



GENE NEWS

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Welcome to the Coordinator's Corner

Aloha all! You may be wondering why the issues of GeneNews stopped last year. You can call it a problem with success. The program has been so busy with its activities to improve genetic and newborn screening services and education in Hawai'i that we didn't have time to keep up with the newsletter. However, we are now getting back on track and continuing GeneNews to keep everyone up-to-date on the latest genetic happenings within the program and elsewhere.

As I mentioned, we've been working hard this past year. Some of the activities include:

- Continuing to provide in-person outreach genetic services and education to the Neighbor Islands;
- Improving the telehealth network and increasing the genetic services provided via telehealth to Neighbor Island families;
- Developing emergency plans for newborn screening;
- Reviewing and trying to improve reimbursement for genetic services;
- Working with other states to improve access to genetic and newborn screening services and education through the Western States Genetic Services Collaborative;
- Implementing quality assurance activities to improve service provision; and
- Partnering with the CDC multi-state program to research the incidence and long-term outcomes of Duchenne and Becker muscular dystrophies.

For more information about our activities, you can go to our website at www.hawaiiogenetics.org.

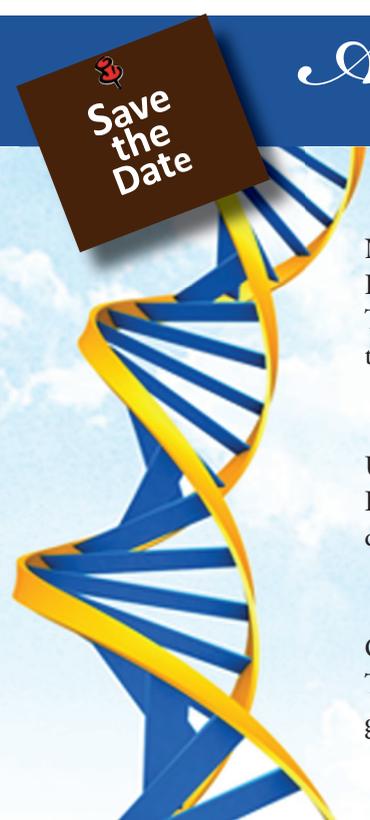
Before I close, I want to mention that Elaine Marr White has left the Hawai'i Genetics Program and will be pursuing her volunteer interests in China with her new husband. We all wish you well in your new adventures! I also want to give a big welcome to Arthur Yu. Arthur is a genetic counselor that comes to us after three years at Kapiolani Medical Center for Women and Children. We are already making sure Arthur has more than enough work in the program. As usual, please drop me a line if you have comments or questions. Please contact Sylvia Au at sylvia@hawaiiogenetics.org

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Announcing the 2009 Genetic Conference

Yes, it's back!

Save the Date

September 2 – Morning

New Healthcare Options: Personalized Medicine
Learn the latest about how genetic testing can personalize medical care. Topics include pharmacogenomics and direct-to-consumer genetic testing.

September 2 – Afternoon

Update on Cancer Genetics for Your Practice
Learn about the newest genetic tools to use for cancer risk assessment, diagnosis, treatment and prevention.

September 3 – Evening

Genetics and Religion – Finding the Right Balance
This is a community forum to discuss how spiritual beliefs interact with genetics healthcare and research.

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Governor of Hawai'i

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GOING THE DISTANCE: CARING FOR OUR NEIGHBOR ISLAND 'OHANA

Dr. Laurie Seaver, the clinical geneticist at Hawai'i Community Genetics, has become an expert at neighbor island traveling. Once a month, she and a genetic counselor from the Hawai'i Genetics Program fly to one of six neighbor island sites to provide a day-long outreach genetics clinic.

Between November 2005 and March 2009, 278 families have been seen in Maui, Kaua'i, Moloka'i, Hilo, Kona, and Waimea. Families are reporting great satisfaction with the outreach clinics. One Hilo mother commented, *"Being able to see a specialist on an outer island versus having to travel to O'ahu makes health care much more accessible for our family, and we are very thankful for that. We have to see another specialist on Oahu and the airfare and rental car costs us about \$675!"*

The Hawai'i Genetics Program and Hawai'i Community Genetics also collaborate to provide half day telemedicine clinics to Maui, Kaua'i, Hilo, and Kona. Using videoconferencing equipment, a geneticist and genetic counselor in Honolulu can "see" a neighbor island family in real-time and can provide a genetics consultation without requiring either the family

or the specialist to travel. Twenty-one families have been evaluated since the telemedicine clinics began in June 2006. Satisfaction surveys similar to those used in the outreach clinics are completed by participating families. Similar to the outreach clinic results, feedback for the telemedicine clinics have been entirely positive:

Information from families, primary care providers, and genetic specialists are collected to help us evaluate the outreach and telemedicine clinics. The data will be used

Family Satisfaction Survey Result

| Statements | Strongly Disagree | Disagree | Agree | Strongly Agree |
|--|-------------------|-----------|----------------|----------------|
| I feel confident in the quality of the care provided today. | 0 0 | 0.4% 0 | 20.5% 28.6% | 70.5% 71.4% |
| I feel confident in the recommendations from today's clinic visit. | 0 0 | 0.4% 0 | 20.8% 23.5% | 70.5% 76.2% |
| I would recommend this clinic to other families. | 0 0 | 0.4% 0 | 20.1% 14.3% | 71.2% 85.7% |

Black = Outreach clinic
Blue = Telemedicine clinic

to help determine the best way to deliver services through Honolulu, neighbor island, and telemedicine clinics so that we can provide comprehensive genetic services to all families in Hawai'i. We hope that our efforts will ensure that any family wanting to receive clinic genetic services will be able to do so, no matter where they live!

Being able to see a specialist on an outer island versus having to travel to O'ahu makes health care much more accessible for our family.....



AUTISM AND GENETIC TESTING

Recently, autism has been receiving widespread attention. Scientific journals and popular media are reporting the latest research, much of which involves genetics. There has been increasing interest from families and health care providers about genetic testing for autism. Before genetic testing is considered, here are the facts:

What is Autism?

Autism is the most common diagnosis in a group of developmental conditions called the autism spectrum disorders (ASD). Autism:

- often presents with communication and social interaction difficulties, and stereotypical behaviors;
- occurs in about 1 in 500 people. If all the ASDs are included, this increases to about 1 in 200 people;
- occurs more often in males than in females;
- usually presents in the first three years of life; and
- is thought to be caused by a mix of genetic and environmental factors.

What is the Recent Research?

Researchers are looking for the causes of autism, including genes that might contribute to the condition:

- In some people with autism, a specific genetic cause is identified. For example, genetic conditions such as Fragile X syndrome share some symptoms with autism. It is not unusual for people with Fragile X syndrome to be diagnosed with autism.
- In other cases, the cause of autism may be genetic, but the exact genetic change has not yet been identified.
- Finally, there are cases in which the cause of autism is still unknown and may involve factors beyond genetics.

Recent research has involved a large number of families with more than one child with autism. These families have participated in genetic research in the hope of finding a genetic cause of autism. Researchers wanted to know if there were changes in the genetic material of people with autism that caused their condition.

What are the Results?

In the recent study described above, six genetic variants, or changes in the DNA, were found to increase the chances that a person will develop autism. Two of the genes identified are involved in the formation of nerve connections in the brain, especially in areas of the brain that regulate speech and interpret social interaction. **It is important to note that these genetic variants are thought to account for only about 15% of autism diagnoses.** There are still other causes of autism that need to be researched, including other regions of DNA with deletions or duplications.

Is Testing Available?

- Testing to identify deletions (missing pieces) or duplications (added pieces) of a person's DNA is available on a clinical basis. The sensitivity of this test may increase due to the new research findings.
- Several laboratories across the USA offer the testing, which costs about \$1600.
- Ideally, a family should seek genetic counseling by a genetics specialist or developmental pediatrician experienced in working with people with autism before being tested. The specialist will help the family evaluate whether or not the genetic testing may be beneficial.





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Hawai'i Joins
National Team to
Learn More About
Duchenne and
Becker Muscular
Dystrophy

The state of Hawai'i recently became the newest addition to the Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet). MD STARnet is a national program developed by the Centers for Disease Control and Prevention (CDC) in several states to identify all children with Duchenne and Becker Muscular Dystrophy (DBMD). Currently, CDC works with Arizona, Colorado, Georgia, Iowa, Western New York State, and now Hawai'i. Locally, the Department of Health Genetics Program has partnered with The Hawai'i Muscular Dystrophy Association and its clinics as well as healthcare organizations and practices in Oahu and the neighbor islands.

With the addition of Hawai'i to the MD STARnet team, the project hopes to learn more about how often DBMD occurs in the Asian and Pacific Islander populations, the long term outcomes of individuals with DBMD, methods of care for children and families with DBMD unique to the culture of Hawai'i, and barriers to care for Hawai'i families with DBMD.

This is a unique opportunity for the families of Hawai'i affected by DBMD to increase the diversity of information known about the condition on a national level. Additionally, it will help strengthen the knowledge we have about DBMD in our community. To learn more about the project, please visit:

<http://hawaii.gov/health/family-child-health/genetics/mdstarnet.html>