



# Spring 2012 Newsletter



## SIMONS VIP CONNECT

### VARIATION IN INDIVIDUALS PROJECT

#### Simons VIP Expands Community and Research Study!

You may have noticed some recent updates to the Simons VIP Connect website; the Simons Variation in Individuals Project has added Class 1 deletions and duplications in 1q21.1 to the research study and the online community!

In general, there are two sizes of variants that may occur in 1q21.1. Class 1 changes are smaller than Class 2, closer to the tip of the long arm of the chromosome, and do not include the TAR syndrome region.

Like individuals with 16p11.2, those with 1q21.1 deletions and duplications may have features that include autistic-like behaviors, developmental delays, and behavior problems. Also like 16p11.2, these features may range from mild to severe.

We will continue to update the Simons VIP Connect website to include more information specific to 1q21.1; please be sure to check back in the coming weeks.

Please join us in welcoming these families to our community!



#### Join us for a Webinar with Jasmine Urquhart, Speech Therapist



We have posted a webinar by Jasmine Urquhart, M.S., CCC-SLP, Pediatric Speech-Language Pathologist. Jasmine will be joining us for a **live question & answer session on Sunday, March 18th at 2 PM EST.**

After the Q&A webinar is posted, you may submit any additional questions until 3/25/12. The answers to any remaining questions will be posted on the website in April.

Please view the webinar prior to the Q&A session and submit your questions to Bethanny (bspackard@geisinger.edu).

You can register for the live Q&A session with Jasmine here: <http://bit.ly/yJUXpA>

Ms Urquhart is the executive director and chief clinician at S&L Therapy. She received both her Master's Degree in Speech-Language Pathology and her Bachelor's Degree in Communication Disorders from Boston University.

She is licensed by the State of Massachusetts and certified by the American Speech-Language and Hearing Association (ASHA), and specializes in the speech and language disorders of young children ranging from early childhood through the middle school years.

Ms Urquhart has a diverse background of clinical experiences including work in settings such as hospitals, public and private schools, and private practice. She has over 10 years of experience working with children in the Boston Area.

#### 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](mailto: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?



## Visiting the Advanced Brain Imaging Sites

In our last newsletter, we featured an article about the advanced brain imaging component of the Simons VIP study. In this newsletter, we focus on what it is like to visit the University of California San Francisco (UCSF) or the Children's Hospital of Philadelphia (CHOP).

### What is a typical visit like?

The visit usually involves one and a half days of assessment. On the first day at UCSF, subjects will have a short session of neuropsychological testing in the morning, a break for lunch and undergo magnetoencephalography (MEG) in the afternoon. On the following morning, subjects travel to UC Berkeley for a functional MRI (fMRI), which takes about 2 hours.

At CHOP, subjects complete an fMRI on the morning of the first day and have the afternoon to explore the city. On the second day, subjects complete a short neuropsychological assessment in the morning and the MEG scan after lunch.

During the fMRI and MEG, subjects can watch a movie for the first half of the session, but will need to respond to questions and do simple tasks for the second half of the session.

**For more information, see the fact sheet on the website: Research Opportunity -> North America -> Advanced Imaging"**

### How can families prepare for their visit?

To prepare for this visit, subjects are sent a behavioral kit for the children to get used to the experience of being in the scanner – This involves reviewing the picture book, practicing wearing earplugs and listening to the MRI sounds.

### Who will families meet during their visit?

At UCSF, families will meet clinicians Elliott Sherr (PI), Brianna Paul (neuropsychologist), and Rita Jeremy (developmental psychologist), research assistants, Polina Bukshpun and Nick Poman, and project manager, Hana Lee. Families also meet a Child Life Specialist who can provide additional assistance during the visit and the hospital liaison dog, Lilani, who will visit during breaks in the playroom.

At CHOP, families will meet clinicians Tim Roberts (PI), Lisa Blaskey (neuropsychologist) Sudha Kessler (neurologist), research assistants - Sarah

Khan, Kory Heiken, Matt Lanza and Katherine Hines, and project manager, Saba Qasmieh.

Hana and Saba guide families through the entire process, from travel arrangements to all the details of the study appointments.

Both teams enjoys having SVIP families visit and look forward to creating a bond with them over the several day visit!

*Thank YOU for having us out there! You guys were great and I enjoyed my time even during the study. I think you all did a great job making us feel so welcome. — Deborah (mom to Cinco)*



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### Where do families stay in San Francisco and how can they get around?

Families often stay at a hotel near popular destinations with a lot of tourist attractions. A car service provides transportation to and from the airport and to all appointments.

### What is there to do in San Francisco?

Families often visit the San Francisco Zoo, the Golden Gate Bridge, various museums or the Lucas Film Headquarters to check out the Yoda statue (below). San Francisco is famous for world-renowned restaurants and diverse selection of great food. Whatever your interests, the team can help you find an exciting activity during your visit.



(Clockwise from top left): Family visit at UC Berkeley; Cinco wins a prize after finishing MEG scan at UCSF; MEG scans with EEG coils which measure eye blinking movements; Setting up mirror to start fMRI tasks.

### Where do families stay in Philadelphia?

Families stay at the Hilton Inn at Penn near the site where the neuropsychological assessments are performed and The Children's Hospital of Philadelphia- Main Campus. The hotel is about a 5 minute cab ride away from both sites.

### What is there to do in Philadelphia?

Philadelphia is the 5th largest city in the US and one of the world's most vibrant destinations. Most famous as the birthplace of American independence and home of the Liberty Bell and National Constitution Center, Philadelphia offers a unique variety of attractions, culture, and entertainment.



## Information about the 2012 16p11.2 Family Meeting

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The Simons VIP Connect 16p Family Meeting will be held on the weekend of July 13-15, 2012 at the Westin River North in Chicago, Illinois. The program is open to members of the Simons VIP Connect community and their family members. This meeting will focus on information specific to 16p11.2 members; however, 1q21.1 families are welcome to attend.

### Location

Since Simons VIP Connect families live all over the country, Chicago was chosen as a central site for the family gathering



### Cost & Registration

Reducing cost to families has been a top priority in all planning considerations. A small registration fee reserves your place at the meeting, and will be used to lower the overall cost for all families. The registration fee has been kept very low compared to other national meetings of this kind. In addition, a greatly reduced room rate has been negotiated for attendees. This rate only applies for the nights of July 13 and 14, for conference attendees. The registration and hotel fees also cover the costs of all meals on Saturday and breakfast on Sunday.



### Hotel

The Westin River North was chosen after visiting a variety of hotels in the downtown Chicago area. The Westin best met our space requirements, provided the best layout for our meeting, was accommodating for individual needs, and was competitively priced compared with other area hotels.

If the Westin does not meet your family's needs, you are free to reserve alternate accommodations. Be sure to consider that the rate given for the Westin includes the 16.4% tax charged at any downtown hotel and that there is a charge to park at almost all downtown Chicago properties.

### Meals

On Saturday, breakfast, lunch, dinner, and snacks during breaks in the meeting will be provided. In addition, breakfast on Sunday will be available. When you register, please indicate any special meal accommodations that you or your family members require. We will do our best to meet your needs, and will contact you with any questions.

### Childcare

There is a kids program planned for children ages 5 - 12yrs and will be free of charge to you. This will be offered during the Saturday meeting from 8:00 am to 4:00 pm. Space will be limited and provided on a first come, first served basis. It is best to have alternate plans for your children during the times you will be in session, so you and others are able to get the most out of the information provided. The hotel may have recommendations for other local providers.



### Conference

The conference agenda is posted on the registration website. Presentations will address topics such as current 16p11.2 research findings, speech therapy, and behavior management. Presentations will be videotaped and posted to the Simons VIP Connect website after the meeting. Our team is looking forward to meeting everyone and will be available to answer informal questions; however, we cannot guarantee that your family will receive one on one time to discuss your child's specific needs.



### Research Opportunities

Additional research projects will be offered during the day on Saturday

and Sunday. If you indicate interest during registration, a coordinator will contact you regarding scheduling prior to the conference. Participation in any research projects is voluntary; you may take part of as many or as few of the projects as you wish.

### Chicago Activities

A shuttle bus will be available to take interested families to Navy Pier or Millennium Park on Saturday morning, with pick-up on Saturday afternoon. We have arranged for several shuttle pick-up and drop-off times. You may also use other modes of transportation (bus, taxi). All transportation provided as part of the meeting will depart and return to the Westin River North.



For more information about what to do in Chicago, check out Chicago's official tourism site - [www.explorechicago.org](http://www.explorechicago.org).

For more information about the family meeting and to register to attend, please visit the website: [www.simonsvipconnect.org](http://www.simonsvipconnect.org)

Early bird registration ends April 8th!

Please send additional questions regarding the family meeting to [chicagofamilymeeting@gmail.com](mailto:chicagofamilymeeting@gmail.com)

Please submit questions to be answered at the meeting via the "Ask an Expert" page (bottom left of the Simons VIP Connect home page).

# Updates from Simons VIP Connect

## Simons VIP Connect reaches 300 members!

### All About Simons VIP Connect Families:

- 312 Families— 230 in the USA, including 44 states and 9 countries
- # Families with 16p11.2 Deletion—159
- # Families with 16p11.2 Duplication— 78
- # Families with 1q21.1 Deletion—12
- # Families with 1q21.1 Duplication— 3
- # Families with other genetic changes— 31

### Clinical Evaluation sites still busy!

Site	# Seen	# Scheduled
<b>Baylor College of Medicine</b>	25	14
<b>Children's Hospital of Boston</b>	27	14
<b>University of Washington</b>	30	12

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## Simons VIP Clinical Site Visits



L-R: Jennifer (mom), Ricky (brother), Brent (dad),

We were told the reality of what Sydnee is capable of doing. We found her non-verbal IQ to be in normal range, so our expectations grew. Also, having the doctor sit down with my other children and explain Sydnee's condition to them [was a positive experience]! - Jennifer, mom to Sydnee (16p11.2 del)

This was very important for us. The visit to Houston gave us our first official diagnosis of autism, which is important for school IEPs and also for future treatment. It is also hard to explain how cool it was to celebrate and be rewarded for Grayson's genetic deletion - he was so proud to be selected for the research, one of the first true positive experiences he has had as a result of his genetic deletion. -Nataynya, mom to Grayson (16p11.2 del)

While intense and overwhelming at times, the team was fantastic. They understood our son's fears, gave us breaks when he needed them and were amazing people all around. It also felt good to know that our son is a piece of some really important research that could make a huge difference in the quality of his life and the lives of others. -Julie, mom to Dylan (16p11.2 del)



Benjamin and dad on the plane to Seattle!

The interviews and records gathering prior to and to a degree during the study was somewhat repetitive, but I guess it was different people in different sites needing the information. All in all, I think the process is organized and very considerate of schedules. -Heather, mom to Justin (16p11.2 del)

Everyone was very friendly and professional. It confirmed what we already knew about our child (his speech delay). It did ease our fears regarding autism.—Julie, mom to Benjamin (16p11.2 del)

### Contact the Simons VIP Genetic Counselors:

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### Contact the Simons VIP Newsletter Editor:

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