Greetings!

In this issue of *The Specialist*, we are pleased to highlight our Neonatal Epilepsy Program, our new Frenotomy Clinic, leading researcher Christina Chambers, PhD, MPH, and share some of our latest research findings through our Peds Focus online video publication that you don’t want to miss!

We are excited to introduce our primary care physician outreach pilot project that is dedicated to improving communication with primary care physicians by notifying them when one of their patients has been admitted into the hospital, and also share more about the RCSSD Specialty Shadowing Program we launched earlier this year.

In addition, we’d like to give a warm welcome to Dr. Lance Prince, new Division Chief of Neonatology and Dr. Brian Fagan, our newest physician to join the Cardiology division.

I hope you enjoy this issue and welcome your feedback.

Herbert C. Kimmons, M.D., M.M.M.
Neonatal Epilepsy Program Aims to Prevent Brain Injury in Infants

Courtesy of the San Diego Union-Tribune

A new program underway at Rady Children’s Hospital gives neurologists unprecedented access to sick newborns in hopes of spotting brain injuries that can last a lifetime if left untreated.

Rady launched the program, which it is calling the “Neuro NICU,” on Oct. 25, becoming one of only about 10 hospitals in the nation to take such aggressive aim at detecting and preventing seizures in the infants admitted to its 50-bed neonatal intensive care unit.

Program director Dr. Mary “M.J.” Harbert, a Rady Children’s Specialists of San Diego neonatal neurologist, said the medical evidence is clear: Seizures, especially those that attack a child in the first month of life, kill neurons and appear to hurt the way the brain wires itself for the future.

“This disruption of brain connectivity is thought to give rise to learning difficulties, developmental problems, memory difficulty and even a predisposition to epilepsy later in life,” Harbert said.

The Neuro NICU, modeled after programs at other hospitals, including one created at UC San Francisco in 2008, uses modern technology and specially trained nurses to get a jump on the problem. Nurses are trained to spot warning signs that could lead to a seizure. Those signs include babies with unstable vital signs, those undergoing certain therapies and those who are not as alert as they should be.

Babies exhibiting any of those telltale signs, Harbert said, are hooked up to an electroencephalogram, commonly known as an EEG, that can measure the brain’s electrical activity.

“We put newborns at risk of seizure on the EEG right away. This way, we can see exactly what their brains are doing and treat with anticonvulsants if they begin to have seizures,” Harbert said. Rady has also invested in new EEG equipment that allows doctors to monitor brain signals remotely, allowing them to spot a seizure even if they are not physically present in the hospital.

Dr. David Rowitch, chief of neonatology at UCSF, is a pioneer in the still-new practice of creating collaborations between neurologists and neonotologists. He applauded Rady’s approach, especially the fact that Harbert, and other Rady Children’s Specialists, are physically present in Rady’s neonatal intensive care unit, making rounds twice a week.

He said that neurological problems are going unrecognized in intensive care units across the nations, and having brain specialists present can help spot problems before they grow.

EEG technician Rita Sullivan prepares an unidentified baby boy for electrode placement in the neonatal intensive care unit at Rady Children’s Hospital on Tuesday, Oct. 30. — Nelvin C. Cepeda
“When we look at the NICU population, I would say about 20 percent of infants are in that high-risk category,” Rowitch said.

One common source of brain injury for babies, Rowitch said, is perinatal asphyxia, which can occur when a newborn is deprived of oxygen at birth. For these patients, the standard of care is treatment with a cooling blanket. By dropping a newborn’s temperature by about eight degrees Fahrenheit, studies have shown that brain injury due to lack of oxygen can be significantly reduced.

Harbert said Rady has been using induced hypothermia treatment for these babies for several years. But recently, as part of a study enabled by the new Neuro NICU program, the doctor said hypothermia treatment is being started sooner, in the ambulance while babies are being transferred to Rady from the hospitals where they were born. That is critical, she said, because no babies are born at Rady itself. Infants in distress are routinely transferred to the hospital only if their condition is too severe to be handled at the hospital where they were born.

Moving forward, Rady and UC San Diego will also collaborate on research into the use of anti-convulsant medication to limit seizures and into a new type of wireless EEG that uses a stamp-sized wearable sensor patch that was developed in San Diego County.

In the end, Harbert said she hopes that Rady’s newly sharpened focus on the brain will pay dividends that show up when NICU patients return for follow-up visits with their pediatricians.

“Immediate and appropriate intervention can make a difference in the outcomes of these infants,” Harbert said. “Having the right tools and being able to act quickly is crucial.”

Neonatal Epilepsy Program Aims to Prevent Brain Injury in Infants

“Families want to feel hopeful about the future, and they want to have the best possible outcome for their child. By focusing on the brain, we’re one step closer to that.”

Harbert said the neonatal epilepsy program has the potential to make a significant impact on the lives of infants and their families.

“By focusing on the brain, we’re one step closer to that,” Harbert said. “It’s a new frontier, and we’re excited to be at the forefront of this new field.”

Article excerpt updated for accuracy. Original article is available at www.UTSanDiego.com.
Frenotomy Clinic

In the last 10 years, there has been a wonderful and meaningful focus on breast feeding. With this increased awareness and direction there has also been a significantly positive impact of lactation consultancy for nursing moms. As a result of these developments, we have seen a huge increase in the number of newborns being identified with and referred for management of ankyloglossia that is causing feeding difficulties. What used to be the very occasional referral in the 1980’s and 90’s has markedly increased to an average of 10 newborns a week being referred for assessment and management. When indicated, a frenulotomy is completed, often times immediately improving the baby’s latch, mother’s milk flow and comfort for the mother. Up until now we have tried to see these patients within a day or two of the call given the nature of the problem and the desire to ensure ongoing breast feeding. With the increase in volume and requests for intervention, and need to provide the service urgently, we are establishing a frenotomy clinic that will take place on Wednesday mornings and Friday afternoons at our main San Diego ENT office. The Frenotomy Clinic will be run by our Physician Assistant, Denise Lago, along with Dr. Magit on Wednesdays and Dr. Pransky on Fridays. We will, however, continue to provide this service in our satellite clinics for those who cannot travel to our main campus.

Your patient will be given specific instructions to pre-medicate with acetaminophen before the office visit and to not feed her infant one-to-two hours before the scheduled office visit. They will be given a post frenotomy instruction sheet as well as a form to fill out and mail back giving us feedback as to the effectiveness of the procedure.

In addition, you are more than welcome to accompany your patient to our clinic if you are interested in learning how to perform a frenotomy.

For your office staff or the patient to schedule an appointment for the Frenotomy Clinic, please call the main ENT number at (858) 309-7701.

For physicians, you may call the physician back line at (858) 309-7708, or fax your fast track referral to (858) 966-8038.
Christina D. Chambers, PhD, MPH, an epidemiologist and leading researcher on birth defects at the UC San Diego, has moved her research team to 7910 Frost Street, home of Rady Children’s and UC San Diego’s new Clinical and Translational Research Center.

At the new research building, Dr. Chambers is the co-director, along with Ken Lyons Jones MD, (Chief of Rady Children’s Division of Dysmorphology) of the new Center for Promotion of Maternal Health and Infant Development. The Center, among other things, studies the effects of drugs, foods and alcohol during pregnancy.

Dr. Chambers’ and Dr. Jones’ research in this area, particularly on fetal alcohol syndrome, has developed a reputation of excellence in data quality and scientific results.

Dr. Chambers is the project director for UC San Diego and Rady Children’s in the National Children’s Study, the largest and longest study of environmental effects on children’s health ever conducted in the United States. The study will examine effects of air, water, diet, sound, family dynamics, community and cultural influences, and genetics on the growth, development, and health of 100,000 children, following them from before birth until age 21.
The Benefits of Human Milk Oligosaccharides for the Breastfed Infant

The Bode laboratory at the University of California, San Diego School of Medicine is dedicated to research on human milk oligosaccharides. These complex sugars comprise the third most abundant component of human breast milk and are not present in infant formula. The lab is working to understand how these complex sugars benefit the breastfed infant and potentially, the breastfeeding mother.

Recently, the lab discovered in animal studies that specific oligosaccharides protect from necrotizing enterocolitis and is now testing whether these results translate to the human infant. The lab has also discovered that certain oligosaccharides correlate with a reduced risk of mother-to-child HIV transmission through breastfeeding; the researchers are now testing whether these results can predict and reduce the risk of transmission from an HIV-infected mother to her uninfected child. Additionally, the lab has discovered that oligosaccharides block certain viruses, bacteria and protozoan parasites that cause severe infections and kill thousands of infants and young children around the world and is now testing whether these oligosaccharides can be used to treat these deadly infections or prevent them from occurring. Finally, the lab has begun to use human milk stem cells to elucidate how oligosaccharides are synthesized in the mammary gland—one of the few biosynthetic pathways in humans that remains to be explored.

For more information visit www.bodelab.com.

Contact: Lars Bode, PhD, 619-543-7545 or lbode@ucsd.edu

Mutations in BCKD-kinase Lead to a Potentially Treatable Form of Autism with Epilepsy

Joseph Gleeson, MD, and colleagues at the University of California, San Diego and Yale University Schools of Medicine have identified mutations in the BCKDK gene as the cause of a potentially treatable recessive syndrome characterized by autism, intellectual disability and epilepsy (Science, published online 6 September 2012; doi:10.1126/science.1224631). The scientists performed exome sequencing in two consanguineous families, each presenting with two siblings with autism and other neurodevelopmental phenotypes and a segregation pattern consistent with autosomal recessive inheritance. In both families, they found that the affected siblings harbored homozygous loss-of-function mutations in BCKDK. Further analyses of in-house exome data led to the identification of a third consanguineous family with two affected siblings harboring a homozygous missense mutation in BCKDK. The kinase encoded by BCKDK acts as a negative regulator of the branched-chain ketoacid dehydrogenase complex, which catalyzes the degradation of branched-chain amino acids.

Consistent with the human findings, adult BCKDK-knockout mice developed neurological phenotypes, including tremors and seizures, accompanied by reduced levels of branched-chain amino acids in the brain and other tissues. Notably, placing these knockout mice on a diet enriched in branched-chain amino acids reversed these neurological phenotypes, suggesting that humans with BCKDK mutations could benefit from similar dietary supplementation.

Contact: Joseph G. Gleeson, MD, 858-822-3786 or jogleeson@ucsd.edu

Molecular Regulation of Fibroblast Phenotypes in Pulmonary Fibrotic Disease

The Hagood Laboratory investigates the molecular regulation of fibroblast phenotypes in pulmonary fibrotic disease and lung alveolarization, undertaking basic cellular and molecular investigation of processes in chronic lung remodeling. James Hagood, MD, also has a special interest in childhood interstitial lung disease, a collection of rare disorders that cause diffuse alterations in the lungs. Recent data has shown that many of the molecular pathways that control the development of the lung in infancy and childhood are re-expressed in idiopathic pulmonary fibrosis (IPF) and other lung diseases in adults.

Dr. Hagood’s lab recently made the observation that epigenetic regulation, which has been shown to control such diverse processes as obesity and cancer, is also important in lung fibrosis. In particular, the researchers found that hypermethylation of the promoter for Thy-1, a critical modulator of cellular phenotypes and regulator of vascular permeability at sites of inflammation, affects the lung fibroblast fibrogenic process. These epigenetic modifications appear to represent a reversible mechanism in fibrosis that offers the possibility of new therapeutic options for IPF and related disorders.

Contact: James Hagood, MD, 858-822-7566 or jhagood@ucsd.edu
Rady Children’s Specialists Division of Neonatology
Welcomes: Lance Prince, MD, PhD
Division Chief of Neonatology

Education and Training:
Medical Degree received from University of Alabama School of Medicine
Residency completed at University of Iowa
Fellowship in Internal Medicine completed at University of Iowa
Fellowship in Neonatology/Pediatrics completed at University of Iowa

Accomplishments & Awards:
National Staff Research Award, American Lung Association (2005)
Trainee Investigator Award, Midwest Society for Pediatric Research;
“Exposure of Human Airway Epithelia to Air-Liquid Interface Changes
the Redox State and Promotes Differentiation” (2000)
Fellow Basic Research Award, Society for Pediatric Research (1999)

Rady Children’s Specialists Division of Cardiology
Welcomes: Brian Fagan, MD

Education and Training:
Medical Degree received from University of California, San Diego
Internship completed at University of California, San Diego
Residency completed at University of California, San Diego
Cardiology Fellowship completed at Children’s Hospital of Los Angeles

Accomplishments & Awards:
The Paige Bronchick Clinical Scholar Fellowship Award (2006)
Merck Award for Outstanding Contributions to the Medical School Community; University of California, San Diego (2001)
Improving Communication with Primary Care Physicians

Rady Children’s Division of Pediatrics and Hospital Medicine has launched a pilot project aimed to help improve communication with referring primary care physicians. A survey conducted in fall 2012 showed “communication back to the referring physician” needed improvement. Primary care physicians were often unaware that one of their patients had been admitted to Rady Children’s Hospital.

In February 2013, the marketing staff of Rady Children’s Hospital began calling primary care physicians who had a recent admission to the Hospital Medicine service at Rady Children’s.

The purpose of these calls is to determine:

- If the primary care physician is receiving their patient’s admission and discharge reports. If not, staff from marketing are helping to figure out why not and fix the problem.
- Whether the primary care physician wants the Hospitalist to call them either during the child’s stay or upon discharge.

Issues that have been identified and resolved on a case-by-case basis include:

- The primary care physician’s preference for communication was incorrect in Epic (i.e., Epic In Basket versus fax).
- The primary care physician’s information was inaccurate, not provided or not obtained during admission.
- The primary care physician’s name, fax number and/or office location was incorrectly listed.

How can you help?

Inform your office staff that we will be calling when one of your patients has been admitted to Rady Children’s Hospital. While it may not be necessary for our representative to speak directly to you, it is important that the information left with your staff not only be added to your patient’s chart, but also that you be informed of their admission. We are working with several large groups to streamline sharing of this information. As this project is further developed we will be offering suggestions on how we can work together to ensure that you are kept well informed.

CareLink

CareLink at Rady Children’s is a web-based portal designed for community physicians to have real-time access to their patient’s electronic medical record. Benefits to community providers include the ability to:

- Instantly access clinical information
- Submit referrals
- Order lab and radiology tests
- Receive lab results, transcribed documents and radiology reports
- Proactively monitor patient’s progress
- Receive patient event alerts
- Communicate with Rady Children’s medical staff including provider-to-provider discussions regarding patient care

Sign up today! For online enrollment visit CareLink: www.CareLinkatRadyChildrens.org. For technical questions call the Rady Children’s Service Desk at (877) 902-4278 or email CareLink@rchsd.org.
Rady Children’s Specialists of San Diego (RCSSD) is excited to offer a new educational opportunity for community providers. The RCSSD Specialty Shadowing Program will allow primary care physicians the ability to shadow a specialist of their choice for a session or a day at the Rady Children’s Hospital main campus or at one of our satellite clinic locations.

The benefits of this program include:

- **Interactive Experience**: The program is designed to allow you to learn techniques and ask questions in diagnosing and treating patients in a particular specialty.

- **Develop Professional Relationships**: The program will allow the opportunity to meet new faculty members.

If you or your colleagues are interested in participating in the RCSSD Specialty Shadowing Program, please contact:

**Dr. Seth Pransky**  
Medical Director or Satellite Services  
Division Chief of Otolaryngology  
Email: spransky@rchsd.org  
Phone: 858-576-1700, Ext. 3377

**Rachel Christensen**  
Ambulatory Director  
Email: rchristensen@rchsd.org  
Phone: 858-966-8593
About Rady Children’s Specialists of San Diego

We are committed not only to offering the most advanced diagnostic and therapeutic options, but also to providing responsive customer and patient service. RCSSD’s physicians are pleased to provide health services ranging from one-time consultation to comprehensive treatment and follow-up, and are committed to maintaining effective communication to provide the best care.

Our Mission

To provide specialty services that are state of the art, comprehensive and family-centered in order to improve the health and well-being of infants, children, adolescents and mothers with high-risk pregnancies.

Our Vision

To be the premier provider of healthcare to infants, children, adolescents and mothers with high-risk pregnancies, through excellence in clinical care, research, teaching and advocacy.