JUN 1 0 2003

6-10-03

We thought you might be interested in the attached material.

Julie,

Here is the information on tuberous sclerosis.

Call if questions

Laura

Wisconsin Legislative Council
One East Main Street Suite 401
P.O. Box 2536
Madison, WI 53701-2536
(608) 266-1304
Fax No. (608) 266-3830

LEXSEE 107 H.C.R. 25

FULL TEXT OF BILLS

107th CONGRESS, 1ST SESSION
IN THE HOUSE OF REPRESENTATIVES
REPORTED IN THE HOUSE

H. Con. Res. 25

2001 H. Con. Res. 25; 107 H. Con. Res. 25; Retrieve Bill Tracking Report

SYNOPSIS:
A concurrent resolution expressing the sense of the Congress regarding tuberous sclerosis.

DATE OF INTRODUCTION: February 08, 2001

SPONSOR(S):
Sponsor and Cosponsors as of 08/02/2001
KELLY, SUE W (R-NY) - Sponsor
BEREUTER, DOUGLAS K (R-NE)- Cosponsor
BURTON, DAN (R-IN)- Cosponsor
CARSON, JULIA (D-IN)- Cosponsor
COX, CHRISTOPHER (R-CA)- Cosponsor
COYNE, WILLIAM J (D-PA)- Cosponsor
CRANE, PHILIP MILLER (R-IL)- Cosponsor
DOYLE, MIKE (D-PA)- Cosponsor
ENGLISH, PHIL (R-PA)- Cosponsor
FOSSELLA, VITO J JR (R-NY)- Cosponsor
GUTIERREZ, LUIS V (D-IL)- Cosponsor
HILL, BARON P (D-IN)- Cosponsor
HILLIARD, EARL FREDERICK (D-AL)- Cosponsor
KIND, RONALD J (D-WI)- Cosponsor
LEE, BARBARA (D-CA)- Cosponsor
LUTHER, BILL (D-MN)- Cosponsor
MALONEY, CAROLYN B (D-NY)- Cosponsor
MCDERMOTT, JAMES A (D-WA)- Cosponsor
MCKINNEY, CYNTHIA (D-GA)- Cosponsor
MILLER, GEORGE (D-CA)- Cosponsor
MORAN, JAMES P (D-VA)- Cosponsor
PASCRELL, WILLIAM J JR (D-NJ)- Cosponsor
PRICE, DAVID EUGENE (D-NC)- Cosponsor
RIVERS, LYNN (D-MI)- Cosponsor
ROTHMAN, STEVEN R (D-NJ)- Cosponsor
ROYCE, EDWARD R (R-CA)- Cosponsor

Julie, this first item contains the language I the congressional Resolution on Tuberous Sclerosis.
Laura

SANCHEZ, LORETTA (D-CA)- Cosponsor
SHADEGG, JOHN BARDEN (R-AZ)- Cosponsor
STARK, FORTNEY PETE (D-CA)- Cosponsor
TANCREDO, THOMAS GERARD (R-CO)- Cosponsor
WALSH, JAMES T (R-NY)- Cosponsor
WAMP, ZACH (R-TN)- Cosponsor
WELDON, W CURTIS (R-PA)- Cosponsor
WELLER, GERALD (R-IL)- Cosponsor
WEXLER, ROBERT (D-FL)- Cosponsor
WOLF, FRANK RUDOLPH (R-VA)- Cosponsor

TEXT:
HCON 25 RH

House Calendar No. 67

107th CONGRESS

1st Session

H. CON. RES. 25

No. 107-181ë

Expressing the sense of the Congress regarding tuberous sclerosis.

IN THE HOUSE OF REPRESENTATIVES

February 8, 2001

Mrs. Kelly submitted the following concurrent resolution; which was referred to the Committee on Energy and Commerce

August 1, 2001

Additional sponsors: Mr. Pascrell, Mr. Walsh, Ms. Rivers, Mr. Hilliard, Mr. Price of North Carolina, Mr. George Miller of California, Mr. Doyle, Mr. Shadegg, Mr. Tancredo, Ms. Carson of Indiana, Mr. Gutierrez, Mr. McDermott, Mr. English, Mr. Fossella, Mr. Weldon of Pennsylvania, Mr. King, Mrs. Maloney of New York, Mr. Hill, Mr. Weller, Mr. Stark, Mr. Rothman, Mr. Crane, Mr. Cox, Ms. Sanchez, Mr. Royce, Mr. Wexler, Ms. Lee, Mr. Bereuter, Ms. McKinney, Mr. Moran of Virginia, Mr. Wolf, Mr. Burton of Indiana, Mr. Wamp, Mr. Luther, and Mr. Coyne

August 1, 2001

Reported with an amendment, referred to the House Calendar and ordered to be printed

out the preamble and insert the part printed in italicë

CONCURRENT RESOLUTION

Expressing the sense of the Congress regarding tuberous sclerosis.

Whereas at least two children born each day will be affected with tuberous sclerosis;

Whereas nearly one million people worldwide are known to have tuberous sclerosis;

Whereas tuberous sclerosis affects all races and ethnic groups equally;

Whereas tuberous sclerosis is caused by either an inherited autosomal disorder or by a spontaneous genetic mutation;

Whereas when tuberous sclerosis is genetically transmitted as an autosomal dominant disorder, a child with a parent with the gene will have a 50-percent chance of inheriting the disease;

Whereas two-thirds of the cases of tuberous sclerosis are believed to be a result of spontaneous mutation, although the cause of such mutations is a mystery;

Whereas diagnosis takes an average of 90 days with consultation of at least three specialists;

Whereas tuberous sclerosis is the largest known genetic cause of epilepsy;

Whereas tuberous sclerosis is now the second largest identifiable genetic cause of autism;

Whereas tuberous sclerosis frequently goes undiagnosed because of the obscurity of the disease and the mild form the symptoms may take; and

Whereas the Congress as an institution, and Members of Congress as individuals, are in unique positions to help raise public awareness about the need for increased funding for research, detection, and treatment of tuberous sclerosis and to support the fight against tuberous sclerosis: Now, therefore, be it

Whereas at least two children born each day will be affected with tuberous sclerosis;

(repeats the above)

Whereas nearly one million people worldwide are known to have tuberous sclerosis;

Whereas tuberous sclerosis affects all races and ethnic groups equally;

Whereas tuberous sclerosis is caused by either an inherited autosomal disorder or by a spontaneous genetic mutation;

Whereas when tuberous sclerosis is genetically transmitted as an autosomal dominant disorder, a child with a parent with the gene will have a 50-percent chance of inheriting the disease;

Whereas two-thirds of the cases of tuberous sclerosis are believed to be a result of spontaneous mutation, although the cause of such mutations is a mystery;

Whereas diagnosis takes an average of 90 days with consultation of at least three specialists;

Whereas tuberous sclerosis frequently goes undiagnosed because of the obscurity of the disease and the mild form the symptoms may take; and

Whereas the Congress as an institution, and Members of Congress as individuals, are in unique positions to help raise public awareness about the need for increased funding for research, detection, and treatment of tuberous sclerosis and to support the fight against tuberous sclerosis: Now, therefore, be it

Resolved by the House of Representatives (the Senate concurring), That it is the sense of the Congress that--

(1) all Americans should take an active role in the fight against tuberous sclerosis by all means available to them, including early and complete clinical testing and investigating family histories;

(2) the role played by national and community organizations and health care providers in promoting awareness of the importance of early diagnosis, testing, and ongoing screening should be recognized and applauded;

(3) the Federal Government has a responsibility to--

(A) endeavor to raise awareness about the importance of the early detection of, and proper treatment for, tuberous sclerosis;

(B) increase funding for research so that the causes of, and improved treatment for, tuberous sclerosis may be discovered; and

(C) continue to consider ways to improve access to, and the quality of, health care services for detecting and treating tuberous sclerosis; and

(4) the Director of the National Institutes of Health should take a leadership role in the fight against tuberous sclerosis by acting with appropriate offices within the National Institutes of Health to provide to the Congress a five-year research plan for tuberous sclerosis.

107th CONGRESS

1st Session

No. 107-181ë

CONCURRENT RESOLUTION

Expressing the sense of the Congress regarding tuberous sclerosis.

August 1, 2001

Reported with an amendment, referred to the House Calendar, and ordered to be printed

SUBJECT: LEGISLATORS (90%); DISEASES & DISORDERS (90%); RESEARCH (90%); INVESTIGATIONS (90%); FAMILY (76%); ETHNICITY (76%); MENTAL ILLNESS (76%); SCIENCE NEWS (74%); HEALTH CARE INDUSTRY (72%); EPILEPSY (71%);

LOAD-DATE: August 2, 2001

LEXSEE 107 Bill Tracking H. Con. Res. 25

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Bill Tracking Report

107th Congress

1st Session

U. S. House of Representatives

HCR 25

2001 Bill Tracking H. Con. Res. 25; 107 Bill Tracking H. Con. Res. 25

No Official Title Given

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SPONSOR: Representative Sue Kelly R-NY

DATE-INTRO: February 08, 2001

LAST-ACTION-DATE: December 12, 2001

STATUS: Introduced in the House, 02/08/01

Markup held, 07/11/01

Markup held, 07/19/01

Reported in the House, amended, 08/01/01

Considered in the House, 12/04/01

Agreed to in the House, as amended, 12/04/01

Considered in the Senate, 12/12/01

Agreed to in the Senate, 12/12/01

TOTAL-COSPONSORS: 35 Cosponsors: 21 Democrats / 14 Republicans

SYNOPSIS: A concurrent resolution expressing the sense of the Congress regarding tuberous sclerosis.

Display Major Actions Only

Display All Legislative Actions (default)

ACTIONS: Committee Referrals:
02/08/2001 House Committee on Energy and Commerce

Legislative Chronology:

1st Session Activity:

02/08/2001 147 Cong Rec H 254
Referred to the House Committee on Energy and Commerce

06/27/2001 147 Cong Rec D 646
Hearing held by House Subcommittee on Health

07/11/2001
Markup held by Health Subcommittee

07/11/2001 147 Cong Rec D 687
House Subcommittee on Health approved for full committee action, as amended

07/18/2001 147 Cong Rec D 728
Ordered reported by the House Committee on Energy and Commerce

07/19/2001
Markup held by House Energy and Commerce

08/01/2001 147 Cong Rec H 5124
Report filed by the House Committee on Energy and Commerce, amended (H. Rept. 107-181)

12/04/2001 147 Cong Rec H 8766
House considered, under suspension of the rules

12/04/2001 147 Cong Rec H 8768
House agreed to, as amended (by voice vote)

12/05/2001 147 Cong Rec S 12446
Referred to the Senate Committee on Health, Education, Labor, and Pensions

12/10/2001 147 Cong Rec E 2246
Remarks by Rep. Rivers (D-MI)

12/12/2001 147 Cong Rec S 13078
Senate discharged from committee, considered, and agreed to (by unanimous consent)

BILL-DIGEST: (from the CONGRESSIONAL RESEARCH SERVICE)

Digest :

Expresses the sense of Congress regarding the fight against tuberous sclerosis with respect to the role of: (1) all Americans; (2) national and community organizations and health care providers; (3) the Federal Government; and (4) the National CRS Index Terms:
Access to health care
Budgets
Community health services

Congress
Congressional reporting requirements
Executive departments
Federal aid to medical research
Health education
Health policy
Hereditary diseases
Medical care
Medical research/Planning
Medical screening
Medical tests
Medicine
Quality of care
Research centers/Department of Health and Human Services
Science policy

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Display Co-Sponsor List By Date

Display Co-Sponsor List By Political Party

Display Co-Sponsors (Default)

CO-SPONSORS:
(Display Co-Sponsor List by Date -- ascending order) Added 02/14/2001:
Hilliard D-AL Pascrell D-NJ Rivers D-MI
Walsh R-NY
Added 02/27/2001:
Doyle D-PA Miller D-CA Price D-NC
Shadegg R-AZ
Added 03/06/2001:
Carson D-IN Gutierrez D-IL McDermott D-WA
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Added 03/20/2001:
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Added 03/27/2001:
Maloney D-NY
Added 04/04/2001:
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Rothman D-NJ Stark D-CA Weller R-IL
Added 05/08/2001:
Cox R-CA Crane R-IL Royce R-CA
Sanchez D-CA Wexler D-FL
Added 05/25/2001:
Bereuter R-NE Lee D-CA
Added 06/14/2001:
McKinney D-GA Moran D-VA Wolf R-VA
Added 06/26/2001:
Burton R-IN Wamp R-TN
Added 07/18/2001:

Luther D-MN
Added 07/30/2001:
Coyne D-PA

SUBJECT: HEALTH CARE POLICY (90%); MEDICAL RESEARCH (90%); GOVERNMENT RESEARCH FUNDING (88%); SCIENCE NEWS (86%); HEALTH CARE INDUSTRY (84%);

LOAD-DATE: 01/16/2002

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Citation: **147 Cong Rec S 13078**

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*147 Cong Rec S 13078, **

CONGRESSIONAL RECORD -- *SENATE*

Wednesday, December 12, 2001

107th Congress, 1st Session

147 Cong Rec S 13078

REFERENCE: Vol. 147, No. 172

SECTION: Senate

TITLE: EXPRESSING THE SENSE OF CONGRESS REGARDING TUBEROUS SCLEROSIS

TEXT: [*S13078]

Mr. REID. I ask unanimous consent that the health committee be discharged from further consideration of ◆ H. Con. Res. 25 , and the Senate proceed to its immediate consideration.

The PRESIDING OFFICER. Without objection, it is so ordered. The clerk will report the resolution by title.

The legislative clerk read as follows:

A concurrent resolution (H. Con. Res. 25) expressing the sense of the Congress regarding tuberous sclerosis.

There being no objection, the Senate proceeded to the immediate consideration of the concurrent resolution.

Mr. REID. I ask unanimous consent the resolution be agreed to, the preamble be agreed to, the motion to reconsider be laid on the table, and that any statements be printed in the Record, with no intervening action or debate.

The PRESIDING OFFICER. Without objection, it is so ordered.

The resolution (H. Con. Res. 25) was agreed to.

The preamble was agreed to.

LOAD-DATE: December 13, 2001

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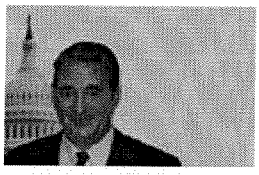
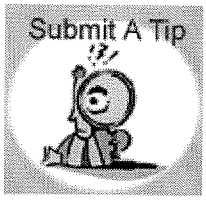
View: Full
Date/Time: Tuesday, June 10, 2003 - 4:31 PM EDT



Committee News

The House Committee on Energy and Commerce
W.J. "Billy" Tauzin, Chairman



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Tauzin Delivers Statement On Addressing Public Health Needs

Contact: Ken Johnson (202.225.5735)

WASHINGTON (June 27) -- House Energy and Commerce Committee Chairman Billy Tauzin (R-LA) is scheduled to deliver the following remarks today at a Health Subcommittee legislative hearing on measures to address various health needs of Americans:

"I commend you for calling this important hearing which will consider a number of bills and resolutions aimed toward advancing the public health. I especially commend you, Mr. Chairman, for calling a hearing which will partially focus on some diseases, which while known only to a few, destroy the lives of those afflicted.

"For example, I imagine that only a very small number of Americans have ever heard of Tuberous Sclerosis. Yet every day, two children will be born with this disease, and many of these children will develop epilepsy and autism. The Congress needs to raise awareness about this disease and declare its responsibility for supporting research into discovering the causes of the genetic mutation which leads to Tuberous Sclerosis.

"Further, while Reflex Sympathetic Dystrophy impacts the lives of the seven million children and adults suffering with the disease, as well as their caregivers, it is still not widely known to many Americans. Individuals suffering from RSD exhibit such painful symptoms as chronic inflammation, spasms, burning pain, stiffness, and discoloration of the skin, muscles, blood vessels and bones. I am pleased that today's hearing will focus attention on this disease, and I commend Mr. Barrett from Wisconsin for introducing the RSD Resolution.

"Tuberous Sclerosis and RSD are just two matters which will be considered today. The Subcommittee will also turn its attention to bills and resolutions intended to better the lives of those suffering with Duchenne Muscular Dystrophy, Juvenile Diabetes, and prostate cancer. Further, we will hear testimony from witnesses who will advocate better access to the flu vaccine, support for drug-free communities, and elevating the Director of the Indian Health Service to the level of Assistant Secretary.

"Last, Mr. Chairman, I am very interested in learning more about your legislation which would allow taxpayers to direct a portion of their tax overpayments directly to the National Institutes of Health for the purpose of increasing biomedical research funding. The Congress as a whole has been working for years to increase NIH funding, and I view your bill as a way of empowering Americans to join us in this cause.

"Again, Mr. Chairman, I commend you for calling this hearing."

####

Related Documents

Full Committee Markup
Markup of H.R. ___, the Project Bioshield Act of 2003.
Full Committee on Energy and Commerce
Thursday, May 15, 2003
10:00 AM

Member Briefing
Member Briefing on SARS
Subcommittee on Oversight and Investigations
Wednesday, May 07, 2003
1:30 PM

SARS: Assessment, Outlook, and Lessons Learned
Hearing by the Subcommittee on Oversight and Investigations
Wednesday, May 07, 2003
2:00 PM
2123 Rayburn House Office Building

Furthering Public Health Security: Project Bioshield
Hearing by the Subcommittee on Health
Subcommittee on Emergency Preparedness and Response of the Committee on Homeland Security
Thursday, March 27, 2003
09:30 AM
2123 Rayburn House Office Building

Full Committee Markup
Markup of H.R. 810, Medicare Regulatory and Contracting Reform Act of 2003
Full Committee on Energy and Commerce
Wednesday, March 26, 2003
11:00 AM

News Release
Tauzin Honored as Distinguished Community Health Champion
March 25, 2003

HIV/AIDS, TB, and Malaria: Combating a Global Pandemic.
Hearing by the Subcommittee on Health
Thursday, March 20, 2003
10:00 AM
2322 Rayburn House Office Building

More On Health

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The Committee on Energy and Commerce
2125 Rayburn House Office Building
Washington, DC 20515
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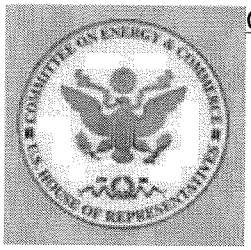

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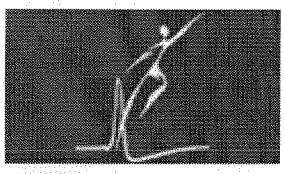

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Tuberous Sclerosis Fact Sheet

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Table of Contents

What is Tuberous Sclerosis?

Tuberous sclerosis-also called tuberous sclerosis complex (TSC)[1] - is a rare, multi-system genetic disease that causes benign tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin. It commonly affects the central nervous system and results in a combination of symptoms including seizures, developmental delay, behavioral problems, skin abnormalities, and kidney disease.

The disorder affects as many as 25,000 to 40,000 individuals in the United States and about 1 to 2 million individuals worldwide, with an estimated prevalence of one in 6,000 newborns. TSC occurs in all races and ethnic groups, and in both genders.

The name tuberous sclerosis comes from the characteristic ***tuber*** or root-like growths in the brain, which calcify with age and become hard or ***sclerotic***. The disorder-once known as ***epiloia*** or ***Bourneville's disease***-was first identified by a French physician more than 100 years ago.

TSC may be present at birth, but signs of the disorder can be subtle and full symptoms may take some time to develop. As a result, TSC is frequently unrecognized and misdiagnosed for years.

What causes Tuberous Sclerosis?

TSC is caused by defects, or mutations, on two genes-TSC1 and TSC2. Only one of the genes needs to be affected for TSC to be present. The TSC1 gene, discovered in 1997, is on chromosome 9 and produces a protein called ***hamartin***. The TSC2 gene, discovered in 1993, is on chromosome 16 and produces the protein ***tuberin***. Scientists believe these proteins act as tumor growth suppressors, agents that regulate cell proliferation and differentiation-the

processes in which nerve cells divide to form new generations of cells and acquire individual characteristics.

Is TSC inherited?

Although some individuals may inherit the disorder from a parent with TSC, most cases occur as spontaneous mutations. In these situations, neither parent has the disorder or the faulty gene(s). Instead, a faulty gene first occurs in the affected individual.

In other cases, TSC is an autosomal dominant disorder, which means that the disease is carried by a dominant gene. In those cases where it is passed from parent to child, only one parent needs to have the gene in order to produce the disease in a child. If a parent has the TSC gene, each offspring has a 50 percent chance of developing the disorder. Children who inherit TSC may not have the same symptoms as their parent and they may have either a milder or a more severe form of the disorder.

Some individuals acquire TSC through a process called ***gonadal mosaicism***. These patients have parents with no apparent defects in the two genes that cause the disorder. Yet these parents can have a child with TSC because a portion of one of the parent's reproductive cells (sperm or eggs) can contain the genetic mutation without the other cells of the body being involved. In cases of gonadal mosaicism, genetic testing of a blood sample might not reveal the potential for passing the disease to offspring.

What are the signs and symptoms of TSC?

TSC can affect any or all systems of the body, causing a variety of signs and symptoms. Signs of the disorder vary depending on which system and which organs are involved. The natural course of TSC varies from individual to individual, with symptoms ranging from very mild to quite severe. In addition to the benign tumors that frequently occur in TSC, other common symptoms include seizures, mental retardation, behavior problems, and skin abnormalities. Tumors can grow on any organ, but they most commonly occur on the brain, kidneys, heart, lungs, and skin. Malignant tumors are rare in TSC. Those that do occur primarily affect the kidneys.

Kidney problems such as ***cysts*** and ***angiomyolipomas*** occur in an estimated 40 to 80 percent of individuals with TSC, usually occurring between ages 20 and 30. ***Cysts*** are usually small, appear in limited numbers, and cause no serious problems. Approximately 2 percent of individuals with TSC develop large numbers of cysts in a pattern similar to polycystic kidney disease[2] during childhood. In these cases, kidney function is compromised and kidney failure occurs. In rare instances, the cysts may bleed, leading to blood loss and anemia.

Angiomyolipomas - benign growths consisting of fatty tissue and muscle cells- are the most common kidney lesions in TSC. These growths, which are not rare or unique to TSC, are found in about 1 in 300 people without TSC. Angiomyolipomas caused by TSC are usually found in both kidneys and in most cases they produce no symptoms. However, they can sometimes grow so large that they cause pain or kidney failure. Bleeding from angiomyolipomas may also occur, causing both pain and weakness. If severe bleeding does not stop naturally, there may severe blood loss, resulting in profound anemia and a life-threatening drop in blood pressure, warranting urgent medical attention.

Other rare kidney problems include renal cell carcinoma, developing from an angiomyolipoma, and oncocytomas, benign tumors unique to individuals with TSC.

Three types of **brain tumors** are associated with TSC: ***cortical tubers***, for which the disease is named, generally form on the surface of the brain, but may also appear in the deep areas of the brain; ***subependymal nodules***, which form in the walls of the ventricles-the fluid-filled cavities of the brain; and ***giant-cell astrocytomas***, a type of tumor that can grow and block the flow of fluids within the brain, causing a buildup of fluid and pressure and leading to headaches and blurred vision.

Tumors called cardiac **rhabdomyomas** sometimes are found in the hearts of infants and young children with TSC. If the tumors are large or there are multiple tumors, they can block circulation and cause death. However, if they do not cause problems at birth-when in most cases they are at their largest size-they usually do not grow and probably will not affect the individual in later life.

Benign tumors called **phakomas** are sometimes found in the eyes of individuals with TSC, appearing as white patches on the retina. Generally they do not cause vision loss or other vision problems, but they can be used to help diagnose the disease.

Additional tumors and cysts may be found in other areas of the body, including the liver, lung, and pancreas. Bone cysts, rectal polyps, gum fibromas, and dental pits may also occur.

A wide variety of **skin abnormalities** may occur in individuals with TSC. Most cause no problems but are helpful in diagnosis. Some cases may cause disfigurement, necessitating treatment. The most common skin abnormalities include:

- **Hypomelanic macules** ("ash leaf spots"), which are white or lighter patches of skin that may appear anywhere on the body and are caused by a lack of skin pigment or melanin-the substance that gives skin its color.
- Reddish spots or bumps called ***facial angiofibromas*** (also called ***adenoma sebaceum***), which appear on the face (sometimes resembling acne) and consist of blood vessels and fibrous tissue.
- Raised, discolored areas on the forehead called ***forehead plaques***, which are common and unique to TSC and may help doctors diagnose the disorder.
- Areas of thick leathery, pebbly skin called ***shagreen patches***, usually found on the lower back or nape of the neck.
- Small fleshy tumors called ***ungual or subungual fibromas*** that grow around and under the toenails or fingernails and may need to be surgically removed if they enlarge or cause bleeding.
- Other skin features that are not unique to individuals with TSC, including ***molluscum fibrosum*** or skin tags, which typically occur across the back of the neck and shoulders, ***café au lait spots*** or flat brown marks, and ***poliosis***, a tuft or patch of white hair that may appear on the scalp or eyelids.

TSC can cause seizures and varying degrees of mental disability. Seizures of all types may occur, including infantile spasms; tonic-clonic seizures (also known as grand mal seizures); or tonic, akinetic, atypical absence, myoclonic, complex partial, or generalized seizures.

Approximately one-half to two-thirds of individuals with TSC have mental disabilities ranging from mild learning disabilities to severe mental retardation. Behavior problems, including aggression, sudden rage, attention deficit hyperactivity disorder, acting out, obsessive-compulsive disorder, and repetitive, destructive, or self-harming behavior, may occur in children with TSC. Some individuals with TSC may also have a developmental disorder called autism.

How is TSC diagnosed?

In most cases the first clue to recognizing TSC is the presence of seizures or delayed development. In other cases, the first sign may be white patches on the skin (hypomelanotic macules).

Diagnosis of the disorder is based on a careful clinical exam in combination with computed tomography (CT) or magnetic resonance imaging (MRI) of the brain, which may show tubers in the brain, and an ultrasound of the heart, liver, and kidneys, which may show tumors in those organs. Doctors should carefully examine the skin for the wide variety of skin features, the fingernails and toenails for ungual fibromas, the teeth and gums for dental pits and/or gum fibromas, and the eyes for dilated pupils. A Wood's lamp or ultraviolet light may be used to locate the hypomelantic macules which are sometimes hard to see on infants and individuals with pale or fair skin.

In infants TSC may be suspected if the child has cardiac rhabdomyomas or seizures (infantile spasms) at birth. With a careful examination of the skin and brain, it may be possible to diagnose TSC in a very young infant. However, most children are not diagnosed until later in life when their seizures begin and other symptoms such as facial angiofibromas appear.

How is TSC treated?

There is no cure for TSC, although treatment is available for a number of the symptoms. Antiepileptic drugs may be used to control seizures, and medications may be prescribed for behavior problems. Intervention programs including special schooling and occupational therapy may benefit individuals with special needs and developmental issues. Surgery including dermabrasion and laser treatment may be useful for treatment of skin lesions. Because TSC is a lifelong condition, individuals need to be regularly monitored by a doctor to make sure they are receiving the best possible treatments. Due to the many varied symptoms of TSC, care by a clinician experienced with the disorder is recommended.

What is the prognosis?

The prognosis for individuals with TSC depends on the severity of symptoms, which range from mild skin abnormalities to varying degrees of learning disabilities and epilepsy to severe mental retardation, uncontrollable seizures, and kidney failure. Those individuals with mild symptoms generally do well and live long productive lives, while individuals with the more severe form may have serious disabilities.

In rare cases, seizures, infections, or tumors in vital organs may cause complications in some organs such as the kidneys and brain that can lead to severe difficulties and even death. However, with appropriate medical care, most individuals with the disorder can look forward to normal life expectancy.

What research is being done?

Within the Federal Government, the leading supporter of research on TSC is the National Institute of Neurological Disorders and Stroke (NINDS). The NINDS, part of the National Institutes of Health (NIH), is responsible for supporting and conducting research on the brain and the central nervous system. NINDS conducts research in its laboratories at NIH and also supports studies through grants to major medical institutions across the country. The National Heart, Lung, and Blood Institute and the National Cancer Institute, also components of the NIH, support and conduct research on TSC.

Scientists who study TSC seek to increase our understanding of the disorder by learning more about the TSC1 and TSC2 genes that can cause the disorder and the function of the proteins-tuberin and hamartin-produced by these genes. Scientists hope knowledge gained from their current research will improve the genetic test for TSC and lead to new avenues of treatment, methods of prevention, and, ultimately, a cure for this disorder.

In one study researchers defined the mutations in the TSC1 and TSC2 genes in a large (more than 300) group of individuals with TSC in an effort to find correlations between types of mutations and clinical features of the disorder. Mechanisms of mutation occurrence and the effects of other genes on clinical severity are also being studied. These same scientists are also developing mouse models of TSC, which will provide a unique opportunity to examine how the disease develops, discover the critical cell types that are affected in TSC, and provide the opportunity for therapeutic intervention.

Another study focuses on two major brain disorders-autism and epilepsy-that occur in children with TSC. Information from this study could lead to a better understanding of all three disorders, as well as to the development of new drug treatments.

Other scientists are trying to determine what causes skin tumors to develop in individuals with TSC and to find the molecular basis of these tumors. Findings from this study could shed new light on the genetics of TSC.

Where can I get more information?

Private, voluntary organizations that offer information and services to those affected by TSC include the following:

Tuberous Sclerosis Alliance
801 Roeder Road
Suite 750
Silver Spring, MD 20910-4467
info@tsalliance.org
http://www.tsalliance.org
Tel: 301-562-9890 800-225-6872
Fax: 301-562-9870

Epilepsy Foundation
4351 Garden City Drive
Suite 500
Landover, MD 20785-7223
postmaster@efa.org
http://www.epilepsyfoundation.org

Tel: 301-459-3700 800-EFA-1000 (332-1000)
Fax: 301-577-2684

National Organization for Rare Disorders (NORD)
P.O. Box 1968
(55 Kenosia Avenue)
Danbury, CT 06813-1968
orphan@rarediseases.org
http://www.rarediseases.org
Tel: 203-744-0100 Voice Mail 800-999-NORD (6673)
Fax: 203-798-2291

For information on other neurological disorders or research programs funded by the National Institute of Neurological Disorders and Stroke, contact the Institute's Brain Resources and Information Network (BRAIN) at:

BRAIN
P.O. Box 5801
Bethesda, MD 20824
(800) 352-9424
www.ninds.nih.gov

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[1]Tuberous sclerosis is often referred to as tuberous sclerosis complex (TSC) in medical literature to help distinguish it from Tourette's syndrome, an unrelated neurological disorder.

[2]Polycystic kidney disease is a genetic disorder characterized by the growth of numerous fluid-filled cysts in the kidneys.

Prepared by:
Office of Communications and Public Liaison
National Institute of Neurological Disorders and Stroke
National Institutes of Health
Bethesda, MD 20892

NINDS health-related material is provided for information purposes only and does not necessarily represent endorsement by or an official position of the National Institute of Neurological Disorders and Stroke or any other Federal agency. Advice on the treatment or care of an individual patient should be obtained through consultation with a physician who has examined that patient or is familiar with that patient's medical history.

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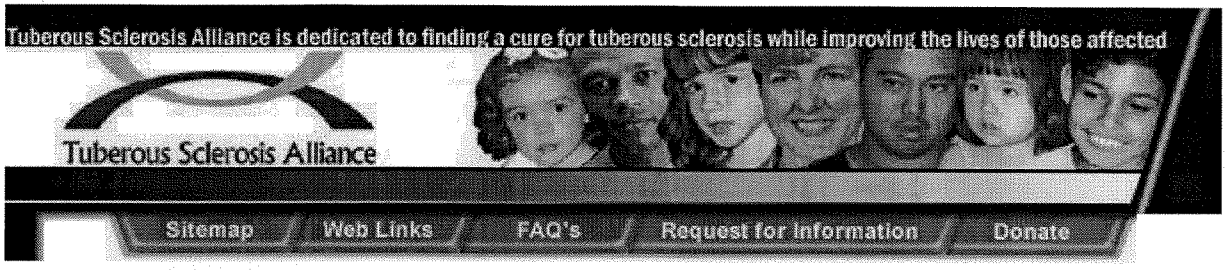
Reviewed May 16, 2003



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TS Alliance Advocates for Increased Federal Support for Research Congress endorses TSC Priorities for NIH

The TS Alliance is pleased to report that Congress was persuaded to include special TSC-related language in an omnibus spending bill enacted Feb. 20, calling on the National Institute of Neurological Disorders and Stroke (NINDS) to expand its TSC research portfolio to include a comprehensive patient registry, epidemiological studies and the development of animal models and cell lines. The same measure boosts overall NIH funding to $27.2 billion for fiscal year 2003, marking the completion of a congressional commitment to double the agency's budget over five years.

This marks the second time Congress agreed to boost the federal investment in TSC research for this fiscal year, the first coming last summer when $2 million was earmarked for TSC research in the Defense Department appropriations bill.

While we will follow up with NINDS to ensure that the congressional directive is implemented, the TS Alliance is already preparing for what is shaping up to be a contentious battle over the fiscal year 2004 budget.

In total, the 107th Congress provided $3 million in new funding ($1 million for fiscal year 2002; $2 million for fiscal year 2003) specifically for TSC research in the Department of Defense Congressionally Directed Medical Research Program.

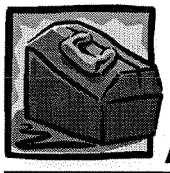
As a first step in this year's government relations program the TS Alliance has already met with key lawmakers to discuss our goal of expanding the Defense Department's investment in TSC research to $10 million in fiscal year 2004.

Your help is needed!

1) Contact the two senators from your state to:

- Urge them to support a $10 million appropriation for the Tuberous Sclerosis Complex Research Program in the Congressionally Directed Medical Research Program. Ask them to make this request to Senate Appropriations Subcommittee on Defense Chairman, Sen. Ted Stevens or Ranking Democrat , Sen. Daniel Inouye.
- Let them know that the current funding level of $2 million, while money well-spent, is not enough to achieve our objective of

finding a way to cure or prevent TSC; note the fact that DoD received more high-quality grant applications than it could fund with the limited amount of money available. Ask them to also request that NIH increase support for tuberous sclerosis research in the Labor, Health and Human Services and Education appropriations bill for 2004.



Advocacy Tool box

Click here to find e-mail addresses for your senator
Click here to find e-mail addresses for your congressional representative
Click here to view the "Dear Colleague" letter currently in circulation
Click here for a sample letter to your representative
Click here for tips on communicating with your representatives
Click here to view a fact sheet that can be used when communicating with your representative.
Click here to view the justification letter (on TS Alliance letterhead)

2) Contact your representative in Congress to:

- Urge them to support a $10 million appropriation to the Tuberous Sclerosis Complex Research Program in the Congressionally Directed Medical Research Program.
- Tell them that at the current funding level of $2 million, while money well-spent, is not enough to achieve our objective of finding a way to cure or prevent TSC; note the fact that DoD received more high-quality grant applications than it could fund with the limited amount of money available.
- Ask them to contact Rep. Jerry Lewis, Chairman of the House Appropriations Subcommittee on Defense, or Rep. John Murtha the Ranking Democrat, to urge support of $10 million for Tuberous Sclerosis Complex research
- Within the next several days we expect that "Dear Colleague" letters will be circulated in Congress, asking lawmakers to co-sponsor our $10 million request. The more who agree to sign on, the better our chances of succeeding in this year of tight budgets. You can help by calling, e-mailing or faxing your senators and representative, and convincing them to join as co-sponsors.
- Ask them to request that NIH increase support for tuberous sclerosis in the bill making appropriations for Labor, Health and Human Services and Education for 2004.

Please check the TS Alliance web site regularly for updates. As soon as

the letters are finalized and ready to be circulated, we will post specific instructions for you.

READ THE RESOLUTIONS

H. CON. RES. 25 - House Concurrent Resolution regarding tuberous sclerosis complex
S. CON. RES. 69 - Senate Concurrent Resolution regarding tuberous sclerosis complex
S. 1379 - Rare Disease Act of 2001

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November 4, 2003

The Honorable Carol Roessler
Wisconsin State Senate
P.O. Box 7882
Madison, WI 53707-7882

Dear Senator Roessler:

On behalf of the Tuberous Sclerosis Alliance (TS Alliance), I want to extend my heartfelt appreciation for your sponsorship of the Wisconsin 2003 Senate Resolution 17. Your efforts to increase awareness of tuberous sclerosis complex (TSC), heighten the importance of early detection and encourage proper treatment mean a great deal to us and to all those that are affected by TSC in the state of Wisconsin and across the nation.

We are most pleased to also share with you that the US Congress has also been supportive of TSC research this year. On September 30, President Bush signed into law a $3 million appropriation for TSC research in the Department of Defense Congressionally Directed Medical Research Program. This program is now a major source of TSC Research funds in the United States and will begin to enable researchers to capitalize on recent advances in molecular, genetic and clinical science. This could also shed new light on the treatment of cancer, kidney disease, epilepsy, diabetes, lung disease and autism.

It is also important for policy makers on the state and local level to encourage awareness of TSC. For that, we thank you for your partnership and for your compassion in assisting those that live with the daily challenges of tuberous sclerosis complex.

Please let me know if there is anything that I can do to assist you. I can be reached directly at 763-434-3455. I'm also including some additional information for you on the TS Alliance and our programs and services.

With best wishes,

Kari Luther Carlson
Vice President of Community Outreach

C: Daryl Duchatschek