



TSC ALERT

Edited by Vicky H Whittemore, PhD

June 2005

Welcome to the June 2005 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 July 2005 *TSC Alert* contact: Vicky.Whittemore@tsalliance.org

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GRANT ANNOUNCEMENTS

NIH RELEASES TUBEROUS SCLEROSIS COMPLEX PROGRAM ANNOUNCEMENT

The TS Alliance is pleased to announce the release of the National Institutes of Health (NIH) Program Announcement with set-aside funds (PAS) entitled "Understanding and Treating Tuberous Sclerosis Complex." This is a joint Program Announcement with NIH and the TS Alliance. The participating organizations intend to commit a total of approximately \$2 million to this PAS in addition to funds available for applications sent in response to this initiative that score within the pay lines of the participating NIH Institutes (NINDS, NIDDK, NIMH, NIAMS and NCI). For more information and the complete text of the Program Announcement see: <http://grants.nih.gov/grants/guide/pa-files/PAS-05-085.html>

NIH-RAID PILOT: Translational Cores Resources Pilot Project The National Institutes of Health Roadmap is establishing a program called the NIH-RAID PILOT (Rapid Access to Interventional Development) to make available, on a competitive basis, certain critical resources needed for the development of new small molecule therapeutic agents. This pilot program is developed as an NIH Roadmap for Medical Research initiative (<http://nihroadmap.nih.gov/>), and it will be administered jointly by the National Institute of Diabetes, Digestive and Kidney Diseases (NIDDK) and the National Cancer Institute (NCI) on behalf of all NIH Institutes.

Services available will include: production, bulk supply, GMP manufacturing, formulation, development of an assay suitable for pharmacokinetic testing, pharmacokinetic studies, and animal toxicology. The resources of NCI's Developmental Therapeutics Program will be used to implement this program. Services actually provided for accepted projects will depend upon the stage of the project and the strength of the preliminary data. Assistance will also be provided in the regulatory process through access to independent product development planning expertise. In the pilot phase, in vitro or animal efficacy studies and synthesis of recombinant proteins, monoclonal antibodies, or reagents for gene therapy will *not* be supported.

Questions regarding this initiative may be directed to the NIH-RAID Office listed below.

NIH-RAID Program Office
National Institute of Diabetes and Digestive and Kidney Diseases
6707 Democracy Boulevard, Room 634
Bethesda, MD 20892-5460
Telephone (301) 594-4660
Email: NIH-RAID@nidk.nih.gov

[HTTP://GRANTS.NIH.GOV/GRANTS/GUIDE/NOTICE-FILES/NOT-RM-05-004.HTML](http://grants.nih.gov/grants/guide/notice-files/not-rm-05-004.html)

NEW TSC PUBLICATIONS

New TSC Publications:

Abbott GF, Rosado-de-Christenson ML, Frazier AA, Franks TJ, Pugatch RD, Galvin JR (2005) From the archives of the AFIP: lymphangioliomyomatosis: radiologic-pathologic correlation. *Radiographics* 25(3):803-28

Bandyopadhyay SK, Das R, Chattopadhyay BK, Dutta A (2004) Tuberous sclerosis with multisystem manifestations. *J Assoc Physicians India* 52:976

Bosincu L, Rocca PC, Martignoni G, Nogales FF, Longa L, Maccioni A, Massarelli G (2005) Perivascular epithelioid cell (PEC) tumors of the uterus: a clinicopathologic study of two cases with aggressive features. *Mod Pathol* 2005 May 13 [Epub ahead of print]

Cartwright MS, McCarthy SC, Roach ES (2005) Hemimegalencephaly and tuberous sclerosis complex. *Neurology* 64(9):1634

Cook JD, Davis BJ, Cai SL, Barrett JC, Conti CJ, Walker CL (2005) Interaction between genetic susceptibility and early-life environmental exposure determines tumor-suppressor-gene

penetrance. Proc Natl Acad Sci U S A Jun 3 [Epub ahead of print]

Curatolo P, Bombardieri R, Verdecchia M, Seri S (2005) Intractable seizures in tuberous sclerosis complex: from molecular pathogenesis to the rationale for treatment. J Child Neurol 20(4):318-25

Dashti SR, Robinson S, Rodgers M, Cohen AR (2005) Pineal region giant cell astrocytoma associated with tuberous sclerosis: case report. J Neurosurg 102(3 Suppl):322-5

Edwards ST, Cruz AC, Donnelly S, Dazin PF, Schulman ES, Jones KD, Wolters PJ, Hoopes C, Dolganov GM, Fang KC (2005) c-Kit immunophenotyping and metalloproteinase expression profiles of mast cells in interstitial lung diseases. J Pathol 2005 May 10 [Epub ahead of print]

Feng Z, Zhang H, Levine AJ, Jin S (2005) The coordinate regulation of the p53 and mTOR pathways in cells. Proc Natl Acad Sci U S A 2005 May 31 [Epub ahead of print]
<http://www.pubmedcentral.nih.gov/picrender.fcgi?artid=1142118&blobtype=pdf>

Jaffe JS, Chambers JT (2005) Endometrial adenocarcinoma presenting in a premenopausal patient with tuberous sclerosis. J Intellect Disabil Res. 2005 Jun;49(Pt 6):463-5.

Johannessen CM, Reczek EE, James MF, Brems H, Legius E, Cichowski K (2005) The NF1 tumor suppressor critically regulates TSC2 and mTOR. Proc Natl Acad Sci U S A Jun 3 [Epub ahead of print]

Khalife WI, Mahmoud F, Larson E, Hardie R (2005) Pulmonary lymphangiomyomatosis in a postmenopausal woman: case report with review of literature. S D J Med 58(4):139-43

Khan N, Wazir MS, Yasin M, Mohammad J, Javed A (2005) Etiology, presentation and management outcome of pneumothorax. J Ayub Med Coll Abbottabad 17(1):62-4

Kobashi Y, Yoshida K, Miyashita N, Niki Y, Matsushima T, Irei T (2005) Multifocal micronodular pneumocyte hyperplasia in a man with tuberous sclerosis. Intern Med 44(5):462-6

Kwiatkowski D (2005) Cancer Genetics: TSC1, TSC2, TSC3? or mosaicism? Eur J Hum Genet 13(6):695-6

Lachhwani DK, Pestana E, Gupta A, Kotagal P, Bingaman W, Wyllie E (2005) Identification of candidates for epilepsy surgery in patients with tuberous sclerosis. Neurology 64(9):1651-4

Lee SY, Wang LJ, Yen TH, Chang CT (2005) Transarterial embolization of ruptured angiomyolipoma associated with tuberous sclerosis. Nephrol Dial Transplant 2005 May 10 [Epub ahead of print]

Terra-Bustamante VC, Fernandes RM, Inuzuka LM, Velasco TR, Alexandre V Jr, Wichert-Ana L, Funayama S, Garzon E, Santos AC, Araujo D, Walz R, Assirati JA, Machado HR, Sakamoto AC (2005) Surgically amenable epilepsies in children and adolescents: clinical, imaging, electrophysiological, and post-surgical outcome data. Childs Nerv Syst 2005 May 19 [Epub ahead of print]

Timotin L, Sarrot-Reynauld F, Lantuejoul S, Pasquier B, Massot C, Ashraf A, Borgel F (2005) [Tuberous sclerosis without mental impairment, diagnosed in adulthood.] Rev Med Interne 26(6):511-3 Epub 2005 Feb 12 [Article in French]

Wilson C, Idziaszczyk S, Parry L, Guy C, Griffiths DF, Lazda E, Bayne RA, Smith AJ, Sampson JR, Cheadle JP (2005) A mouse model of tuberous sclerosis 1 showing background specific early post-natal mortality and metastatic renal cell carcinoma. Hum Mol Genet 2005 May 11 [Epub ahead of print]

Wouters BG, van den Beucken T, Magagnin MG, Koritzinsky M, Fels D, Koumenis C (2005) Control of the hypoxic response through regulation of mRNA translation. Semin Cell Dev Biol 2005 May 13 [Epub ahead of print]

CONFERENCES

From Our German TSC Colleagues:

We would like to invite you to **the Tuberous Sclerosis Complex (TSC) International Research Conference 2006** that will take place from **May 3rd to May 5th, 2006**, in Berlin, Germany. The conference will be organized by the German Tuberous Sclerosis Association (TSD e.V.) together with the Epilepsy Centre of the federal states of Berlin and Brandenburg and the Epilepsy Centre Bethel, both operated by the »von Bodelschwing'sche Anstalten« Bielefeld-Bethel.

Both basic research and applied clinical research will be discussed. Subjects include the molecular functions of TSC, animal models, translational research and clinical problems in different organ systems. There will be enough remaining time for informal discussion. We will also organize a poster exhibition.

We are sure to offer a scientific meeting in an exciting and emerging field and we will be pleased by your participation. The German capital Berlin will invite you to an attractive stay, to which we will contribute with an appealing side program.

For additional information, visit the conference Web site at: <http://www.tsc2006.org>

July 5–July 15, 2005

Summer Institute in Mental Health Research

The Summer Institute in Mental Health Research is offered over the course of a two-week period, July 5–July 15, 2005, by the Department of Mental Health, Johns Hopkins Bloomberg School of Public Health. For more information about courses and enrollment, contact Adriane King at 410-955-3908 or email: aking@jhsp.edu.

http://www.jhsp.edu/Dept/MH/Summer_Institute/index.html

July 21-23, 2005

XXVII Annual Meeting of the Cognitive Science Society

Stresa, Italy

<http://www.psych.unito.it/csc/cogsci05/home.html>

August 28 – September 1, 2005

26th International Epilepsy Congress

Le Palais des Congres de Paris

Paris, France
<http://www.epilepsycongress.org>

September 11-14, 2005

The Second International Conference on Birth Defects and Disabilities in the Developing World

Jiuhua Spa and Resort, Beijing, China
www.chinamed.com.cn/birthdefects

September 26-27, 2005

Access to Quality Testing for Rare Diseases: A National Conference

Doubletree Executive Meeting Center and Hotel
Rockville, MD
<http://rarediseases.info.nih.gov/QTRD/>

October 14-18, 2005

World Congress on Psychiatric Genetics XIII: Advancing on the Pathway to Discovery

The Westin Copley Place, Boston, MA
<http://www.ispg.net>

October 18-23, 2005

American Academy of Child and Adolescent Psychiatry & Canadian Academy of Child and Adolescent Psychiatry: Joint Annual Meeting

Sheraton Centre Toronto
Toronto, Canada
<http://www.aacap.org>

December 2-6, 2005

American Epilepsy Society & American Clinical Neurophysiology Society

Washington, DC Convention Center
Washington, DC
Deadline for submission of abstracts: May 6, 2005
For more information: <http://www.aesnet.org>

****Come to the TSC SIG at this year's meeting!!!**

May 3-5, 2006

TSC International Research Conference 2006

Spandau, Ev. Johannesstift
Hotel "Christophorus Haus"
Berlin, Germany
<http://www.tsc2006.org>

July 2-6, 2006

7th European Congress of Epileptology

Helsinki Fair Centre, Helsinki, Finland

<http://www.epilepsyhelsinki2006.org>

SAVE THE DATE!

July 14-16, 2006

National TSC Conference

[Indian Lakes Resort](#)

Chicago, Illinois

<http://www.tsalliance.org>

NEWS

SECRETARY LEAVITT TAKES NEW STEPS TO ADVANCE HEALTH IT: National Collaboration and RFPs Will Pave the Way For Interoperability

HHS Secretary Mike Leavitt today announced the formation of a national collaboration and four requests for proposals (RFPs) that will advance efforts to reach President Bush's call for most Americans to have electronic health records within ten years. The President's vision would create a personal health record that patients, doctors and other health care providers could securely access through the Internet no matter where a patient is seeking medical care. Today's announcement provides a way for patients, doctors, hospitals, insurance companies, and employers to agree on standards for electronic health records and ways to achieve interoperability -- the ability to access this vital medical information immediately and efficiently.

"The national strategy for achieving interoperability of digital health information is for federal agencies -- who pay for more than one-third of all health care in the country -- to work with private-sector health care providers and employers in developing and adopting an architecture, standards and certification process," Secretary Leavitt said.

An electronic health record is a digital collection of a patient's medical history and could include items like diagnosed medical conditions, prescribed medications, vital signs, immunizations, lab results, and personal characteristics like age and weight.

"The use of electronic health records and other information technology will transform our health care system by reducing medical errors, minimizing paperwork hassles, lowering costs and improving quality of care," Secretary Leavitt said. "We will bring together the public and private health care sectors to transform health care as we know it."

The cornerstone of this effort, a private-public collaboration called the American Health Information Community (AHIC), will help nationwide transition to electronic health records -- including common standards and interoperability -- in a smooth, market-led way. The AHIC, which will be formed under the auspices of the Federal Advisory Committee Act, will provide input and recommendations to HHS on how to make health records digital and interoperable, and assure that the privacy and security of those records are protected.

HHS will solicit nominations for people to serve on the AHIC and Secretary Leavitt will appoint up to 17 commission members, as well as serve as chairperson. The AHIC will be chartered for two years, with the option to renew and duration of no more than five years. The department intends for the AHIC to be succeeded within five years by a private-sector health information community initiative that, among other things, would set additional needed standards, certify new health information technology, and provide long-term governance for health care transformation.

HHS will also issue four RFPs to pave the way for interoperability. These RFPs will create processes for setting data standards, certification, and architecture for an Internet-based nationwide health information exchange, as well as assess patient privacy and security policies. In total, HHS will spend \$86.5 million on health IT in FY 2005, and President Bush has requested \$125 million for health IT in FY 2006.

"Once the market has structure, patients, providers, medical professionals and vendors will innovate, create efficiencies and improve care," Secretary Leavitt said.

The Institute of Medicine estimates that medical errors kill 45,000 to 98,000 Americans each year in hospitals. Technology can help reduce these medical mistakes by making health information more accessible to patients and providers. As important, the information needed to treat patients effectively will be a computer click away, no matter where the patient is receiving care, which will improve patient care and satisfaction. Moreover, health technology can reduce costs by saving time, reducing duplication and waste, and improving efficiency with the potential to reduce costs by as much as 10 percent. Consumers not only benefit by saving money and improved patient care, but also by saving time because they will not have to supply basic information over and over again, or be subjected to duplicative and unnecessary tests.

Secretary Leavitt noted that HHS will do its part by adopting standards and data-sharing processes for Internet-based applications that will help federal programs like Medicaid and Medicare support the use of digital and interoperable health records that are privacy-protected and secure.

AHIC will advise the department on how to accelerate the nationwide adoption of interoperable digital records, while striking the right balance between the public and private sectors. The AHIC will have five specific tasks:

1. Make recommendations on how to protect privacy and security.
2. Identify and make recommendations for prioritizing health information technology achievements that will provide immediate benefits to consumers of health care (e.g., drug safety, lab results, bio-terrorism surveillance, etc.).
3. Make recommendations regarding the creation of a private-sector, consensus-based, standard-setting and harmonization process, and a separate product certification process.
4. Make recommendations for a nationwide architecture that uses the Internet to share health information in a secure and timely manner.
5. Make recommendations on how the AHIC can be succeeded by a private-sector health information community initiative within five years. The sunset of the AHIC, after no more than five years, will be written into the charter.

"We've outlined a powerful vision for health IT and the steps to achieve the President's goal," said Dr. David J. Brailer, National Coordinator for Health Information Technology. "The actions announced today build on the evaluation and strategies that we have developed in collaboration with the broad community of healthcare leaders."

More information is available at www.hhs.gov/healthit

NIAMS APPROPRIATIONS

The 2006 written statement of the director of NIAMS to the appropriations subcommittee is posted.

http://www.niams.nih.gov/ne/reports/congree_rep/cj2006/open2006.htm

PUBLIC ACCESS TO PUBLICATIONS

Implementation of Policy on Enhancing Public Access to Archived Publications Resulting from NIH-Funded Research (NOT-OD-05-045)

<http://grants.nih.gov/grants/guide/notice-files/NOT-OD-05-045.html>

TSC INFORMATION

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