Rare Diseases Day is One Month Away: February 29

Olympic sprinter Priscilla Lopes-Schliep (left) and Jill Viles (right) have a genetic mutation in common. It causes one's muscles to overdevelop, while the other's waste away.

Rare Disease Day is recognized worldwide this year on Monday, February 29, as a day to raise awareness of rare diseases. Here at UAB, the CCTS is proud to sponsor the Rare Disease Genomics Symposium on Friday, February 26, 8 am-3:30 pm at Children's of Alabama's Bradley Lecture Center (4th Floor, Children's Harbor Building). Registration is $15 and you can do so online today.

A recent story on the radio podcast "This American Life" featured a story of a woman who had to convince her own parents that she—and her father—had a rare disease. After a childhood of not understanding why her limbs would stop working, suddenly, and her
muscles seemed too weak to support her own weight, Jill Vines, (pictured above, right) began doing her own research and came across a medical journal describing an extremely rare type of muscular dystrophy called Emery-Dreifuss.

"And then looking at the pictures, it was a very startling thing to realize, I'm seeing my dad's arm-- just an instantaneous lock on something that's in your visual memory," Vines says in the interview, conducted by David Epstein. Epstein is the author of a book called "The Sports Theme," on the intersection of athleticism and genetics.

Vines wound up saving her father's life when she told him about her theory and he admitted that in addition to the muscle wasting, he experienced cardiac issues common among Emery-Dreifuss patients. He went to a cardiac clinic where he learned that his heart rate was only 30 beats per minute. He had a pacemaker put in immediately.

This is just the tip of this tale's iceberg, because Vines continued her research and her fight, arguing with neurologists and genetic experts that she had this rare disease. And further, she posited, a female professional athlete who was so muscular she had been tested for performance-enhancing drugs at a higher rate than most, had a genetic mutation in common with herself.

Priscilla Lopes-Schliep is an Olympic bronze medalist sprinter from Canada. She is so muscle-bound that she has been accused throughout her career of taking steroids. Someone once posted a photo online featuring a male bodybuilder's straining face atop her body.

"That was pretty messed up. And I was really pissed about that. And I got tested," Lopes-Schliep tells Epstein. "I felt like I was targeted because I got tested so much for track and field. And I think a lot of people really honestly truly believed that I was taking steroids."

Vines learned of Lopes-Schliep after her father's death when her sister showed her a photo of the muscle-bound sprinter.

"It was just unmistakable," Vines said. "It's like a computer that can analyze a photograph and get a match and be 100% sure that's the same shoulder, that's the same upper arm, I see the same veins, I see them branching this way. You just know. And that's hard to convey-- how could you just know? But I knew we were very likely cut from the same cloth, a very, very, very rare cloth."
To learn more, read the full story as told on "This American Life." ProPublica features the tale as told by Epstein, "The DIY Scientist, the Olympian, and the Mutated Gene" on their website, which features videos and photos of both women. It is an unique tale, as each story of individuals with rare disease is, and shows no matter how far the science advances in this area, there are always additional discoveries to be made.

Seminars, Symposia and Conferences

***PLEASE NOTE: The CCTS Forum for Wednesday, February 3, will NOT take place in order to better organize next month's event. Please look to future issues of the Digest for updates on the March event.***

Thursday, February 4
Clinical Trials Office Research Seminar
Theme: ClinicalTrials.gov
"SAE Reporting" with Lee Powell

Noon-1pm
PCAMS, 1924 7th Avenue South

Please join us remotely via GoToMeeting from your computer, tablet or smartphone: https://global.gotomeeting.com/join/930891813
You can also dial in using your phone (267) 507-0008; Access Code: 930-891-813

Friday, February 6
First Friday Mentoring Lunch
Noon-1pm
PCAMS, 1924 7th Avenue South

Please RSVP so that we have enough food: ccts@uab.edu; 205-934-7442.

Tuesday, February 9
"Developing a Bioinformatics Environment for Immunoglobulin Repertoire Sequencing"
Alexander F. Rosenberg, PhD, Research Assistant Professor
Division of Allergy, Immunology and Rheumatology
Director of Bioinformatics, Department of Medicine
University of Rochester Medical Center, Rochester, NY
11:30am
Thursday, February 11
UAB Informatics Institute Lecture
"Demystifying Brain Injuries and Beyond: Informatics for Critical Care Medicine"
Vignesh Subbian, PhD
Instructor, Department of Electrical Engineering and Computing Systems
University of Cincinnati

Noon-1pm
Shelby 515
1825 University Blvd

Please register to attend no later than Monday, 02/08/16
Lunch will be served. Questions? Please contact Dr. James Cimino, Director, Informatics Institute (ciminoj@uab.edu; 205.996.1958)

Thursday, February 18
Professional Skills Development Series
"Drug Discovery and Device Development- A Birmingham Partnership Making it Happen"

Presented by:
Maaike Everts, PhD
Associate Professor, Department of Pediatrics, Infectious Diseases
Associate Director Alabama Drug Discovery Alliance
(and)
Robert Hergenrother, PhD
Director, Medical Technology Development, Director, AIMTech
Southern Research

Goals:
- Educate about the drug discovery and device development process
- Describe the CCTS Drug Discovery program and the 'Alabama Drug Discovery Alliance,' as well as the local device development program 'AIMTech'
- Describe how to get support from either program

8-9:30 am
Pittman Center for Advanced Medical Studies (PCAMS)
1924 7th Avenue South

REGISTER: http://drugdiscoverydevicedevelopment.eventbrite.com

The PSDS program is also available via GoToMeeting. If you cannot join us in PCAMS, please join us from your computer, tablet or smartphone via https://global.gotomeeting.com/join/840329197
You can also join us by phone (long distance): +1 (224) 501-3412
Friday, February 26
Rare Disease Genomics Symposium

8:30am-3:30pm
Bradley Lecture Center
4th Floor, Children's Harbor Building
$15 to attend
To register, visit ChildrensAL.org/genetics
Questions? Please contact Shaila Handattu at hande@uab.edu

Friday, February 26
UAB Sponsored Conference on Therapeutic Adherence

A national conference on therapeutic adherence sponsored by the UAB School of Medicine and our University Wide Interdisciplinary Research Centers (UWIRCS) at the Edge of Chaos (http://theedgeofchaos.org/).
Please RSVP at http://amc21adherenceconference.eventbrite.com

The "Understanding and Improving Treatment Adherence: An Interdisciplinary Approach" conference will include several notable speakers (see below) didactic discussions, as well as breakout sessions. The published proceedings will highlight gaps in adherence research and collaborative funding opportunities. More details will be forthcoming.

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<tr>
<th>Speaker</th>
<th>Topic</th>
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<tr>
<td>Rivet Amico, PhD, University of Michigan</td>
<td>Conceptual models and behavioral frameworks in designing studies on adherence: How to employ models in needs assessment, intervention mapping, or continuous quality improvement</td>
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<tr>
<td>Elizabeth McQuaid, PhD, Brown University</td>
<td>Ethnic disparities and therapeutic adherence</td>
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<td>Michael Stirrat, PhD, NIH/NIMH</td>
<td>Taxonomy of adherence research: How models of therapeutic adherence research fit within clinical settings</td>
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<tr>
<td>Jeffrey Curtis, MD, MS, MPH, UAB</td>
<td>Improving therapeutic adherence and utilization of large databases in therapeutic adherence research</td>
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Thursday, February 25
UAB Research Orientation Program
Fourth Thursday of each month
PCAMS, 1924 7th Avenue South

New to the UAB platform of education offerings for research is a program designed for young investigators and research staff that are new to research (possibly within the first 2
Content: A basic review of how to implement a sponsored or investigator initiated study including clinical, regulatory and financial aspects. Designed to provide a basic overview of implementing a study, familiarize attendees with the language, and hopefully provide guidance on avoiding pitfalls when starting out. Will provide basic tools and some of the necessary language in order to successfully conduct a research study.

Who should attend: New faculty, coordinators, budget and regulatory staff

Cost: Free

Registration: Limited, so please REGISTER early by contacting Dharti K. Patel, Dpatel80@uab.edu

SAVE THE DATE:
Friday, March 18
"Infusing Integrative Bioethics into Research Partnerships: Bioethical Dialogue on Respect and Power-Sharing"
Presented at the Tuskegee University Kellogg Conference Center
9:30am-4:30pm

Wednesday, May 4
Center for Genomic Medicine Symposium
Presented by:
The University of Alabama at Birmingham and
HudsonAlpha Institute for Biotechnology
Jackson Center, Huntsville, AL
10am-3pm

Hear talks from specialists and gain the opportunity to ask questions of a distinguished panel of experts.

You will have the chance to tour the innovative HudsonAlpha facility, enjoy a networking lunch with your peers and discuss with colleagues over cocktails at the evening social.

Registration is free of charge. Agenda and registration information to follow.

Funding Opportunity: REACT RFA
Announcing the CCTS Research Voucher Program
Accelerate Discovery with CCTS Research Vouchers

The CCTS Research Voucher Program provides direct research support to investigators throughout the partner network. Investigators may use vouchers to offset expense(s) for clinical and translational research services, including clinical research units, sample handling and biospecimen storage, necessary laboratory tests, study design and methodology, database handling, informatics and other CCTS-related resources across the Partner Institutions.

ICYM! Our January 6 Forum topic focused on the Voucher Program-click to review the presentation.
Apply for up to $5,000 (direct) in research support to facilitate the rapid and efficient implementation of a study by working closely with shared facilities using state-of-the-art methodologies.

Applications must outline:
- Experimental need
- What will be enabled by the investment (e.g., manuscript, grant application, additional aims)
- Provide a budget with justification

Proposals which seek to use CCTS Resources will be given special consideration. All lines of investigation supported by the CCTS Research Voucher Program require appropriate regulatory approvals (IRB, IACUC, as applicable) in advance of award and study implementation.

Voucher Ideas (not an exhaustive list):

- I would like to use CRSP and the Clinical Research Unit to recruit, consent and collect blood specimens from a few participants to generate preliminary data for an R01.

- I need extended biostatistics consultation / methodologic assistance from USA's Translational Research Services Unit, which will inform the research strategy of an investigator-initiated clinical trial.

- I would like to image a few healthy controls at Auburn University's MRI Research Center to establish study feasibility for a potential career development award.

- I need help to develop a REDCap database to aid in the collection, management and archival of clinical research data.

Please visit CCTS Research Commons to learn more about available CCTS resources and capacities across the Partner Network. If you have any questions about eligible opportunities.

Research Voucher Requests will be accepted electronically as a single PDF by CCTS Research Commons (ccts@uab.edu) on a rolling basis. Applications will be reviewed by the CCTS Executive Council for scientific merit, mission alignment, appropriateness of the budget and justification of need.

Questions? Contact the CCTS--call: 205-934-7442 or email: ccts@uab.edu

ACTS Translational Science 2016
Travel Awards Deadline Extended
*Due January 31*
The Center for Clinical and Translation Science (CCTS) is pleased to offer travel awards to investigators and trainees across the CCTS Partner Network who are interested in presenting their work at the upcoming ACTS Translational Science Meeting.

Available for investigators and teams at all levels of career development, with special emphasis on junior faculty, postdoctoral fellows, and graduate students. Requires an abstract submission as a presenter at the Translational Science 2016 Meeting, taking place April 13-15, 2016, at the Omni Shoreham Hotel in Washington, DC.

What You Will Experience at ACTS:

- Translational Science 2016 will offer more continuing education than ever before!
- Hear dedicated plenary sessions, view abstracts during expanded poster-hall viewing hours with dedicated oral presentation time, and over thirty 60-minute breakout sessions covering the following topic areas:
  - Translational Science in the Digital Age
  - Dissemination and Implementation Science
  - Team Science
  - Learning and Development Science
  - Innovation and Entrepreneurial Science
  - Integrity & Conduct of Clinical and Translational Science
  - Translational Science Acceleration
  - Informative Examples of Translational Science
- The Translational Science Meeting spotlights cutting-edge achievements in clinical and translational investigation, research impact, networking and scientific community, exposure to novel methods, best practices and important resources, and career development programs.

Learn more about the meeting and REGISTRATION INFORMATION for Translational Science 2016.

Travel awards of up to $1,000 will be available from the CCTS to match funds from the researcher's department.

Please submit the following materials to the CCTS to apply for your travel award:

- Copy of submitted abstract
BIOSTATISTICS & BIOREPOSITORY*(new!)

Mondays
Epidemiology/Biostatistics Clinic in The Edge of Chaos
This clinic provides the UAB community access to epidemiology and biostatistics resources. Available most Mondays, 10am-2pm in The Edge of Chaos (Lister Hill Library, 4th floor). Check the Edge of Chaos calendar for confirmation of meeting dates and times.

UAB CCTS Biostatistics & Biorepository Clinics

Biostats: Weekly on Wednesdays, 11:30am-1pm
Biorepository: First Wednesday of each month, 11:30am-1pm
PCAMS Building 1924 7th Avenue South

We've expanded our Biostats Clinic to include members of our CCTS Biorepository Team. On the FIRST WEDNESDAY of each month, experts from our CCTS Core Lab will join the CCTS Biostats experts to answer questions and provide consultative services on any aspect of specimen collection, specimen management, specimen processing and biorepository needs.

The Biostatistics & Biorepository Clinics are open to all faculty members, post-docs, fellows, residents, and students. Attendees are invited to bring statistical and methodological questions about on-going research projects, projects being planned, manuscripts in progress, responses to peer reviewers, and published articles. They are also invited to bring their laptops. Assistance will be provided by members of the CCTS BERD & Biorepository groups. A light lunch is served.

BERD Design Consultation Available Today
If you can’t visit the clinic, please contact the BERD or the Core Lab at your convenience by emailing ccts@uab.edu, calling (205) 934-7442, or connecting through Research Commons on the CCTS home page.