



# Women & Children's Hospital of Buffalo

*A Kaleida Health Facility*

ISSUE 2 | SPRING 2012

## IN THIS ISSUE:



Dr. Arie Weinstock  
*Pediatric Epilepsy:  
Regional Epilepsy Monitoring Center  
in Women & Children's Hospital*



Dr. Bianca Weinstock-Guttman  
*Pediatric Multiple Sclerosis:  
One of six Centers of Excellence  
nationally*



Dr. Richard Erbe  
*Pediatric Genetics:  
State's Newborn  
Screening Program*



Dr. Michelle Hartley-McAndrew  
*Pediatric Neurology:  
Children's Guild Foundation  
Autism Spectrum Disorder Center*

PEDIATRICS

## PHYSICIANS ON A MISSION

I am incredibly proud and inspired by the accomplishments and expertise of our medical staff at Women & Children's Hospital of Buffalo.

Our physicians have a long and proven history of research and clinical activity resulting in the highest level of care available. Our physician leaders drive our plans for the development and design of how specialized pediatric and women's health care will be delivered for future generations of families in our region. Throughout our history and far into the future, our medical staff will have a significant impact on the health and lives of women and children throughout Western New York and beyond.

In this edition of *Physicians on a Mission*, you'll read about how at our hospital Dr. Robert Guthrie developed the simple and effective test for detecting phenylketonuria (PKU) in 1958. This finding led to our Division of Genetics today as the regional referral center for the New York State Newborn Screening Program led by Richard Erbe, M.D., a founding fellow of the American Board of Medical Genetics.

You'll also learn about our physicians who lead teams of multidisciplinary specialists, including one of the six Pediatric MS Centers of Excellence in the U.S. designated by the National Multiple Sclerosis Society. Our Regional Epilepsy Monitoring Center also provides the broadest range of complex medical and surgical treatments available. Similarly featured is The Children's Guild Foundation Autism Spectrum and Disorder Center, among the first of its kind to use a family-focused and multidisciplinary approach to screen for autism.

Another exciting endeavor is our physician-led plans to construct a new and free-standing children's hospital by 2015. Intensive planning is underway to create a state-of-the-art facility as the newest development on the Buffalo Niagara Medical Campus, a world-class destination for clinical care, research, education and entrepreneurship in downtown Buffalo, New York.

These and other related efforts have led to recognition by *US News & World Report* in its special edition of Best Children's Hospitals. This is not only a great achievement by our medical staff, but also by our nursing, other clinical and support staff within our organization.

Our physicians and everyone else here at Women & Children's Hospital of Buffalo have many accomplishments to be proud of and significant plans to look forward to. On behalf of all of them, I'd like to share with you this edition of *Physicians on a Mission*.

Sincerely,

Cheryl A. Klass  
President





*Women & Children's Hospital of Buffalo has been a regional, national and international leader in providing comprehensive and specialized care to women, children and infants for nearly 125 years. A partnership with the University at Buffalo School of Biomedical Sciences has allowed the hospital to offer its patients the latest medical advancements available. Women & Children's Hospital of Buffalo also serves as the teaching site for University at Buffalo Medical School and has trained hundreds of physicians practicing across the county.*



 **Women & Children's  
Hospital of Buffalo**  
A Kaleida Health Facility

## Dr. Michelle Hartley-McAndrew

*Michelle Hartley-McAndrew, MD is a neurologist and clinical professor of pediatric neurology at the University of Buffalo and Women & Children's Hospital of Buffalo.*

### PEDIATRIC NEUROLOGY:

## Cutting-edge research and care in autism spectrum disorder

When it comes to detecting autism, time is of the essence. Numerous studies find that children diagnosed with the condition early in life have an important head start on therapies that can significantly enhance their development.

At Women & Children's Hospital of Buffalo, the turnaround time to diagnosis is being accelerated, thanks to the Children's Guild Foundation Autism Spectrum Disorder Center. Opened in 2010, the center is among the first of its kind to use a family-focused, multidisciplinary approach to screen for a disorder that affects one out of every 110 youngsters.

During an initial, 90-minute assessment, each child is seen separately by a developmental pediatrician, child neurologist and child psychologist. According to the center's medical director, this coordinated team approach allows physicians to expeditiously diagnose children and then triage them to appropriate treatment services as efficiently as possible.

"All three of us look at the child through a different lens," explains Michelle Hartley-McAndrew, MD, a neurologist and clinical professor of pediatric neurology at the University of Buffalo. "Collectively, we're trying to get a very good sense of that child – their strengths, what's happened as they've grown and at different milestones, and where their parents have seen major changes, regressions and delays. Because there are many factors involved in an autism diagnosis, it's valuable to have each of us offering our perspectives and making an educated assessment."



In New York State, community agencies for children with autism abound. Too often, though, the key that opens the door to such services – namely, a timely, accurate diagnosis – eludes parents.

The diagnosis can be difficult to obtain precisely because the disorders along the autism spectrum are so challenging to pinpoint. Diagnosing a condition characterized by deficits in social interaction and communication can be a formidable task given that young children are still developing speech, social and reasoning skills.

“It’s not a nice, clean test, like if you were a diabetic. A lot of things can look like autism,” Hartley-McAndrew says. “If a child has hyperactivity, for example, they will have a lot of difficulty engaging with you, just as a child with autism would.” Such factors are examined more closely during the Autism Diagnostic Observation Schedule – ADOS, for short – widely considered the gold standard of autism testing.

If a child’s primary-care physician is uncertain whether his patient has autism, he might delay the diagnosis until the disorder definitively declares itself. If the child is truly autistic, valuable time for early intervention is lost.

“Getting that diagnosis out there a lot sooner gives kids a leg up on therapies and an improved outcome,” says Hartley-McAndrew. “Neurologically speaking, synapses are still forming in younger kids – the brain is more malleable, so it’s easier to manage their behaviors and train them in social and language interactions.”

Since it opened in January of 2010, the center has been briskly productive. Some 211 patients between the ages of 2 and 17 were assessed during the first year, half of whom were diagnosed with an autism spectrum disorder. Of those diagnosed, 84 percent were male – not surprising, given that boys are four times more likely than girls to be affected by autism. In the cases where children were not diagnosed with autism, they were found to have behavioral disorders, ADHD, anxiety or other conditions.

While it’s common for pediatricians to refer children to the center, many parents have discovered it on their own. As word has gotten out, families from all over Western New York’s eight-county region, and even New Jersey and Pennsylvania, have found their way here.

“Since we first announced the center’s formation, everyone has been so enthusiastic,” Hartley-McAndrew says. “We have been working collaboratively with many community agencies, and because we know so much about their programs out there, we can pass that information on to families.”

There are a lot of wonderful resources for children with autism,” she adds, “but we want to be the source for their diagnosis. We want to be a funnel for patients to go through, and then we can send them back out into the community with the information they need to get care.”

At the time of diagnosis, the center’s specialists arm parents with everything they need to get their child the requisite services. All receive an extensive guide on how to proceed in the first 100 days after an autism diagnosis as well as the New York State Guide to Special Education, which informs parents of their rights and teaches them how to advocate for their kids at school. In some cases, the center’s social worker offers guidance on school placements and Individual Education Plans. Parents also are invited to attend a monthly support group that the center cosponsors.

At the same visit, they receive individualized recommendations about interventions for their child, both in the community and at the hospital itself.

The Children’s Guild Foundation Autism Spectrum Disorder Center at Women & Children’s Hospital of Buffalo is the only provider of medically based diagnostic services for autism in Western New York; these are complemented by a comprehensive range of pediatric specialty services post-diagnosis, including occupational therapy, speech therapy and genetic testing. As many children with autism experience food aversions, some will be referred to the hospital’s feeding clinic. Those who have obstructive sleep apnea or difficulty regulating their circadian rhythm might be seen by specialists in the sleep lab.

“Everyone at the center is so motivated to help families and their children and to plug them into the resources they need,” Hartley-McAndrew says. “We’re always here to help if they feel like they’re running into a wall. We never want them to feel abandoned.”

Although the center’s primary mission is patient care, physicians are actively engaged in research aimed at better understanding the complex, and increasingly prevalent, autism spectrum disorders. Hartley-McAndrew and her colleagues have embarked on the early stages of a study with the radiology department at Women & Children’s Hospital and the Neuroimaging Analysis Center at nearby Buffalo General Hospital that will study the pathology of autism using the latest imaging techniques

This year, the center hopes to expand on its mission by adding more follow-up sessions to monitor children’s progress and by starting a program for newly diagnosed patients.

“Everybody has been excited about the impact we’ve had so far,” she says. “Parents will tell me, ‘I’m so glad we did this. Now we know what to do.’”

“When you get feedback like that, you know they feel very supported. You know that you’re helping.”



## Dr. Arie Weinstock

*Arie Weinstock, MD, is director of pediatric epilepsy at the Regional Epilepsy Monitoring Center in Women & Children's Hospital and associate professor of clinical neurology at the University of Buffalo.*

Veetai Li, M.D.  
Pediatric Neurosurgery

## Dr. Veetai Li

*Veetai Li, MD, is chief of pediatric neurosurgery at Women & Children's Hospital and clinical associate professor of neurosurgery at the University of Buffalo.*

### PEDIATRIC EPILEPSY:

## Epilepsy is the most prevalent pediatric neurological condition

In the United States, nearly three million individuals live with epilepsy. Despite the dozens of antiepileptic medications available – many that have come to market in the past decade – a third of epileptic patients don't respond to any of them. Among these drug-resistant individuals, doctors estimate, 250,000 are children.

One such youngster came to the Regional Epilepsy Monitoring Center in Women & Children's Hospital of Buffalo. The girl had been diagnosed with epilepsy at age 4. Now 11, she had moved to Buffalo with her family to seek treatment for refractory seizures that had confounded the girl's physician in her native Puerto Rico and wreaked havoc on her life.

"This child was having daily seizures – terrible, out-of-control seizures," recalls Arie Weinstock, MD, director of pediatric epilepsy at the center and associate professor of clinical neurology at the University of Buffalo (UB). "This was a 'catastrophic epilepsy,' so severe that it affected the child's development. In this case, she'd lost her ability to speak."

An MRI revealed extensive lesions on different areas of the girl's brain, but physicians determined that the seizures originated at the left

temporal lobe. Convinced that the largest lesion on the lobe could be removed safely and would dramatically improve the girl's condition, they recommended surgery.

Veetai Li, MD, chief of pediatric neurosurgery at the hospital and clinical associate professor of neurosurgery at UB, performed the left temporal lobectomy. The lesion was found to be a low-grade tumor with surrounding developmental abnormalities. Immediately, the girl's seizures ended. Within months, she'd started speaking again.

### Creating Solutions

Among children, epilepsy is the most prevalent neurological condition, with between 30,000 and 45,000 new pediatric cases diagnosed each year. The Regional Epilepsy Monitoring Center was founded 12 years ago to care for such patients, the only facility of its kind between Cleveland, OH and Rochester, NY.

For the second consecutive year, the unit has been named a Level 4 epilepsy center by the National Association of Epilepsy Centers (NAEC). The highest designation given by the NAEC, a Level 4 center offers the

broadest range of complex medical and surgical treatments for epilepsy.

“We all work together to create solutions for our patients,” Weinstock says of the care team. “There is such a wide spectrum of epilepsy disorders, but there are so many therapies that we can offer – such as safe, effective medications, a specialized diet and epilepsy surgery – that it’s very rewarding to treat.”

In response to patient demand, the center has grown from a three-bed unit to eight beds, but its mission remains the same: to diagnose, evaluate and develop individualized treatment plans for people with epilepsy.

Epileptic seizures take many forms, and medication that suppresses one type may be ineffective on the other. For example, partial seizures, which originate at one part of the brain, respond to some antiepileptic medications while generalized seizures, which affect both cerebral hemispheres, respond to others.

That’s why a primary goal of the center’s staff is to pinpoint the kind of seizure a child experiences. To that end, children are admitted to the center for up to five days, during which time they stop taking anti-epileptic medications. Each of the center’s patient rooms, which a child shares with his parent or another caregiver, is equipped with digital video and EEG monitoring equipment to record brain and physical activity.

Nurses or technicians monitor the video screens 24 hours a day, vigilant to the possibility of a seizure. In the event of one, members of the care team are on hand to help, administering Ativan if necessary. “This diagnostic set-up is the most conclusive to manage and monitor a patient with epilepsy,” Weinstock says. “If a child is to have a seizure, this is the best place to have it.”

In some cases, doctors find that a patient’s spells are not seizures, but actually stress events; these children work with the center’s psychiatrist to improve their symptoms. In other cases, doctors conclude that spells that were believed to be sleep disturbances are, in fact, epileptic seizures.

Once center specialists have evaluated and diagnosed a patient, they make recommendations on a course of care. This may call for tweaks to a child’s medication regimen. For some patients, Vagus Nerve Stimulation (VNS) – in which a pacemaker-

like battery delivers regular, mild pulses of electricity to the brain via the vagus nerve – is an option when medications don’t work.

An increasingly popular method of controlling pediatric seizures requires neither medication nor surgery. It’s all in the diet – albeit an extremely onerous eating plan that must be professionally administered and vigilantly monitored. Women & Children’s Hospital is one of about 150 hospitals in the United States, Canada and other countries whose staff includes specialists – in Buffalo’s case, both physicians and a dietician – trained to counsel parents about the ins and outs of the Ketogenic Diet, a high-fat regimen that’s frequently as effective, and sometimes more effective, than antiepileptic drugs.

The diet works by drastically reducing the amount of carbohydrates a child consumes, thereby tricking the body into a state of ketosis, causing it to burn fat, rather than carbohydrates, for fuel. “The diet simulates fasting,” explains Weinstock. “The fat turns to ketones and the body starts using ketones as energy instead of glucose. And the fact that it’s using ketones reduces seizures.”

The diet is particularly effective for children with seizures resistant to pharmacological treatment, Weinstock notes. Recently, the center’s physicians presented their findings on the diet’s success rate to the American Epilepsy Society. “We found that more than 60 percent of the children on the diet saw significant improvements in their condition and tolerated it very well.”

## Regaining Childhood

For a minority of pediatric patients with refractory epilepsy, surgery is considered. “This is mainly for those who don’t respond to medication and in cases where doing the surgery will likely eliminate the seizures but not cause any harm,” Weinstock says. As it turns out, children make particularly good candidates for this surgery: Research shows that when performed on pediatric patients, surgery may not only end seizures but prevent or reverse the developmental delays that so often accompany epilepsy.

Common procedures include a temporal lobectomy or lesionectomy, the removal of an abnormal lesion in the brain that causes the seizures. In certain cases, the surgeon must perform a two-phase procedure. Initially, electrodes are placed on the brain to pinpoint

## Regional Epilepsy Monitoring Center at a Glance

- Founded in 1999
- Expanded from three beds to eight in response to patient need
- Cares for pediatric patients as well as adults
- Individualized treatment options include medication management, specialized diets, vagus nerve stimulation and surgery
- The care team includes four epileptologists, a specialized epilepsy neurosurgeon, a team of specialists dedicated to advanced imaging and testing, a pediatric neuropsychologist, a nutritionist, a psychiatrist and numerous nurse practitioners and EEG technicians

the seizure foci and map important brain functions, like speech and motor skills. Several days later, the surgeon safely removes the part of the brain from where the seizures arise.

“In patients with temporal lobe epilepsy, there is a 70 percent chance of becoming seizure-free after surgery,” Weinstock notes. “That’s huge because it can greatly improve the quality of one’s life while eliminating the fear associated with epilepsy, the stigma and the fact that you have to be on medication.”

One of the center’s patients, a 16-year-old named Casey, was diagnosed with tuberous sclerosis in early childhood, a condition characterized by numerous brain lesions that resulted in different types of seizures for many years. Casey had tried many medications without success. After a two-stage surgery, however, her seizures ended and she began to plan for the future.

“She told me, ‘Now I can do what other kids do. I can get my driver’s license,’” Weinstock recalls.

From Weinstock’s vantage point, seizures are only the tip of the iceberg for pediatric patients with epilepsy. “You see the seizures, but what you don’t always see is the stigma children experience, the fear of having another seizure, the associated depression and the other comorbidities that occur. So often, the burden of epilepsy is not what you see at first. But we focus on every aspect of epilepsy. Our main goal is to make a difference – to greatly improve a child’s quality of life.”



## Setting the standard for childhood MS care

The late Buffalo neurologist Lawrence Jacobs, MD, devoted his career to improving the lives of adults with multiple sclerosis. From his pioneering research he developed the interferon drug Avonex, the most prescribed treatment for individuals with relapsing MS.

Today, specialists at Women & Children's Hospital of Buffalo are building on Jacobs' legacy with an eye to the youngest MS patients. At the Pediatric MS Center of the Jacobs Neurological Institute – one of six sites that the National Multiple Sclerosis Society has designated a Center of Excellence – they are setting the standard for childhood MS care while untangling the disease's lingering mysteries.



The Center was established in 2006 with a \$1.8 million grant from the National MS Society. Collectively, the six

facilities have built a registry of patients that is yielding important clues into the pathogenesis, demographics and natural history of a disorder that, until recently, was thought to occur only in adults.

"What really revolutionized the recognition of multiple sclerosis in children was magnetic resonance imaging," explains E. Ann Yeh, MD, assistant professor of neurology at the University at Buffalo (UB), who directs the center with Bianca Weinstock-Guttman, MD, also a UB neurology professor. "As MRI technology has been more readily available, we have been able to make the diagnosis earlier."

Yeh, Weinstock-Guttman and their colleagues are now using the same technology to distinguish pediatric MS from the adult-onset disease. In 2009, they reported in the journal *Brain* that children with MS had more brain lesions than their adult counterparts. Measuring two types of brain tissue damage, they found that the overall disease burden was greater in children in the early stages despite the fact that children develop disabilities at a slower pace than adults.

"What we now understand is that while children may look very good on the outside, their brain is affected from the beginning. They will progressively experience loss of reserves," says Weinstock-Guttman.

"They may recover very fast from their initial episodes, so they appear to be doing very well superficially," she adds. "However, because of these brain lesions, progressive deterioration may occur in the long run, after the reserves are depleted. Therefore, we know we have to follow pediatric patients very closely and provide effective preventive therapies."

### Comprehensive Care

In addition to Weinstock-Guttman and Yeh, the Pediatric MS Center staff includes a case manager, a pediatric neuropsychologist and pediatric physical, occupational and speech therapists. Physicians

in pediatric urology, otolaryngology, psychiatry and neuro-ophthalmology provide care on a case-by-case basis.

"I don't think that families realize before they come here that they'll receive this many services," Yeh says. "Those who self-refer to us are worried, and they're trying to understand more about multiple sclerosis. When they come here, they have the pleasant surprise of seeing all areas of their child's illness addressed."

Pediatric-onset MS comprises up to 5 percent of the 400,000 total cases of multiple sclerosis in the U.S. Extremely rare in children younger than 10, the disorder usually emerges during the stormy years of adolescence.

"Being a teenager without a chronic disease is difficult enough, so many of our patients struggle a great deal," says Weinstock-Guttman. "A few are very optimistic and go through without problems, but others become depressed, complaining of sleeping problems and headaches. It is a challenging time, so we are there to hold their hands, to counsel them and to encourage healthy habits."

That includes discouraging smoking, a potential risk factor in the development and progression of MS. Another concern among teenage patients: medication compliance. The center recently produced a DVD for the National MS Society in which youngsters share their experiences with the injectable drugs.

Because MS can compromise a child's ability to think, reason and remember, school can be especially trying. The center's social worker, Mary Karpinski, who co-wrote a handbook about pediatric MS for school personnel, serves as a liaison between families and schools, advocating for educational accommodations based on the child's neuropsychology assessment.

"Schools may not be aware that kids are suffering cognitively because their gross motor skills are intact," Yeh points out. "Motor disability progresses at a much slower rate in kids with MS than in adults, so what their teachers see is a student who, from outward appearances, looks fine."

When symptoms do present themselves, teachers don't usually recognize them as such. Lassitude due to impaired nerve conduction may be mislabeled laziness. Illegible handwriting may be attributed to sloppiness when it, in fact, could be a byproduct of optic neuritis, often the heralding manifestation of MS.

"If kids have been significantly affected by optic neuritis they can't see the lines on notebook paper very well," Yeh says. "As a result, their handwriting may be difficult to read because they don't know where to place the words. It's these kinds of things that we help the schools recognize."

While many of the center's patients hail from Buffalo – which has the second-highest rate of MS in the country – children travel from all over the country, and beyond, for treatment. More than 200 pediatric patients receive MS care at Women & Children's Hospital of Buffalo, about half of whom have multiple sclerosis or another demyelinating





## Dr. E. Ann Yeh

*E. Ann Yeh, MD, director of the Pediatric MS Center of the Jacobs Neurological Institute and assistant professor of neurology at the University of Buffalo.*

## Dr. Bianca Weinstock-Guttman

*Bianca Weinstock-Guttman, MD, director of the Pediatric MS Center of the Jacobs Neurological Institute and a neurology professor at the University of Buffalo.*

disorder, such as acute disseminated encephalomyelitis, transverse myelitis and Devic's disease.

"The majority of our base is from Ohio, Pennsylvania and New York, but we have seen patients from all over the world, including England and Kuwait," Yeh says. "If they experience exacerbations, local patients will come see us in person, but we usually manage those who are farther away via telephone with their doctors. We prefer it if out-of-town patients can make it here for routine visits twice a year, but because of the time commitment, some families can only make it once a year."

When pediatric patients turn 18, they transfer to the William C. Baird MS Center, also part

of the Jacobs Neurological Institute. "Our established adult center was critical to us getting the grant for the Pediatric MS Center," says Weinstock-Guttman, who also directs the Baird Center. "One of the considerations that our pediatric center was built on was that there would be neurologists here with a specialty in adult MS responsible to take over their care."

Just as patients benefit from this seamless care continuum, it's proving advantageous to researchers. Until recently, much of the data about pediatric MS was drawn from adults reporting symptoms they experienced in childhood.

"Everything that we've seen to this point has been retrospective data," Weinstock-Guttman

says. "Although you trust a 40-year-old patient when she tells you that she had her first symptoms at 15, from a research standpoint the data is much stronger when you can follow patients from the get-go."

Doing so, researchers anticipate, will produce far-reaching findings about the disease, in children and adults.

"Kids have had fewer environmental exposures at the time of their disease, so they can teach us about the environmental causes of MS and its progression," Weinstock-Guttman says. "We're turning to this special group of patients, who have started with us so early, to help us find the answers we seek."

## State's newborn screening program

In 1958, Robert Guthrie, MD, PhD – a bacteriologist and physician at what was then known as Children's Hospital of Buffalo – developed a simple and effective test for detecting phenylketonuria (PKU), a genetic disorder that results in mental retardation and other neurological problems when treatment is not initiated within the first few weeks of life.

Guthrie's test came to be used throughout the United States and in dozens of countries worldwide to screen for PKU; what's more, his blood collection system made possible the expanded newborn screening programs conducted today.

Fittingly, in the eight-county region of Western New York, the referral center for the state's Newborn Screening Program is the Division of Genetics at Women & Children's Hospital of Buffalo, located just steps from where Guthrie prepared his revolutionary test.

Division chief Richard Erbe, MD, a founding fellow of the American Board of Medical Genetics and a professor of pediatrics at the University of Buffalo (UB), spoke to us about the division's tripartite mission of clinical care, research and education.

### What kind of clinical care does the Genetics Division offer?

The activities are organized along two lines: One is birth defects and genetic counseling, and the other is inherited metabolic disorders. Within birth defects and genetic counseling, we evaluate and test outpatients for such problems as developmental delay, unusual features, chromosomal disorders such as Down syndrome, autism and other conditions before returning them to their primary physicians for continuing care.

Another outpatient aspect of our birth defects and genetic counseling that's very important is cancer gene testing. For individuals seeking this type of testing, we go through the careful assembly of family history, reviewing the records and then selecting appropriate tests. In metabolic disorders, we receive referrals from family physicians and pediatricians, but most of our referrals come from the New York State Newborn Screening Program.

In addition to these outpatient programs, both our birth defects and genetic counseling program and our inherited metabolic disease program see many inpatients in consultation at Women & Children's Hospital of Buffalo or other hospitals in the region or in nearby regions of Pennsylvania.

### Tell us about the division's staff.

Our staff includes MDs certified in pediatrics and in internal medicine, as well as genetic counselors with specialty board certification. We provide care from conception to advanced age, bringing the full range of genetic services to our region. As treatment of disorders such as cystic

fibrosis has improved, patients with this and other serious disorders have "aged out" of their relationship with their pediatricians. The fact that we're staffed in this multidisciplinary way allows us to continue seeing patients from birth to advanced age. We also have the special capability of seeing adults for conditions that come along a little later. These would include breast and ovarian cancers, metabolic disorders of adulthood, neurologic disorders such as Alzheimer's disease and others.

### Although newborn screening now detects infants who may have any of 30 or more disorders, the most common referral you receive from it continues to be infants with a positive screen for PKU, the progressive disease characterized by the body's inability to metabolize the amino acid phenylalanine. What kind of research into PKU are specialists within your division engaged in?

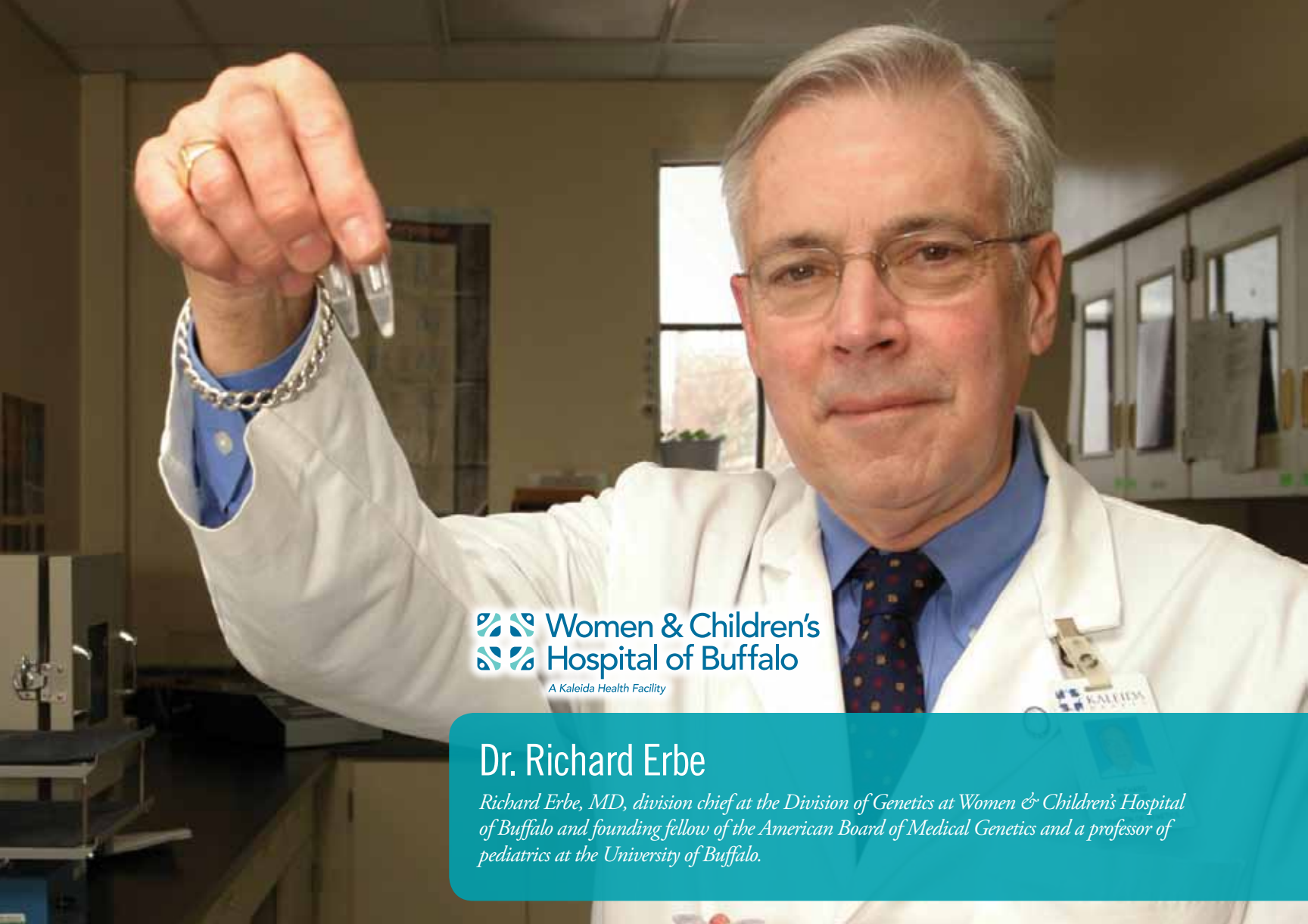
The treatment of PKU has changed little in the past 25 years. Although effective in preventing mental retardation in children, it has been found to be less than fully effective by the time the person is an adult. Even worse, many older children and adults abandon the therapeutic diet because of its severe limits on dietary choices. We are joining in a large, nationwide study to find improved treatment methods for PKU while allowing for a more palatable diet. All told, we have about 80 different families in Western New York that we're following who have kids with PKU. Although there's considerable understanding of the disease, there are still many unanswered questions, like why siblings affected with the same genetic defect seem to have quite different outcomes, or why some kids with high phenylalanine concentrations still seem to do fine in school and in others areas.

### In 2006, screening for Krabbe disease, a rare lysosomal disorder, was added to the New York State Newborn Screening Program. This was largely due to the advocacy work of former-Buffalo Bills quarterback Jim Kelly, whose son died of the disease at the age of 8. How is the Genetics Division helping us better understand this disorder?

We are part of the Krabbe Consortium of New York State, which brings together the eight inherited metabolic disease centers across the state to refine the diagnosis and management of infantile Krabbe disease and enhance long-term follow-up of patients. The consortium was established by Dr. Patricia Duffner, a pediatric neurologist at Women's and Children's Hospital of Buffalo and UB. This is the first time that new testing mandated by a state has been evaluated by a consortium of physicians.

Lysosomes, structures that are present in most cells, contain 40 enzymes that are important in metabolism. Any of these can be made deficient





 **Women & Children's  
Hospital of Buffalo**  
A Kaleida Health Facility

## Dr. Richard Erbe

*Richard Erbe, MD, division chief at the Division of Genetics at Women & Children's Hospital of Buffalo and founding fellow of the American Board of Medical Genetics and a professor of pediatrics at the University of Buffalo.*

by inherited mutations. Replacement enzymes that can be given intravenously to slow progression of symptoms have been developed for four of the 40 disorders, so it's key to find patients as early as possible through newborn screening. Krabbe Disease is not one of the disorders for which enzyme therapy can be used, but bone marrow transplant has been used. Screening of more than a million newborns in New York State has identified 300 considered at high risk for Krabbe Disease, of which three were referred to Duke University for transplant and one was successfully transplanted.

### What kind of outreach is the genetics division involved in?

We're a key part of the hospital's Lung and Cystic Fibrosis Center, and Sickle Cell and Hemoglobinopathy Center, as well as the Muscular Dystrophy Association. Our genetic counselors participate in these clinics and share our expertise, partnering

with the physicians at these centers to educate and provide care to patients. We also provide prenatal diagnosis and genetic counseling to the Women's Health Center at the hospital.

### How is your division helping educate the next generation of physicians understand genetics?

We teach in the first- and second-year courses at the University at Buffalo's medical school, and we have a month-long elective for fourth-year medical students, pediatric residents and internal medicine residents. We also teach in quarterly sessions for the half of the region's internal medicine residents in the Catholic Health System.

### How has the completion of the Human Genome Project, our better understanding of DNA and other advances transformed genetics?

There are many more tests than ever before, and they are much more powerful. We have progressed from only being able to count chromosomes and tell one chromosome from another to being able to identify defects in the function of nearly every gene. Although our ability to diagnose based on single genes is enormously improved, the major challenges are now focused on how these genes work together under environmental influences to produce predispositions and disorders, and how we can translate this new knowledge into prevention and treatment. The new molecular techniques give us almost too much detail, and this will require a lot of digesting before its full benefit can be realized. The Division of Genetics aims to facilitate the transfer of this new knowledge to all of our professional colleagues and patients.

© 2012 Kaleida Health. All rights reserved.

## Women & Children's Hospital of Buffalo

219 Bryant Street  
Buffalo, NY 14222  
wchob.org  
(716) 878-7000



## IN THIS ISSUE:

### 2 **Pediatric Neurology:**

Children's Guild Foundation  
Autism Spectrum Disorder  
Center

### 4 **Pediatric Epilepsy:**

Regional Epilepsy  
Monitoring Center in Women  
& Children's Hospital

### 6 **Pediatric Multiple Sclerosis:**

One of six Centers of  
Excellence nationally

### 8 **Pediatric Genetics:**

State's Newborn  
Screening Program