



**Dyskeratosis Congenita (DC) and other Telomere Biology Disorders (TBD) are complex multi-system disorders resulting from mutations in genes responsible for telomere maintenance. The conditions cause premature aging of cells and organs but the clinical symptoms of TBDs are varied. A person diagnosed with a TBD may not experience all of the symptoms listed below.**

### Ophthalmology

Blepharitis, corneal dryness /scarring/ ulceration/ perforation. Conjunctivitis, epiphora, exudative retinopathy (CP). Sparse eye lashes/ectropion/entropion/trichiasis, stenosis/absence of tear duct, retinal haemorrhages, retinal vascular changes / atherosclerosis / neovascularisation /proliferation.

### Gastroenterology

Enteropathy, gastrointestinal bleeding, oesophageal stenosis/stricture/webbing, telangiectasias, ulceration, varices, vascular ectasias, enterocolitis.

### Hepatology

Hepatopulmonary syndrome, steatohepatitis, fibrosis, cirrhosis, hepatocellular carcinoma, non-cirrhotic portal hypertension, nodular regenerative hyperplasia.

### Dermatology

Alopecia, dystrophic finger & toenails, early greying of hair, hyperhidrosis, lacy reticular pigmentation.



### Neurology

Ataxia/spasticity/hypotonia (HH), developmental delay, cerebellar hypoplasia (HH), intracranial calcifications (RS), microcephaly. Structural brain and neurodevelopmental abnormalities.

### Dentistry/Oral Medicine

Atrophy of papillae on dorsum of tongue, decreased crown/root ratio, dental caries/tooth loss, oral leukoplakia, periodontal disease, taurodontism, tongue/mouth squamous cell cancer.

### Respiratory

Pulmonary disease including idiopathic pulmonary fibrosis, interstitial disorders, organising pneumonia, restrictive, arteriovenous fistulus /malformations, hepatopulmonary syndrome.

### Haematology

Bone marrow failure: aplastic anaemia, myelodysplastic syndrome, leukaemia, acute myelogenous leukaemia.

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

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



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## Audiology

Hearing loss (rare).

## Orthopaedics

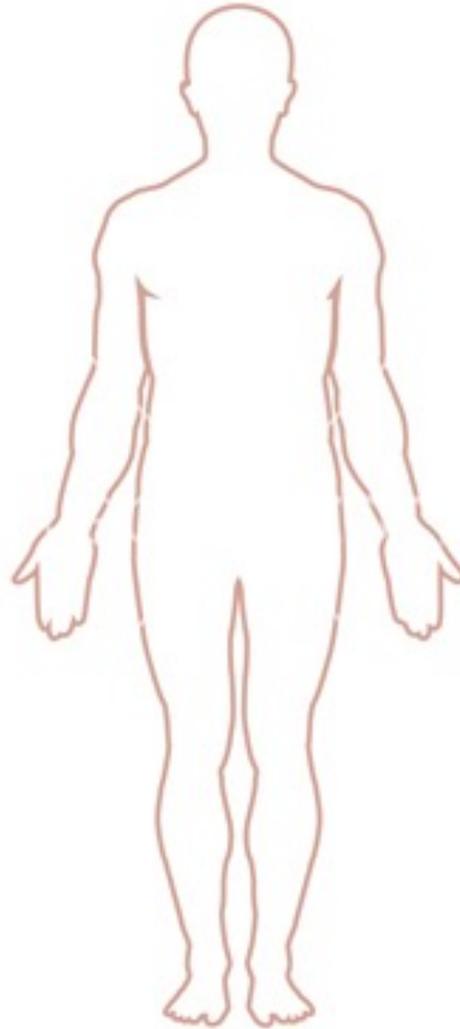
Avascular necrosis of the hips & shoulders, osteopenia, osteoporosis, scoliosis.

## Oncology

Squamous cell head, neck & anogenital cancers.

## Gentourinary

Anogenital squamous cell cancers/epithelial cancers, haematuria due to haemorrhagic cystitis, meatal/urethral stenosis/hymenal stricture, microscopic phimosis.



## Psychology/Psychiatry

Neurologic and psychiatric symptoms common inc mental health issues secondary to diagnosis. ADHD, schizophrenia described in case reports.

## Cardiology

Cardiovascular fibrosis, dilated cardiomyopathy, atrial or ventricular septal defects (rare).

## Endocrinology

Hypogonadism, intrauterine growth restriction, short stature.

## Dietetics

Failure to thrive, feeding difficulties, poor weight gain.

## Immunology

Immunodeficiency.

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/>  
Tummala H, Walne A, Dokal I. The biology and management of dyskeratosis congenita and related disorders of telomeres. Expert Rev Hematol. 2022 Aug;15(8):685-696. doi: 10.1080/17474086.2022.2108784

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

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