Tuberous Sclerosis Complex (TSC) is a Multiorgan Genetic Disorder

- It is characterized by the formation of hamartomas, which are noncancerous tumor-like masses.
- These tumors can form in major organs including the brain, skin, eyes, and kidneys. Tumors in the heart often occur in children, while lung tumors can occur in adults.
- Symptoms of TSC can range from mild to severe and can change over time.
  - TSC may not be noticeable. Because symptoms vary and may not be immediately recognized by a health care provider, TSC is often undiagnosed for years.

Depending on the body organs affected by TSC, different specialists may be involved, such as:

- **Nephrologist or urologist** for kidney manifestations, such as renal angiomyolipoma
- **Neurologist** for brain manifestations, such as subependymal nodules (SENs) and subependymal giant-cell astrocytomas (SEGAs)
- **Dermatologist** for skin manifestations
- **Pulmonologist** for lung complications, such as lymphangioleiomyomatosis (LAM)
- **Psychiatrist or psychologist** for issues related to cognition, mood, and behavior disorders

**Approximately 50,000 people in the United States have TSC.**

TSC occurs in all races and ethnic groups, and in both genders.

The disease affects an estimated 1 in 6,000 newborns.

Diseases with similar US prevalence rates include cystic fibrosis (approximately 30,000 people) and amyotrophic lateral sclerosis (ALS), or Lou Gehrig’s disease (up to approximately 30,000 people).

About 1/3 of all people with TSC genetically inherit the disease, while in the remaining individuals, the disease is acquired as a result of spontaneous genetic mutation.

Tuberous Sclerosis Complex (TSC): An Overview
TSC affects multiple organs

TSC SIGNS, SYMPTOMS AND RESULTING DISORDERS

**Skin lesions 90%**
Skin lesions can appear in many different forms, including reddish raised lesions or light, oval patches of skin

**Seizures 85%**
Seizures can start in infancy and may increase in frequency and severity throughout childhood

**Kidney tumors 80%**
Kidney tumors (renal angiomyolipomas) can be difficult to manage and can be a major factor of morbidity and mortality in adult patients

**Developmental disorders ~60%**
Developmental disorders can range from mild learning disabilities to severe mental retardation

**Heart tumors up to 50%**
Heart tumors (cardiac rhabdomyomas) frequently occur in infancy and are often used to help diagnose TSC

**Autism ~50%**

**LAM up to 80% of women**
Lymphangioleiomomatosis (LAM) is a lung disease that can cause shortness of breath, occurring mostly in women and a small number of men with TSC

**SEGAs ~20%**
Subependymal giant cell astrocytomas (SEGAs) are noncancerous brain tumors that may cause potentially life-threatening brain swelling if they grow

References:

Novartis Pharmaceuticals Corporation
East Hanover, New Jersey 07936-1080

© 2017 Novartis

AGM-1165541