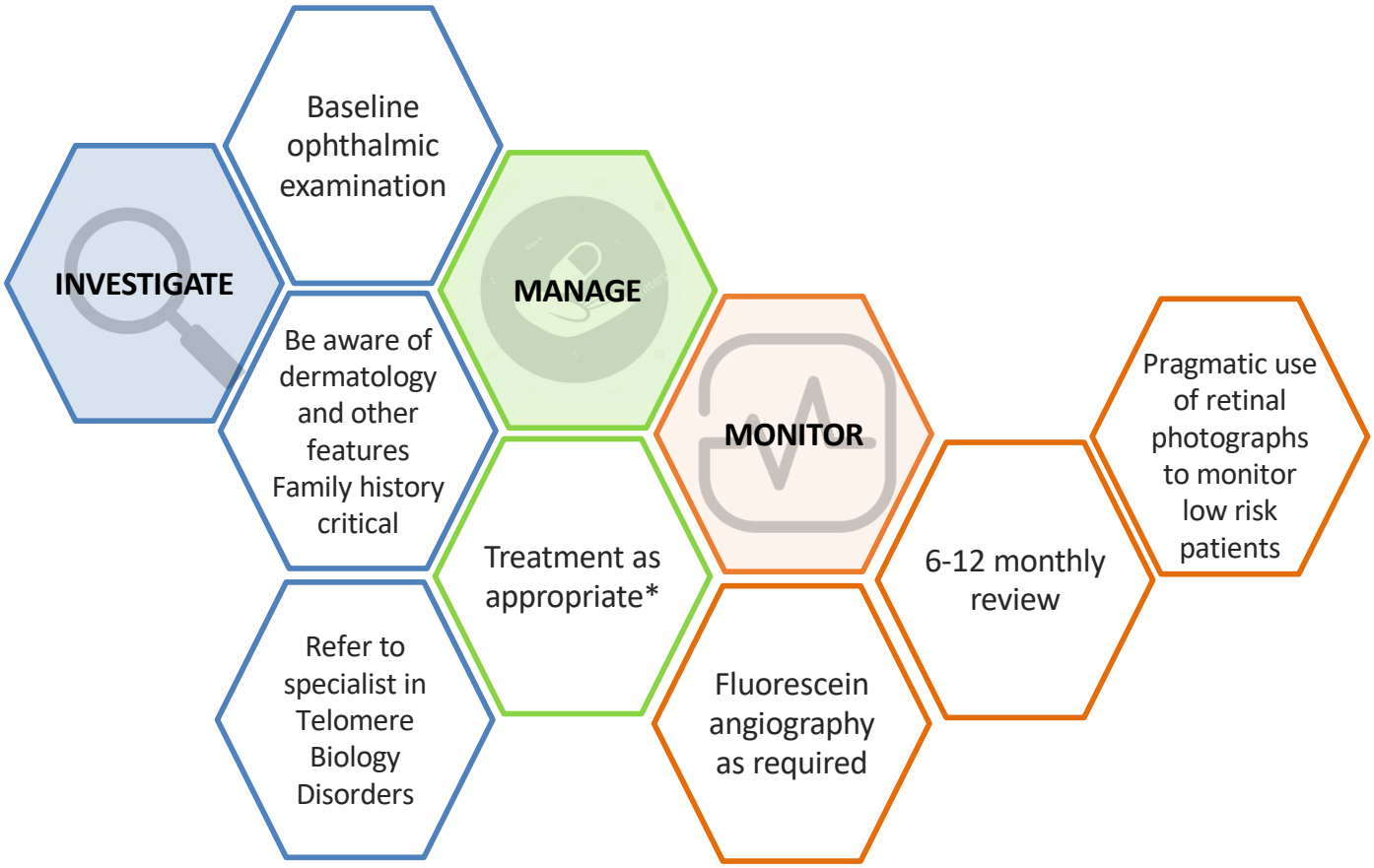
Telomere Biology Disorders (TBDs), including Dyskeratosis congenita, are complex multi-system disorders that cause premature aging of cells and organs. The clinical symptoms of TBDs are varied. A person diagnosed with a TBD may not experience all of the symptoms described below.

Ophthalmology

Epiphora, lachrymal duct stenosis/absence of tear duct, blepharitis, conjunctivitis, sparse eye lashes/ectropion/entropion/trichiasis, corneal dryness/scarring/ulceration/perforation, retinal vascular changes/atherosclerosis/neovascularisation/proliferation, retinal haemorrhages, exudative retinopathy.

DC variants: Høyeraal-Hreidarsson (HH), Coats Plus (CP), Revesz Syndrome (RS) at high risk of ocular involvement.

Secondary manifestations: Post transplant Graft Versus Host Disease (GVHD) and increased sensitivity to treatments such as corticosteroid or radiation-related changes likely to be more severe e.g. cataract.



Marena R. Niewisch & Sharon A. Savage (2019) An update on the biology and management of dyskeratosis congenita and related telomere biology disorders, Expert Review of Hematology, 12:12, 1037-1052, DOI: 10.1080/17474086.2019.1662720

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/

*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**

Please contact us. We welcome your feedback.

Severe Dyskeratosis Congenita (DC) / Telomere Biology Disorder (TBD) variants Høyeraal-Hreidarsson Syndrome, Revesz Syndrome (RS) and Coats' Plus Syndrome (CPS) may present with serious ocular problems. In inherited bone marrow syndromes associated with Telomere Biology Disorders, common findings include punctal stenosis with epiphora, entropion, and trichiasis. Retinal changes may not be rare and warrant close evaluation.

In a cross-sectional study, 75 patients (2001-2007) with an inherited bone marrow failure syndrome and 121 of their first-degree relatives had complete ophthalmic investigations. In the 28 patients with DC, abnormalities of the lacrimal drainage system (29%) were the most prevalent findings, followed by retinal abnormalities (pigmentary changes, retinal neovascularization, retinal detachment, exudative retinopathy) in 21%, cicatricial entropion with trichiasis and blepharitis in seven percent each, sparse eyelashes and congenital cataract in three and a half percent each.
Tsilou ET *et al*. Ocular and orbital manifestations of the inherited bone marrow failure syndromes: Fanconi anemia and dyskeratosis congenita. *Ophthalmology*. 2009;117(3):615–622.

CASE STUDIES

Condition	Ocular findings	Reference
Autosomal dominant DC	Bilateral retinal vasculopathy and proliferative retinopathy with vitreous hemorrhage in the right eye, in the absence of pancytopenia	Vaz-Pereira S *et al*. Bilateral retinal vasculopathy associated with autosomal dominant dyskeratosis congenita. Eur J Ophthalmol 2013;23(5):772-5
DC	Two siblings presenting with exudative retinopathy, thrombocytopenia, and macrocytosis with markedly shortened telomeres and a previously unreported, inherited mutation in TERT, c.2603A>G	Sharma A *et al*. Dyskeratosis Congenita Caused by a Novel TERT Point Mutation in Siblings With Pancytopenia and Exudative Retinopathy. Pediatr Blood Cancer. 2014; 61(12):2302-4.
HH (DKC1 mutation)	Sclerotic retinal vessels and peripheral non-perfusion and neovascularisation.	Allingham MJ. Bilateral Proliferative Retinopathy Associated With Høyeraal-Hreidarsson Syndrome, a Severe Form of Dyskeratosis Congenita. *Ophthalmic Surg Lasers Imaging Retina*. 2016;47(4):366–368.
CPS	Vitreous haemorrhage and peripheral retinal arteriovenous anastomosis.	Mansukhani S *et al*. Cerebroretinal microangiopathy with calcifications and cysts (CRMCC) or "Coats Plus": when peripheral retinal vasculature signals neurologic disease. J AAPOS.2017;21(5):420-422.
DC	Siblings diagnosed with familial exudative vitreoretinopathy managed with laser photocoagulation. Eight years later, both developed pancytopenia secondary to bone marrow failure. Both had severely shortened telomere length caused by a missense mutation in the gene encoding reverse transcriptase component of telomerase	Thanos A *et al*. Retinal vascular tortuosity and exudative retinopathy in a family with dyskeratosis congenita masquerading as familial exudative vitreoretinopathy. Retin Cases Brief Rep. 2017;11 Suppl 1:S187-S190.