

SEEDLINGS

News from the UNIVERSITY OF CHICAGO DEPARTMENT OF PEDIATRICS
the UNIVERSITY OF CHICAGO COMER CHILDREN'S HOSPITAL

Autumn 2007

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Little-known genetic disorder causes debilitating headaches and other neurological symptoms

Heather Sosenheimer stands at the side of her 7-year-old son's hospital bed. Kedrick's long eyelashes are at rest. His left leg is fully extended, his right slightly crooked. A picture of a dog rests at the foot of the bed for when he awakens. For the time being, he's at peace.

Kedrick is still sedated after an MRI. His mom hopes the test will reveal the cause of his continuing headaches, pain so awful he has to lie down every few hours. She has lived through this scene so often that she can't recall how many times. She's seen her four children through at least 11 MRIs during the past two years, as well as 11 surgeries.

Kedrick, his brothers Drake and Hunter, older sister Markee, mom and dad all have a genetic condition called Chiari malformation, a brain protrusion at the base of the skull and the top of the neck that restricts the flow of cerebrospinal fluid.

First documented in the late 19th century, the condition remains largely a mystery. There's no known cure, just means of relieving symptoms. For the time being, surgeries and shunts are the only methods for giving Chiari sufferers relief. David Frim, MD, PhD, chief, pediatric neurosurgery at the University of Chicago and an expert in treating Chiari (pronounced kee-ar'-ee), takes care of the entire Sosenheimer family.

"There's no research except for what Dr. Frim and a few others are doing," says Heather, who finds the dearth of research frustrating. "It seems hopeless because there's no information there. No laid out treatment and plan of care, a way to say, 'If you do this, you'll get better.'"

Chiari symptoms vary. They can include headaches caused by coughing or laughing, muscle weakness, eye dysfunction, numbness, difficulty swallowing, trouble articulating words and loss of consciousness because of lack of blood flow to the brain. During periods of headache, some children must wear rose-colored glasses to go outdoors. Others



The Sosenheimer family (clockwise from front: Hunter Rick, Markee, Rase, Heather, Drake and Kedrick). Photo by Dan Dry

must turn their bedrooms into dark caverns for days at a time to dull the pain.

Each member of the Sosenheimer family endures the disease differently. For father Rick, pain usually settles in his back and neck. Kedrick, who's autistic, has excruciating headaches that lead to behavioral changes. Drake suffers from sleep apnea, spina bifida and restless leg syndrome.

Heather can tell when Hunter is suffering because she can see it in his eyes. Hunter's Chiari led to pseudotumor cerebri, a condition in which the brain is unable to fully absorb the cerebrospinal fluid, causing pressure in the brain. In Hunter's case, this condition led to inserting a shunt to remove the extra fluid; the syndrome also has caused partial loss of vision.

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COMER
CHILDREN'S
HOSPITAL



THE UNIVERSITY OF
CHICAGO



Letter from David M. Frim, MD, PhD
Associate Professor of Surgery and Pediatrics,
Chief, Pediatric Neurosurgery
Director, Pediatric Neurosciences Center

Dear Fellow Physicians,

Every year we see more than 6,000 young patients with neurosurgical and neuromedical disease. Physicians from across the country and around the world refer the usual, the unusual and the most complex cases to the University of Chicago Comer Children's Hospital because they trust our medical decision-making, our surgical skills and our interdisciplinary approach to each individual child.



Our pediatric neurosciences team includes neurosurgeons, neurologists, neuro-oncologists, orthopedic surgeons, otolaryngologists, plastic surgeons, neuropsychologists, geneticists, radiologists, specially trained nurses and nurse practitioners, social workers, and pediatric rehabilitation specialists. Our Neurogenetics Clinic is staffed by experts in neurofibromatosis and tuberous sclerosis and is a home to one of the finest pediatric neurogeneticist in Chicago.

We are one of a select few centers in the country with full array of noninvasive diagnostic testing. Our sophisticated 3-D imaging techniques – pioneered at the University of Chicago – provide detailed and accurate information about the brain. This advanced technology enables us, for example, to accurately pinpoint and more completely remove brain tumors. Our Pediatric Epilepsy Center is the only pediatric center in the country to offer a full range of noninvasive imaging tools that diagnose the cause of seizures.

After diagnosis, we develop a comprehensive treatment plan, uniquely tailored to each individual patient. Our approach reflects the University of Chicago Medical Center's broad research efforts and clinical experience. In fact, we are often in a position to give patients access to new treatments years before they are available at other medical centers. If surgery is required, we always use the most minimally invasive techniques available.

Our young patients and their families are also reassured by our state-of-the-art Children's Hospital facility and family support services and compassionate care. We look forward to working with you and responding to your patients' neurological challenges.

Sincerely,

A handwritten signature in cursive script that reads "David M. Frim".

David M. Frim, MD, PhD
Associate Professor of Surgery and Pediatrics
Chief, Pediatric Neurosurgery
Director, Pediatric Neurosciences Center

**Receive CME Credit at Our
Pediatric Grand Rounds**

Pediatric Grand Rounds at the University of Chicago is a weekly Regularly Scheduled Conference (RSC) sponsored by the Department of Pediatrics. It provides an open forum in which to discuss advances in pediatric research and their translation into clinical care. Speakers include U of C faculty as well as distinguished faculty from all over the world. Topics range from basic science research and its potential for bedside translation to new clinical programs and advances. All are welcome. Complimentary parking is available and lunch is served.

The University of Chicago Pritzker School of Medicine is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians. The University of Chicago Pritzker School of Medicine designates this educational activity for a maximum of 1 AMA PRA Category 1 Credit™. Physicians should only claim credit commensurate with the extent of their participation in the activity.

Pediatric Grand Rounds located at:
Wyler Pavilion
5839 S. Maryland Avenue -P117
Chicago, IL 60637

Below is our schedule for November / December 2007, please visit our website www.pediatrics.uchicago.edu for the current schedule, or call (773) 702-6602 for more information.

November 15, 2007

Presenter: Sharon Hirsch, MD
Assistant Professor
Residency Training Director
Child and Adolescent Psychiatry
University of Chicago Medical Center
Topic: "ADHD: Focus on Comorbidity and Treatment"

November 22, 2007

Happy Thanksgiving!

November 29, 2007

Presenter: Deborah Burnet, MD
Professor of Medicine and Pediatrics
Topic: "Community-Based Research to Decrease Diabetes Risk in African American Youth"

December 13, 2007

Presenter: Holly J. Benjamin, MD, FAAP, FACSM
Associate Professor of Pediatrics and Surgery
Director of Primary Care Sports Medicine
Topic: "When Are Children Ready for Organized Sports?"

International patients, please call toll-free 1-877-482-8318

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Markee, whose biological father does not have Chiari, has only an occasional headache. Her Chiari seems to have resolved itself. Heather, on the other hand, anticipates surgery. Chiari has impacted her thyroid, leading her to put on weight, which makes the headaches worse. Frim tells her that she should lose the weight to relieve the symptoms, but the cycle is a catch-22. “Chronic pain destroys their lifestyle and quality of life,” Frim said. “Everyone has a story, but you don’t know it unless you listen.”

Despite long-known evidence of Chiari, the scientific community is only beginning to understand it. In 1883, Scottish physician John Cleland, chairman of anatomy at Glasgow University, published “Contribution to the study of spina bifida, encephalocele, and anencephalus” in the *Journal of Anatomy and Physiology*. It’s believed to be the first public observations of the deformity.

In 1891, Austrian pathologist Hans Chiari advanced Cleland’s findings and classified different variations. In cadavers, he noticed four variations, which he categorized as types I, II, III and IV. The four types involve the cerebellum, but that’s where the similarities end:

- In Chiari malformation Type I, the brain tissue protruding from the bottom of the cerebellum, called “cerebellar tonsils,” extends beyond the structure at the bottom back of the brain (the foramen magnum). This extra brain tissue exerts pressure on the brain stem, which affects speech, swallowing, vomiting and other functions, and often impedes the flow of cerebral spinal fluid, causing a host of other possible symptoms.
- Type II, which always accompanies open spina bifida, consists of tonsils longer than those in Type I and also involves the brainstem, fourth ventricle and the vermis, another part of the cerebellum separate from the tonsils.
- Types III and IV are extremely rare. In Type III, the cerebellum and brainstem descend into the spine and an external sac, creating a fluid-filled cyst. A small or underdeveloped cerebellum characterizes Type IV.

In terms of understanding the condition, “we are where Pasteur was when he found out that you could pasteurize milk,” Frim said. “We know that if you operate, most of the patients get better. We don’t know why,” he said. “We don’t know why some people have the same looking MRIs and don’t have any symptoms at all. We don’t know why some children of parents with Chiari are affected and some aren’t. We don’t know why some people have headaches and some don’t. We don’t know any of those things.”

Frim would like to study many other aspects of Chiari treatment too, but for now, he spends his typical 12-hour day treating patients.

*The previous excerpt is from an article published in the spring 2007 issue of **Medicine on the Midway**. To read the entire article, go to www.uchospitals.edu/news/publications/midway.*

Patient Baylie Owens turns misfortune into gold (bracelets)

Madelyn Kahana, MD, section chief of pediatric critical care at the University of Chicago, owns a star sapphire bracelet given to her by a dear friend who noticed she seldom wears it. Instead, the doctor opts to don beaded bracelets from a patient named Baylie Owen.

Baylie isn’t just any patient. She is perhaps Dr. Kahana’s most famous. Last year, the 7 year old garnered a page in *PEOPLE* magazine in recognition of her efforts to raise



Photo by Audrey Wancket

funds to combat Chiari malformation, a rare condition thought to be genetic. In Baylie’s case, Chiari causes the brain tissue near the base of her skull to extend outside of her head and crown her brain, prompting crippling headaches and other neurological dysfunctions. For many, brain surgery is the only option to stop the pain.

Chiari is relatively unknown. Scientists understand little about the disease. To raise money for research, Baylie started making bracelets, which she sells online for \$5. She, her mother Tressie and supporters have raised more than \$105,000, which they have donated to the research program of David Frim, MD, PhD, chief of pediatric neurosurgery at the University of Chicago Comer Children’s Hospital.

The bracelets proved so successful, Baylie and Tressie recently expanded their product line to include T-shirts, candles and car magnets. They market the awareness-raising merchandise online at www.baylieforbrains.com.

*The previous article was published in the spring 2007 issue of **Medicine on the Midway**. To read *Medicine on the Midway* in its entirety, go to www.uchospitals.edu/news/publications/midway.*

Unusual Case Raises Alert on Smallpox Vaccine

The recent case of a two-year-old boy referred to the University of Chicago Comer Children's Hospital with an unusual rash escalated into a life-threatening situation for the child and raised a red flag for healthcare professionals regarding smallpox vaccination.

A Rare Diagnosis

The affected boy has a history of severe eczema. In late February, he developed a pustular rash on his face, neck and upper extremities, with weeping lesions and an associated fever. On March 3, the boy was seen at a community hospital emergency department near his home in Indiana and then transferred the same day to the University of Chicago Comer Children's Hospital for specialty care.

By March 7, the lesions progressed to umbilicated lesions with an erythematous base. Pustules covered 50% of the boy's skin, including his hands, forearms, neck, chest, face and knees. Eventually, the lesions spread over 80% of the boy's body, and he developed signs of sepsis. The boy's mother also developed pustular lesions on her face and hand, although much less severe. Both mother and child were under stringent infection control measures: they were quarantined together within the University of Chicago Comer Children's Hospital; the air in their room was vented inward; and other measures were implemented to prevent the spread of a potentially communicable disease. In the meantime, fluid samples from the pustules analyzed in the Diagnostic Microbiology Laboratory at the University of Chicago Medical Center were found to be positive for a virus not normally isolated in the laboratory.

At the University of Chicago Comer Children's Hospital, pediatric infectious disease specialists John Marcinak, MD, and Surabhi Vora, MD, pediatric intensivist Madelyn Kahana, MD, and pediatric dermatologist Sarah Stein, MD, led the team providing care for this child. Adult infectious disease physician Stephen Weber, MD, provided care for the mother. In all, many physicians and nurses contributed to care for the child and mother.

As the child's vesicles became larger and puffier, a diagnosis remained elusive. The extensive quantity and uniformity of the lesions was unusual. In fact, the lesions closely resembled photos of smallpox found in medical textbooks. However, the last known case of smallpox occurred 30 years ago in Somalia.

A revelation occurred when doctors learned that the boy's father had received the smallpox vaccine in late January before his scheduled military deployment. After immunization, the father's planned deployment was delayed, so he enjoyed an unplanned visit with his family for three weeks. Suddenly, the puzzle pieces fit together; perhaps the boy's rash was due to vaccinia, the virus used in the smallpox vaccine.



*Dr. John Marcinak, associate professor,
section of infectious disease*

Specimens from the boy's lesions were analyzed at the Illinois Department of Health's Public Health Laboratory using a real-time polymerase chain reaction (PCR) assay for orthopox viruses. Results were positive for vaccinia virus DNA. This finding, in conjunction with the boy's clinical presentation, led to a diagnosis of eczema vaccinatum. Clinical samples were sent to the Centers for Disease Control and Prevention (CDC), and the diagnosis was confirmed: the child had a vaccinia infection.

Treating an Unknown

Since the World Health Organization declared smallpox officially "eradicated" in 1979, today's physicians are not experienced with smallpox or the related vaccinia virus. The only FDA-approved treatment for eczema vaccinatum is Vaccinia Immune Globulin Intravenous (VIGIV), which contains antibodies to vaccinia. VIGIV is held in the Centers for Disease Control's strategic national stockpile for use in the event of a bioterrorist attack. It was delivered to the University of Chicago Comer Children's Hospital by U.S. Marshals.

The University of Chicago Medical Center was the first to give VIGIV to a pediatric patient, the mother was treated with VIGIV also. Her lesions were limited in number and began healing a few days after treatment.

However, the boy's condition was much more severe than his mother's. Doctors at the University of Chicago Comer Children's Hospital were in daily contact with the CDC about the boy's case. Despite the VIGIV treatment, the child's condition worsened. He developed systemic inflammatory response syndrome with renal and respiratory failure. The CDC turned to Siga Technologies, a small research and development drug firm working with a federal grant to develop a drug against smallpox. Designated ST-246, the experimental antiviral drug blocks pox virus replication.

After a Saturday teleconference among Dr. Marcinak, CDC, Siga Technologies and the FDA, an emergency investigational new drug approval was issued for the child's care. Siga's chief scientific officer, Dennis Hruby, flew on a private plane to Chicago and hand-delivered the ST-246 to Comer Children's Hospital. Treatment began Sunday morning. "This was the first infected person to take ST-246, and the first child to use this experimental therapy," said Kenneth Alexander, MD section chief of pediatric infectious diseases at Comer Children's Hospital. After one week of aggressive antiviral therapy, the child's condition began to improve.

Coordination among the parties involved in the child's care was pivotal to a successful outcome. "For over two weeks, we had daily and sometimes twice-daily conference calls with the CDC," says Dr. Marcinak. Every day, the boy's blood samples were sent to the CDC for PCR analysis to measure virus load and anti-vaccinia antibody levels, and to Siga to measure levels of tST-246.

Beyond saving this child's life, the pediatric physicians and nurses at the University of Chicago Comer Children's Hospital were focused on assuring the best long-term outcome for the boy. Like smallpox, the vaccinia virus causes painful lesions that reach deep into the dermis and can affect internal organs. The Pediatric Intensive Care Unit team, led by Dr. Ka-

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Celiac Disease – Awareness and Diagnosis Essential to Treatment

The biggest obstacle facing people with celiac disease is not lack of treatment; it's lack of awareness. A study published in the *Archives of Internal Medicine* [Feb. 10, 2003] found that as many as one in 133 people in the U.S. may have celiac disease, yet only one in 4,700 have been diagnosed – leaving more than 95% undiagnosed and at risk for complications such as osteoporosis, infertility, type 1 diabetes, thyroid disease and cancer. An estimated 1% of the general population may have the disease (and about 35% carry the gene), making celiac disease one of the most common genetic diseases among Caucasians – more common than cystic fibrosis, Crohn's disease and ulcerative colitis combined.

This high rate of prevalence raises the likelihood that pediatricians and other primary care physicians will encounter a patient with celiac disease at some point in their practice, and suggests the need for routine screening among populations at highest risk. The University of Chicago Celiac Disease Center is committed to raising awareness among the medical community and the public about this often unrecognized yet manageable disease. It's not uncommon for children and adults with celiac disease to endure symptoms for months or even years before an accurate diagnosis is made and effective intervention can begin. With children, earliest diagnosis and intervention is essential to limit impairment of the child's growth and development because of nutrient malabsorption.

The University of Chicago Medical Center has one of only two pediatric celiac disease programs in the U.S. Stefano Guandalini, MD, director of the Celiac Disease Center and section chief of pediatric gastroenterology, hepatology and nutrition, has more than 30 years of experience focused on celiac disease in adults and children, and is recognized worldwide as one of the leading authorities on this auto-immune disease – particularly as it affects children. He urges pediatricians and other primary care physicians to “be suspicious” and consider celiac disease when probing for the cause of a patient's symptoms.

Among children, the disease usually presents with classic symptoms that include: recurrent abdominal pain, diarrhea or constipation, frequent vomiting, abdominal bloating and failure to thrive. In older children, delayed puberty, short stature and fatigue are common presentations.

Diagnosis among adults is more elusive. Celiac disease has nearly 250 potential symptoms and rarely presents with “classic” symptoms. Adults' presenting symptoms may not be gastrointestinal in nature, but can appear as skin problems, fatigue, infertility, miscarriage, anemia or a host of other symptoms that commonly are attributed to other causes, such as irritable bowel syndrome. “Celiac disease can be very subtle – especially in adults,” says Dr. Guandalini.

Understanding Celiac Disease

Celiac disease is a genetic autoimmune disease of the small intestine. The impact of celiac disease, however, can reach far beyond the GI tract and affect multiple organ systems. Individuals with celiac disease cannot process gluten, a protein found in wheat, rye and barley. The body's auto-immune response damages villi in the intestine which, in turn, limits the intestine's ability to absorb nutrients. Once damaged, intestinal villi can only be healed by avoiding gluten. In fact, strict,

life-long adherence to a gluten-free diet can completely and definitively restore villi to normal.

Screening for Celiac Disease

Although universal screening is not standard protocol at this time, Dr. Guandalini recommends screening every three years for certain groups at high risk for developing celiac disease. “Don't wait until symptoms develop in those at high risk,” he says. Common risk factors include:

- First-degree relative with celiac disease (gene must be present to develop celiac disease)
- Down syndrome (estimated 10-12% prevalence)
- Type 1 diabetes (estimated 6-10% prevalence)
- Thyroiditis
- Presence of another autoimmune disease
- Sjögren syndrome and other connective tissue diseases (estimated 5% prevalence)
- Turner syndrome
- Williams syndrome

Because of its link to wheat, celiac disease often presents in babies shortly after cereals are introduced to the diet. However, the disease can lie dormant and not become symptomatic until a carrier reaches adolescence or even middle-adulthood (typically 40s).

Primary care physicians can screen with a blood serum test to detect anti-tissue transglutaminase (tTG) and assess total serum IgA level. Any positive blood serum test should be followed up with an esophagogastroduodenoscopy (EGD) and multiple biopsies of mucosal tissue from the duodenum and jejunum. Dr. Guandalini recommends that biopsies are analyzed by pathologists experienced in identifying the subtle tissue changes associated with celiac disease, such as increase in intraepithelial lymphocytes. The EGD with biopsy is considered the “gold standard” for diagnosing celiac disease.

Multidisciplinary Specialists

Adult and pediatric patients referred to the University of Chicago Celiac Disease Center benefit from expert diagnosis and focused multidisciplinary care. The Center currently sees the largest population of celiac patients in the Midwest and maintains referral relationships with nearly every hospital in the Chicago metropolitan area and northwest Indiana. The University of Chicago Celiac Disease Center has one of only two major pediatric celiac programs in the U.S.

The Center's multidisciplinary team is equipped to address a full range of symptoms and potential complications of celiac disease. The Center includes pediatric and adult gastroenterologists with expertise in celiac disease, as well as neurologists, endocrinologists who specialize in type 1 diabetes, dermatologists, hepatologists, infertility specialists, obstetrician/gynecologists and specialists in Down syndrome. Additionally, dietitians and nurses with extensive specialized training are key members of the team.

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Team approach, early intervention saves NF1 patient's sight

Sitting in her pediatrician's office in June 2002, Ann Jongsma watched the doctor count small brown spots on her two-month-old daughter, Jenna.

"When the doctor reached eighteen spots, I guessed I would be taking Jenna to a dermatologist," says Mrs. Jongsma. "Instead, I was referred to a neurologist at the University of Chicago Comer Children's Hospital because the pediatrician suspected that Jenna had neurofibromatosis."

Neurofibromatosis type 1 (NF1) causes hyperpigmented spots on the skin and may lead to a wide variety of additional problems including tumors on nerves. It is one of the most common neurological genetic disorders. One in every 3,000 babies is born with NF1. Like Jenna, most patients first present with café-au-lait spots. Other complications of NF1 can occur later including small subcutaneous neurofibromas and Lisch nodules of the iris in teenagers and young adults. NF1 may also cause problems with bone growth, learning disabilities and short attention span.

James Tonsgard, MD, associate professor of pediatrics and neurology and director of the University of Chicago Ambulatory Program for Neurofibromatosis, saw Jenna shortly after the visit to her pediatrician.

"Jenna had multiple café-au-lait spots, one of the major criteria for the diagnosis of NF1," says Dr. Tonsgard. (See inset for diagnostic criteria). "I suspected that she had NF1 and immediately started her on a protocol of regular exams to check her vision, hearing, spine, long bones, blood pressure, growth and development."

In January 2004, Jenna's ophthalmologist, Michael Kipp, MD, a former University of Chicago medical student and faculty member, now a full-time staff member at the Wheaton Eye Clinic, saw a mild pallor on Jenna's right optic nerve and a preference for using her left eye. Her vision was normal, but an MRI revealed an optic nerve glioma.

At that time, Charles Rubin, MD, associate professor of pediatric hematology/oncology and director of the Brain and Spinal Cord Tumor Center at the University of Chicago Comer Children's Hospital, was asked to join the team treating Jenna.

"Because Jenna had no functional compromise of her vision and because an optic glioma is a low-grade tumor that does not necessarily affect vision, we decided to take a 'wait and see' approach," said Dr. Rubin. Jenna was monitored very frequently with eye exams and regular MRI scans.

Ten months later, in October 2004, Dr. Kipp saw Jenna and noted worsening of her vision. The team decided to treat the tumor with chemotherapy. After receiving twelve months of therapy, Jenna's tumor size decreased, and her vision improved.



Jenna Jongsma on her first day of kindergarten, courtesy of Ann Jongsma

"We feel so lucky that Jenna's tumor was detected when it was asymptomatic," says Mrs. Jongsma. "Because of early intervention and research, our daughter is more likely to enjoy a lifetime of normal activities."

Because signs of NF1 vary significantly from patient to patient and can develop as people grow, Jenna will continue to be monitored closely by all the physicians involved in her care.

"Jenna has had fifteen MRIs and two surgeries at the University of Chicago Comer Children's Hospital," says Mrs. Jongsma. "Because the doctors, nurses and Child Life specialists made her feel her so comfortable through all of her procedures, she looks forward to her visits."

New research directed at diagnosis and treatment of NF

The University of Chicago Comer Children's Hospital is one of nine centers in a newly formed consortium devoted to the understanding and treatment of the complications of Neurofibromatosis. The government-funded Neurofibromatosis Research Program (NFRP) is currently focused on development of new drugs for optic pathway tumors, plexiform neurofibromas and malignant peripheral nerve sheath tumors. Another focus is on the neurocognitive disabilities caused by NF1. Clinical trials will be opening soon and available to patients.

The Neurofibromatosis Program at the University of Chicago is one of the only programs in the country that treats both adults and children. The benefit is that one physician can care for a family affected with NF1; and that physician can follow the child's progress through adulthood. Throughout its 20 year existence, the program has attracted highly specialized physicians such as Dr. Rubin, Dr. Kipp and Mark Greenwald, MD. The physicians of the NFRP have extensive NF1 experience that enables them to anticipate complications and begin appropriate early intervention.

To refer a patient or for a physician consultation, please call (773) 702-6808.

Diagnostic Criteria for Neurofibromatosis Type 1 (2 or more are diagnostic)

1. Six or more café-au-lait spots, greater than 5 mm in diameter in prepubertal and over 15 mm in diameter in postpubertal individuals.
2. Two or more neurofibromas or 1 plexiform neurofibroma
3. Axillary and/or inguinal freckling
4. Two or more iris Lisch nodules.
5. Optic nerve glioma
6. Osseous lesions, such as dysplasia of the sphenoid wing and thinning of long bones that can lead to fracture.
7. First-degree relative (parent, sibling or offspring) with NF1 as indicated by criteria

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hana (the PICU medical director and section chief of pediatric critical care), kept the child sedated through most of his time in the hospital to alleviate the pain and discomfort while the medical team focused on his treatment.

The lesions covered nearly every inch of this child's body. Historically, standard treatment for pox-like lesions is a "dry" approach that allows the blisters to dry, scab over and fall off. However, because the child's lesions were so extensive and deep, physicians decided upon an alternative approach, adapting burn-wound techniques to the treatment of the pox lesions. Dr. Kahana and Lawrence Gottlieb, MD, plastic and reconstructive surgeon at the University of Chicago Medical Center, used a "wet" approach to keep the skin very moist while the lesions healed.

The comprehensive therapy was successful. The lesions healed, and two months later the child had almost no scarring. He was discharged after 48 days in the hospital. "There is little evidence of the war his skin went through," says Dr. Alexander. Today, the boy continues to be monitored by his primary pediatrician in Indiana, with periodic follow-up visits to Dr. Marcinak and Dr. Stein at the University of Chicago Comer Children's Hospital.

Multi-specialty Coordination Crucial to Success

The successful outcome of this challenging case hinged on multidisciplinary coordination among diverse experts in Chicago and across the U.S. At the University of Chicago Medical Center, physicians, nurses and specialists in pediatric infectious disease, pediatric dermatology, pediatric critical care, pathology, microbiology, plastic and reconstructive surgery, infection control and adult infectious disease all contributed to the care of child and mother. Other key participants in the process included scientists and physicians at the CDC; the U.S. Department of Defense; Vincent Fulginiti, MD, (emeritus professor at the University of Arizona and poxvirus expert); the Departments of Public Health from Illinois, Indiana, Chicago and Hammond; the FDA; and Siga Technologies.

A Call for Physicians to be Alert

"There is nothing like this in the literature," says Dr. Kahana. "The military has reported a few cases of seriously ill complications from vaccinia virus, but none to the life-threatening magnitude that this child experienced."

Having observed what this child went through, Dr. Kahana has strong words of warning to physicians. "Modern physicians have no experience with smallpox or smallpox vaccination, yet an index of suspicion should be an important part of the thought process," when encountering a patient with an unusual, pox-like rash. "Ordinarily, we don't think of vaccinia or smallpox. But, now, there are so many people (military and health personnel) being vaccinated for smallpox that we need to ask about a patient's exposure to the vaccine or to a recently vaccinated person." Following the child's diagnosis, public health officials contacted 23 family members and 73 healthcare workers who came in contact with this family during the infectious period to determine if there was additional transmission of vaccine virus.



*Dr. Madelyn Kahana, section chief,
pediatric critical care*

Dr. Alexander reminds health professionals that the military and public health officials have established criteria for smallpox vaccination. Smallpox vaccination is contraindicated in persons with severe eczema or those who are immunosuppressed.

Noting that, "Dr. Marcinak and the team at Comer Children's Hospital brought to bear new technology to address an old problem," Dr. Alexander adds: "If the vaccine can be this bad, heaven help us if the disease (smallpox) ever reappears."

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Supporting Your Patients' Health

Ongoing communication with referring physicians supports effective, long-term management of celiac disease. Pediatricians who refer a child for diagnosis typically receive a letter within two business days regarding the patient's assessment, dietary treatment plan and continuing needs. The patient returns to his or her primary pediatrician for ongoing care, but usually comes to the University of Chicago Celiac Disease Center for initial dietary education and annual follow-up visits.

Living with celiac disease can be a daily, life-long challenge. The Center here offers resources to help children and adults live comfortably and healthfully with this chronic disease. After diagnosis, every patient receives a gift basket with gluten-free foods, a cookbook and resource materials to begin their new gluten-free lifestyle. Our website – www.celiacdisease.net – provides guidance on food choices and accurate information about celiac disease on a 24/7 basis. During business hours, patients and families can call our Information Hotline for personalized answers to their questions.

Research: Search for a Cure

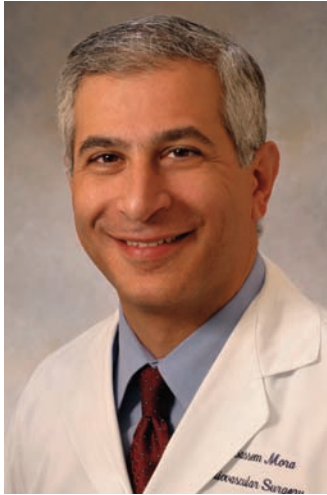
While a gluten-free diet can effectively control symptoms, specialists here continue to search for a medical intervention. Bana Jabri, MD, PhD, – a world-renowned researcher in gut immunology – is studying the disease pathogenesis, including the mechanisms that trigger the auto-immune response to gluten and the cascade of events that follow gluten ingestion. Dr. Jabri's research focuses in particular on the early phases of celiac disease, before extensive damage and complications occur. This dedicated celiac disease research lab is identifying markers that appear normal in mucosa tissue yet signal an inflammatory reaction to gluten.

To Refer a Patient

To refer a patient for diagnosis or consultation at the University of Chicago Celiac Disease Center, please contact: Stefano Guandalini, MD (pediatric patients) or Carol Semrad, MD (adult patients). The Celiac Disease Hotline is 773-702-7593.

Bassem Mora, MD, MBA, is named Chief, Pediatric Cardiac Surgery

Bassem N. Mora, MD, MBA, is a highly regarded expert in pediatric cardiac surgery and congenital heart disease. Dr. Mora performs the full range of surgical procedures to treat congenital heart problems, including corrective and palliative surgery for complex congenital heart disease, hybrid surgery for hypoplastic left heart syndrome, and pediatric heart and lung transplantation. He treats people of all ages with congenital heart disease- from infants and children, to adults with repaired or unrepaired heart defects.



Dr. Mora has extensive expertise in minimally invasive and robotic pediatric heart surgery. These new techniques significantly reduce the recovery time, scarring, blood loss and tissue damage associated with surgery.

Dr. Mora's research interests include the application of genomics and proteomics to congenital heart disease, the study of nitric oxide in the setting of lung transplantation, and outcomes research with respect to pediatric cardiac surgical procedures.

A popular speaker, Dr. Mora has given presentations at numerous medical conferences both in the United States and overseas. In addition, he has published several book chapters and more than 30 peer-reviewed journal articles.

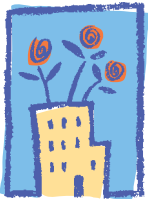
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We would like to extend a personal invitation to visit the all-new University of Chicago Comer Children's Hospital. Our hospital combines a kid-friendly, family-focused environment with advanced diagnostics and state-of-the art medical techniques. Teams of pediatric specialists from multiple specialties work closely together to develop individualized treatment plans for each child, providing the highest quality, often innovative care for conditions ranging from routine to complex.

Comer Children's Hospital is also the home of the newest and most advanced pediatric emergency room in Chicago. Our emergency room includes programs and equipment that are dedicated to the specialized needs of children with cancer, epilepsy, cardiac disease and severe trauma, making it the most prepared pediatric ER in the region to handle the emergency needs of children with complicated diseases.

Come take a look for yourself, please call 773-702-6239 to schedule a personal tour.

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