

www.region4genetics.org

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 and click on the "Region 4" tab, under "Additional Resources" click on "Member Directory". 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 released first.

Thank you!

Upcoming Events...

The Children's Hospital Boston Teen Challenge 2009 - a program for teens ages 13-18 with any metabolic disorder. Event held on Aug 5-8, 2009 in Boston. For more information go to http://www.region4genetics.org/information_pages/TeenChallengeBrochure2009.pdf

Upcoming Workgroup Meetings...

Medical Home
6/12, 7/10, 8/14

Care Coordination
6/23, 7/28, 8/25

Inborn Errors of Metabolism Information System - IBEM-IS
6/19, 7/17, 8/21



Region 4 Genetics Collaborative Stakeholder State Meetings

As many of you know the Region 4 staff has been visiting each of the seven states in our region for stakeholder meetings. We started in Lansing, Michigan in March and just completed the last visit in Minneapolis. The meetings have been a wonderful opportunity to provide updates on the work going on in your state, region and nationally. There are many exciting projects happening in our region,

including some tools and resources that will be available to parents and families soon. One example is the Medical Home Workgroup's [Family is the Center for the Medical Home: A Guide for Families with Children who have Heritable Conditions](#). The goal of the guide is to encourage families to work with primary care providers to establish medical homes for their children. Another is MEMSCIS (Midwest

Emergency Medical Services for Children Information System), which makes Emergency Information Forms (EIF) available via the internet in a secure, private fashion, improving emergency care for children with genetic conditions. Watch for more information coming out soon about these exciting projects!

What is the Region 4 Genetics Collaborative all about?

For those who are new to us, the Region 4 Genetic Collaborative brings together representatives from diverse stakeholder groups to realize our mission:

- 1) Increase access to information about newborn screening and genetic resources, services and family support
- 2) Facilitate data collection and analysis to guide decision-making regarding screening cut-offs, diagnosis and long term treatment of heritable disorders
- 3) Support state public health agencies in improving infrastructure for genetic service delivery to children with heritable disorders
- 4) Provide a forum for families, public health, and clinical providers to share best practices and models for improving newborn screening, follow-up and genetic care coordination
- 5) Link Region 4 states with regional and national initiatives for improving the quality of newborn screening and genetic service delivery

Parents and family members are essential partners in the Region 4 Genetics Collaborative. If you would like more information about opportunities for getting involved, contact Sarah Wedepohl at s.wedepohl@yahoo.com.

Finding a support group

On April 20, 2009 Chicago Parent published the following article by Marla Davishoff:

At my son's early intervention therapist's office, I commiserated with other parents about the challenges of raising a child with special needs, learned who the "in" neurologist was and what alternative treatments were popular and how to navigate through the insurance maze. Although talking to parents in the waiting room reminded me I wasn't alone, I often walked out of the clinic more anxious than when I entered. I wondered if I was doing enough.

Finding a support group that is a good fit is worth investigating. Research what is available in your area. Most support groups have an open door policy that allows parents to shop around. Before visiting, talk to the facilitator and ask questions. How often do they meet? How big is the group? Who leads the group? Is it OK to observe? Do the parents have kids of similar ages or with similar issue?

Once you find the right support, you may be surprised at how much easier it is to breathe.

Support groups for emotional needs

Parent-led support groups try to provide empathy and understanding.

"We all come because of who we are rather than who our children are," voiced a mother at a recent support group started four years ago by a mother, Kristen Scott, who has a son with autism. At these meetings every parent has a chance to talk uninterrupted about their experiences. New parents may learn from listening to more seasoned ones that they can still carve out a life for themselves. In turn, more seasoned parents often recognize how far they have come in their journey when they talk to parents of younger children.

Diagnosis-based support groups

When a support group is offered to parents whose children share a diagnosis, the focus tends to be more on sharing tips about how to manage common problems. Sometimes this means parents ask ahead of time to be put on the agenda to seek help in resolving a specific issue. It could also mean a more formal presentation about a topic of concern.

"I like knowing there are others in the same boat as me," one mother shared at a group I visited. "I don't necessarily need to vent. I simply want to connect with other parents who get it."

Media-based support

Although most support groups are free and accessible, many parents have neither the time nor the desire to meet face to face with other parents. Internet support groups in the form of message boards, chat rooms and blogs are another valuable resource for support. Reading about the journeys of other parents who have raised a child with special needs can be therapeutic. In fact, I have contributed to several anthologies in this genre and have found that even the writing and journaling process can be rewarding.

Individual counseling

Although support groups can be helpful, they are not a replacement for therapy. Individual counseling is recommended when your stress interferes with daily life. Significant changes in eating and sleeping habits, difficulty concentrating and feelings of worthlessness are all symptoms that should be referred to a social worker, psychologist or physician if they continue for more than two weeks.

Marla Davishoff is a licensed clinical social worker, freelance writer and mom living in Deerfield.



NOTE: For more information and resources about family support please go to www.region4genetics.org and look under "Family Resources" on our home page. If you know of any support groups that are not listed there is a place to submit information about family support resources that are not included in the directory.

This past year, Wondertime magazine (no longer in publication) published a four part series which followed the Foard family whose child Johnny was born with Alfi's Syndrome or 9p minus, a rare chromosomal disorder. Johnny also exhibits signs for autism. Wondertime was published by Disney) and the link to the series of article is still working. The link is below. If it doesn't connect please copy and paste into your web browser.

http://wondertime.go.com/parent-to-parent/article/jimmy-foard_SP.html

Meet one of our Region 4 Parent Participants!

Every quarter we will be highlighting one family in our collaborative. Here is one parent's story as told to us:

My husband and I are parents to two children. Our child G. has MCAD and our other child is a carrier. Everything was normal after G.'s birth until his blood sugar dropped at three days old. He was unresponsive and would not react. I was nursing him but finally got him to take a bottle and his blood sugar returned to normal. A couple of days later we got a call saying that his newborn screening was positive. We saw a specialist when he was 8 days old and it was confirmed that he has MCAD--the most severe and common form. He had 8-10 hospitalizations in the first two years ranging from stomach bugs to fevers. Now he is almost six years old and he has not been in the hospital since near his second birthday. He is doing great in school and at home!

I became involved with the Region 4 Genetics Collaborative through one of the specialists who saw G. She made a huge difference in our lives. If it had not been for her, G. would not have survived. His doctors and nurses saw that I am a good advocate for G. and they recommended I get involved with the

collaborative. I have been active ever since and I want to stay involved for as long as I can.

Some advice I can give is to try and get support. It gets very stressful and a support person can be really helpful. Looking back, I was really stressed and sick from it, so had I been able to take a break for myself once in a while that would have helped. Also, it may sound strange but you have to give yourself a chance to grieve and accept that the baby you were expecting to be perfect is not. It's a process and it's not going to happen overnight, but be patient.

Another piece of advice is to be an advocate for your child. I have learned to be a good advocate and have even made suggestions or done something that has influenced how my son and others are cared for! I still have a good relationship with my doctor even though I challenge her sometimes. The doctors do pay attention to what parents do and sometimes they even thank us for tips. You are the one in the trenches--you are the one that has to live this.

Tell me a bit more about....genetic services providers

Here are some questions taken from our website about genetic services. Please visit the FAQs (Frequently Asked Questions) section on the Region 4 website for more information at

http://www.region4genetics.org/family_resources/faq.aspx

Who provides genetic services?

Generally, medical genetic services are provided by health professionals called genetic counselors or medical geneticists (doctors who specialize in genetics). Medical genetics is a branch of medicine specializing in detecting both inherited conditions, that is things that result from our genetic make-up; and genetic changes that may show an increased risk of certain conditions, such as cancer or heart disease.

What is a medical geneticist?

A medical geneticist is a doctor who has special training in diagnosing and treating patients with genetically linked conditions.

What is a genetic counselor?

A genetic counselor is a health professional with a graduate degree and experience with medical genetics and counseling. Genetic counselors provide information and support to people who may have, or are at risk for, a variety of inherited disorders. They also discuss genetic testing options so people can make fully informed decisions about whether to undergo genetic testing.

Are genetic counselors and medical geneticists different?

Yes. Medical geneticists are the doctors who specialize in diagnosing and treating medical conditions that have a genetic basis. Genetic counselors provide information, counseling and support to families with birth defects or other genetic conditions. Medical geneticists and genetic counselors often work closely together while performing genetic evaluations or genetic counseling for patients and families.