

**DEPARTMENT OF HEALTH AND HUMAN SERVICES
OFFICE OF DIRECTOR**

ACTION REFERRAL

TO <i>Waldrep</i>	DATE <i>10-12-12</i>
----------------------	-------------------------

DIRECTOR'S USE ONLY	ACTION REQUESTED
1. LOG NUMBER <i>10-105</i>	<input type="checkbox"/> Prepare reply for the Director's signature DATE DUE _____
2. DATE SIGNED BY DIRECTOR <i>cc: Mr. Kech, Singleton</i> 	<input type="checkbox"/> Prepare reply for appropriate signature DATE DUE _____
	<input type="checkbox"/> FOIA DATE DUE _____
	<input checked="" type="checkbox"/> Necessary Action

APPROVALS (Only when prepared for director's signature)	APPROVE	* DISAPPROVE (Note reason for disapproval and return to preparer.)	COMMENT
1.			
2.			
3.			
4.			

October 10, 2012

Mr. Tony Keck
Director
Department of Health and Human Services
PO Box 8206
Columbia, SC 29202

RECEIVED

OCT 12 2012

Department of Health & Human Services
OFFICE OF THE DIRECTOR

Dear Mr. Keck:

For many years, the Greenwood Genetic Center (GGC) has placed a high priority on understanding the genetic contribution to autism spectrum disorders, a diagnosis that affects 1 in 88 children in South Carolina. GGC's addition of a next-generation sequencing panel of 62 genes associated with syndromic autism has captured the attention of national media in a recent article in *The Wall Street Journal*. GGC was noted as one of only three organizations in the US offering such testing.

The enclosed article highlights the cutting-edge technology and available genetic testing that provides answers to families seeking a diagnosis, and hope for advances in treatment options. GGC's dedication to providing research breakthroughs, accurate diagnoses and viable treatment options places our institution on the cutting-edge of technological advances, while at the same time maintaining an unparalleled commitment to quality patient care and family support.

We are proud to be the premier provider of genetics services in South Carolina. Our faculty and staff strive daily to ensure that our patients and their families have access to the newest technologies and highest quality clinical and diagnostic services.

If you have any questions regarding the Greenwood Genetic Center's programs and services, please feel free to contact me directly at sas@ggc.org or (864)941-8164.

Sincerely,



Steven A. Skinner, MD
Director

Enclosure: Beck, Melinda. "Progress in Identifying the Genetic Roots of Autism." *The Wall Street Journal* [New York] 25 Sept 2012, D5.

HEALTH JOURNAL | Updated September 24, 2012, 7:45 p.m. ET

Progress in Identifying the Genetic Roots of Autism



By MELINDA BECK

One of the most agonizing questions that parents of children with autism ask is—why?

Now, a growing number of genetic tests are providing some answers.



Melinda Beck on Lunch Break looks at how a growing number of genetic tests for autism are providing answers for some families. Plus, the parent of an autistic child joins us to explain how a new drug for the condition is having an impact on her son. Photo: Brian Harkin for The Wall Street Journal.

Scientists say that roughly 20% of autism cases can be linked to known genetic abnormalities, and many more may be discovered.

Pinpointing a genetic explanation can help predict whether siblings are likely to have the disorder—and even point to new, targeted treatments. Last week, for example, researchers reported that an experimental drug, arbaclofen, reduced social withdrawal and challenging behaviors in children and adults with Fragile X syndrome, the single most common genetic cause of autism.

Cari Wheeler of Madera, Calif., says her 11-year old son, Max, who has both Fragile X and autism, used to get "sensory overload" just sitting in a restaurant, and had to withdraw into himself or a DVD player. Now, he can order food, look at the waiter and greet people with "What's up?" says Ms. Wheeler. "These are things he never did, ever."

A cheerleading mascot for the local youth football team, Max is now even comfortable having the entire stadium yell, "How's it going, Max?" to which he responds, "I feel good—like a Hawk should."

Autism spectrum disorder (ASD) is a collection of conditions that can range in severity from the social awkwardness and narrow interests seen in Asperger's to severe communication and intellectual disabilities. ASD now affects 1 in every 88 U.S. children—nearly double the rate in 2002—according to the Centers for Disease Control and Prevention.

More

The Fragile X Connection

Experimental Drug to Treat Mental Disorder Shows Promise 9/19/12

Finding the Best Doctor for You

School Nurses' New Role in Children's Health

No Breakfast Affects Students' Performance

Several other academic medical centers offer their own gene-sequencing tests for autism, looking at different suspect genes. The tests typically cost about \$2,000 and generally are covered by insurance.

Other kinds of genetic tests may predict the likelihood that a child will develop autism before a clinical diagnosis is made.

Researchers at the University of Melbourne, Australia, have developed a test that looks for 237 genetic markers called single-nucleotide polymorphisms (SNPs). Some are thought to raise the risk of autism; others seem to protect against it. The test correctly predicted autism with more than 70% accuracy in people of Central European descent, but only 54% in those of Chinese descent, according to a study in the journal *Molecular Psychiatry* this month.

Autism Quick Facts

1 in 88 U.S. children has been diagnosed with autism spectrum disorder (ASD)

ASD diagnoses have increased 78% since 2002

Symptoms typically appear by 18 months of age

Average age of diagnosis: after age 4

ASD is 5 times more common among boys than girls

Symptoms, which can range from mild to severe, typically include social, communication, behavior and intellectual challenges

Early interventions can help children walk, talk, behave and socialize better and raise IQ

In April, IntegraGen Inc., a Cambridge, Mass., biotech company, announced a test, called ARISK, that predicts the likelihood that children aged 6 to 30 months who have older siblings with autism will develop the disorder themselves. The company is also developing a test to assess the risk of autism in children with no family history of the condition.

The goal of such tests, says IntegraGen general manager Larry Yost, is to have children at very high risk for autism referred to specialists for a definitive diagnosis earlier. Studies show that early intervention can significantly improve a child's IQ, language ability and

social skills. But many children aren't diagnosed until after age 4, according to the CDC.

Autism experts say the disorder should never be diagnosed based on gene tests alone; some studies suggest that environmental factors may play an equally important role.

The largest-ever study of twins with ASD—192 pairs—reported last year that when one identical twin has autism, there is only a 70% chance that the other twin will, despite their identical genetic makeup.

Among fraternal twins, the likelihood that a second twin will have autism is 35%—nearly twice the risk other siblings face, the study found. "That suggests there was something about their shared prenatal environment that really increases the risk," says Clara Lajonchere, vice president of clinical programs for Autism Speaks, a nonprofit science and advocacy group.

Environmental factors—premature delivery, low birth weight, maternal infections and maternal nutrition—have been implicated in autism, as well as advanced parental age. Some experts suspect that the older the father, the greater the chance for spontaneous genetic errors in sperm.

Most cases of autism probably involve some combination of genetic and environmental factors, experts say, and research is burgeoning.



**Greenwood
Genetic Center**
101 Gregor Mendel Circle
Greenwood, South Carolina 29646

GREENVILLE
SC 296

OCT 12 2012
PM 3:1

Mr. Tony Keck
Director, DHHS
PO Box 8206
Columbia, SC 29202

RECEIVED

OCT 12 2012

Department of Health & Human Services
OFFICE OF THE DIRECTOR



U.S. POSTAGE PITNEY BOWES
ZIP 29646 \$ 000.45⁰
02 1W
0001372037 OCT. 10 2012

29202820606

