



Simons VIP report helps guide therapies



Jack, age 12, and his family have benefited from improved speech therapy following their Simons VIP site visit. His mother, Michelle, answers some questions about their experience.

What therapy was Jack getting prior to your site visit?

Jack was getting speech and occupational therapy in school and he was seeing a private counselor.

What was recommended following your site visit?

The most useful recommendation was to fight one battle at a time. Having an autistic child can be very overwhelming when you try to "fix" everything at once. While in Houston, we were asked what the most important thing we felt needed to be addressed with Jack. The team and we agreed that it was Jack's communication. Jack has been in speech therapy within the public school system for 7 years and we never saw any improvement. The team recommended that we seek speech therapy from someone who works with autistic children outside the school.

How did you use this information?

As soon as we got home from Houston, we started looking for a speech therapist. It took a few weeks to find one and get an appointment but it was worth the wait. Jack started showing improvement immediately with some useful tricks and reminders. His teacher is using the ideas that the speech therapist is giving us in the classroom.

What changed in his therapies?

Jack was getting speech therapy from a

school therapist before. His speech problems aren't like those of a typical child. In school, they always worked on speech sounds. Jack had the ability to make each sound but when he put his thoughts together, he spoke too quickly for most people to understand. As his parents, we could understand him but Jack couldn't communicate effectively with the outside world. Jack's new speech therapist is giving him very simple tricks to slow down and gather his thoughts before speaking - we also give him non-verbal cues to remind him to slow down.

Words of Wisdom

I just wanted to add that we have always been frustrated with Jack's speech therapy in school but we didn't know how to find anyone better. We also knew that any additional speech therapy would be expensive. Since we already have a lot of expenses for Jack (counseling, medication, doctors) and we are living on a single income, we just never thought it would be possible to do anything else. When we sat down with the team in Texas, they put things in perspective for us. They reminded us that if Jack can't communicate with the world, he can't be a part of it. It seems like a simple idea but when you are dealing with so many different problems, you tend to bundle them into one big unsolvable problem. We have learned to step back and tackle one thing at a time. This has been much



L-R: Jack, (16pdel); Anna (Simons VIP Houston), Brandon (Jack's brother)

easier for us and certainly much easier for Jack. His anxiety is greatly reduced; usually, the new school year is plagued with sleepless nights, seizures and lots of pacing and tics. This year hasn't been that way. I attribute this to Jack's counseling and his confidence being boosted by his ability to communicate better.

Thanks to Jack and his family for sharing their story!

Update: When using the tools the speech therapist has given him, Jack's articulation was at 95%! When we went to Houston it was 50%. Now our focus will be on him using these tools automatically - without the need of verbal or non-verbal cues. We are so pleased with his achievement!

16p11.2 Family Event Chicago, Illinois ~ July 13 - 14, 2012

Highlights to include:

- Friday evening—An informal family gathering at the hotel with light refreshments
- Saturday—Educational sessions/presentations
- Saturday evening—A family dinner event
- Sunday—Additional research opportunities for interested families

The hotel will be close to attractions such as Navy Pier, Miracle Mile, Shedd Aquarium, the Field Museum, Millennium Park, and more!

Registration & additional information will be available mid-January 2012
Registration will include a small administrative fee.

Watch the website for more details!

SIMONS VIP CONNECT

VARIATION IN INDIVIDUALS PROJECT



Advanced Brain Imaging to Better Understand 16p11.2



The University of California San Francisco (UCSF) and the Children's Hospital of Philadelphia (CHOP) have begun seeing eligible patients with 16p11.2 deletions or duplications for advanced brain imaging. Mari Wakahiro, project manager for the Simons VIP Imaging Core at UCSF, answers a few questions!

What can we learn from this part of the study? The advanced brain imaging will focus on obtaining a more in depth understanding of the structure and function of the brain and how it works and connects in individuals with 16p11.2.

Who are the subjects and how they are chosen? We are inviting individuals with 16p11.2, over the age of 7 years, who completed the MRI scan at their initial visit. We expect that some people will not be able to understand or respond to tasks or pay attention long enough to finish.

How many subjects have been seen and how many total do you expect to see? UCSF has seen 10 subjects and CHOP has seen 3. Our goal is to have 40 subjects each year (20 per site).

Who will arrange travel? Like the first visit, Pro Travel will arrange travel from the participant's home to San Francisco/Philadelphia along with hotel ac-

commodations. Local travel arrangements including transportation will be arranged by the site coordinators.

How long is the visit and what tasks are required? The visit includes two days of assessments and one to two days for travel.

During the visit, subjects will undergo three assessments: neuropsychological testing which takes about 2-4 hours and two different brain imaging scans—each take about 2-3 hours, during which participants look at pictures and listen to recordings and are asked to respond. Both scans are non-invasive procedures.

The feedback that we have received from participants has been positive—subjects have enjoyed their visits and found the assessments to be interesting.

Do subjects receive results of their testing? All participants will receive a CD of MRI images that they can share with their personal physician.



The UCSF Imaging Team: (l to r) Maxwell C, Pratik M, Hana L, Rita J, Mari W, Elliott S (Director of the Imaging Core), Julia O, Polina B, Brianna P, Tracy L, Nick P, Katie M; **Not pictured:** Sri N, Corby D, Elysa M, Alistair M, Vivek S, Jane Y, Leighton H

For more information, see the fact sheet on the website under "Research Opportunity -> North America"

Watch for our next newsletter which will introduce the teams at UCSF and CHOP and discuss what it is like to visit an imaging center!

SIMONS VIP CONNECT

VARIATION IN INDIVIDUALS PROJECT

16p11.2 Family Slideshow

A selection of photos from the 16p11.2 family slideshow that was shown at the 2011 family gathering has been posted on the website.

To request a copy of the full slideshow set to music, please email Bethanny (bspackard@geisinger.edu) with the address that you would like this disc sent.

Many thanks to the families who shared their pictures and to 16p mom Belinda Chatman for putting this together!

Family Pictures Needed!

The Global Rare Diseases Registry (GRDR) has requested pictures of 16p families to rotate through the banner on their home page. If you would like to submit a photo, please email it to Bethanny (bspackard@geisinger.edu).

GRDR is registry database created by the NIH Office of Rare Diseases to collect de-identified patient clinical information to accelerate clinical research. Simons VIP Connect is cooperating with the GRDR to increase research opportunities and has agreed to put a subset of our data in the global database.

For more information, see their website at: <http://www.grdr.info>

Check Out What is Going on with Other 16p Families!

Make sure to check the discussion boards on the website to see what other families are talking about and share your own experiences!

Recently discussed topics include:

- * Weight
- * Behaviors and educational therapies
- * Diabetes and webbed toes
- * Seizures
- * ...and more...!

FAMILY SPOTLIGHT -TELL US YOUR STORY!-

If you would like your family to be featured in an upcoming newsletter, email answers to the following questions to **Bethanny** at bspackard@geisinger.edu.

Families will be featured in the order they are received. You may also attach a picture.

- Name
- Deletion or Duplication
- Age(s) of family member(s) with 16p
- How was your child diagnosed?
- What are your child's strengths?
- What problems does he/she have?
- What therapies does he/she receive?
- What does your child/family do for fun?



Updates from Simons VIP Connect



SIMONS VIP CONNECT

VARIATION IN INDIVIDUALS PROJECT

Clinical Evaluation sites still busy!

Site	# Seen	# Scheduled
Baylor College of Medicine	18	6
Children's Hospital of Boston	24	12
University of Washington	25	4

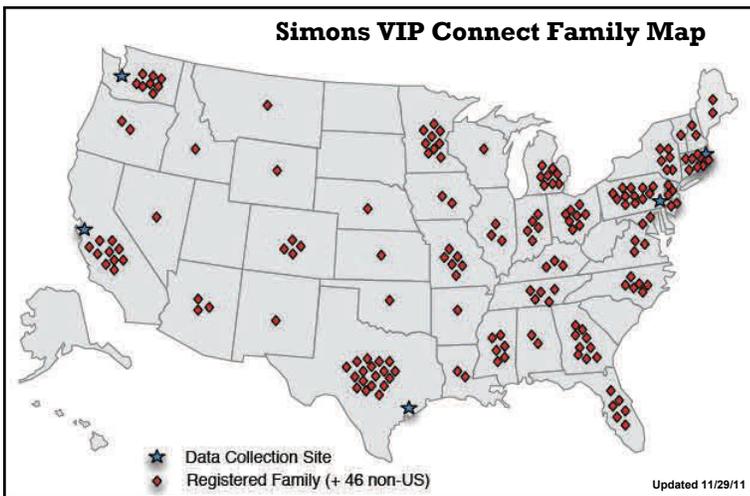
All About Simons VIP Connect Families:
 233 Families— 175 in the USA,
 including 42 states and 46 internationally
 # Families with 16p11.2 Deletion—132
 # Families with 16p11.2 Duplication— 60
 # Families with other genetic changes— 25

In the last week...

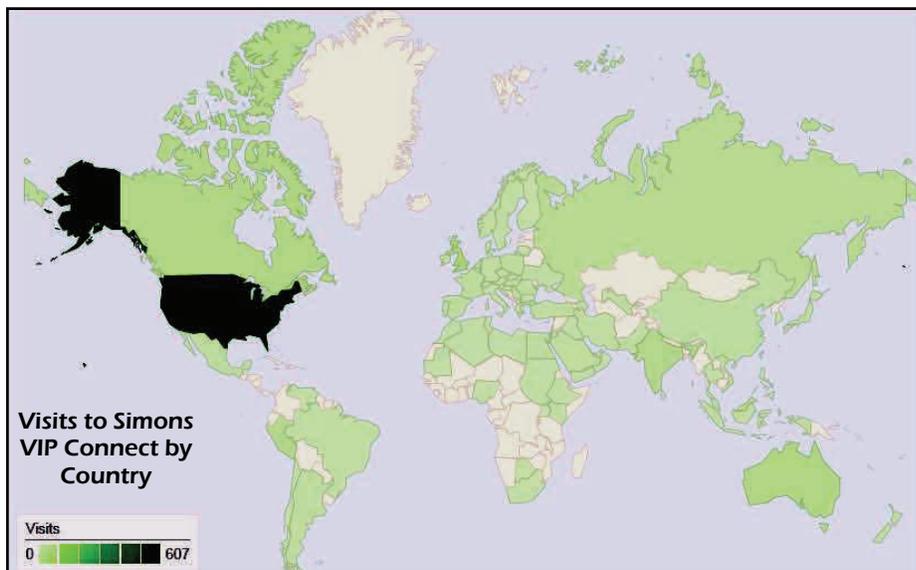
There have been 1006 visits to Simons VIP Connect

By 755 visitors

59% are first time visitors!



People all over the world visit Simons VIP Connect



Top 10 Countries to Visit Simons VIP Connect

1. United States
2. United Kingdom
3. Australia
4. Canada
5. India
6. Peru
7. Lebanon
8. Russia
9. Malaysia
10. Vietnam



Contact the Simons VIP Genetic Counselors:

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