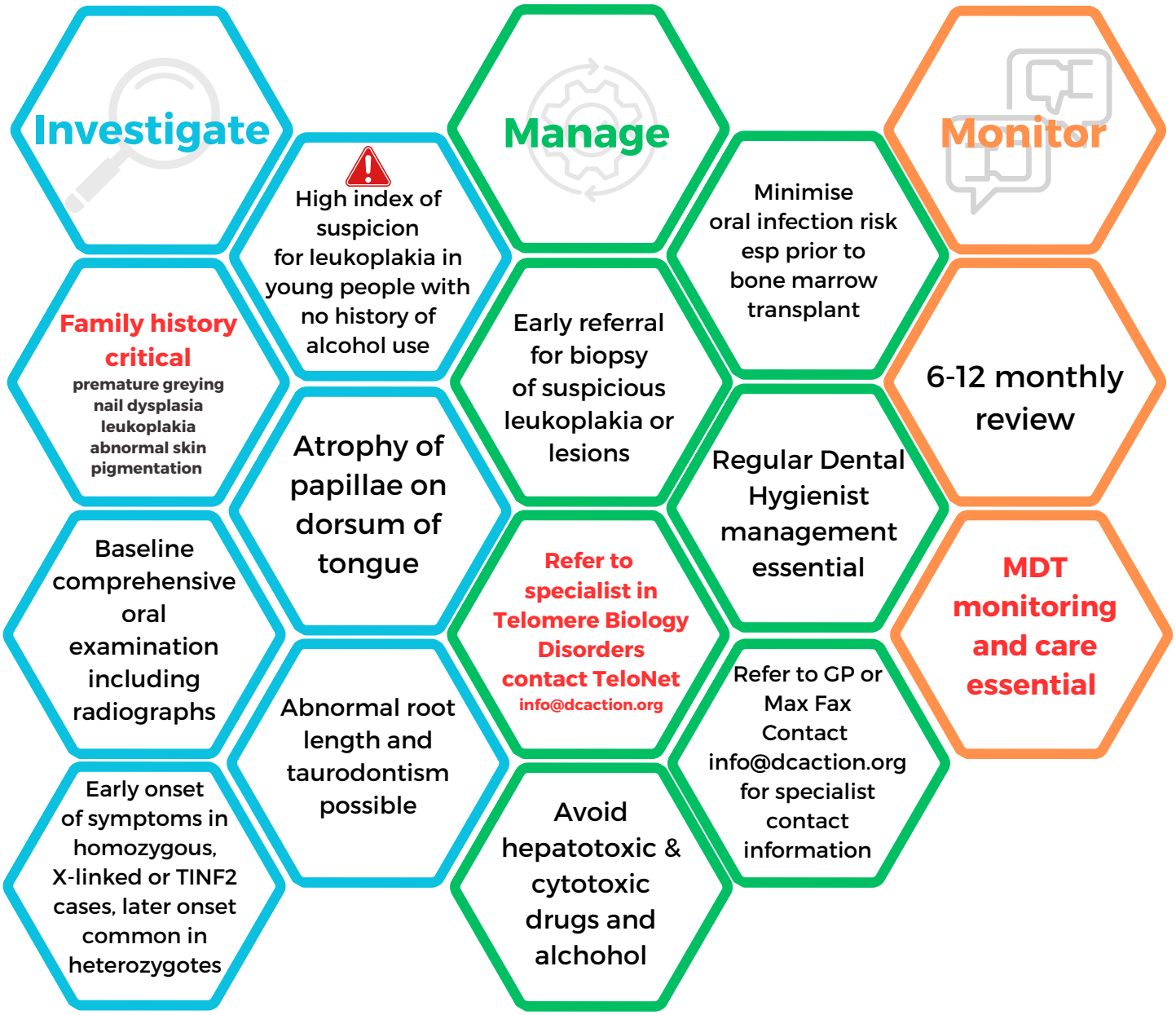
Telomere Biology Disorders (TBD), including Dyskeratosis Congenita (DC), are complex, genetic, multi-system disorders that cause premature aging of cells and organs. The clinical symptoms of TBDs are varied and require multi-disciplinary diagnosis, monitoring and management.

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 indicated in TBD with bone marrow failure. Take steps to minimise oral infection prior to transplant.

Team Telomere Telomere Biology Disorders: Diagnosis and Management Guidelines 2022 Chapter 8
https://teamtelomere.org/telomere-biology-disorders-diagnosis-and-management-guidelines-downloads/



*Atkinson JC, Harvey KE, Domingo DL, Trujillo MI, Guadagnini JP, Gollins S, Giri N, Hart TC, Alter BP. Oral and dental phenotype of dyskeratosis congenita. Oral Dis. 2008 Jul;14(5):419-27. doi: 10.1111/j.1601-0825.2007.01394.x.

Savage SA, Niewisch MR. Dyskeratosis Congenita and Related Telomere Biology Disorders. 2009 Nov 12 [Updated 2023 Jan 19]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023. Available from: https://www.ncbi.nlm.nih.gov/books/NBK22301/

The US National Cancer Institute (NCI) monitors a large cohort of inherited bone marrow failure patients for cancer incidence. Fifteen year follow up data suggests that people with Dyskeratosis Congenita and Telomere Biology Disorders may be more likely to develop cancer at a higher frequency and at a younger age than the general population.

A cohort study including 230 individuals withTBDs found a 3-fold increased risk of cancer in individuals with TBDs compared with the general population. Individuals with autosomal recessive or X-linked TBDs had the highest risk of cancer, at approximately 19-fold in those without prior organ transplant.

Regular monitoring and early treatment is critical.

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.

Niewisch MR, Kim J, Giri N, Lunger JC, McReynolds LJ, Savage SA. Genotype and Associated Cancer Risk in Individuals With Telomere Biology Disorders. JAMA Netw Open. 2024 Dec 2;7(12):e2450111. doi: 10.1001/jamanetworkopen.2024.50111.

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Chapter 8 Dental and Oral Complications.
https://teamtelomere.org/telomere-biology-disorders-diagnosis-and-management-guidelines-downloads/

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 et al. Squamous cell carcinoma of the tongue in a patient with Dyskeratosis congenita: a rare entity. British Journal of Oral and Maxillofacial Surgery: 57(2019) 79-81
DC	Two case studies of Dyskeratosis Congenita (DC) and a brief review of the literature.	Karunakaran A et al. Dyskeratosis Congenita: A Report of Two Cases. Case Reports in Dentistry Volume 2013.
DC	A case study and recommendations for dental management of a child with Dyskeratosis Congenita undergoing bone marrow transplantation.	Koruyucu M et al. Oral and Dental Findings of Dyskeratosis Congenita Case Reports in Dentistry 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. ASH Education Book. December 10, vol. 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 et al. Oral and Dental Phenotype of Dyskeratosis Congenita. Oral Dis. 2008 July; 14(5): 419–427.
DC	Oral and Dental Changes of Dyskeratosis Congenita: A Case Report with Literature Review.	Serindere G. Journal of Advanced Oral Research 2018 : 9(1-2) 20–23 DOI: 10.1177/2320206818789761