

Developments in science help families have healthy children

Addison Strobel is a happy and healthy baby. But the outcome might have been very different had she been born prior to the development of a new screening process called preimplantation genetic diagnosis. Addison's parents, Jane and Tim, both carry a recessive gene that can cause autosomal recessive polycystic kidney disease. Addison was the first baby born after using preimplantation genetic diagnosis.

Polycystic kidney disease is a rare genetic disorder that affects 1 in 10,000 people. The disease is chronic and progressive, and eventually causes kidney failure and liver abnormalities. There is no cure for the disease. It often is lethal in newborns. Nearly 50 percent of newborns with this problem die after birth – not from kidney failure, but rather from complications of underdeveloped lungs. If newborns survive, they often struggle with large, cystic kidneys.

Researchers at Children's Research Institute and The Medical College of Wisconsin developed preimplantation genetic diagnosis as a screening program to identify healthy embryos and eliminate the disease. This groundbreaking procedure creates a worldwide resource for families to deliver a healthy infant, rather than accept

(over)



The Strobel family



2009 research highlights

-  More than 940 clinical trials in 2009.
-  The Neurology Program at Children's Hospital of Wisconsin began offering a new pharmacogenetic test for patients with new onset seizures. Children's Hospital is one of only a few institutions in the world offering the test.
-  Investigators at Children's Hospital were featured in the *New England Journal of Medicine* for discovering a new type of inflammatory disease in newborns.

the 25 percent possibility of having an infant born with this polycystic kidney disease. In a laboratory, embryos are formed using in-vitro fertilized eggs and sperm. Then, polycystic kidney disease embryos are analyzed using preimplantation genetic diagnosis to ensure they are disease-free. If so, they are implanted into the mother.

When both parents, such as Jane and Tim, have the recessive gene, they can pass the disease on to their child. Tragically, polycystic kidney disease caused the death of Addison's brother. Had the Strobels not used preimplantation genetic diagnosis, they would have run the risk of having another child born with the disease.

The preimplantation genetic diagnosis screening program resulted from an 18-month collaboration between Children's Research Institute and The Medical College of Wisconsin. "This research gives parents the peace of mind to know they have done everything possible to ensure their baby is born healthy," said Ellis D. Avner, MD, director, Children's Research Institute; professor, Pediatrics and Physiology, associate dean, Research, the Medical College.

While preimplantation genetic diagnosis is key in screening for autosomal recessive polycystic kidney disease, it also can be used to identify other serious medical conditions such as cystic fibrosis, Down syndrome and sickle cell disease. Identifying the embryos that carry these disorders can have a significant impact on the health of children and their families.

In 2009, Children's Hospital invested more than \$10 million in research to help improve the health of children.

The preimplantation genetic diagnosis program collaboration included:

- Ellis D. Avner, MD, the senior editor of the newly released sixth edition of *Pediatric Nephrology*, the leading reference on childhood kidney diseases.
- David Bick, MD, medical director, Genetics, Children's Hospital; professor, the Medical College.
- Eduardo C. Lau, PhD, investigator, Genetics, Children's Research Institute, assistant professor, the Medical College.
- Estil Y. Strawn Jr., MD, director, Reproductive Medicine and Fertility, Froedtert & the Medical College; professor, Obstetrics and Gynecology, the Medical College.

How you can help

Children often are impacted by local, state or federal policies that affect their health care. They need others to be their voice, which is why these efforts rely on the strength and action of people who want to make a difference for children.

Join Children's Hospital of Wisconsin's Children's Advocacy Network today and help us make a difference in the lives of children and families in our communities. Through CAN, you help ensure that children's concerns are heard when policies are being made. The network sends members action alerts and legislation updates and provides tools that you need to help you take action on issues important to children's health.

Join CAN at chw.org/can.

