Chapter 27

Routine Healthcare for Adults with Telomere Biology Disorders

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Introduction

Health care maintenance for people with telomere biology disorders (TBDs) can be complex due to the multisystem nature of these illnesses. This chapter summarizes current guidance based on expert opinion and experience of the authors.
Hematology

Hematologic abnormalities are very common in individuals with TBDs, from isolated (asymptomatic) macrocytosis or thrombocytopenia to severe aplastic anemia and myeloid neoplasia (see Chapter 10, Medical Management of Bone Marrow Failure in Telomere Biology Disorders). When an adult individual is diagnosed with a TBD, hematopoiesis should be assessed to identify marrow failure and/or malignant transformation:

- Complete blood count (CBC) with differential and reticulocyte count
- Bone marrow aspiration
- Bone marrow biopsy
- Flow cytometry of bone marrow mononuclear cells (BMMCs)
- Conventional and molecular cytogenetics
- When available, Next Generation Sequencing (NGS) myeloid panel to assess somatic variants/clones

If evaluation provides evidence of aplastic anemia or myeloid malignancy (excess blasts, abnormal cytogenetics, marrow clonality), the patient should be referred to a hematologist/specialist to consider specific therapies (androgen, hematopoietic cell transplant, etc.).

If the CBC is normal or demonstrates mild cytopenias/abnormalities (e.g., macrocytosis, platelets >100/μL), blood and marrow may be monitored annually. However, if cell counts are falling, blood and marrow should be monitored more frequently.

Immunology

Immune dysregulation is often seen in patients with inherited or acquired marrow failure, including those with telomere-biology disorders. Thus, adult individuals
diagnosed with telomere diseases should be evaluated at presentation for cellular and humoral immunologic parameters:

- Flow cytometry for peripheral blood leukocytes including lymphocyte subsets (T CD4+, T CD8+, B cells, NK cells);
- Serum immunoglobulin levels (total and fractions, IgG, IgA, IgM);
- Depending on the patient’s history, determine serum levels of tetanus/diphtheria/poliomyelitis/pneumococcal antibodies.

When immunodeficiency is detected, the patient should be referred to an immunologist.

**Dermatology**

Because of the increased risk of skin cancer, prevention strategies are highly recommended for individuals with TBDs/Dyskeratosis congenita (DC) (see also Chapter 6, Dermatologic Manifestations). Recommended strategies include:

- Regular use of sunscreen or sunblock when outdoors, and use of a daily moisturizing lotion with sunblock
- Wear hats and sun-protective clothing when outdoors to prevent excessive sun exposure
- Limit outdoor time during hours of peak sun exposure (between 10am and 4pm)
- Be mindful of reflected sun from water and snow when engaging in outdoor activities
- Avoid tanning beds
- Perform regular skin self-examinations to look for new or changing skin growths

In addition, an annual full body skin examination by a dermatologist is recommended.
**Ophthalmology**

Ophthalmic abnormalities are common and variable in children with DC/TBDs [1] and more severe forms, including Revesz syndrome and Coats plus syndrome [2], but are less frequent among adults with TBDs (see also Chapter 7, Ophthalmologic Complications) [3]. When the diagnosis of a telomere-biology disorder is confirmed in an adult individual, careful ophthalmic examination should be performed covering anterior and posterior segments and adnexa.

In addition to visual acuity, examination should address corneal changes, nasolacrimal duct obstruction, trichiasis in the adnexa; corneal lesions in the anterior segment; and cataracts, and retinal changes (vasculopathy, exudate, telangiectasias) in the posterior segment [2]. Unusual cases of exudative retinopathy may present late at adulthood [4]. If changes are observed, careful evaluation and follow-up with an ophthalmologist is warranted.

**Dental**

Individuals with TBDs are at increased risk of oral head and neck squamous cell carcinoma (see also Chapter 8, Dental and Oral Complications and Chapter 9, Solid Tumors). Adults with TBDs are advised to establish routine care and annual visits with an otolaryngologist (ear, nose, and throat physician) to screen for oral cancers. Monthly oral self-examination is recommended and can be done by the patient or a family member after ENT education. Patients should have a low threshold for evaluation if they note oral changes lasting more than two or three weeks.

Some patients with TBDs have dental anomalies, including short roots and widened pulp chambers. General hygiene recommendations include brushing teeth two to three times a day with fluoridated toothpaste and flossing once a day at a minimum to help prevent tooth decay. Some dentists recommend using prescription strength fluoride toothpaste or antibacterial mouth rinse to aid in reducing oral disease. Biannual dental
checkups and cleanings are recommended to monitor for the presence of oral pathology and prevent the development of significant dental decay and gum disease. Precautions during routine dental treatment may be necessary in the presence of low platelet counts and white blood cell levels.

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**Pulmonary**

Large, prospective studies of pulmonary screening for asymptomatic individuals with TBDs have not yet been conducted (see also Chapter 14, Pulmonary Fibrosis). Figure 1 provides an algorithm for consideration of screening and routine follow up (as carried out at Mayo Clinic). Please note that these recommendations vary from institution to institution as there is lack of prospective evidence on which to base these screening recommendations. Several pulmonary screening methodologies are available including high-resolution chest computed tomography (HRCT), chest x-ray, and pulmonary function tests (PFTs). The two principal decision points in asymptomatic patients are long-term safety, primarily related to radiation exposure (measured in millisieverts [mSV]), versus sensitivity and specificity of the testing modality to identify interstitial lung disease (ILD). To put it into perspective, the average annual exposure to radiation in the environment is 3 mSV, compared to 7-15 mSV with chest CT scans and 0.02-0.1 mSV with chest X-rays [5] (see also Chapter 23, Radiation and Telomere Biology Disorders).
Figure 1: Pulmonary function assessment in telomere biology disorders. These recommendations should be tailored for each specific patient in consultation with their medical team.*

*These are the recommendations of the Mayo Clinic team and the authors of this chapter. Other approaches may vary. Each patient is strongly encouraged to work closely with their medical team to develop the best approach for them.

CT = computed tomography
PAVM = pulmonary arteriovenous malformation
PFTs = pulmonary function tests
ILD = interstitial lung disease

The sensitivity and specificity of each modality is variable and relative to the diagnostic value of the test. While PFTs are noninvasive and relatively accessible, they lack the sensitivity and specificity to identify early stages of ILD. A new ILD-Screen tool using age, height, total lung capacity, FEV1, diffusion capacity, and PFT indication demonstrated a sensitivity of 79% and specificity of 83% when validated with more
precise imaging [6]. Chest imaging is considered more sensitive but varies depending
upon the technique used. While HRCTs have the best ability to identify early changes of
ILD, they are associated with significant radiation exposure, as noted earlier; thus,
questioning the long-term safety of repeat usage. They, however, do provide the best
likelihood of identifying ILD with a sensitivity of 95% and specificity approaching 100%
[7]. Chest x-rays on the other hand do not have the same sensitivity (80%) or specificity
(82%) and diagnostic confidence is low (23%) [7].

Pulmonary function tests should include measures of FEV₁, FVC, FEV₁/FVC ratios and
DLCO estimates. We also obtain inspiratory and expiratory flow loops along with
bronchodilator responsiveness, especially if the patient endorses hyperreactive airway
disease like symptoms. Oximetry studies including the 6-minute walk test are carried
out in patients with established ILD.

At Mayo Clinic, based on the age of the patient, smoking history (primary or secondary
exposure), personal history of lung disease (e.g., asthma) and family history of
pulmonary involvement, we consider performing a baseline HRCT with inspiratory and
expiratory views. If there is no ILD, we follow patients clinically with PFTs and chest
x-rays annually. We have a low threshold to obtain a HRCT in the event of decline in
PFTs, cardiopulmonary symptoms or radiological abnormalities on the chest x-ray. In
asymptomatic patients without exposure history or family history, a risk versus benefit
discussion is warranted to evaluate patient’s desire for more aggressive baseline
assessment (HRCT) versus less sensitive or specific testing (PFTs and chest X-ray)
(Figure 1).

If pulmonary findings are present, we refer our patients to an ILD Clinic to meet with
Pulmonary experts and decide whether a bronchoscopy with transbronchial biopsies or
open lung biopsy are needed for histopathological confirmation, or to rule out
alternative etiologies. Occasionally, a bronchoscopy with bronchoalveolar lavage using
an immunocompromised host protocol is needed to rule out atypical infections that
could mimic ILD. Tissue biopsies are often not helpful if a strong diagnosis of a
Individuals with TBDs can have pulmonary arteriovenous malformations (PAVMs) that can often mimic clinical features of ILD (see also Chapter 16, Vascular Complications). They may also be present in the absence of lung parenchymal involvement and should be suspected if there is a progressive decline in the DLCO without any change in the FEV₁ or FEV₁/FVC ratio [8]. Multiple diagnostic options exist though the most sensitive and specific is a transthoracic contrast echocardiogram (TTCE) or bubble echocardiography [9]. Injection of agitated saline while imaging the right and left ventricles allows visualization of microbubbles in the left ventricle that are not filtered out by the pulmonary vasculature suggest right-to-left shunting. An alternative and often less readily accessible evaluation may include a radioisotope (e.g. technetium-99m) ventilation/perfusion lung scan.

All individuals with TBDs should be informed of the pulmonary exposure risk with inhalants through recreational use and secondary exposure including nicotine (cigarettes, cigars, vaping) and inhaled marijuana. Counseling on cessation and referral to dependency centers for assistance with interventional and behavioral modifications is critical.

Cardiology

Little is definitively known on the association between TBDs and atherosclerotic cardiovascular disease (ASCVD) and associated coronary heart disease (CHD), myocardial infarction, angina pectoris, heart failure, stroke, transient ischemic attack, and peripheral artery disease. However, atherosclerosis has been demonstrated by senescence of vascular smooth muscle and inhibition of telomerase activity with marked telomere shortening, suggesting a potential for increased risk [10]. Additionally, observational studies of people without DC/TBDs suggest an association of short
telomeres with CVD and cardiovascular mortality including a 3-fold higher risk of MI and stroke [11].

Baseline assessment at diagnosis should include evaluation of blood pressure and blood cholesterol. Management of hypertension and hyperlipidemia should be in accordance with nationally recognized comprehensive care guidelines, including risk calculation and use of primary prevention where warranted [12, 13]. Androgen therapy, including danazol, increases the risk for hypertension, elevated LDL, and reduced HDL levels. A baseline lipid panel should be obtained prior to androgen treatment initiation and every 6 months while on therapy. Initiation of antihypertensives and lipid lowering therapy may be required though drug-drug interactions should be considered [14, 15].

Additionally, there is no clear data on the risk of congestive heart failure (CHF) in patients with TBDs. Data from individuals without TBDs suggest that cardiac myocyte telomere length is shortened in hypertrophic hearts, independent of age [16]. A baseline echocardiogram is not routinely recommended unless presenting symptoms warrant further investigation. Cardiopulmonary symptoms may manifest secondary to PAVMs as noted earlier and should be evaluated at baseline and throughout disease management when present.

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**Gastroenterology/Hepatology**

**Gastroenterology**

Baseline evaluations should include physical assessment for the primary GI manifestations of TBDs, including esophageal stenosis, a celiac-like enteropathy, and B-cell immunodeficiency with enterocolitis (see also Chapter 17, Gastrointestinal Disease) [17]. Only if symptoms of esophageal stenosis are present (difficulty swallowing, intolerance of solids), should evaluation move forward with a video contrast swallow study (esophagram) or esophagogastroduodenoscopy (EGD). If stenosis is
present, intermittent balloon dilation and rarely stenting can alleviate symptoms. In the absence of symptoms, no routine evaluation is warranted.

Small intestine celiac-like enteropathy and immunodeficiency-mediated enterocolitis require exclusionary work up of other etiologies including infection or malignancies. Evaluation should include a celiac disease cascade; assessment of immunoglobulin levels; and quantification of T-cell, B-cell, and NK-cell subsets. Recommended diagnostic procedures include EGD and/or colonoscopy with biopsies [17].

Hepatology

The most common hepatic manifestations of TBDs include cryptogenic cirrhosis and nodular regenerative hyperplasia leading to non-cirrhotic portal hypertension (see also Chapter 18, Hepatic Complications) [18]. Baseline assessment of all patients include physical exam, family history, social history (alcohol and drug use), body mass index (BMI) and liver function test (Figure 2). In patients who are asymptomatic, lack family or social history concerns, have a normal BMI and liver function tests (LFTs), a Fibroscan (transient hepatic elastography) can be done at baseline and repeated every 2 years. If screening is positive, we prefer proceeding with magnetic resonance elastography (MRE) for evaluation of liver and spleen stiffness and anatomy. In patients with TBDs, the use of liver biopsy should be done judiciously, particularly if bone marrow failure and risk for infection and hemorrhagic complications are present.
**Figure 2: Hepatic function assessment in telomere biology disorders.** These recommendations should be tailored for each specific patient in consultation with their medical team.

BMI = Body mass index
INR = International normalized ratio
PFTs = pulmonary function tests
EGD = esophagogastroduodenoscopy

If baseline assessments are negative, patients may revert to monitoring with Fibroscan every 2 years. If baseline assessment is concerning for fibrosis, we refer to Hepatology for consideration of EGD, evaluation and management of portal hypertension and assessments for hepatopulmonary syndrome. At times a liver biopsy may be necessary if there is clinical suspicion of portal hypertension with preserved synthetic liver function, absence of other causes for chronic liver disease and no risk factors for alcohol or non-alcoholic liver disease [18]. Meta-analysis of MRE as an imaging modality for liver fibrosis has demonstrated a sensitivity of 73% and a specificity 79% [19].
Patients on androgen therapy, such as danazol, should have their liver function tests evaluated pre-treatment and every 3-6 months while on therapy (or sooner if indicated).

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**Gynecologic Health**

As described in detail in Chapter 20, Genitourinary Complications and Chapter 21, Gynecologic and Obstetric Considerations, females with TBDs may have specific gynecologic and genitourinary manifestations requiring close follow-up with a gynecologist. Annual routine gynecologic care is recommended, including cervical cancer screening with human papilloma viral (HPV) testing, beginning when they become sexually active or at age 21 years, whichever is first.

HPV vaccination is recommended for females and males between ages 9-27 years and is FDA approved up to age 45.

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**Endocrinology**

There is no specific association with endocrinopathies and TBDs except for osteopenia and osteoporosis. We recommend comprehensive baseline screening of endocrinopathies including bone health with bone mineral density, 25-hydroxyvitamin-D total, thyroid hormone cascade, and diabetes mellitus assessment with fasting blood glucose and hemoglobin A1c if clinically appropriate. These tests are repeated annually or more frequently if clinically indicated.

In individuals of reproductive age, there is limited data on pregnancy outcomes. A recent cohort demonstrates an increased risk of progressive cytopenias, preterm and cesarean deliveries, and recurrent pregnancy loss in women with autosomal dominant inheritance [20]. Referral to a multidisciplinary center experienced in management of patients with hematologic conditions is recommended.
Orthopedics

Osteopenia and osteoporosis are not uncommon in individuals with TBDs (see also Chapter 22, Endocrine and Skeletal Disorders) [21]. Some data suggest that telomere shortening may induce osteoblast abnormalities associated with osteoporosis [22]. Thus, an adult individual with TBDs should have a bone density scan at baseline, and monitoring will depend on the findings. Vitamin D and calcium supplementation may be considered depending on findings.

Table 1. Screening recommendations for adult patients with Telomere Biology Disorders (TBD) by speciality.

<table>
<thead>
<tr>
<th>Specialty</th>
<th>Evaluation</th>
<th>Recommendation</th>
</tr>
</thead>
</table>
| Cardiology  | Hypertension | • Baseline blood pressure  
• Repeat vital signs with routine visits  
• Manage per nationally recognized guidelines  

*Patients on androgens (e.g. danazol) may warrant more frequent monitoring and treatment* |

<table>
<thead>
<tr>
<th>Specialty</th>
<th>Evaluation</th>
<th>Recommendation</th>
</tr>
</thead>
</table>
| Pulmonology | Pulmonary fibrosis | • Assessment of baseline risk  
(see Figure 1)  
• Testing may include:  
  o Pulmonary function tests (PFTs)  
  o High-resolution chest CT with inspiratory & expiratory views  
  o Chest X-ray  
  o Early referral for shortness of breath or unexplained cough  
  o Nicotine dependency & secondary exposure counseling |
<table>
<thead>
<tr>
<th>Specialty</th>
<th>Condition</th>
<th>Procedures/Tests</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pulmonary</td>
<td>Arteriovenous malformation</td>
<td>Transthoracic echocardiogram with contrast (bubble echocardiography)</td>
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<td></td>
<td></td>
<td>Nuclear medicine scan</td>
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<tr>
<td>Gastroenterology</td>
<td>Esophageal stenosis</td>
<td>Esophagogram and/or upper endoscopy (esophagogastroduodenoscopy, EGD), if symptom</td>
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<tr>
<td></td>
<td></td>
<td>Medical management with balloon dilation and rarely stenting</td>
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<tr>
<td></td>
<td>Enteropathy or Enterocolitis</td>
<td>Imaging studies, EGD/colonoscopy with biopsies, and evaluation for infection and malignancy</td>
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<tr>
<td>Hepatology</td>
<td>Cirrhosis</td>
<td>Assessment of baseline risk (see Figure 2)</td>
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<tr>
<td></td>
<td></td>
<td>Annual liver function tests including total/direct bilirubin, AST, ALT, alkaline phosphatase, albumin, PT, aPTT, and INR</td>
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<td></td>
<td>Baseline transient elastography (Fibroscan ultrasound)</td>
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<td>Magnetic resonance (MR) elastography if risk warrants</td>
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<td></td>
<td></td>
<td>Referral to Hepatology for variceal assessment and liver biopsy as clinically indicated for concern of non-cirrhotic portal hypertension</td>
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<tr>
<td></td>
<td></td>
<td>*Patients on androgens may warrant more frequent monitoring and treatment</td>
</tr>
<tr>
<td>Hematology</td>
<td>Cytopenias; Aplastic anemia; Myeloid neoplasia</td>
<td>Complete blood counts, reticulocyte counts and white blood cell differential at baseline</td>
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<tr>
<td></td>
<td></td>
<td>Bone marrow assessment at baseline: bone marrow aspiration and biopsy, flow cytometry, conventional and molecular cytogenetics, and myeloid panel by NGS for somatic variants</td>
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<tr>
<td>Immunology</td>
<td>Immunodeficiency</td>
<td>Flow cytometry of peripheral blood lymphocyte subsets</td>
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<td></td>
<td></td>
<td>Serum immunoglobulin levels (total and fractions)</td>
</tr>
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</table>
### Ophthalmology

<table>
<thead>
<tr>
<th>Segment</th>
<th>Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anterior segment</td>
<td>• Corneal assessment</td>
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<tr>
<td>Posterior segment</td>
<td>• Retinal evaluation</td>
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<tr>
<td>Adnexa</td>
<td>• Nasolacrimal duct, eyelid assessment</td>
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</table>

### Orthopedics

- **Osteoporosis**
  - Bone density scan at baseline and then as needed

### Table 2: Cancer Screening Considerations†

<table>
<thead>
<tr>
<th>Cancer Screening</th>
<th>American Cancer Society</th>
<th>United States Preventive Services Task Force</th>
<th>National Comprehensive Cancer Network</th>
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<tbody>
<tr>
<td><strong>Breast Cancer</strong></td>
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<tr>
<td></td>
<td><strong>Average</strong> risk women</td>
<td>Biennial screening 50 to 74 Y</td>
<td><strong>Average</strong> risk women 40 Y and older, annual screening mammogram</td>
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<tr>
<td></td>
<td>screening mammogram:</td>
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<td>Risk assessment by 25 and counseling on benefits, risks, and limitations of screening.</td>
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<tr>
<td></td>
<td>· 40 to 44 Y optional</td>
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<td>Clinical breast exams with provider visits.</td>
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<td></td>
<td>to start annual</td>
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<td>Individuals should be familiar with their breasts and promptly report changes to their health care provider.</td>
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<tr>
<td></td>
<td>screening</td>
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<td>· 45 to 54 Y annual</td>
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<td>screening</td>
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<td>· 55 Y and older every</td>
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<td>other year OR yearly</td>
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<td>as long as health in</td>
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<td>good standing and</td>
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<td>live expectancy &gt; 10</td>
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<td></td>
<td>years</td>
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<td>Women should be familiar with how their breasts normally look and feel and should report any changes to a health care provider right away.</td>
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<tr>
<td><strong>Cervical Cancer</strong></td>
<td>Initiate screening at 25 Y</td>
<td>Initiate screening at 21 Y</td>
<td>None</td>
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<td></td>
<td>· HPV testing every 5</td>
<td>Cytology along 21 to 29 Y</td>
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<td></td>
<td>Y until 65 Y (preferred)</td>
<td>Age 30 to 65 Y:</td>
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<tr>
<td></td>
<td>· Cytology + HPV</td>
<td>· HPV testing every 5 Y</td>
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<tr>
<td></td>
<td>testing every 5 years</td>
<td>· Cytology + HPV testing every 5 years</td>
<td></td>
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<tr>
<td></td>
<td>· Cytology alone</td>
<td>· Cytology alone every 3 years</td>
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<tr>
<td></td>
<td>every 3 years</td>
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<tr>
<td><strong>Colon and Rectal Cancer</strong></td>
<td><strong>Average</strong> risk people start screening at 45 Y with a stool-based test or visual exam. Screening until 75 Y; The decision to screen between 76 and 85 Y should be individualized. Over 85 Y no screening required.</td>
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</tbody>
</table>
| **Test examples:**       | · Highly sensitive fecal immunochemical test every year  
  · Highly sensitive guaiac-based fecal occult blood test every year  
  · Multi-targeted stool DNA test every 3 years  
  · Colonoscopy every 10 years  
  · CT colonography every 5 years  
  · Flexible sigmoidoscopy every 5 years |
| **Recommend** age 50 to 75 Y (Grade A); Recommend 45 to 49 Y (Grade B); Recommend 76 to 85 Y, based on patient's overall health, prior screening, and preference (Grade C) |
| **Test examples:**       | · High-sensitivity guaiac fecal occult blood test or fecal immunochemical (FIT) test every year  
  · Stool DNA-FIT every 1 to 3 years  
  · Computed tomography colonography every 5 years  
  · Flexible sigmoidoscopy every 5 years  
  · Flexible sigmoidoscopy every 10 years + annual FIT  
  · Colonoscopy screening every 10 years |
| **Average** risk people start screening at 45 Y with a stool-based test or visual exam. Screening until 75 Y; The decision to screen between 76 and 85 Y should be individualized. |
| **Test examples:**       | · Highly sensitive fecal immunochemical test (FIT) every year  
  · Highly sensitive guaiac-based fecal occult blood test every year  
  · Multi-targeted stool DNA test every 3 years  
  · Colonoscopy every 10 years  
  · CT colonography every 5 years  
  · Flexible sigmoidoscopy every 5-10 years |

<table>
<thead>
<tr>
<th><strong>Lung Cancer</strong></th>
<th>Currently undergoing revision; recommend USPSTF guidelines</th>
</tr>
</thead>
</table>
| **Low dose CT screening in adults 50 Y to 80 Y with a 20 pack-year smoking history and currently smoking OR have quit in the past 15 years.**  
Screening may be discontinued once a person has not smoked for 15 years, develops a life limiting illness, or is no longer willing to undergo lung surgery. |
| **Low dose CT screening in adults ³ 50 Y with a 20 pack-year smoking history** |
Prostate Cancer
Average risk, begin at 50 Y if expected to live at least 10 more years. Screening modality: prostate specific antigen (PSA) blood test; digital rectal exam, optional (DRE).
- If PSA < 2.5 ng/mL may only need to be retested every 2 years.
- If PSA 2.5 ng/mL or higher, screen yearly.

Age 55 Y to 69 Y, individual decision in concert with provider.
Screening modality: prostate specific antigen (PSA) blood test;
digital rectal exam, optional (DRE).
- If PSA < 1 ng/mL repeat testing at 2 to 4 year intervals.
- If PSA 1-3 ng/mL, repeat testing at 1 to 2 year intervals.

Average risk, begin at 45 Y
Screening modality: prostate specific antigen (PSA) blood test; digital rectal exam, optional (DRE).

‡: This table is reflective of select guidelines and statements from those guidelines have been selected to articulate standard situations. It is not meant to reflect the guidelines in their entirety and nor do they reflect all guideline options. Clinicians are encouraged to review these documents in their entirety when making clinical decisions for patients with TBDs/DC.

Table 3: Cardiovascular Disease Screening†

<table>
<thead>
<tr>
<th>United States Preventive Services Task Force</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Hypertension</strong></td>
</tr>
<tr>
<td>Begin screening at 18 Y with office blood pressure measurement. Confirm with home measurement before starting treatment. Screen every 3-5 years for adults 18-39 Y who are not at increased risk and have a documented normal blood pressure. Screen annually in adults &gt; 40 Y and adults with increased risk.</td>
</tr>
</tbody>
</table>

| **Diabetes mellitus type 2**               |
| Offer screening to adults 35 Y to 70 Y who are overweight or obese |

| **Hyperlipidemia**                        |
| Screen initially at age 20 for familial disorders; Resume screening no later than 40 years of age. Screen approximately every 5 years or as clinically indicated |

†: This table is reflective of United States Preventive Services Task Force recommendations. It is not meant to reflect the recommendations in their entirety and nor do they reflect all available consensus guidelines. Clinicians are encouraged to review any guideline in its entirety when making clinical decisions for patients with TBDs/DC.
### Table 4: Vaccination

<table>
<thead>
<tr>
<th></th>
<th>Ages 21 to 39 years</th>
<th>Ages 40 to 49 years</th>
<th>Ages 50 to 64 years</th>
<th>Age ≥ 65 years</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>COVID-19</strong></td>
<td>Recommended for everyone, follow schedule and doses per vaccine brand and current CDC guidance.</td>
<td></td>
<td></td>
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</tr>
<tr>
<td><strong>Influenza</strong></td>
<td>Recommended annually for everyone, inactivated or recombinant vaccine recommended for immunocompromised.</td>
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</tr>
<tr>
<td><strong>Pneumococcus (PCV13 and PPSV23)</strong></td>
<td><strong>Immunocompromised:</strong> 1 dose PCV13 followed by 1 dose PPSV23 at least 8 weeks later, then another dose PPSV23 at least 5 years after previous PPSV23; at age 65 years or older, administer 1 dose PPSV23 at least 5 years after most recent PPSV23</td>
<td><strong>Immunocompetent:</strong> PPSV23 1-time dose. May consider PCV13 in shared decision making effort.</td>
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<tr>
<td><strong>Tetanus diphtheria acellular pertussis (Tdap and Td)</strong></td>
<td>1-time dose of Tdap, then boost with Tdap or Td every 10 years</td>
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<td><strong>Human papilloma virus (HPV)</strong></td>
<td><strong>Immunocompetent:</strong> Initiate series for adults up through age 26 years, consider series initiation for ages 27 through 45 based on shared decision making <strong>Immunocompromised:</strong> Initiate series for adults up to age 45 years.</td>
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<tr>
<td><strong>Herpes zoster or shingles (HZV)</strong></td>
<td>2 doses of Shingrix (regardless of previous Zostavax) at age ≥ 50 years unless contraindicated</td>
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<tr>
<td><strong>Meningococcal Serogroup B Meningococcal conjugate</strong></td>
<td><strong>Immunocompetent:</strong> Meningococcal conjugate vaccine (MenACWY) single dose + Meningococcal vaccine serogroup B (MenB) 2 or 3-dose series based on shared decision making. <strong>Immunocompromised:</strong> Meningococcal conjugate vaccine (MenACWY) 2-dose series, revaccinate every 5 years + Meningococcal vaccine serogroup B (MenB) 2 or 3-dose series every 2 to 3 year, based on risk re-evaluation.</td>
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</table>

*: This table is an abbreviation of the CDC recommendations for vaccinations. Guidelines are subject to change and update. Please refer to the full guidelines noted below for details and the most up to date guidance on vaccinations.
Guidelines
United States Preventive Services Task Force Recommendations:
https://www.uspreventiveservicestaskforce.org/uspstf/recommendation-topics

American Cancer Society Guidelines:

National Comprehensive Cancer Network Guidelines:
https://www.nccn.org/guidelines/category_2

Center for Disease Control and Prevention Vaccinations Schedules:
https://www.cdc.gov/vaccines/schedules/index.html

References


