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For Immediate Release:

Delayed recognition of tuberous sclerosis complex in adult women has life-threatening consequences

Bethesda, Md. – Women with tuberous sclerosis complex (TSC) are often misdiagnosed because the condition, commonly recognized in early childhood when it presents with seizures, manifests differently later in life, typically with renal angiomyolipomas – a benign tumor of the kidney – and pulmonary lymphangiomyomatosis (LAM) – a rare lung disease that affects almost exclusively women. This diagnostic delay places women with TSC at increased risk for morbidity and mortality. TSC is a genetic disease associated with tumor development in the brain, retina, kidney, skin, heart, and lung.

The findings, released online June 20, 2011 in the *Annals of Internal Medicine*, suggest that education of primary care providers of the warning signs early could speed early treatment and improve outcomes.

According to Diane Seibert, Ph.D., CRNP, associate professor, Uniformed Services University of the Health Sciences (USU) Graduate School of Nursing, the lead author, “We noticed that some patients had symptoms or skin lesions for years before someone finally ‘figured’ out their diagnosis. Others did not appear to have enough manifestations to receive a TSC diagnosis until adulthood. Clinicians need to know more about TSC so that they can recognize the disease earlier in life.”

The study looked at 45 patients who received a diagnosis of TSC in adulthood, 21 of whom had symptoms of LAM, 19 who had renal angiomyolipomas and 10 who had a history of seizures. Of these 45 patients, 30 met clinical criteria for TSC in childhood that remained undiagnosed for a median of 21.5 years. Another 15 were older than 18 years before they met the clinical criteria for a TSC diagnosis. Patients diagnosed in adulthood and those diagnosed in childhood had similar occurrences of pneumothorax, shortness of breath, hemoptysis, nephrectomy and death.

This study found that women who received a TSC diagnosis in adulthood had minimal morbidity during childhood, but were still at risk for life-threatening pulmonary and renal manifestations.

“When people think of patients with TSC they tend to think about a child with cognitive dysfunction, skin lesions and seizures,” said Thomas N. Darling, Ph.D., associate professor of Dermatology, USU, the senior author. “However, some patients with TSC have minimal signs or symptoms in childhood and present later in life. We hope that our paper will increase awareness about this subgroup of patients, resulting in earlier recognition and diagnosis along with better outcomes.”

This retrospective study was conducted at the National Institutes of Health Clinical Center in Bethesda, Md.

The Uniformed Services University of the Health Sciences, or USU, is the nation’s federal health sciences university. USU students are primarily active-duty uniformed officers in the Army, Navy, Air Force and Public Health Service who are being educated to deal with wartime casualties, emerging infectious diseases and other public health emergencies. Of the university’s more than 4,500 physician and more than 900 advanced practice nursing alumni, the vast majority are supporting operations in Iraq, Afghanistan and elsewhere, offering their leadership and expertise. For more information, visit www.usuhs.mil.