World's Leading GSD Researcher Joins Connecticut Children's

Leading pediatric endocrinologist and scientist David Weinstein, MD, and his world-renowned Glycogen Storage Disease (GSD) Program have relocated from the University of Florida to join Connecticut Children's Medical Center and the University of Connecticut School of Medicine.

Weinstein's GSD Program is the largest clinical and research program of its kind in the world. Pediatric and adult patients with the rare genetic liver disease travel from around the globe for his team's expert care. They currently treat more than 500 patients from 49 states and 45 countries.

Weinstein is a professor in the Department of Pediatrics at the UConn School of Medicine and director of the GSD Program, a joint venture of Connecticut Children's and UConn Health. The program's research laboratories are at UConn Health, while the multidisciplinary team provides comprehensive clinical care at Connecticut Children's.

Weinstein and his team are launching a clinical trial of the first gene therapy for GSD, a therapy that has shown great promise.

Internationally Renowned Pediatric Endocrinologist Chooses Connecticut

Emily L. Germain-Lee, MD, a world-renowned pediatric endocrinologist and researcher, has been recruited to be the new division chief of Pediatric Endocrinology and Diabetes at Connecticut Children's Medical Center and professor of pediatrics at UConn School of Medicine. Her research laboratory is located at UConn Health, and her clinical care is being performed at Connecticut Children's. Germain-Lee was recruited to Connecticut Children's and UConn Health from Johns Hopkins School of Medicine and the Kennedy Krieger Institute in Baltimore, Maryland.

Germain-Lee is an NIH-funded physician-scientist specializing in the clinical care and translational research of genetic and metabolic bone diseases. Her research at UConn Health and Connecticut Children's focuses on Albright hereditary osteodystrophy (AHO) and osteogenesis imperfecta (OI), two rare genetic bone disorders.

Germain-Lee has a distinguished history of caring for children and adults from around the world with Albright hereditary osteodystrophy. Over the past two decades, she has built extensive clinical and research programs focused on AHO and

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The work is the result of the Crohn’s & Colitis Foundation’s RISK Stratification study, the largest new-onset study completed on pediatric Crohn’s disease patients. It is the culmination of almost a decade of work by the leading pediatric inflammatory bowel disease centers in North America. Hyams is one of the seven-member steering committee that developed and guided the groundbreaking research.

Using patients’ genetic makeup, researchers discovered a new way to predict if a child newly diagnosed with Crohn’s disease will develop disease-related complications that will require major surgery within three to five years. The study of clinical data, demographics, baseline gene expression, immune reactivity and intestinal bacteria has identified distinct biological signatures capable of predicting, at diagnosis, stricturing and penetrating disease—the two most common complications requiring surgery.

“The course of Crohn’s disease and its response to current therapies is very variable. The ability to predict natural history and response to therapy is invaluable,” says Hyams. “The RISK study provides new and exciting findings that genetic signatures in bowel tissue, along with analysis of the intestinal microbiome, may help doctors treat the disease specific to the patient and predict how a patient will respond to therapy.”

Jeffrey Hyams, MD, is also a professor of pediatrics at the University of Connecticut School of Medicine. Contact him at jhyams@connecticutchildrens.org.
An 8-month old female presented to the Connecticut Children’s Emergency Department (ED) for evaluation after a reported episode of coughing, gagging and vomiting several hours earlier that led to family concern of possible foreign body ingestion. In the ED, she was in no distress though was mildly fussy with the exam. Her vital signs were normal for age. Notably, there were no lesions in her mouth, and her breath sounds were clear and equal bilaterally. Her abdomen was soft and nondistended with no palpable masses. Radiographs were obtained and demonstrated a round radiopaque foreign body in the mid-esophagus at the level of the carina. Additional views confirmed it to be a disc battery. The Gastroenterology service was consulted, and the patient was taken to the OR for removal of the battery. At endoscopy, the button battery was successfully removed and mucosal “caustic” lesions without active bleeding were visualized at the level of the impaction. Postoperatively, the patient appeared well, tolerated plenty of oral liquids and was discharged home with close follow-up.

The following day, the patient returned to the ED due to fever, fussiness and decreased oral intake that had started during the day and was worsening. She was seen by both the GI and Surgery services and admitted for observation and IV antibiotics. Plain radiographs of the chest and abdomen showed no evidence of perforation, and an initial contrast esophagram revealed irregularity, thickening and narrowing at the site of the foreign body ingestion. In the ED, she was doing well and was worsening. She was made NPO and monitored with intermittent CT scans performed the following day demonstrated a contained perforation of the esophagus. She was transferred to the pediatric ICU. She was treated with parenteral nutrition, pantoprazole and glycopyrrolate. After her cough and tachypnea resolved, she underwent gastrostomy tube placement. She was fed via the G-tube for five weeks until the inflammation regressed and a subsequent esophagram showed no leak. At follow-up, approximately four months post-ingestion, she was doing well and had a normal esophagram.

Button battery ingestion, both intentional and not, has increased markedly as they have become more ubiquitous. According to the National Battery Ingestion Hotline (202.625.3333), approximately 3,500 such ingestions are reported nationally each year. Of those, approximately 50 to 60 individuals experience moderate to major complications or death (such as from aortoesophageal fistula). Major complications are associated with lithium batteries, which produce higher voltage. The majority of ingestions occur in children under age 6 with a peak incidence at ages 1 to 2.

Although pressure and leakage of contents may occasionally contribute to tissue damage, the major mechanism is thought to be the continuation of electrical current flow with production of sodium hydroxide at the anode. This effect can raise the adjacent pH to 11 in minutes, resulting in liquefaction necrosis of the adjacent tissue.

The symptoms of button battery ingestion may be nonspecific initially, and a high index of suspicion should be maintained, especially for infants and toddlers, with the acute onset of wheezing or stridor in the absence of viral symptoms; chest pain; abdominal pain; coughing, choking or gagging; with attempts at or refusal of oral intake.

Children under age 12 who are suspected of having ingested a button battery > 12 mm in diameter (or unknown size) should undergo radiographic localization, imaging the nasopharynx to the anus. Those who have esophageal impaction or are younger than 5 with a battery > 20 mm in diameter in the stomach should be sent to the Emergency Department for immediate evaluation and endoscopic removal. Those older than 5 and asymptomatic may be followed as outpatients with repeat radiographs in 48 hours and instructions to be seen in the Emergency Department at the development of any symptoms.

REFERENCES

Small, Common—and DANGEROUS

Have you had an interesting case involving Connecticut Children’s?
Contact Medical News Managing Editor Dennis Crean, RN, at 860.837.6248 or dcrean@connecticutchildrens.org.
Aerodigestive Team Expands

Connecticut Children’s multidisciplinary Aerodigestive Team recently made changes that are enabling its clinicians to double the number of patients they see each month.

The team, composed of subspecialists from Otolaryngology, Gastroenterology, Pulmonology, Speech-Language Pathology and Pediatric Surgery, has hired a nurse practitioner and a medical assistant. The additional staff make scheduling and seeing patients more efficient. The physicians now hold two clinics a month: one in Hartford and one in Farmington. In addition, they have reserved operating room time, so patients have the procedures they need more quickly.

The Aerodigestive Team focuses on diagnosing and treating patients with complex upper-airway disorders and those whose respiratory or digestive problems have not responded adequately to treatment.

“Our team functions in two ways,” explains pediatric otolaryngologist Nicole Murray, MD. “We see patients together in clinic, and when patients need diagnostic procedures for ENT, GI or pulmonary, we do them all at the same time so the patient only has anesthesia once.”

“Many problems involve more than one discipline,” says pediatric gastroenterologist Bella Zeisler, MD. “Because we’re seeing the patient at the same moment and looking at the issue from different angles, we can develop a coordinated plan instead of having patients see several different doctors at different times.”

Zeisler notes that the team’s approach contributes to better outcomes because patients have a diagnosis and begin treatment sooner. Referring providers benefit because they can have patients seen by multiple specialists with just a single referral.

For more information, or to refer a patient, contact Claribel Vega at 860.545.8100 or Cvega01@connecticutchildrens.org.

RESEARCH

Toward a Better Understanding of CMT Disease

A team of clinicians at Connecticut Children's is using a grant from the Harold and Rebecca H. Gross Foundation to conduct groundbreaking research on the impact of orthopaedic surgery on patients with Charcot-Marie-Tooth disease. CMT is the most common inherited neuromuscular disease affecting the peripheral nervous system. It affects one in 2,500 people worldwide.

Principal Investigator Sylvia Õunpuu, MSc, of Connecticut Children’s Center for Motion Analysis, says she and her team are comparing pre- and post-operative gait analyses of patients in order to objectively evaluate surgery’s impact on gait. At the same time, they are studying the natural progression of the disease by measuring gait changes over time among adolescents with CMT.

“We have to separate disease progression from surgical outcome,” Õunpuu says. “We want to better understand that so surgeons can make more informed treatment decisions.”

CMT is often underdiagnosed. The first signs of CMT in children frequently include foot deformity and the inability to run as fast as children their age.

Connecticut Children’s Neurology Clinic, headed by Gyula Acsadi, MD, is designated by the international Inherited Neuropathy Consortium as a Pediatric CMT Center of Excellence. It is one of only three consortium-member sites collaborating with a pediatric gait laboratory to study the natural history and surgical outcomes in the pediatric population.

To discuss referring a patient, contact the Center for Motion Analysis at 860.837.9201 or the CMT Clinic at 860.837.7500.

Adolescent Consultation Service Opens

Connecticut Children’s has launched a new Adolescent Health Consultation Service, providing consultative outpatient services to adolescent and young adult patients (ages 12 to 24 years) for the diagnosis and management of various conditions and disorders. The service is at 505 Farmington Ave. in Farmington. Please visit the Adolescent Medicine page of connecticutchildrens.org to learn more about our program.

To make a referral, please contact our Adolescent Medicine referral line at 860.837.7681.
Child in Surgery? Now Waiting is EASE-ier

Connecticut Children’s is the first hospital in Connecticut to offer EASE, an application that enables clinicians to send HIPAA-compliant updates to a surgical patient’s loved ones before, during and after a surgical procedure. The app, available for download in the iOS and Android app stores, is designed to improve communication, reduce anxiety and increase patient and family satisfaction. Physicians can now send families text updates.

Responses from families have been overwhelmingly positive. On surveys, 93 percent said EASE demonstrates the hospital’s compassion and caring, 95 percent said EASE would prompt them to recommend the hospital to others, and 95 percent said EASE showed the hospital was committed to transparency and better communication. Respondents rated their overall EASE experience at 9.7 out of 10.

Faster GI Appointments in Fairfield County

Patients will be seen more quickly at Connecticut Children’s Danbury and Fairfield locations this fall when two new pediatric gastroenterologists join the medical staff. The addition will allow new patients to be seen within about two weeks at the Danbury or Fairfield location. In the meantime, some openings are available in Fairfield. Request that location when submitting your referral.

Connecticut Children’s Named an MDA Care Center

The national Muscular Dystrophy Association (MDA) has designated Connecticut Children’s an MDA Care Center. The designation brings grant funding for 2017 – 2019. In awarding the designation, reviewers cited strong institutional support, very innovative concepts around care delivery and the expertise and experience of Gyula Acsadi, MD, PhD, head of the Division of Neurology at Connecticut Children’s and professor of pediatrics and neurology at the UConn School of Medicine.

Physicians Provide Services at Baystate

Connecticut Children’s continues to expand services at Springfield’s Baystate Medical Center. Chief of Neurosurgery Jon Martin, MD, is at Baystate every Wednesday to see patients and operate. He also provides consultation and peer support to subspecialists in Baystate’s NICU, PICU and Pediatric Hospital Medicine service. Recently, Connecticut Children’s pediatric surgeon Brendan Campbell, MD, MPH, performed Baystate’s first patent ductus arteriosis ligation in collaboration with its cardiology, surgery and NICU clinicians. Connecticut Children’s provided support for pulmonology during a recent staffing shortage and may soon introduce nephrology services there. The relationship helps ensure that children can receive expert care close to home.

First in Prescription Drug Monitoring Program

Connecticut Children’s earlier this year became the first health care organization in Connecticut to successfully integrate its electronic health record system with the state prescription drug database.

The achievement means that the Medical Center can now provide its physicians access to patients’ controlled-substance prescription histories in real time, at the point of care, to improve patient safety and prevent overprescribing.

New Vestibular and Balance Program

Connecticut Children’s Audiology Department has opened a Vestibular and Balance Program at 505 Farmington Ave., Farmington, for the diagnosis and treatment of inner-ear and balance problems.

Children with such problems may have difficulty crawling, walking, swimming or bicycle riding. They may have trouble academically, because poor coordination between the eyes and inner ear can affect reading ability. Other symptoms include headaches and stomach upset.

Connecticut Children’s Vestibular and Balance program provides a comprehensive team approach to balance and dizziness disorders in children. Children receive expert care from specialists trained in the diagnosis of such conditions in pediatric patients. The program’s multidisciplinary team includes specialists from Audiology, Otolaryngology, Neurology, Physical Therapy and Sports Medicine.

For more information, or to schedule an appointment, call 860.545.9642.
New Website Now Live

Connecticut Children’s recently unveiled a new website boasting a fresh look, mobile-friendly design and exciting new features, including:

- Improved navigation – Find information faster! The "For Medical Professionals" section is now under the main menu. A drop-down menu includes quick links to key topics.
- New search functionality – Search the website and health information library using keywords.
- Health information library – Refreshed content and a great resource for your patients.

Visit the new website today at www.connecticutchildrens.org.

Get Grand Rounds via Podcast

Podcasts of Connecticut Children’s Pediatric Grand Rounds are at your fingertips. There are three ways to listen: 1) Through the Podbean app on your phone (Download the app and search "CT Children’s grand rounds); 2) Through Connecticut Children’s website (Go to www.connecticutchildrens.org/cme, click Podcast and listen to or download the recording); or 3) Key in the URL (Type in http://podcast.connecticutchildrens.org, listen or download.)

Billboards Feature Physicians

Connecticut Children’s recently began advertising on eye-catching electronic billboards near I-84 and I-91. The first message congratulates our physicians of the year, Drs. Harry Weinerman and Jessica Zimmerman.

Lau Is U.S. Lead on IBM Global Initiative

Ching Lau, MD, PhD, of Connecticut Children’s and the UConn School of Medicine, is spearheading the United States portion of a worldwide research initiative designed to accelerate the discovery of new treatments for childhood cancer.

The research project, called Smash Childhood Cancer, involves a team of international researchers and IBM. IBM provides scientists with free access to its World Community Grid, which harnesses computing power “donated” by volunteers around the globe. The grid is the equivalent of a free, virtual supercomputer that enables scientists to more quickly conduct millions of virtual experiments to help pinpoint promising drug candidates for further study. While otherwise idle, volunteers’ devices automatically perform virtual experiments on behalf of the research team. Results are transmitted back to researchers for analysis.

EpicCare Link: Information at Your Fingertips

Referring providers can access patients’ EMR information and test results from Connecticut Children’s online through the Medical Center’s free Web-based patient management portal, EpicCare Link.

EpicCare Link gives registered users 24/7, secure, read-only access to clinical and administrative information on their patients seen at Connecticut Children’s facilities. The application also streamlines ordering tests, referring patients and scheduling appointments.

Contact Trish Masse (Tmasse@connecticutchildrens.org).

On the “Best Doctors” List

One hundred ten providers affiliated with Connecticut Children’s Medical Center have been named to the 2017 Hartford Magazine’s Best Doctors list—a great honor for both the individual providers and the Medical Center.
Faculty and Community Provider Awards

Congratulations to these physicians who recently received Connecticut Children’s Faculty and Community Provider Awards.

Jessica Zimmerman, MD
Primary Care Center

Christine Finck, MD
Pediatric surgeon-in-chief

Steve Rogers, MD
Emergency Medicine

Harry Weinerman, MD
Community pediatrician, Bloomfield

Welcome Aboard

The newest additions to our medical staff!

Jonathan Bernstein, MD
Division of Hematology/Oncology
- Medical director, Children’s Specialty Center of Nevada
- Fellowship in hematology/oncology, James Whitcomb Riley Hospital for Children
- Internship/residency, Children’s Hospital of Los Angeles
- MD, University of Rochester School of Medicine

Melissa Fernandes, MD
Division of Digestive Diseases, Hepatology and Nutrition
- MAS in clinical research, University of California, San Francisco
- Fellowship, University of California, San Francisco
- Internship/residency, Tufts University
- MD, Saint Louis University
- BA, history, Saint Louis University

Carlos Medina, MD
Clinical Director
Division of Pediatric Urology
- Attending, pediatric urology, New York Methodist Hospital, New York Presbyterian Hospital
- Assistant professor, pediatric urology, Weill Cornell Medical College
- Master’s degree program, clinical investigation, Weill Cornell Medical College
- Fellowship, pediatric urology, San Diego Children’s Hospital
- Surgical residency in urology, University of Connecticut
- BA, philosophy and history, Columbia University

Sunitha Sura, MD
Division of Endocrinology
- Fellowship in pediatric endocrinology, University of Connecticut
- MS in clinical and translational research, University of Connecticut
- Residency in pediatrics, New York University School of Medicine
- Internship, Sri Devaraj Urs Medical College
- MD, Sri Devaraj Urs Medical College

Expanded Services

Sleep Medicine (860.837.6643)
Lynelle Schneeberg, PsyD, board-certified sleep psychologist, now sees patients several days a month in Fairfield.

Nephrology (860.545.9395), Pulmonology (860.545.9440) and Pediatric Surgery (860.545.9520)
BayState Medical Center

Infectious Diseases (860.545.9490)
Farmington and Danbury

50th Anniversary Celebration

A symposium and celebration will be held this fall to mark the 50th anniversary of the University of Connecticut School of Medicine’s Department of Pediatrics. More information coming soon!
Ask the Specialist

This column features questions referring providers have asked Connecticut Children’s faculty at Lunch & Learn talks. Priya Phulwani, MD, a pediatric endocrinologist and head of Connecticut Children’s Gender Identity Program, is this issue’s contributor.

Q: When and how should I discuss gender identity with patients and families?

A: For a prepubertal child, often the parent will bring up the topic. For example, “My son wants to wear a dress. Will he be gay or transgender?” Or “My daughter insists that she is a boy. What should I do?” One study shows that one-third of such children will be transgender. Of the other two-thirds, about one-third will not be heterosexual. So the PCP response should be along these lines: “We cannot predict outcomes with certainty, but we know that suppressing or punishing gender expression can cause anxiety and depression. Allow your child to safely express their preferred gender, and here’s a list of therapists who can help you.” At Tanner stage 2, the child should be referred to Endocrinology and a therapist. The incidence of persistent gender dysphoria after puberty has begun is over 90 percent, with an over 40 percent rate of suicide attempts if the adolescent is not supported. Therefore, ALL adolescents should be asked about gender as part of general pediatric practice. As part of your standard questionnaire or confidential interview with adolescents (before the question regarding sexual preference and activity), ask “Are you happy with your assigned gender?” If they have a history of depression or suicidal ideation, also ask confidentially if they have EVER desired to be another gender.

For more information, please visit the Gender Identity Program section of connecticutchildrens.org.