Section X Tuberous Sclerosis Complex Research Program

Vision: To lessen the impact of tuberous sclerosis.

Mission: To encourage innovative research aimed at understanding the role and function of proteins produced by the TSC1 and TSC2 tumor suppressor genes.

Congressional Appropriations for Peer-Reviewed Research: $1M in FY02

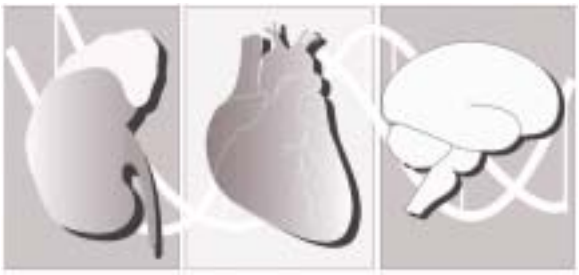
Funding Summary: ~2–3 awards anticipated from the FY02 appropriation

The Disease

Tuberous sclerosis is a genetic disorder that can affect any or all systems of the body. The disorder is characterized by seizures, developmental delays, kidney disease, behavioral problems, and the growth of benign tumors (tubers) on vital organs such as the brain, kidneys, and heart. These tumors typically calcify with age, becoming hard (sclerotic). Children with tuberous sclerosis may have autistic-like symptoms. Tuberous sclerosis affects as many as 25,000 to 40,000 individuals in the United States and about 1 to 2 million individuals worldwide. Although this disorder can be inherited as an autosomal dominant trait, two-thirds of cases are the result of a spontaneous genetic change on one of two genes, TSC1 or TSC2.[1] The TSC1 gene is located on chromosome 9 and produces the protein hamartin. The TSC2 gene is located on chromosome 16 and produces the protein tuberin. Hamartin and tuberin are believed to act as tumor growth suppressors. Therefore, their dysfunction may underlie the appearance of tumors that characterize tuberous sclerosis. There is currently no cure for this disease; however, surgical intervention and a number of treatments can help affected individuals.

Program Background

The DOD Tuberous Sclerosis Complex Research Program (TSCRP) was established in FY02 by Joint Appropriations Conference Committee Report No. 107-350, which provided $1M for tuberous sclerosis research. In accordance with the directives received from Congress, the TSCRP will support innovative research directed toward a better understanding of the role and function of proteins produced by the TSC1 and TSC2 tumor suppressor genes.



The kidneys, heart, and brain are all susceptible to the development of tumors in patients with Tuberous Sclerosis complex.

The Vision for the FY02 Program

The goal of the FY02 TSCRP is to promote research in the field of TSC. One award mechanism, Idea Development Awards, was offered to support innovative research aimed at understanding the role and function of proteins produced by the TSC1 and TSC2 tumor suppressor genes.

In response to the TSCRP Program Announcement, 13 proposals were received electronically. Because of its similarity to neurofibromatosis and in an effort to reduce management costs, proposals received in response to the FY02 TSCRP will be reviewed in conjunction with proposals received in response to the FY02 NFRP. Scientific peer review will be conducted in November 2002, and programmatic review is scheduled for January 2003. Approximately 2 to 3 awards are anticipated.

Summary

The TSCRP was established in FY02 with a $1M congressional appropriation for research on tuberous sclerosis. Projects funded by this program are anticipated to support research that will lead to the substantial improvement in the understanding, diagnosis, and treatment of tuberous sclerosis, and enhance the quality of life of persons with the disease.

FY02 Ad Hoc Programmatic Reviewers[2]

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[1] *National Institute of Neurological Disorders and Stroke Fact Sheet, 2001, and Harrison's Principles of Internal Medicine, 15th Edition, McGraw-Hill, 2001.*

[2] *FY02 TSCRP Programmatic Reviewers serve as ad hoc members to the FY02 NFRP Integration Panel.*