

Hunting newborn tests for super-rare gene diseases

WASHINGTON (AP) — At his first birthday, John Klor couldn't sit up on his own. A few months later, he was cruising like any healthy toddler — thanks to a special diet that's treating the North Carolina boy's mysterious disease.

What doctors initially called cerebral palsy instead was a rare metabolic disorder assaulting his brain and muscles, yet one that's treatable if caught in time.

Urged by John's family, Duke University researchers are working on a way to test newborns for this disease, called GAMT deficiency. It's part of a growing movement to add some of the rarest of rare illnesses — with such names as bubble-boy disease, Pompe disease, Krabbe disease — to the battery of screenings given to U.S. babies hours after birth.

"There's other children out there that can be helped and be saved," says Melissa Klor, John's mother.

"Families go through these odysseys of diagnosis" to learn what's wrong with a child, says Dr. Alan Fleischman of the March of Dimes, who's part of a government advisory committee studying what to add to the national screening list. Often, "they argue that they would have been better off knowing even if there were no treatments."

Since 2004, specialists have urged that every U.S. newborn be tested for 29 rare but devastating genetic diseases, using that single heel-prick of blood, to catch the fraction who need fast treatment to avoid retardation, severe illness, even death. States gradually adopted those recommendations, and federal



In this photo provided by the family, John Klor, then-16-months old, of Pine Knoll Shore, N.C., pushed a toy in the photo taken Sept. 12, 2009. (AP Photo/Klor Famil. HO)

health officials say the testing catches about 5,000 babies a year with disorders ranging from sickle cell anemia to maple syrup urine disease and others with such tongue-twisting names that they go by acronyms like LCHAD.

John Klor's illness is too new for that list. By the time her son was 6 months old, Melissa Klor knew something was wrong. John missed developmental milestones, unable to sit, stop his head from wobbling, or babble. He regressed, quitting rolling over. He stared blankly for moments at a time, a kind of mini-seizure.

A neurologist diagnosed cerebral palsy. But John never had an MRI scan to prove the diagnosis, and Klor eventually sought a second opinion. Right after John's first birthday came the news: His brain scan showed no sign of

cerebral palsy, but he might have any of a number of degenerative metabolic disorders.

In a lucky break, John's blood and urine were sent to Duke's genetics laboratory for specialized testing that found he couldn't process protein correctly. John's body wasn't producing a substance called creatine that's crucial for providing energy to the brain and muscles, leading other protein metabolites to basically clog his system and damage his brain.

Creatine deficiency syndromes weren't discovered until 1994; Duke is one of the few labs able to diagnose them. Fortunately, John's version — called GAMT deficiency for the enzyme, guanidinoacetate methyltransferase, that his body lacks — is treatable in the young.

Doctors ordered a vegan diet — only fruits, vegetables and specially processed pastas — with no more than 6 grams of protein daily. John drinks a formula containing creatine and other missing nutrients.

"Within days, we started to see him getting stronger," says Klor, of Pine Knoll Shores, N.C.

Today at 19 months, John runs and climbs stairs. He's starting to make sounds like "ma" but speech is coming more slowly; doctors are optimistic but make Klor no promises.

Only 40 cases of GAMT deficiency have been reported in medical journals, but Duke specialists say creatine disorders probably are underdiagnosed, with symptoms similar to other metabolic diseases. GAMT deficiency may eventually be a candidate for newborn screening, although it's not yet clear if the troublesome substances will show up in blood at birth or if a different test will be required, cautions medical geneticist David Millington. His lab is studying that now.

HEALTH TIP

Managing eczema

Eczema is a condition that results in red, itchy and scaly patches of skin — often on the scalp, forehead, face or extremities.

The American Academy of Dermatology offers these suggestions help manage the uncomfortable symptoms of eczema:

Make an appointment with a dermatologist to confirm your diagnosis and get treatment.

If your doctor recommends so, use a topical prescription medication on the skin, such as a corticosteroid or a medication that affects the immune system.

An antihistamine also may help. Ask your doctor about ultraviolet light (UV) therapy.

Apply a skin moisturizer. (Source: HealthDay News)

Sleep disorders

plague cancer patients

Insomnia and sleep disorders affect more than three-quarters of cancer patients undergoing chemotherapy, a rate nearly three times higher than that of the general population, a new study finds.

The problem is more common in younger patients and in those with lung and breast cancers, said University of Rochester Medical Center researchers, who assessed sleep problems in 823 cancer patients.

Questionnaires completed by patients after their first two chemotherapy treatments revealed that 37 percent suffered from insomnia symptoms and another 43 percent had insomnia syndrome, which means they had difficulty falling asleep and staying asleep at least three nights per week, the researchers reported in the Nov. 23 online edition of the Journal of Clinical Oncology.

"These numbers are very high, and something we can't ignore," study author Oxana Paless, a research assistant professor of radiation oncology, said in a news release from the medical center. "The good news is that insomnia is a very treatable problem that can be addressed quickly so it doesn't compound other symptoms."

Sleep problems — which are generally combined with fatigue and depression — haven't typically been studied to determine their causes and impact on patients' quality of life, she noted. (Source: HealthDay News)

Missing gene tied to bone marrow transplant problems

Researchers are reporting that a genetic variation — a missing gene — boosts the risk that someone who's had a bone-marrow transplant will develop a complication called graft-versus-host disease.

The immune system-related complication occurs after surgery when the recipient has the variation and the donor does not.

When that's the case, the immune cells from the donor go on the attack against the recipient's tissues.

"This finding gives us a glimpse into the genetic incompatibilities that can complicate transplants," the study's lead author, Steven McCarroll, an assistant professor at Harvard Medical School and an associate member of the Broad Institute of MIT and Harvard, said in an institute news release. "There are likely many other compatibility loci left to be found, and with the enhanced capabilities for surveying human genetic variation, it will become increasingly feasible to find them." (Source: HealthDay News)

Human trials of artificial artery

An artificial artery is set to be tested in human trials early this year.

London's Royal Free Hospital used nanotechnology to develop the small bypass graft from a polymer material.

The material enables the graft to mimic the natural pulsing of human blood vessels, which enables them to deliver nutrients to the body's tissues.

The ultimate aim is to use the graft in coronary artery and lower-limb arterial surgery, which doctors say could reduce amputations and heart attacks.

If the trials — funded by a grant of £500,000 — are successful, the device could potentially help thousands of patients with vascular disease.

The wall of the artery is designed to be able to withstand blood pressure throughout a person's lifetime and is normally very strong.

If it is damaged by disease such as hardening of the arteries, the artery can become blocked or in some patients the wall can weaken, becoming an aneurysm, and it may rupture.

The current surgical treatment is to bypass or replace the damaged vessel using a plastic graft or preferably a vein taken from the patient's own leg.

But many patients do not have suitable veins.

The plastic grafts were originally made with the same nylon used to make "drip-dry" shirts. But although they work well for larger grafts, they are less successful for grafts of less than 8mm.

This is because these materials cannot pulse and their surfaces stimulate clotting of the blood in the graft.

Researcher Professor George Hamilton said: "There is a high



Professor Hamilton with the artificial artery

failure rate using these rigid, small diameter bypass grafts.

"Many patients who have needed smaller bypass grafts, but have not had suitable veins, have had limbs amputated and some patients unable to have coronary bypass surgery have had heart attacks and died."

The new artificial artery has been designed to mimic the natural version as closely as possible.

It is strong, flexible, resistant to blood clotting and pulses rhythmically to match the beat of the heart. Using nanotechnology, the researchers incorporated specific microscopic molecules into the graft.

Some aid circulation, while others encourage specialized stem cells to coat its lining, boosting its ability to repair damaged blood vessels still further.

Professor Hamilton said: "This will be hugely beneficial to patients in the NHS as we will be able to reduce heart attacks, reduce amputations and ultimately save lives."

In the long-term, the team hope to develop a range of "off the shelf" grafts, stents and other devices.

Judy O'Sullivan, of the British Heart Foundation, said: "We welcome this interesting development which could potentially be of enormous benefits to patients who need a bypass operation to treat their coronary heart disease."

"The availability of suitable artificial arterial grafts would mean that more patients could benefit from the bypass operation whilst avoiding having a wound following removal of a patient's own veins or arteries to be used as grafts, as current practice requires." (Source: BBC)

Many ignorant on waist fat risk

Almost nine in 10 people are not aware of the risks of carrying extra fat around their waistline.

A survey of 12,000 Europeans found most had no idea that a thick waist was a sign of a build-up of a dangerous type of fat around the internal organs.

The report from GlaxoSmithKline, who make weight loss drug Alli, said this "visceral fat" is strongly linked with type 2 diabetes and heart disease.

Most people would lose weight once they found out the risk, the survey found.

Report author Dr. Terry Maguire, honorary senior lecturer at Queen's University in Belfast, said people did not know that visceral fat, which you cannot see or feel and which sits around the organs in the abdomen, is there or that it poses a problem.

It is thought that the danger of visceral fat is related to the release of proteins and hormones that can cause inflammation, which in turn can damage arteries and enter the liver, and affect how the body breaks down sugars and fats.

Only a quarter of those questioned in the Europe-wide study thought being overweight was a risk to long-term health at all.

"Most overweight people still see themselves as having a body image issue not a health problem and they need to understand the health benefits of weight loss as well as the cosmetic results," he said.

Research has shown that waist circumference is a good indicator

of visceral fat and therefore of a person's risk of diseases associated with being overweight, such as type 2 diabetes.

The report pointed out that when weight is lost visceral fat is more easily broken down for energy than the fat immediately under the skin and even a small amount of weight loss can cause a difference.

When asked about losing weight, two-thirds of respondents said they would go on a diet in the New Year.

But the report's co-author Professor David Haslam, chair of the UK National Obesity Forum, cautioned that steady sustainable weight loss is important and that crash diets were likely to be unsuccessful.

"They can actually do more harm than good," he said.

It comes as the Department of Health announced that more than 300 of the 1,500 babies who were likely to have been born this New Year's Day could be overweight or obese by the time they start school unless action is taken.

Professor Steve Field, chair of the Royal College of GPs, said most of the focus in recent years had been on weight.

"It is the weight around your belly which really does the harm. A lot of these things take a while to get into people's heads especially as there has been so much focus on weight and body mass index."

"I'm not surprised at the findings because it will take more than a few academic papers to really change people's minds." (Source: BBC)

25,000 Britain-trained doctors to leave for India

NEW DELHI (AFP) — Nearly 25,000 British doctors of Indian origin are set to emigrate to India within two to four years, a report said Monday quoting a British medical association.

"There are around 15,000 young Indian-origin doctors undergoing training in different parts of Britain who will return to India," Ramesh Mehta, president of the British Association of Physicians of Indian Origin,

told media in New Delhi. "Also, at least 10,000 senior doctors of Indian origin who are retiring from their jobs in the UK are set to return to India," the doctor said, according to India-based The Economic Times.

Mehta said many of them were expected to take up positions in seven new elite medical centers for which the Indian health ministry is looking for quality doctors.

For baby and mom alike, breast-feeding may be best

Reports on the benefits of breast-feeding continue to accumulate as researchers evaluate the breast-over-bottle option.

It's been shown to help a baby's later performance in school, to reduce the odds of problem behavior and to help kids cope with stress. And moms stand to benefit later on as well, studies show.

But what is it about breast-feeding that's so helpful and healthy?

For starters, breast milk is loaded with health-promoting nutrients. "It's not just one mechanism," said Melinda Johnson, a spokeswoman for the American Dietetic Association, a lecturer in nutrition at Arizona State University and a dietitian in private practice in nearby Chandler, Ariz.

"The nutrition (provided by breast-feeding) is perfect for the growing child," Johnson said. Take, for example, DHA (docosahexaenoic acid), an omega-3 fatty acid. "DHA is critical for brain development and also for nervous system development," Johnson said.

The presence of DHA in breast milk, she said, might explain the finding that breast-fed kids do better academically.

Breast milk also contains the amino acid taurine, considered important for neurological development, said Dr. Ruth Lawrence, who chairs the American Academy of Pediatrics' section on breast-feeding and is a professor of pediatrics and obstetrics-gynecology at the University of Rochester School of Medicine and Dentistry in New York.

"Newborns and preemies cannot manufacture taurine," Lawrence said, although adults do. "Taurine is one of the amino acids needed for brain growth. The brain will double in size in the first year of life." That makes it critical to have nutrients that help brain growth.

"We in the breast-feeding field have been focusing on brain growth (and its importance) for a number of years," she said. Those who manufacture formula, Lawrence said, focus more on how much weight babies can gain with their product.

Breast milk also has been shown to jump-start a baby's immune system, and researchers think that's due at least in part to a protein found in breast milk. Called soluble CD14, it helps develop beta cells, a type of immune cell that helps produce antibodies, which are needed to protect against illnesses.

Breast milk also contains live and active organisms that can never be duplicated in formula, Johnson said. In one of the newer areas of research, experts have found that breast-fed babies' guts have different bacteria than those of formula-fed babies, and that the breast-fed babies' gut bacteria appears to be healthier, she said.

Other research has found that the intestinal bacteria present early in life play a role in whether a person will suffer from allergies, have an overactive immune system or tend to put on excess weight later in life, Johnson said. Breast-feeding also has emotional and bonding benefits, according to Lawrence and Johnson, although they say it's harder to explain the "why" and "how" of those.

Though a mother who bottle-feeds also holds her baby, the child has actual physical attachment while breast-feeding. "Certain hormones, feel-good hormones, are released when a woman is breast-feeding," Johnson said, citing oxytocin and prolactin as examples. "The theory is, that's how the moms bond."

(Source: HealthDay News)