Surveillance and management recommendations for newly diagnosed or suspected tuberous sclerosis complex (TSC)

Organ System or Specialty Area	Recommendation
Genetics	• Obtain three-generation family history to assess for additional family members at risk of TSC • Offer genetic testing for family counseling or when TSC diagnosis is in question but cannot be clinically confirmed
Brain	• Perform magnetic resonance imaging (MRI) of the brain to assess for the presence of tubers, subependymal nodules (SEN), migrational defects, and subependymal giant cell astrocytoma (SEGA) • Evaluate for TSC-associated neuropsychiatric disorder (TAND) • During infancy, educate parents to recognize infantile spasms, even if none have occurred at time of first diagnosis • Obtain baseline routine electroencephalogram (EEG). If abnormal, especially if features of TAND are also present, follow-up with a 24-hr video EEG to assess for subclinical seizure activity
Kidney	• Obtain MRI of the abdomen to assess for the presence of angiomyolipoma and renal cysts • Screen for hypertension by obtaining an accurate blood pressure • Evaluate renal function by determination of glomerular filtration rate (GFR)
Lung	• Perform baseline pulmonary function testing (pulmonary function testing and 6-minute walk test) and high-resolution chest computed tomography (HRCT), even if asymptomatic, in patients at risk of developing lymphangioleiomyomatosis (LAM), typically females 18 years or older. Adult males, if symptomatic, should also undergo testing • Provide counsel on smoking risks and estrogen use in adolescent and adult females
Skin	• Perform a detailed clinical dermatologic inspection/exam
Teeth	• Perform a detailed clinical dental inspection/exam
Heart	• Consider fetal echocardiography to detect individuals with high risk of heart failure after delivery when rhabdomyomas are identified via prenatal ultrasound • Obtain an echocardiogram in pediatric patients, especially if younger than 3 yr of age • Obtain an electrocardiogram (ECG) in all ages to assess for underlying conduction defects
Eye	• Perform a complete ophthalmologic evaluation, including dilated funduscopy, to assess for retinal lesions and visual field deficits