

### Aloha, Hawai'i!

The Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet) welcomes a new site! Hawai'i joins Arizona, Colorado, Georgia, Iowa, and western New York.

Hawai'i MD STARnet, led by the Hawai'i Department of Health Genetics Program, is a collaboration between:

- Department of Health program
- The Muscular Dystrophy Association and its clinics
- Other health care organizations and medical practices in Hawai'i

We hope Hawai'i's addition to MD STARnet will increase knowledge and awareness of Duchenne and Becker muscular dystrophy (DBMD) in the Asian and Pacific Islander populations. We also want to learn more about the barriers to services for families in remote geographic areas. Other interests are methods of care unique to Hawai'i and care for a population where almost everyone has health care coverage.

The Hawai'i MD STARnet team is made up of many key players. Sylvia Au leads the Hawai'i team. A certified genetic counselor, Ms. Au also serves as the Hawai'i state genetics coordinator. In addition to Ms. Au, the State Genetics Program includes four other certified genetic counselors: Sarah Scollon, Arthur Yu, Lianne Hasegawa, and Kirsty

*Editorial Statement: this newsletter is published for people with Duchenne and Becker muscular dystrophy and their families. The Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet) is funded by the Centers for Disease Control and Prevention.*



**From left to right: Lianne Hasegawa, Kirsty McWalter, Sarah Scollon, Pauline Mui, Sylvia Au, and Arthur Yu.**

McWalter, who also work on Hawai'i MD STARnet. Ms. Scollon is the project coordinator. She and Mr. Yu perform many roles within the project, including data management, abstraction, and interviewing. Ms. Hasegawa and Ms. McWalter support the project. Other Hawai'i team members include: Pauline Mui, administrative assistant; Don Hayes, project epidemiologist; and Dr. Keith Abe, Hawai'i's clinical reviewer. The team is also being supported by its project partners and families.

The goal of the Hawai'i State Genetics Program is to improve the quality of genetic services for the families of Hawai'i and the nation. The program has examined the issues related to access to care in many states and to expanding newborn screening. They are also looking into the cause of hearing loss in newborns and providing genetic follow-up services. To learn more about the Genetics Program and its projects, please visit the program's web site at [www.hawaiigenetics.org](http://www.hawaiigenetics.org).

# Hot Off the Press

## Tracking Clinical Trial Involvement

An increasing number of treatments are under study for DBMD. Some persons with DBMD have had the chance to participate in clinical trials or research projects test medications and medical devices used to treat the disorder.

We interviewed primary caregivers in MD STARnet and found that 1 in 3 persons with DBMD participated in clinical trials or research projects. Most of these individuals were involved in only one trial or project; a small number participated in many trials or projects.

Nearly all of the trials or research projects were for medications. A steroid, such as prednisone or deflazacort, was the most frequently reported medication. People ranged in age from 2 to 21 at the time of research participation. More information about clinical trials can be obtained from <http://clinicaltrials.gov>.

**This year, the *Journal of Pediatrics* accepted an article featuring our research on diagnostic delay in Duchenne muscular dystrophy (DMD).** We wanted to learn the main reasons for delayed diagnosis of DMD. The medical records of 453 boys in MD STARnet were reviewed. We found 156 boys who were the first in their family to be diagnosed with DMD. We looked at their records to see how and when they were diagnosed.

Among boys with no family history of DMD, the average age of first signs or symptoms was 2.5 years of age. A doctor or health care provider first examined them for these concerns at an average age of 3.6 years. The first creatine kinase (CK) test was done around 4.7 years of age. The diagnosis of DMD was not confirmed until 4.9 years of age on average.

We found a delay of 2.5 years between first signs or symptoms and diagnosis of DMD in boys with no family history of the disease. We recommend that doctors check CK early when seeing boys with developmental delay that has no clear cause. This includes delayed walking, delayed speech, and difficulty running, jumping, or climbing stairs. Getting an earlier diagnosis is very important. Families will have answers sooner. They can avoid the cost and time of many doctor visits. An earlier diagnosis will also allow for genetic counseling and access to therapies and services sooner.

**The *Journal of Child Neurology* published MD STARnet findings about testing and type of DMD mutations.** We looked at the kinds of tests used among persons in MD STARnet. We found the frequency of muscle biopsies has dropped greatly. About 66% of boys born between January 1982 and September 1987 received muscle biopsies. However, only about 33% of boys born between April 1999 and September 2004 had them. On the other hand, the use of DNA testing rose.

DBMD is caused by mutations in the *DMD* gene. We looked at the types of *DMD* mutations of persons in MD STARnet. *DMD* mutation was documented for 73% of boys. Deletion of one or more exons was found in 57% of all boys and 78% of boys who have a known *DMD* mutation. Duplication was identified in 8% of all boys and 11% who have a known *DMD* mutation. Point mutation, small insertion, or small deletion was found in about 8% of all boys and 10% who have a known *DMD* mutation. Point mutation analysis was done in only 29% individuals with negative deletion and/or duplication testing. Knowing the type of mutation is important. It can help persons with DBMD to find out if they are eligible for any clinical trials.

# MD STARnet Activities Update

We have completed initial telephone interviews with more than 200 families, and this information is being analyzed. Findings will help us answer many questions, such as:

- How common is DBMD?
- What are the early signs and symptoms of DBMD?
- Does the type of care received affect the severity or progression of DBMD?
- What medical and social services are families receiving?
- Do different populations receive different levels of care?

Last fall, we mailed needs assessments surveys to study participants. The next step will be to review the answers. We will identify the most important needs of families with DBMD and how these needs change as children grow. The data will help us answer questions such as which needs are unmet and what kinds of services are the most helpful for families.



# Meet Jacob Conover



*Rebecca, Jacob, and Jenny Conover. Fuzzy the Yorkshire Terrier is 9 years old. (Not pictured is Goldie, Jacob's 15 year old Golden Retriever.)*

Jacob is 14 years old and lives in Tucson, Arizona. He's a very cool guy!

**What's your favorite school subject?** Social studies – I like to learn about the wars and stuff.

**Do you have any siblings?** I have a younger sister, Jenny, who is 12. My older sister, Jessica, is 18. And my brother, Josh, is 23 and lives in Scottsdale.

**You are a Muscular Dystrophy Association (MDA) ambassador. What does that mean?**

In Wyoming, I was the MDA state ambassador when I was 6 and 7. Two years ago, I was the ambassador for southern Arizona. Then last year, I was goodwill ambassador for the whole state of Arizona.

I go to all the MDA events and talk about MDA. I have these cool cards I hand out to people.

**Have you been to the MDA summer camp?** I've gone since I was 6 years old. My brother is my counselor every year. My sister Jessica is also a counselor at camp.

**What's your favorite thing to do at camp?** Wheelchair soccer. They divide us into four teams. Two teams play each other. Each team has a goalie, two people on defense (who can't cross over to other side), and three people on offense. The first year I went to this camp, we got first place. The second year, second place. The third year, third place. So I'm planning on fourth place this year. As a matter of fact, I like soccer so much that I joined the team that just started in Tucson.

**Your ambassador card says the Hummer is your favorite car.** I'm gonna buy a Hummer when I'm older, because I'll be rich. I have ideas lined up. It has something to do with video games, but I can't tell you.

**Does that mean you like videogames?** I like to have my friends over to play videogames. I usually help my friend, Sharky, play. He got that name because he eats like a Shark. He was with me when I was interviewed for the MDA telethon on TV. Sharky was taking this big bite out of a sandwich when I said in my interview "My best friend Sharky – he got that name because he eats like a shark." So right as I said that, they showed him taking that big bite. It was perfect.

**Did you just get your DNA testing results?** Duchenne muscular dystrophy runs in the family, so they knew I had it when I was 6 months old. The genetic testing was done a long time ago, but we didn't get the results until recently. It was done so long ago; it said my name was Fetus on the paper! I have exon deletion 5-45.

**Why is it important to know that?** There are two different mutations: genetic where it runs in your family or a new mutation where no one else in your family has it. If you know the deletion and all that, you can find out what tests and trials you can get on. There are different drugs they want to try like PTC124. Right now there are not really any trials for me that have started yet.

**What would you like to be changed for persons with Duchenne?** I think research is important. Find a cure. America's a great country. I am American and I like America. But they're not good about one thing. If people want to test drugs and say they won't sue or anything, they should let them do it. It takes forever to pass new drugs. By the time they do it, it might be too late. In other countries, if people want to do it, they can test it. (Jacob's mom, Rebecca, agrees, "I don't see why they can't let people take things if it's there and they choose to.")

**In addition to camp, are you doing anything else over the summer?** We go on vacation over the summer. My favorite places so far are New York and San Diego. We drove from here to New York. This summer we're going to Wyoming where we used to live. After that we're going down to Colorado for a family reunion.

Jacob's mom says, "We're working on getting a new minivan. He's too tall now and his chair has the tilt feature on it that makes it higher. So he can't sit up all the way. He has to ride tilted back and then he can't breathe that well. So we need to get a new one for the trip."

**And then you start 9th grade this fall?**  
Yeah. I don't wanna go to high school – it's lots of work!

# Kid's Corner

Summer is here! Can you find the words? They might be backwards, forwards, or diagonal. See answer key on page 8.

Y	L	E	W	O	T	E	L	O	N	U
L	O	C	N	L	S	M	V	I	C	I
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F	S	U	N	S	H	I	N	E	S	S

- Beach
- Camp
- Fourth of July
- Ice cream
- Jacob
- Picnic
- Summer
- Sunshine
- Swimming pool
- Towel
- Vacation
- Watermelon

# Q&A With...

## Stacey Hockett Sherlock

Stacey Hockett Sherlock is the survey coordinator for MD STAR*net*.

### **What is your professional background?**

I work for the College of Public Health at the University of Iowa. My work experience includes research on health and environmental topics. I have a Master of Applied Anthropology from the University of Maryland. Anthropology is the study of the culture of humankind.

### **What do you do within MD STAR*net*?**

I am the survey coordinator for MD STAR*net*. I train the interviewers at each site. I also conduct quality control and quality assurance for the interviews and survey data. I oversee the day-to-day recruitment activities and answer questions from participants. I help develop and test the interview application and instrument.

### **Why is this work important to you?**

We hope the interviews and survey research will directly improve health care. We also want the results to help people with muscular dystrophy and their family members in getting their needs met. As a parent, it is meaningful to me to work on a project that can help children and families.

We are very appreciative of the time caregivers and adults with DBMD have given to the project. We are hearing many suggestions from families for improvement of local resources and areas where they need assistance.

### **What is the most unique project in which you've been involved?**

I conducted field work in West Africa. There I learned traditional arts and crafts techniques from local artisans such as potters, weavers and sculptors. I used to live in Maryland and worked on projects with watermen families on the Chesapeake Bay. I researched the roles and economic decisions of women in the fishery. I also worked on a learning project where watermen and policy makers shared knowledge to improve environmental policy.

### **Do you have any hobbies?**

In my free time, I enjoy reading books with my children, making chocolates and candies, and gardening.



# MD STARnet Print & Posters

## PRINTED PUBLICATIONS

Journal of Pediatrics, April 2009. Epub ahead of print. **Delayed diagnosis in Duchenne muscular dystrophy: Data from the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet)**. Ciafaloni E, Fox DJ, Pandya S, Westfield CP, Puzhankara S, Romitti PA, Mathews KD, Miller TM, Matthews DJ, Miller LA, Cunniff CC, Druschel CM, Moxley RT, and the MD STARnet.

Journal of Child Neurology, April 2009. Volume 24, issue 4, pages 425-30. **Mutation analysis in a population-based cohort of boys with Duchenne or Becker muscular dystrophy**. Cunniff C, Andrews J, Meaney FJ, Mathews KD, Matthews D, Ciafaloni E, Miller TM, Bodensteiner JB, Miller LA, James KA, Druschel CM, Romitti PA, Pandya S.

Birth Defects Research Part A: Clinical and Molecular Teratology, November 2006. Volume 76, issue 11, pages 793-7. **The Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet): surveillance methodology**. Miller LA, Romitti PA, Cunniff C, Druschel C, Mathews KD, Meaney FJ, Matthews D, Kantamneni J, Feng ZF, Zemblidge N, Miller TM, Andrews J, Fox D, Ciafaloni E, Pandya S, Montgomery A, Kenneson A.

## POSTER PRESENTATIONS

Poster presentation at Meeting of Association for European Paediatric Cardiology, May 2008. **Novel genetic associations with delayed cardiomyopathy onset in muscular dystrophy utilizing MD STARnet data**. Barber BJ, Andrews JG, Cunniff CM, Klewer SE, Miller TM, Meaney FJ, and MD STARnet.

Poster presentation and special forum discussion at American Academy of Neurology Annual Meeting, April 2008. **How frequently are corticosteroids used in boys with dystrophinopathy in the USA: Data from the MD STARnet**. Ciafaloni E, Pandya S, Matthews D, James K, Mathews K, Miller T, and MD STARnet.

Poster presentation at American Academy of Neurology Annual Meeting, April 2008. **Palliative care services in families of patients with Duchenne muscular dystrophy**. Pandya S, Meaney FJ, Andrews J, Davis M, and Palliative Care Group MD STARnet.

Poster presentation at the 12th International Congress of World Muscle Society Meeting, October 2007. **Palliative care services in families of males with Duchenne muscular dystrophy**. Meaney F, Pandya S, Andrews J, Davis M, and Palliative Care Group of MD STARnet.

Abstract and poster presentation at the American Academy of Neurology Annual Meeting, May 2007. Abstract in Neurology 2007; 68 (Suppl 1):A243. **MD STARnet: case definition in surveillance for childhood onset Duchenne and Becker muscular dystrophy (DBMD)**. Mathews K, Cwik V, Ciafaloni E, Miller T, Matthews D, Pandya S, Romitti P, Cunniff C, Druschel CM, Miller L, Meaney FJ.

Abstract and poster presentation at American Academy of Neurology Annual Meeting, May 2007. Abstract in Neurology 2007; 68 (Suppl 1):A244. **Diagnostic delay in Duchenne and Becker muscular dystrophy (DBMD): Data from the MD STARnet cohort**. Ciafaloni E, Pandya S, Fox D, Matthews D, Mathews K, Miller T, and the MD STARnet.

Poster presentation at the National Birth Defects Prevention Network Annual Meeting, February 2007: **The muscular dystrophy surveillance tracking and research network (MD STARnet): Population-based surveillance and research on Duchenne and Becker muscular dystrophy**. Meaney FJ, Montgomery A, Miller LA, Romitti PA, Druschel CM, Cunniff C, Matthews D, Mathews K, Pandya S, Vatave A, and the MD STARnet.

Poster presentation at the Colorado Public Health Association meeting, September 2006: **MD STARnet – The Muscular Dystrophy Surveillance Tracking and Research Network**. Miller LA, Matthews D, James K, Montgomery A, and the MD STARnet.

Poster presentation at 35th Annual Meeting and National Health Conference of the Association of American Indian Physicians, August 2006: **MD STARnet: The Arizona experience. The Muscular Dystrophy Surveillance, Tracking, and Research Network**. Damon S, Arias R, Cunniff C, Meaney FJ, Pettygrove S.

# Resource Corner

## Centers for Disease Control and Prevention (CDC)

MD STARnet is a program of CDC's National Center on Birth Defects and Developmental Disabilities. You can learn more about DBMD, other research, and read one family's personal story on the web site at <http://www.cdc.gov/ncbddd/duchenne/index.htm>.

## General Questions

If you have general questions about MD STARnet, please contact your state's program:

### ARIZONA

Jennifer Andrews, MBA, Study Coordinator  
Department of Pediatrics  
The University of Arizona College of Medicine  
1501 N. Campbell Avenue  
Tucson, AZ 85724-5073  
(520) 626-6816

### COLORADO

April Montgomery, MHA, Study Coordinator  
Colorado Responds to Children with Special Needs  
Colorado Department of Public Health and Environment  
4300 Cherry Creek Drive South  
Denver, CO 80246  
(303) 692-2620

### GEORGIA

Natalie Street, MS, Study Coordinator  
Centers for Disease Control and Prevention  
1600 Clifton Road, N.E., E-88  
Atlanta, GA 30333  
(404) 498-3001

### HAWAII

Sarah Scollon, MS, CGC, Study Coordinator  
Hawai'i Department of Health  
Genetics Program  
741 Sunset Avenue  
Honolulu, HI 96816  
(808) 733-9055

### IOWA

Paul Romitti, PhD, Principal Investigator  
University of Iowa  
C21-E GH, 200 Hawkins Drive  
Iowa City, IA 52242  
(866) 834-9676

### NEW YORK

Charlotte Druschel, MD, MPH, Principal Investigator  
Congenital Malformations Registry  
New York State Department of Health  
547 River Street, Room 200  
Troy, NY 12180  
(518) 402-7990

Y	L	E	W	O	T	E	L	O	N	U
L	O	C	N	L	S	M	V	I	C	I
U	O	G	E	J	S	B	E	A	C	H
J	P	A	E	E	U	H	O	E	B	I
F	G	M	C	P	M	A	C	C	T	V
O	N	O	L	E	M	R	E	T	A	W
H	I	L	J	A	E	O	I	C	W	J
T	M	N	M	A	R	E	A	M	C	M
R	M	Y	M	C	C	T	A	O	H	U
U	I	H	A	P	I	C	N	I	C	A
O	W	L	I	O	M	A	I	M	C	A
F	S	U	N	S	H	I	N	E	S	S

Beach  
Camp  
Fourth of July  
Ice cream  
Jacob  
Picnic  
Summer  
Sunshine  
Swimming pool  
Towel  
Vacation  
Watermelon

# Kid's Corner

Summer is here! Answer key.