



Winter 2012-2013 Newsletter



Newsletter Editor:

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SIMONS VIP CONNECT

VARIATION IN INDIVIDUALS PROJECT

Save the Date — 2013 Family Meeting!

The 2013 Simons VIP Connect Family Meeting will be held in Orlando, Florida July 19th-21st, at the family friendly Hilton Orlando Lake Buena Vista hotel!

The Hilton is in the Walt Disney World Resort, situated next to the Downtown Disney Marketplace and Downtown Disney West Side and Cirque de Soleil. The hotel offers complimentary transportation to and from Disney theme parks and access to Disney's exclusive Extra Magic Hours benefit — providing longer fun and shorter queues.



Registration will open in early March. Hotel reservations will be made through the registration website. Please do not call the hotel directly. There will be ample time to make your reservations. Simons VIP Connect family guests will be extended a special discount room rate of \$99/night and \$129/night per suite. On Friday night the Simons Foundation will host a dinner and a movie at the hotel. The meeting will be all day Saturday, concluding around 4:00pm. Saturday night families will be on their own to enjoy the parks. Research projects will also be available for participation, starting as early as Friday afternoon and finishing up mid-day on Sunday.

Since July is one of the busiest months at Walt Disney World, we know many of you want to get busy planning your stay, including the possibility of adding days before and/or after the meeting itself. We are happy to say the negotiated room rate is in effect up to 3 days before and three days after the meeting dates.

We are very fortunate to have families directly participating in the meeting planning and presentations this year! We know of 2 Simons VIP Connect parents who are Disney travel specialists and have offered to help families navigate the decision making for park pass and dining plan purchases. We also have a parent who specializes in advocacy as a presenter for Saturday, so stay tuned!



One item that may be of interest to some families is the Disney character dining experience. This is not a part of the Simons VIP Connect family event (though we hope to have a character pop in to say hello at some point!), however we wanted to highlight this since it sounds like spaces go quickly and the reservation period will be opening soon. As with any park pass purchases, Disney character dining reservations would be arranged and paid for directly by you.

Here is the information from one of our VIP family travel specialists:

Guests can make dining reservations 180 days in advance. The two most popular restaurants are Cinderella's Royal Table (in the castle in the Magic Kingdom and requires park admission) and Chef Mickey's (in the Contemporary Resort and does not require park admission). Dining reservations can be made online at <https://disneyworld.disney.go.com/dining/#/reservations-accepted>. You can also call 407-WDW-DINE. All character restaurant reservations will require a credit card to hold them and some such as Cinderella's Royal Table will require payment in full. All reservations can be cancelled for a full refund within 24 hours of the reserved dining time.

We're very excited for this summer's event! Watch your email and the website for more details!

Help Needed: EEG Tracings!

Reviewing medical records is an important part of Simons VIP research. We are especially in need of EEG tracings, which are the print-outs that allow doctors to determine if someone is having a seizure, and what kind. We are interested in looking at tracings from both people who have seizures and those who do not. This is important so that the neurologists working on the project can learn more about the relationship between 16p variations and seizures.

To obtain these records, we need your permission in the form of a signed medical records release. Releases have been sent to families where our records indicate that an EEG was done. These have been sent via email and snail mail. About 60% of these forms have been signed and returned, but we are still waiting for many of them to come back.

For a copy of the release form, please email coordinator@simonsvipconnect.org



Simons VIP Connect Updates

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Check out our New Webinar!

“All I Really Need is an iPad, right?” Myths & Realities of iPads for Families of Individuals with Special Needs Kelly A. Johnson, PhD; with contributions from Jo Ristow MS, CF-SLP



During this presentation, Dr. Johnson shares lessons learned through an iPad pilot project, including points to consider before deciding to buy an iPad for your child, guidelines for making the iPad even more helpful for your child, and resources related to using the iPad with individuals who have special needs. Dr. Johnson has also included video demonstrations of several different Apps, as well as the "Guided Access" capability on newer iPads.

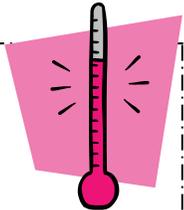
**“Awesome webinar! A great resource for parents and teachers.”
See page 4 for additional resources!**

Dr. Johnson is a licensed clinical psychologist at the University of Washington Autism Center, where she provides diagnostic evaluations, individual and family consultation. She is also involved in research and training activities. Dr. Johnson has led the UW Autism Center’s Autism Interactive Project which, with the help of a donation from Easter Seals, was a pilot project involving the use of iPads with children who have Autism Spectrum Disorders.

Watch for our next webinar with Dr. Elliott Sherr, principle investigator of the brain imaging portion of the study, at the end of January!

Survey on Infections, Temperature and Fever

During the Simons VIP Family meeting in July, several families raised concerns about the number of infections their children had experienced and stated that their children’s daytime or night time temperature was higher than normal. In response we posted an online survey in September to determine the frequency of these problems. 109 families responded to the survey. Survey answers were completed for 75 16p11.2 deletion individuals along with 70 of their siblings. Survey answers were completed for 35 16p11.2 duplication individuals along with 27 of their siblings.



The survey about infections showed that children with the 16p11.2 deletion AND duplication more frequently have minor infections compared to their brothers and sisters without the deletion. The most common of these infections were colds, and 24% 16p11.2 deletion carriers and 26% of duplication carriers had 5 or more colds in the last year. There were slight increases in other types of infections including ear infections, bronchitis, pneumonia, sinusitis, urinary tract infections, and gastrointestinal infections. Overall 68% of deletion carriers and 60% of duplication carriers required antibiotics at least once last year, at rates about 15% higher than their brothers and sisters.

Parents of 16p11.2 deletion carriers report that 35% of their children’s temperature is abnormal during the day and 17% are abnormal at night with 27% sweating excessively during the day and 35% sweating excessively at night. Parents of 16p11.2 duplication carriers report that 46% of their children’s temperature is abnormal during the day and 40% are abnormal at night with 31% sweating excessively during the day and 57% sweating excessively at night.

	16p Del Probands	16p Del Siblings	16p Dup Probands	16p Dup Siblings
Temp during day different than normal	34.7%	5.7%	45.7%	20%
Temp at night different than normal	17.3%	2.9%	40%	25%
Sweating excessively during day	26.7%	2.9%	31.4%	10%
Sweating excessively at night	34.7%	2.9%	57.1%	15%



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Advanced Brain Imaging Update

The University of California, San Francisco (UCSF) and the Children’s Hospital of Philadelphia (CHOP) are inviting eligible participants to take part in the next major phase of the Simons VIP. The investigators at UCSF and CHOP are using cutting edge brain imaging to help us understand the connection between changes in brain structure and their functional consequences in individuals with 16p11.2 and 1q21.1 deletions and duplications. This study may also help develop biomarkers for autism diagnosis and treatment.

This portion of Simons VIP includes 2 days of assessment and 1 to 2 days of travel. During the visit, participants will complete three study components: neuropsychological testing (2-4 hours) and two different brain imaging scans (about 2-3 hours each), in which participants play games, look at pictures and videos, and listen to different recordings.

Almost 50 participants with a 16p11.2 copy number variant have already completed this portion of the Simons VIP study, and we are excited to welcome several families with 1q21.1 copy number variants in early 2013. Our goal is to recruit an additional 50 participants to complete the advanced brain imaging. Like the first Simons VIP visit, hotel accommodations and travel will be arranged and paid for by the study. Although we are still recruiting families for this study,

we are already seeing novel and exciting preliminary group results that we hope will be published in the near future (we plan to share these preliminary results in more detail at the family meeting in Orlando this summer and will make the published manuscripts available to all participants). These initial findings suggest that the brain “wiring” is altered (but in different ways) in both 16p11.2 duplication and deletion carriers. Enrolling and testing more participants will help us confirm these results and will enable us to ask whether these changes in brain wiring also correlate with how clinically affected deletion and duplication carriers may be.

We are proud and happy to say that we have received kind feedback from the families who have participated in this further imaging phase. Everyone has found the testing to be interesting and unique. In addition, visiting families have enjoyed the sightseeing and culinary opportunities offered in San Francisco and Philadelphia. The teams in San Francisco and Philadelphia are excited to meet more families and welcome you to participate. We are grateful for the important contribution that your family is making to better understand the biology of copy number variations.

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

Find other families on Facebook: “Gathering Group – 16p VIPS”

“When myself and other families first started talking about meeting up, we were just kicking around what to call ourselves and “Gathering of the Gremlins” just sort of evolved. It was never meant as anything derogatory. But as new families continued to join our group, there were many that found the name distasteful. So, out of respect for our new and older families, we decided on a new name.

16p VIPS was a great blend of who we are & the link we have to Simons. So many of us have felt truly blessed to be in the research and to learn so much from the research that is being done on our kids. VIPS can stand for the Simons VIP – or it also means our Very Important Persons!”

- Dawn Miller



A Note from Andrea—

After almost two and a half rewarding years with the Simons VIP project, I will soon begin a new chapter in my career. I have accepted a clinical genetic counseling position at an Atlanta hospital where I will begin mid-January. I have truly valued the relationships that I have developed with so many of you and your families - both over the phone and in person at the meetings in Gatlinburg and Chicago. I wish each of you and your children the best and know that Simons will continue to serve as an unparalleled resource and advocacy group for this community.





RESOURCES

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Interested in learning more about how to fundraise?

Check out some of these websites for fundraising tips and ideas!

- <http://www.thefundraisingauthority.com/>
- <http://www.gofundme.com/>
- <http://www.partylite.com/en-us/Pages/fundraisers/fundraisers.aspx>
- http://www.squidoo.com/101_ways_to_fundraise

Fundraising Resources

Web Resources!

HerSelf First: A resource for the caregiver, designed to provide support, strengthen resilience and enhance self-worth. This website features articles about issues that impact caregivers, articles written by caregivers, practical tips and daily motivational resources. <http://herselffirst.com/>

NICHCY—National Dissemination Center for Children with Disabilities: Provides information for children, parents and family members with disabilities, as well as for the early intervention and education professionals caring for these individuals. <http://nichcy.org/>

Resources include:

- Fact sheets about specific disabilities
- Materials to raise awareness of disability
- Easy to read articles about education and disability laws
- Lists of state and national disability agencies and organizations
- Monthly newsletters—past newsletters have addressed behavior, rare disorders, effective teaching practices, inclusion and access to general curriculum, and much more!



Genetic Alliance: A nonprofit health organization focused on advocacy in genetics, including education, building awareness, and developing registries, biobanks, medications and tests. The network includes over 1,000 disease specific advocacy groups, thousands of universities, private companies and public policy organizations who share resources, tools and programs. Subscribe to their “Weekly Bulletin” to receive announcements regarding current news topics, upcoming events and more! <http://www.geneticalliance.org/>



Technology and Autism: What’s available and What Works From the Autism Consortium

This article discusses currently available assistive technology, how these options may help your child, and how to determine what is the best fit for your child. There are also tips included about how to get an assistive technology evaluation and what your rights to request this evaluation are. Examples of available devices/apps/options are provided, as well.

<http://www.autismconsortium.org/blog/detail/technology-and-autism-whats-available-and-what-works>

Testing Autism and Air Travel

From the New York Times

In the past couple years, a number of airports have begun to offer families with autistic children the opportunity to participate in “mock boarding” situations where they are able to practice buying tickets, going through security, and boarding the plane. This article provides information, tips, and other resources to help passengers with disabilities and their caretakers navigate the airport.

http://travel.nytimes.com/2012/10/28/travel/testing-autism-and-air-travel.html?pagewanted=all&_r=0

Contact the Simons VIP Genetic Counselor:

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