Study Seeks to Improve Early Detection of Joint Bleeds in Children with Hemophilia

For patients with hemophilia — an inherited disorder marked by the absence or reduced levels of certain proteins, or clotting factors, instrumental to the body’s ability to control bleeding — recurrent internal bleeding, especially into the knees, hips, ankles and elbows, is a common and often painful problem. If left untreated, it can lead to severe joint disease and, eventually, arthrosis.

Fernando F. Corrales-Medina, M.D.

Fernando F. Corrales-Medina, M.D., assistant professor of pediatrics at the University of Miami Miller School of Medicine, pediatric hematologist at the Hemophilia Treatment Center, and associate program director of the Miller School’s Pediatric Hematology/Oncology Fellowship Program, is leading a five-year, longitudinal study that aims to minimize the lifelong joint impairment caused by hemophilia, a condition known as debilitating chronic hemophilic arthropathy.

Over the past few decades, doctors have made great strides in treating and preventing clinically evident joint bleeds by using lifelong prophylaxis regimens, a regular schedule of intravenous infusions of the clotting factor that patients lack. But even with a lifetime of prophylaxis and with seemingly normal joints, a number of young adults with hemophilia are now developing arthropathy in their 20s and 30s.

"Many joint bleeds are obvious: the joint becomes swollen, hot and red, and patients are unable to extend the joint or bear weight," Corrales-Medina said. "On the other hand, hard-to-detect microbleeds, which are invisible and even asymptomatic, need to be identified and treated as soon as possible to prevent long-term joint damage."

Corrales-Medina said the study, titled “Evaluation of Joint Arthropathy Using Ultrasound Technique in Children with Severe Hemophilia Undergoing Prophylaxis Regimen,” is the first of its kind. "We're using ultrasound for early detection of microbleeds and to monitor their progression in our youngest patients — newborns and infants up to 18 months old — who have severe hemophilia and are undergoing routine prophylaxis regimen."

The multicenter study, funded by a $750,000 grant from Baxalta, now part of Shire, a global biotechnology company, is being conducted at different sites, including the University of Florida, University of Kentucky, and Tulane University, with the University of Miami Miller School of Medicine serving as the primary site.

Patients enrolled in the study are followed every six months for five years, at no cost to the families, to monitor the progression of the joint disease and determine if ultrasound is the most efficient imaging
method for identifying joint abnormalities. The study is already providing valuable data for Corrales-Medina and his team.

“Ultrasound has been shown to be extremely accurate in detecting both microbleeds in hemophilia patients and early changes in the tissues lining the joints,” he said. “It’s a far better option than X-rays, which don’t pick up the first variations in the joint itself and require radiation, or MRI scans, which are too costly and, for small children, require intravenous sedation or even anesthesia. With ultrasound, parents can remain with their toddlers and keep them calm and still during the procedure.”

Corrales-Medina says this study can even potentially establish a new standard for factor infusion dosing. “By detecting early changes in a patient’s joints, we can adjust their infusion therapy and, hopefully, prevent irreparable joint damage before it’s too late,” he said. “What we’re learning from this study has the potential to change the way hemophilia is treated worldwide.”

The study is one of many underway at the UM Hemophilia Treatment Center (HTC), established in 1987 as part of the Department of Pediatrics at the Miller School of Medicine. Led by its founder, Joanna Davis, M.D., associate professor of clinical pediatrics, UM-HTC is the only federally funded pediatric hemophilia center in South Florida. Its physician-scientists are engaged in leading-edge research efforts and are currently treating more than 450 patients of all ages, from across the United States and around the world.

Davis says that the HTC offers patients, with bleeding and thrombosis disorders, state-of-the-art, comprehensive care and access to clinical trials available only at an academic medical center such as the Miller School of Medicine.

“Here at HTC we have, under one roof, a multidisciplinary team of hematologists, social workers, nurses, physical therapists, psychologists and orthopaedic physicians,” says Davis. “We’re all working together to help our patients better manage their disease and improve their quality of life.”

More information about the Hemophilia Treatment Center is available here. To schedule an appointment, call 305-243-5302 and select option #3.