TELONET

DCACTION

DCAction.org
Supporting people living with Telomere Biology Disorders

Recommended surveillance for adults with Telomere Biology Disorders

General recommendations for patients	Use sunscreen regularly and avoid excessive sun exposure Avoid exposure to cigarette smoke, vaping and alcohol Seek specialist clinic guidance on how to perform a monthly self-examination for oral, head and neck cancer Maintain good oral hygiene Take vitamin D and calcium as needed to optimise bone health
Basic Surveillance	
Haematology	Baseline FBC with differential and reticulocyte count BM aspiration and biopsy Conventional and molecular cytogenetics Consider NGS myeloid panel to assess for somatic variants/ clones Monitoring FBC normal / no cytogenetic abnormality: FBC every 6-12 months BM evaluation if cytopenia develops Mild cytopaenia / no cytogenetic abnormality: FBC every 3-4 months BM evaluation based on clinical development, consider regular intervals (eg, every 1-3 years Abnormal cytogenetics: Clonal cytogenetic abnormalities require more frequent FBC/BM evaluation to evaluate potential leukaemic or MDS progression; intervals depend on development of FBC counts High-risk abnormalities such as chromosome 7 change need immediate referral 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
Respiratory	Baseline Pulmonary function test Consider HRCT based on results and risk factors Bubble echocardiogram to check for AV malformations Monitoring Annual pulmonary function test HRCT as clinically indicated Bubble echocardiogram for pulmonary symptoms in the absence of pulmonary fibrosis
Dermatology	Baseline and monitoring Perform regular skin self-examination for new or changing skin growth Annual dermatologist evaluation. Regular dermatologic advisable starting before the age of 30 years Cutaneous squamous cell carcinomas have been described in young adults with TBDs
ENT	Baseline and monitoring Annual cancer screening by an Ear Nose and Throat specialist
Dental	Baseline and monitoring Dental hygiene and oral screening every 6 months
Gastroenterology/ hepatology	Baseline Evaluate for risk factors for hepatic disease (alcohol, drug use, viruses) Liver function tests Liver ultrasound and/or 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 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
Gynaecology/ obstetrics	Baseline and monitoring: Annual gynaecological evaluation with HPV testing starting at 18 years of age or at start of sexual activity HPV vaccination in females and males <27 years of age if not adequately vaccinated in childhood Leukoplakia / VIN / CIN assessment Pre-pregnancy- consider referral for counselling re genetics or preimplantation embryo selection Pregnancy: referral to maternal- foetal medicine specialist for high-risk pregnancy
Orthopaedics	Bone density scan at baseline Younger TBD patients have an increased risk of avascular osteonecrosis and unexplained fractures A baseline bone density scan is advisable even in young adults
Genetics	Genetic screening for family members, counselling and preconception planning. See also haematology section
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
Additional surveillance based on individual clinical presentation	
Cardiology	Regular assessment for hypertension Baseline lipid level
Neurology	MRI assessment for cerebellar hypoplasia in individuals with developmental delay or learning problems

Immunology ‡	**Suspected immunodeficiency - 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**
Ophthalmology *	**Annual examination to detect/correct vision problems** **Check for abnormally growing eyelashes, lacrimal duct stenosis, retinal changes, bleeding, cataracts, and glaucoma**
Colour coding	**Patient self-care with help of practice nurse / community pharmacist where necessary** **GP or community dentist/ optometrist/nurse-led screening services etc. where results can be electronically shared with specialist clinic Specialist care required if concerns/abnormalities** **Can be monitored by specialist clinic unless abnormal** **Expert specialist care required**

Notes

‡ Immunodeficiency (commonly with lymphopaenia) and developmental delay (often cerebellar hypoplasia) is predominantly observed in TBD cases with onset in early childhood, specifically Hoyeraal-Hreidarsson syndrome.

* Ophthalmologic manifestations are frequent in childhood- onset TBDs, including classic Dyskeratosis congenita, Hoyeraal-Hreidarsson syndrome, Revesz syndrome or Coats plus.

BM – Bone marrow MDS – Myelodysplasia, FBC – Full blood count, HPV – Human Papilloma Virus, NGS - Next Generation Sequencing, ENT – Ear Nose and Throat, HRCT – High Resolution CT Scan, AV Malformation – Arteriovenous malformations, CIN - Cervical Intraepithelial Neoplasia, VIN - Vulvar Intraepithelial Neoplasia

Surveillance recommendations are adapted from:
Niewisch MR, Beier F, Savage SA. Clinical manifestations of telomere biology disorders in adults. Hematology Am Soc Hematol Educ Program. 2023 Dec 8;2023(1):563-572. doi: 10.1182/hematology.2023000490. PMID: 38066848; PMCID: PMC10726987

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