

# Region 4 Family Newsletter

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## Enter Your Contact Information on the Region 4 Genetics Collaborative Website!

If you have not entered or updated your contact information please go to [www.region4genetics.org](http://www.region4genetics.org) and click on the "Region 4" tab, then "Member Directory" under Additional Resources. From there you can update or add your contact information. Keep in mind that your new information will not show-up right away, it needs to be approved first.

Thank you!

## Region 4 Stakeholder Meetings

Michigan – March 23

Kentucky – April 14

Ohio – April 15

Wisconsin – April 22

Indiana – April 29

Illinois – April 30

Minnesota – May 15

## Upcoming Workgroup Meetings...

Priority 2 3/20

Care Coordination 3/24



## Calling all parents who have children with heritable conditions!

Parents who have children with heritable conditions are invited to participate in an all day stakeholder meeting in their state on genetics and newborn screening. The stakeholder meetings, hosted by the Region 4 Genetics Collaborative, will be held in Illinois, Indiana, Kentucky, Ohio, Michigan, Minnesota and Wisconsin. They will include discussions on a variety of topics about how our region is

working to improve the lives of children with heritable conditions and their families.

Professionals such as genetic counselors, pediatricians, physicians and genetic specialists will also be attending. After the stakeholder meeting, parents are invited to participate in a facilitated discussion focusing on how having a child with a heritable condition effects your family. The experiences and thoughts of families

are very important to help professionals understand family needs and issues. The Region 4 Genetics Collaborative will reimburse parents for travel costs to and from the summit and will be given \$50 for their participation. Breakfast and lunch will be provided. For more information or if you are interested in registering, please contact Sarah Wedepohl at [s.wedepohl@yahoo.com](mailto:s.wedepohl@yahoo.com) or (248) 761-2553.

## Access to genetic services survey for families

Region 4 recognizes the importance of learning about perceived family needs before developing a plan to improve access to genetic services. Region 4 is partnering with the Michigan's Birth Defects Registry and Children's Special Health Care Services programs to survey families of children with heritable conditions in Michigan.

The survey will assess families' knowledge of and perceived need for clinical genetic services. Region 4 will provide incentives to parents to complete the survey. The survey will be sent to a sample of families in Michigan.

If you do receive the survey, please take some time to complete it and to help our region better understand the

needs of families! The information families provide will be used by the genetic expertise work group to explore family issues on access to genetic services.

## Genetics in the Media...

**On February 18, 2009**  
**The New York Times**  
**published the following**  
**article by Roni Caryn**  
**Rabin:**

When David Swift's baby girl Giana was born at Santa Monica U.C.L.A. Medical Center six years ago, a nurse approached to ask if the baby could participate in a pilot program to screen newborns for dozens of rare genetic disorders. It involved little more than a heel prick blood test, but Mr. Swift, a 33-year-old first-time father, declined.

The nurse persisted, asking the question again and again, until Mr. Swift relented. And he's been trying to find that nurse ever since, because the test, which identified an extremely rare metabolic condition in his newborn daughter, may have saved her life.

"Had we not been at that particular hospital, with that particular pilot program and that particular nurse..." he said, his voice trailing off. "It's miraculous."

A March of Dimes report released today says all

50 states and the District of Columbia now require newborn screening for 21 or more so-called core disorders recommended for testing. These core disorders, 29 in all, include many rare but potentially disabling or fatal metabolic disorders. Although all states have rules or laws requiring the screenings, Pennsylvania and West Virginia have yet to implement their expanded programs, according to the organization.

The increase represents a big change since 2000, when most states screened for only four conditions and testing practices varied widely from state to state, according to March of Dimes officials.

"It's a milestone," said Jennifer L. Howse, president of the March of Dimes, which advocated for expanded testing. Screening rates in newborns rose from 38 percent in 2005, when the organization began monitoring, to 96 percent by the end of 2008, she said. "That's very, very dramatic."

Twenty-four states do all of the tests, and seven do 28 of the 29 tests, she said. The tests include screens for Phenylketonuria, or PKU, a potentially devastating illness that strikes about one in 25,000 newborns, and maple syrup urine disease, so-named for the infant's sweet-smelling urine, which affects one in 100,000. More common ailments like sickle cell anemia, which affects one in 400 African-Americans, and cystic fibrosis and hearing loss, each of which affect one in 5,000 children, are screened for as well.

The American College of Medical Genetics developed the list of the 29 disorders recommended for testing. (A list of tests provided by each state can be found at [www.marchofdimes.com/peristats](http://www.marchofdimes.com/peristats)).

Dr. Howse emphasized that the metabolic disorders on the list are rare but treatable, and "if they're not detected and treated, they're catastrophic and can lead to infant death." In most cases, she added, the babies look

perfectly healthy.

New technology, specifically the advent of a technique called tandem mass spectrometry, provided the means to identify many conditions from just a tiny sample of blood, she said.

Mr. Swift's daughter was found to suffer from a condition that makes her unable to break down leucine, an essential amino acid in protein. Only about 30 cases of the disorder have ever been documented and, undiagnosed, it could have led to mental or physical disability, or even death.

Instead, Giana was put on a modified diet with very little protein and takes a special protein formula free of leucine, Mr. Swift said.

"She is beautiful, healthy, precocious, 99th percentile height and weight, just wonderful," Mr. Swift said. "That test saved her life."