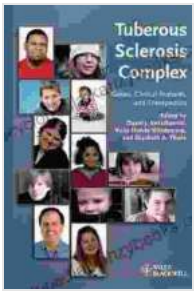
Tuberous Sclerosis Complex: An In-Depth Exploration of Genes, Clinical Features, and Therapeutic Approaches

Tuberous Sclerosis Complex (TSC) is a rare genetic disFree Download characterized by the formation of benign tumors in multiple organs,

including the brain, kidneys, heart, lungs, and skin. It is caused by mutations in either the TSC1 or TSC2 gene, which are responsible for encoding proteins involved in the regulation of cell growth and proliferation. TSC affects an estimated 1 in 6,000 individuals worldwide, with equal distribution between males and females.



Tuberous Sclerosis Complex: Genes, Clinical Features and Therapeutics

★★★★★ 5 out of 5

Language	: English
File size	: 5746 KB
Text-to-Speech	: Enabled
Screen Reader	: Supported
Enhanced typesetting	: Enabled
Print length	: 432 pages
Lending	: Enabled

Genetic Basis of TSC

TSC is an autosomal dominant disFree Download, meaning that the affected gene is located on one of the non-sex chromosomes. Mutations in either the TSC1 or TSC2 gene can cause TSC. These mutations result in the production of defective proteins, which leads to the uncontrolled growth of cells and the formation of tumors.

Approximately two-thirds of TSC cases are caused by mutations in the TSC2 gene, located on chromosome 16. The remaining one-third of cases are caused by mutations in the TSC1 gene, located on chromosome 9. Mutations in these genes lead to the disruption of the TSC1-TSC2

complex, which plays a critical role in regulating cell growth and proliferation.

Clinical Features of TSC

TSC can manifest in a wide spectrum of clinical features, depending on the severity of the mutations and the organs affected. The most common clinical features include:

- **Neurological features:** Seizures, autism spectrum disFree Download, intellectual disability, behavioral problems, developmental delays
- **Renal features:** Renal cysts, angiomyolipomas (benign kidney tumors),end-stage renal disease
- **Cardiac features:** Cardiac rhabdomyomas (benign heart tumors),arrhythmias, heart failure
- **Pulmonary features:** Lymphangioleiomyomatosis (LAM),a rare lung disease that affects women
- **Skin features:** Facial angiofibromas (small red bumps on the face),hypomelanotic macules (white spots on the skin)
- **Ocular features:** Retinal hamartomas (benign tumors in the retina),glaucoma
- **Dental features:** Enamel pits, intraoral fibromas

Diagnosis of TSC

TSC is typically diagnosed based on a combination of clinical features, physical examination findings, and genetic testing. Magnetic resonance imaging (MRI) and computed tomography (CT) scans are often used to

visualize the tumors in different organs. Genetic testing can confirm the diagnosis of TSC by identifying mutations in the TSC1 or TSC2 genes.

Treatment of TSC

There is currently no cure for TSC, but treatment focuses on managing the symptoms and preventing complications. Treatment options may include:

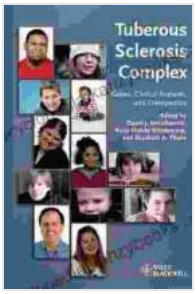
- **Medical management:** Medications to control seizures, prevent kidney and heart problems, and treat LAM
- **Surgical intervention:** Surgery to remove or reduce the size of tumors in the brain, kidney, heart, or lungs
- **Laser therapy:** To remove facial angiofibromas
- **Psychotherapy:** To address behavioral and developmental issues
- **Regular monitoring:** Regular check-ups with specialists to monitor the progression of the disease and detect any potential complications

Prognosis of TSC

The prognosis for individuals with TSC varies depending on the severity of the mutations and the organs affected. The most common cause of death in TSC is complications related to the brain tumors, such as seizures and epilepsy. However, with early diagnosis and proper management, most individuals with TSC can live full and productive lives.

Tuberous Sclerosis Complex (TSC) is a complex genetic disFree Download that can affect multiple organs. The clinical manifestations of TSC vary widely, but early diagnosis and proper management are crucial for improving outcomes. Ongoing research is focused on understanding the

genetic basis of TSC, developing new treatments, and improving the quality of life for individuals affected by this condition.



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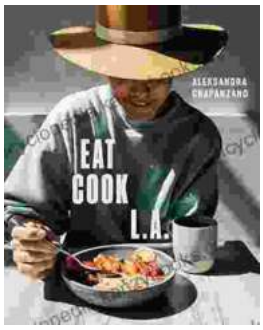

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