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

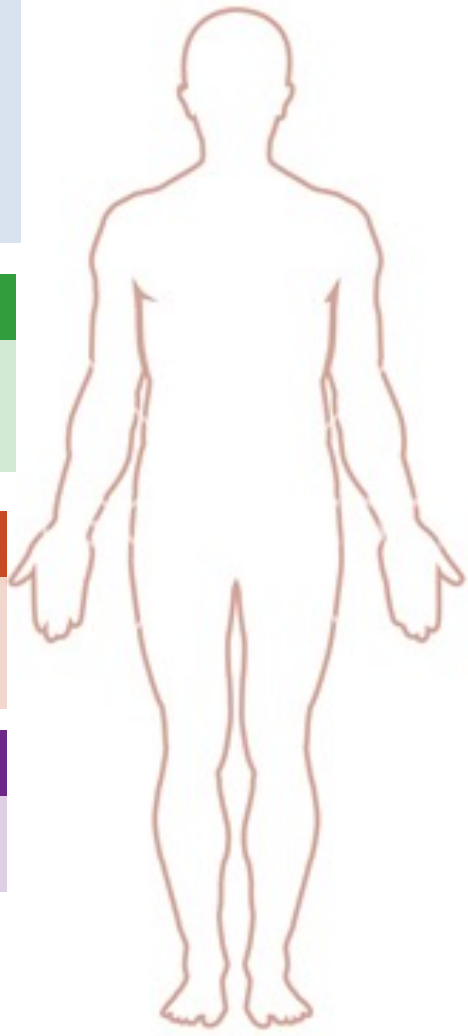
Enteropathy, gastrointestinal bleeding, oesophageal stenosis/stricture/webbing, telangiectasias, ulceration, varices, vascular ectasias, enterocolitis.

Hepatology

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

Dermatology

Alopecia, dystrophic finger & toenails, early greying of hair, hyperhydrosis, 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, peridontal 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 = Høyeraal-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.

ADVOCACY **EDUCATION** **SUPPORT**

Please contact us.
We welcome your feedback.

Charity No. 1167150 | www.dcaction.org | info@dcaction.org

August 2023

Audiology

Hearing loss (rare).

Orthopaedics

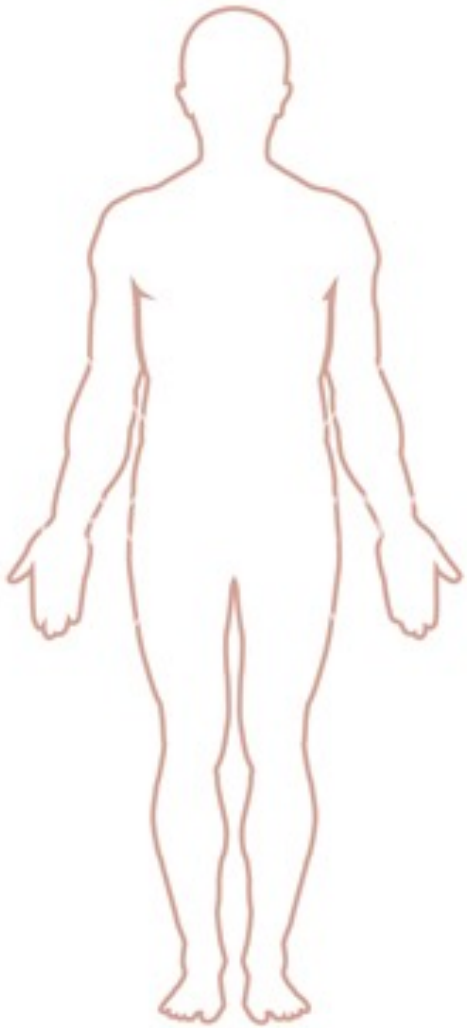
Avascular necrosis of the hips & shoulders, osteopenia, osteoporosis, scoliosis.

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/

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

DC ACTION

ADVOCACY EDUCATION SUPPORT

Charity No. 1167150 | www.dcaction.org | info@dcaction.org

Please contact us .
We welcome your feedback.