



Telomere Biology Disorders (TBD) including Dyskeratosis Congenita (DC) 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 listed below.

Ophthalmology

Corneal dryness/scarring/ulceration/perforation.
Conjunctivitis, epiphora, exudative retinopathy (CP), blepharitis.
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.

Hepatology

Hepatopulmonary syndrome, liver disease, non-cirrhotic portal hypertension.

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 neuro-developmental 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, leukemia, acute myelogenous leukemia.



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



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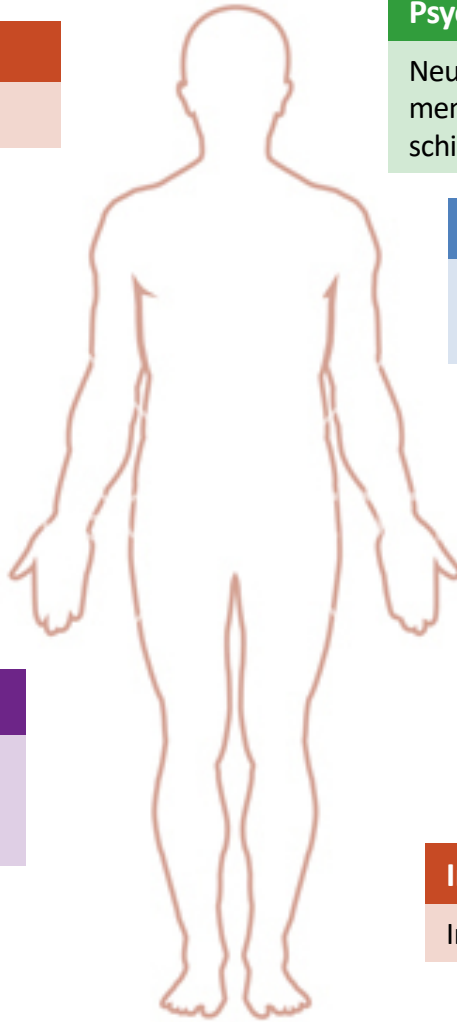


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Audiology

Hearing loss (rare).

Psychology/Psychiatry

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

Orthopaedics

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

Cardiology

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

Oncology

Squamous cell head, neck & anogenital cancers.

Endocrinology

Hypogonadism, intrauterine growth restriction, short stature.

Gentourinary

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

Dietetics

Failure to thrive, feeding difficulties, poor weight gain.

Immunology

Immunodeficiency.

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

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/>

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](https://doi.org/10.1080/17474086.2019.1662720)

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