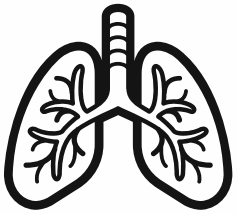
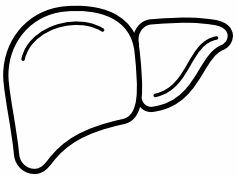
TELOMERE BIOLOGY DISORDERS MONITORING RECOMMENDATIONS

Respiratory



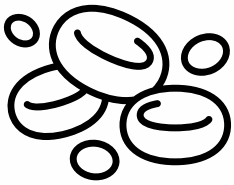
Baseline
Pulmonary function test
Consider HRCT based on results and risk factors
Bubble echocardiogram (agitated saline contrast echocardiography) to check for AV malformations
Monitoring
Annual pulmonary function test
HRCT as clinically indicated but avoid excessive radiation exposure
Bubble Echo for pulmonary symptoms in the absence of pulmonary fibrosis

Hepatology



Baseline
Evaluate risk factors for hepatic disease Liver function tests, ultrasound and / or transient elastography fibroscan
Evaluate for clinical signs of oesophageal stenosis
Monitoring
Liver function tests annually
Consider imaging (fibroscan /ultrasound) every 2 years
On androgen therapy Check liver function tests prior to starting and every 1-2 weeks for first month, then every 6-12 weeks or 3-6 months for lower doses
Check lipid profile prior to starting and every 6-12 months
Perform liver ultrasound examination prior to starting androgens and then every 2 years to evaluate for adenomas, carcinomas or fibrosis

Haematology



Baseline
FBC with differential and reticulocyte count.
BM aspiration and biopsy.
Conventional and molecular cytogenetics - assess for somatic variants / clones
Monitoring
FBC normal / no cytogenetic abnormality:
FBC every 6-12 months - BM if cytopenia develops
Mild cytopaenia / no cytogenetic abnormality:
FBC every 3-4 months BM based on clinical development, consider regular intervals (eg, every 1-3 years)
Abnormal cytogenetics:
More frequent FBC / BM evaluation to evaluate potential leukaemic or MDS progression; intervals depend on development of FBC counts
Refer high-risk abnormalities such as chromosome 7 change immediately to specialist centre
Progressive decline or rise in blood counts require FBC and BM evaluation based on clinical situation
On androgen therapy:
FBCs prior to therapy; repeat FBCs every 4-6 weeks to assess response,
When counts are stable -every 2-3 months

Contact info@dcaction.org for information on TELONET a clinical network aiming to improve diagnostic pathways, facilitate expert referrals, coordinate MDT care, share expertise and enable research into Telomere Biology Disorders

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ENT and Oral Medicine

Baseline and monitoring

Annual cancer screening by an Ear Nose and Throat specialist

Dental

Baseline and monitoring

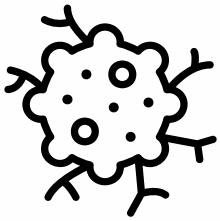
Advise on dental hygiene

Oral screen every 6 months

Refer leukoplakia to an Oral Medicine specialist

The risk of head and neck cancer is raised in Telomere Biology Disorders

Oncology and Dermatology



Dermatology baseline and monitoring

Regular skin self-examination for new or changing skin growth and annual dermatologist evaluation starting before the age of 30 years

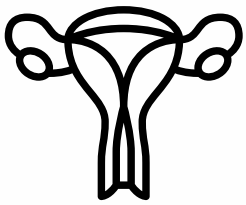
Oncology

Follow surveillance guidelines for breast cancer, cervical cancer, colon / rectal cancer, lung cancer, prostate cancer

In case of cancer diagnosis: increased sensitivity to therapeutic radiation and chemotherapy may require dose reductions / regimen modification

Cutaneous squamous cell carcinomas have been described in young adults with TBDs

Gynaecology/ obstetrics / urology



Baseline and monitoring:

Annual gynaecological evaluation with HPV testing - start at 18 years of age or at onset of sexual activity

HPV vaccination in females and males < 27 years of age if not adequately vaccinated in childhood

Leukoplakia / VIN / CIN assessment

Baseline assessment for genitourinary anomalies, including symptoms of urethral stenosis, penile / or vulval leukoplakia

Pre-pregnancy- refer to genetics for counselling re: preimplantation embryo selection

Pregnancy: referral to maternal- foetal medicine specialist for high-risk pregnancy.

Immunology

If immunodeficiency is suspected eg: increased sinus / lung infections:

Serum immunoglobulin levels (total and fractions)

Flow cytometry for peripheral blood leukocytes including lymphocyte subsets

Consider evaluating childhood vaccine antibody titres

T cell immune deficiency may be linked to development of SCC

Neurology and opthalmology

MRI assessment for cerebellar hypoplasia in individuals with developmental delay or learning problems.

Annual examination to detect/correct vision problems.

Check for abnormally growing eyelashes, lacrimal duct stenosis, retinal changes, bleeding, cataracts, and glaucoma.