



TSC ALERT

February 2007

Table of Contents

[Clicking on one of the headings takes you directly to that section of TSC Alert]

ABSTRACT SUBMISSION DEADLINES	1
FUNDING OPPORTUNITIES	2
NEW TSC PUBLICATIONS	3
RESEARCH RESOURCES	5
CONFERENCES AND SEMINARS	7
TSC CLINICAL TRIALS AND STUDIES	8
TSC INFORMATION	10

ABSTRACT SUBMISSION DEADLINES

April 20–22, 2007

**The LAM Foundation Lymphangiomyomatosis International Research Conference
The Hilton Netherlands, Downtown Cincinnati**

Deadline for Submission of Abstracts: Extended to Friday, February 9, 2007

For abstract submission forms, go to: www.thelamfoundation.org and click on Conferences/Physicians & Scientists/Call for Abstracts. Please email your abstracts and abstract form to Paul Yoder at pyoder@thelamfoundation.org

Online conference registration: www.thelamfoundation.org

May 24-26, 2007

Tuberous Sclerosis International Research Conference in Rome (Italy)

Organized by the Italian Tuberous Sclerosis Association

Convenor: Paolo Curatolo

Venue: Grand Hotel Palazzo Carpegna, Rome, Italy

Deadline for Submission of Abstracts: Extended to February 15, 2007

Information for registration and hotel accommodation: www.ptsroma.it/tsc2007

September 23-25, 2007

Tuberous Sclerosis Complex: From Genes to New Therapeutics

International TSC Conference organized by the Tuberous Sclerosis Alliance

Loews Annapolis Hotel, Annapolis, MD

Deadline for Submission of Abstracts: July 1, 2007

Information, Abstract Submission Forms, Registration Forms at: www.tsalliance.org

FUNDING OPPORTUNITIES

Understanding and Treating Tuberos Sclerosis Complex (R01) (PAS-07-190)

National Institute of Neurological Disorders and Stroke

National Cancer Institute

National Institute of Arthritis and Musculoskeletal and Skin Diseases

National Institute of Diabetes and Digestive and Kidney Diseases

National Institute of Mental Health

Application Receipt/Submission Date(s): Multiple dates, see announcement.

<http://grants.nih.gov/grants/guide/pa-files/PAS-07-190.html>

Research on Psychopathology In Intellectual Disabilities (Mental Retardation) (R01) (PA-07-212)

National Institute of Mental Health

Application Receipt Dates: Multiple dates, see announcement.

<http://grants.nih.gov/grants/guide/pa-files/PA-07-212.html>

Research on Sleep and Sleep Disorders (R01) (PA-07-140)

Letters of Intent Receipt Date: Not applicable

Application Receipt Dates: Multiple dates, see announcement.

<http://grants.nih.gov/grants/guide/pa-files/PA-07-140.html>

Tools for Zebrafish Research (R01) (PAR-07-145)

Letters of Intent Receipt Date: August 19, 2007

Application Receipt Date: September 19, 2007

<http://grants.nih.gov/grants/guide/pa-files/PAR-07-145.html>

Innovations in Biomedical Computational Science and Technology Initiative (SBIR [R43/R44]) (PAR-07-160)

Letters of Intent Receipt Date: Not applicable

Application Receipt Dates: Multiple dates, see announcement.

<http://grants.nih.gov/grants/guide/pa-files/PAR-07-160.html>

Innovations in Biomedical Computational Science and Technology Initiative (STTR [R41/R42]) (PAR-07-161)

Letters of Intent Receipt Date: Not applicable

Application Receipt Dates: Multiple dates, see announcement.

<http://grants.nih.gov/grants/guide/pa-files/PAR-07-161.html>

Funding Opportunities for Small Business Innovation Research (SBIR) and Small Business Technology Transfer (STTR) Grant Programs for 2007 (NOT-OD-07-036)

National Institutes of Health

Centers for Disease Control and Prevention

Food and Drug Administration

<http://grants.nih.gov/grants/guide/notice-files/NOT-OD-07-036.html>

NIH Pathway to Independence (PI) Award

The primary, long-term goal of the Pathway to Independence (PI) Award program is to increase and maintain a strong cohort of new and talented NIH-supported independent investigators. The PI award program is designed to facilitate a timely transition from a mentored postdoctoral research position to a stable independent research position with independent NIH or other independent

research support at an earlier stage than is currently the norm. For more information regarding this award, follow the link below:

<http://grants.nih.gov/grants/guide/pa-files/PA-07-297.html>

NIH Fiscal Policy for Grant Awards – FY 2007 (NOT-OD-07-030)

<http://grants.nih.gov/grants/guide/notice-files/NOT-OD-07-030.html>

CDC Mentored Public Health Research Scientist Development Award (K01) (RFA-CD-07-003)

Office of Public Health Research

Application Receipt Date(s): March 30, 2007

<http://grants.nih.gov/grants/guide/rfa-files/RFA-CD-07-003.html>

Request for Proposals: Autism Spectrum Disorder Research

The Nancy Lurie Marks Family Foundation has announced the availability of research funds for projects aimed at investigating communication difficulties, capacities, and options for individuals with autism spectrum disorders. Letters of intent are due March 15, 2007. For more information, visit www.nlmfoundation.org

NEW TSC PUBLICATIONS

Al-Ateeqi A, Ali RH, Kehinde EO, Mujaibel K, Al-Hunayan A, Al-Harmi J (2006) Increasing Severity of Haematuria with Successive Pregnancies in a Woman with Renal Angiomyolipoma. *Int Urol Nephrol* 2006 Dec 14 [Epub ahead of print]

Al Nazer M, Ashraf MA (2001) Fine-needle aspiration cytology of hepatic angiomyolipoma: Case report with histological, immunohistochemical and electron microscopic findings. *Ann Saudi Med* 21(5-6):324-33

Culty T, Molinie V, Lebret T, Savareux L, Souid M, Delahousse M, Botto H (2006) TSC2/PKD1 contiguous gene syndrome in an adult. *Minerva Urol Nefrol* 58(4):351-4

Dabbeche C, Chaker M, Chemali R, Perot V, El Hajj L, Ferriere J, Ballanger P, Chabbert V, Cimpean A, Otal P, Huyghe E, Grenier N, Joffre F (2006) [Role of embolization in renal angiomyolipomas.] *J Radiol* 87(12 Pt 1):1859-67 [Published in French]

Darling TN (2006) Hitting the mark in hamartoma syndromes. *Adv Dermatol* 22:181-200

de Ribaupierre S, Dorfmueller G, Bulteau C, Fohlen M, Pinard JM, Chiron C, Delalande O (2007) Subependymal giant-cell astrocytoma in pediatric tuberous sclerosis disease: when should we operate? *Neurosurgery* 60(1):83-90

de Vries PJ, Hunt A, Bolton PF (2007) The psychopathologies of children and adolescents with tuberous sclerosis complex (TSC): A postal survey of UK families. *Eur Child Adolesc Psychiatry* 2007 Jan 31 [Epub ahead of print]

de Vries PJ, Prather PA (2007) The tuberous sclerosis complex. *N Engl J Med* 356(1):92; author reply 93-4

Erlich S, Alexandrovich A, Shohami E, Pinkas-Kramarski R (2007) Rapamycin is a neuroprotective treatment for traumatic brain injury. *Neurobiol Dis* 2007 Jan 30 [Epub ahead of print]

Findlay GM, Yan L, Procter J, Mieulet V, Lamb RF (2007) A MAP4 kinase related to Ste20 is a nutrient-sensitive regulator of mTOR signalling. *Biochem J* 2007 Jan 26 [Epub ahead of print]

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- Hernandez O, Way S, McKenna J 3rd, Gambello MJ (2007) Generation of a conditional disruption of the Tsc2 gene. *Genesis* 2007 Jan 23 45(2):101-106 [Epub ahead of print]
- Herry I, Neukirch C, Debray MP, Mignon F, Crestani B (2007) Dramatic effect of sirolimus on renal angiomyolipomas in a patient with tuberous sclerosis complex. *Eur J Intern Med* 18(1):76-77
- Hossain Z, Ali SM, Ko HL, Xu J, Ng CP, Guo K, Qi Z, Ponniah S, Hong W, Hunziker W (2007) Glomerulocystic kidney disease in mice with a targeted inactivation of Wwtr1. *Proc Natl Acad Sci U S A* 104(5):1631-6
- Huang Y, Kang BN, Tian J, Liu Y, Luo H, Hester L, Snyder SH (2007) The Cationic Amino Acid Transporters CAT1 and CAT3 Mediate NMDA Receptor Activation-Dependent Changes in Elaboration of Neuronal Processes via the Mammalian Target of Rapamycin mTOR Pathway. *J Neurosci* 27:449-458
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- Konstantinopoulos PA, Papavassiliou AG (2007) The tuberous sclerosis complex. *N Engl J Med* 356(1):92-3; author reply 93-4
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- Le Bacquer O, Petroulakis E, Paglialunga S, Poulin F, Richard D, Cianflone K, Sonenberg N (2007) Elevated sensitivity to diet-induced obesity and insulin resistance in mice lacking 4E-BP1 and 4E-BP2. *J Clin Invest* 117(2):387-96
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- Roma AA, Magi-Galluzzi C, Zhou M (2007) Differential expression of melanocytic markers in myoid, lipomatous, and vascular components of renal angiomyolipomas. *Arch Pathol Lab Med* 131(1):122-5
- Somani BK, Nabi G, Thorpe P, Hussey J, McClinton S (2006) Therapeutic transarterial embolisation in the management of benign and malignant renal conditions. *Surgeon* 4(6):348-52
- Sparling MJ, Hong CH, Brahim JS, Moss J, Darling TN (2007) Oral findings in 58 adults with tuberous sclerosis complex. *J Am Acad Dermatol* 2007 Jan 18 [Epub ahead of print]
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- Vaidyanathan S, Soni BM, Hughes PL, Mansour P, Singh G (2006) Pitfalls in radiologic and histopathologic diagnosis of urologic disease-report of 4 cases. *Adv Ther* 23(6):1030-9
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- Wheless JW, Ramsay RE, Collins SD (2007) Vigabatrin. *Neurother* 4(1):163-172
- Winterkorn EB, Pulsifer MB, Thiele EA (2007) Cognitive prognosis of patients with tuberous sclerosis complex. *Neurology* 68(1):62-4

RESEARCH RESOURCES

TSC1 and TSC2 Variation Database

The TS Alliance announces that the TSC1 and TSC2 Variation Databases are now accessible to all researchers and clinicians. The TS Alliance is supporting the development of the comprehensive

TSC1 and TSC2 variation databases. These databases are curated by Professor Sue Povey and Dr. Rosemary Ekong at the University College London. Researchers and clinicians, as well as the genetic testing laboratories, will be interested in these databases containing all of the known variations (mutations and polymorphisms) in the TSC genes that will be regularly updated and revised as new variations are reported. The TSC1/2 Variation Databases can be accessed at:

TSC1 Variation Database:

http://chromium.liacs.nl/lovd/index.php?select_db=TSC1

TSC2 Variation Database:

http://chromium.liacs.nl/lovd/index.php?select_db=TSC2

NIH Announces Licensing Opportunities for Rare Disease Technologies

The National Institutes of Health (NIH) launched a new website today offering technologies available for commercial licensing that are related to rare diseases or conditions. The listing can be found at <http://www.ott.nih.gov/rarediseases> and currently consists of more than 500 such technologies, including drugs, biologics, and devices, available to be transferred from the NIH and the U.S. Food and Drug Administration (FDA) to the private sector for further research and development and potential commercialization.

The new resource was developed by the Office of Rare Diseases (ORD) and the Office of Technology Transfer (OTT) at the NIH. "By making it much easier for pharmaceutical companies and academic institutions to identify licensing opportunities, this new site will help facilitate the transfer of research advances from bench to bedside where the interventions can ultimately benefit patients," said NIH Director, Elias Zerhouni, M.D.

A rare disease is defined as one with prevalence less than 200,000 in the United States*. There are an estimated 25 to 30 million people in the United States with one of over 6,500 known rare diseases. Though technically "rare," some rare diseases are familiar, such as meningitis (inflammation of membranes of the brain and spinal cord most commonly caused by a bacterial or viral infection) and Lou Gehrig's Disease (amyotrophic lateral sclerosis, or ALS, a chronic, progressive disease marked by gradual degeneration of the nerve cells in the central nervous system that control voluntary muscle movement).

Stephen Groft, Pharm.D., Director of ORD, explained that, "Because relatively few people are affected by any one rare disease, finding therapies for each poses unique challenges and requires innovative approaches." He added, "We're excited about this new mechanism to foster collaboration with the private sector and the potential to make a real difference for patients."

The website module was developed by OTT and ORD in an attempt to provide a more collaborative, consolidated, and systematic approach to the development of products for rare diseases and conditions. "In addition to the technologies already available on the site, we encourage not-for profit organizations, academic research centers and foundations in the U.S. and abroad to submit technologies available for licensing from their institutions," said Mark Rohrbaugh, Ph.D., J.D., Director of OTT. Parties interested in licensing will be directed to the institution owning the technology. More information about submitting additional technologies can be found at <http://www.ott.nih.gov/rarediseases/submit>.

The NIH Office of Technology Transfer evaluates, protects, licenses, monitors, and manages the NIH and FDA intramural invention portfolios to carry out the mandates of the Federal Technology Transfer Act of 1986. For more information about OTT and its programs, visit www.ott.nih.gov.

The NIH Office of Rare Diseases stimulates and coordinates research on rare diseases and supports research to respond to the needs of patients, healthcare providers and the research

communities involved in the care, treatment, and evaluation of products for the preventions, diagnosis, or treatment of these conditions. For more information about ORD and its programs, visit www.rarediseases.nih.gov.

The National Institutes of Health (NIH) — *The Nation's Medical Research Agency* — includes 27 Institutes and Centers and is a component of the U.S. Department of Health and Human Services. It is the primary federal agency for conducting and supporting basic, clinical and translational medical research, and it investigates the causes, treatments, and cures for both common and rare diseases. For more information about NIH and its programs, visit www.nih.gov.

*The term "rare (or orphan) disease," as defined in the Orphan Drug Act, is a condition affecting fewer than 200,000 in the United States or a disease with a greater prevalence but for which no expectation exists that the costs of developing or distributing a drug can be recovered from the sale of the drug in the United States.

CONFERENCES AND SEMINARS

March 15, 2007 5:00 – 7:00 PM

LAM/TSC Seminar Series: "New Developments in the Neurobiology of Tuberous Sclerosis Complex and Molecular Insights into the Effects of Therapy on Malignant Gliomas"

Presented by Peter Crino, M.D., Ph.D., University of Pennsylvania and David Louis, M.D., Harvard Medical School & Massachusetts General Hospital
New Research Building, Room 350
Harvard Medical School
Boston, MA
Please RSVP to: amy_farber@hms.harvard.edu

March 29-30, 2007

Curing Epilepsy 2007: Translating Discoveries into Therapies

Natcher Conference Center, Bethesda, MD
Follow-up to 2000 Conference "Curing Epilepsy: Focus on the Future"
<http://curingepilepsy.ninds.nih.gov/>

April 19-22, 2007

2007 LAM International Research Conference

The LAM Foundation
Cincinnati, OH
<http://www.thelamfoundation.org>

May 9-12, 2007

68th Annual Meeting of the Society for Investigative Dermatology

The Century Plaza Hotel, Los Angeles, CA
For more information: www.sidnet.org

May 24-26, 2007

Tuberous Sclerosis Complex International Research Conference 2007 in Rome (Italy)

Venue: Grand Hotel Palazzo Carpegna, Rome, Italy
Information: <http://www.ptsroma.it/tsc2007/>

September 23-25, 2007

Tuberous Sclerosis Complex: From Genes to New Therapeutics

International TSC Research Symposium organized by the Tuberous Sclerosis Alliance
Loews Annapolis Hotel, Annapolis, MD
More information coming soon on the TS Alliance website at www.tsalliance.org

November 3-4, 2007

Advances in Tuberous Sclerosis: From Pathway to Therapy

Sydney Children's Hospital, Randwick, NSW, Australia

For more information, contact Dr. David Mowat at d.mowat@unsw.edu.au or Dr John Lawson at John.Lawson@unsw.edu.au

September 11-14, 2008

International TSC Conference

Organized by the Tuberous Sclerosis Association, U.K.
Brighton, U.K.

More information coming soon!

NEWS

Tiny Laboratory Enables Huge Leaps in Mapping Protein Function

Howard Hughes Medical Institute researchers have designed a laboratory about the size of a quarter that is capable of conducting thousands of experiments simultaneously to measure how specialized proteins bind their DNA targets. This tool provides a new way of measuring the activity and function of proteins. Having those measurements may help scientists predict the behavior of individual proteins in biological systems without making any direct measurements on model organisms. This research was published by Stephen R. Quake, D.Phil., HHMI investigator, Stanford University, in the January 12, 2007, issue of Science. For the full story, go to:

<http://www.hhmi.org//news/quake20070112.html>

Turning a Cellular Sentinel into a Cancer Killer

HHMI researchers have developed two strategies to reactivate the p53 gene in mice, causing blood, bone and liver tumors to self destruct. The p53 protein is called the "guardian of the genome" because it triggers the suicide of cells with damaged DNA.

The researchers' findings show for the first time that inactivating the p53 gene is necessary for maintaining tumors. While the researchers caution that cancers can mutate to circumvent p53 reactivation, they believe their findings offer ideas for new approaches to cancer therapy. This research was published in the January 24, 2007, issue of Nature by Tyler Jacks, Ph.D., HHMI investigator at Massachusetts Institute of Technology and Scott W. Lowe, Ph.D., HHMI investigator at Cold Spring Harbor Laboratory. For the full story, go to:

<http://www.hhmi.org//news/jackslowe20070124.html>

TSC CLINICAL TRIALS AND STUDIES

1. Sirolimus in Treating Patients with Angiomyolipoma of the Kidney

Official Title: Phase II Study of Sirolimus in Patients with Angiomyolipoma of the Kidney Secondary to Tuberous Sclerosis or Lymphangiomyomatosis

Study Purpose:

Rationale: Drugs used in chemotherapy, such as sirolimus, work in different ways to stop the growth of tumor cells, either by killing the cells or by stopping them from dividing.

Purpose: This phase II trial is studying how well sirolimus works in treating individuals with angiomyolipoma of the kidney.

Eligibility:

Ages Eligible for Study: 18 Years - 65 Years
Genders Eligible for Study: Both

Location and Contact Information:

Please refer to this study by ClinicalTrials.gov identifier NCT00126672

Connecticut

Connecticut Children's Medical Center, Hartford, Connecticut, 06106, United States; Recruiting Francis J. DiMario, MD 860-545-9460

Massachusetts

Massachusetts General Hospital, Boston, Massachusetts, 02114, United States; Recruiting Elizabeth Thiele, MD, PhD 617-726-6540

New York

New York University Medical Center, New York, New York, 10016, United States; Recruiting Daniel K. Miles, MD 212-263-8318

Ohio

Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio, 45229-3039, United States; Recruiting David Franz, MD 513-636-4222 david.franz@chmcc.org

Pennsylvania

University of Pennsylvania Medical Center, Philadelphia, Pennsylvania, 19104-4283, United States; Recruiting Peter Crino, MD, PhD 215-349-5312

Texas

University of Texas Southwestern Medical Center at Dallas, Dallas, Texas, 75390, United States; Recruiting Arthur I. Sagalowsky, MD 214-645-8797 arthur.sagalowsky@utsouthwestern.edu

Study chairs or principal investigators: Sandra Dabora, MD, PhD, Study Chair, Brigham and Women's Hospital

For more information about this clinical trial, visit:
<http://www.cancer.gov/clinicaltrials/DFCI-04298>

2. Siblings Wanted for a Modifier Gene Research Study



The Herscot Center for Tuberous Sclerosis Complex and the Center for Human Genetic Research at the Massachusetts General Hospital are trying to identify additional genes that play a role in neurological problems in TSC.

We would like to enroll TSC siblings over 2 years of age that differ in their expression of
autism or **infantile spasms** or **uncontrollable seizures**

Participants will have:

- clinical evaluation by a neurologist specialized in TSC (Dr. Elizabeth Thiele)
- blood drawn
- brain MRI
- EEG
- neuropsychological testing (only for siblings differing in autism)

The whole study will take about 2 days to complete. There is no direct cost to the participants.

Travel and lodging will be reimbursed for participants who live more than 2 hours away from Boston.

For more information contact Susana Camposano 617-726 0240 (M-F, 8-5),
scamposano@partners.org

3. TSC and TSC/LAM Protocols at the National Institutes of Health

The Clinical Research Nurses can be reached toll-free at 1-877-NIH-LUNG, # 3 on the menu or locally through 301-496-3632.

4. Individuals with Seizures Sought

Scientists at the National Institute of Neurological Disorders and Stroke (NINDS) seek people age 5 and older with seizures for participation in research studies. People with seizures that are not controlled by standard antiepileptic drugs are eligible. However, those with other medical conditions, particularly if on-going therapy is needed, may be excluded. The scientists will record seizures with video-EEG (electroencephalogram) monitoring, and will conduct non-invasive brain imaging tests such as positron emission tomography (PET) and magnetic resonance imaging (MRI) scans. The studies may last several months, with an inpatient stay of up to 2 weeks and 10-15 outpatient visits of about an hour each.

Before patients enter the study, they will be screened in the outpatient clinic. The screening will include a history, a neurological examination, an EEG, and an MRI scan, if needed. Even if patients decide not to enter the study after the initial screening, the investigators may be able to make suggestions for further seizure evaluation or treatment.

The studies will take place at the National Institutes of Health (NIH) Clinical Center in Bethesda, MD. All study-related expenses will be paid by the NIH. There is no cost for participation or for any tests associated with the research.

For further information, contact Dr. William Theodore, Chief, Clinical Epilepsy Section, NINDS, NIH, Building 10, Room 5N250, 10 Center Drive MSC 1408, Bethesda, MD 20892-1408; telephone: 301-496-1923. Please refer to study number 01-N-0139.

TSC INFORMATION

For information about TSC and the Tuberous Sclerosis Alliance, visit the TS Alliance Web site at: <http://www.tsalliance.org> or contact the TS Alliance at info@tsalliance.org or by telephone: 1-800-225-6872 or 301-562-9890.

This is the February 2007 edition of *TSC Alert* – an online research newsletter for individuals interested in Tuberous Sclerosis Complex (TSC) research and clinical care. This online newsletter contains information of interest to the TSC research and health care community. Please forward this newsletter to colleagues who are interested in TSC. To be added/deleted to/from the mailing list for *TSC Alert* and/or to submit information for the March 2007 *TSC Alert* contact: vwhitemore@tsalliance.org

Archived issues of the TSC Alert can be found at: <http://www.tsalliance.org/pages.aspx?content=25>