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Improving newborn screenings and care for rare diseases

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The birth of a new child is an exciting, anxiety-inducing, life-changing event. Among the flurry of physical health tests and introductions to your new family addition, your child is screened for a litany of rare diseases in his or her first hours of life. For most parents, this newborn screening is an imperceptible blip on life's radar, but for a minority, it can mark the beginning of the most difficult journey of their lives.

Newborn screening for rare and often fatal diseases is surrounded by issues that University of Rochester Medical Center child neurologist Jennifer Kwon, M.D., M.P.H. will tell you are anything but simple. One issue Kwon finds concerning is that patients with rare diseases are given care that is variable across the nation; some care may be good, but some may not.

"My goal is to make sure that the care of children identified by newborn mandatory screening is optimal and reasonable," says Kwon, associate professor in the departments of Neurology and Pediatrics. "If we are going to have a mandatory newborn screening program, then we really need to pay attention to how identified patients are cared for."

Cystic fibrosis (CF) is a rare genetic disease that is universally screened for at birth across the US, and thanks to the establishment of a CF registry we've seen consistent improvements in the health of patients over the years. In cystic fibrosis, abnormally thick bodily fluids cause frequent lung infections and difficulty breathing, and death in early adulthood. CF patients are living much longer than they could have

expected just 15 years ago, and Kwon credits the CF registry program, which ensures "very consistent care, and very tight control of care."

Patient registries compile information on patients' health that is shared amongst doctors across the US, allowing them to see which treatments are working and to aid them in the design of clinical trials to test new treatments. In the end, registries help set guidelines for treatment that translate to patients receiving the same level of care, no matter where they are in the US.

Pompe disease, another rare genetic disorder characterized by progressive weakening of muscles and damage to other organs starting in infancy (early onset) or between late childhood and early adulthood (late onset), is currently screened for in a handful of states and has been recommended for nationwide newborn screening. While there are national efforts to develop a newborn screening clinical follow-up registry for Pompe, they are still in the early stages.

Pompe patient care after diagnosis is very variable in those states that screen for Pompe at birth.

Without a registry to guide them, doctors are left guessing at the best ways to treat their patients. Patients' families also have no way of knowing if their child's care is optimal. They can't simply Google their treatment, as you could for a urinary tract infection, to see how it compares to how other people are usually treated for that condition.

Kwon believes that developing a national registry for Pompe and other rare diseases would give patients and their medical providers the opportunity to compare and improve their medical care.

Establishing a registry for Pompe is a particular focus of an upcoming meeting organized by Kwon titled, "Newborn Screening for

Neuromuscular Diseases: Improving Patient Outcomes and Evaluating Public Health Impact". This University of Rochester-sponsored, international, two-day conference will bring clinicians together to discuss how best to implement newborn screenings and care for patients after diagnosis.

The meeting, which starts today in St. Louis, MO, showcases Kwon's dedication to her patients with rare diseases at UR Medicine's Golisano Children's Hospital. Though registries are expensive to set up and maintain, she believes they "are the best way to help us understand what happens to these children over their lives."

Provided by University of Rochester Medical Center