



# GenomeConnect Fall 2017 Newsletter

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Dear GenomeConnect,

Thank you for your participation in GenomeConnect!

Since our last newsletter, GenomeConnect has continued to grow because of all our wonderful participants (like you!) who want to share their data, connect with others, and help promote genomic discovery!

## In this Newsletter:

GenomeConnect Enrollment Update!

What do your genetic test results really mean?

Celebrate Family History Day!

Meet the Genetic Counselors on our GenomeConnect Team!

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Connect with GenomeConnect via social  
media, phone, and email!

email: [info@genomeconnect.org](mailto:info@genomeconnect.org)

phone: 570-214-1721 (toll-free 855-322-7683)





# What do your genetic test results really mean?

## Copy Number Variant

A change is found in the amount of genetic material a person has. They have too much (a duplication) or too little (a deletion) of a particular genetic region.

If you think of all our genetic instructions as a library, a copy number variant means a person is missing some books or has some extra books.



## Single Nucleotide Variant

A spelling change is found within a gene. A person has the right number of books in their library, but there is a spelling mistake in one of the books.

Sometimes, we have spelling changes that our body can overcome. Other times, spelling changes cause health or developmental differences.



## Pathogenic

A copy number or sequence variant that is known to cause an individual's symptoms or features

The genetic change has been seen before or is known to impact the body's ability to use that region of the genetic code



## Variant of Uncertain Significance

There is not enough evidence that the copy number or sequence variant causes an individual's symptoms or features

The variant may eventually be reclassified as pathogenic or benign



## Benign

A copy number or sequence variant that is not known to cause any health or developmental differences

The variant has been seen in healthy individuals or is known not to impact the body's ability to grow and develop



If you have questions about your genetic test results, reach out to the healthcare provider that ordered your testing or a genetics expert in your area. To find a genetic counselor near you, you can use the following website:

<http://www.nsgc.org/page/find-a-genetic-counselor>

# Celebrate National Family History Day!

Did you know that Thanksgiving is also National Family History Day?

This year, celebrate by collecting your family history. Gathering information about the illnesses your parents, grandparents, and other blood relatives had can help your doctor understand your risks and whether a genetic condition may run in your family.

Here is one tool to help you collect your history:

<https://familyhistory.hhs.gov/FHH/html/index.html>



# Meet the Genetic Counselors on our GenomeConnect Team!

November 9th is Genetic Counselor Awareness Day!

Genetic counselors have specialized training in genetics and counseling so they can interpret genetic test results and support patients. Genetic counselors work in many settings including clinics, hospitals, and research! Learn more about our genetic counselors here:

<http://www.nsgc.org/page/aboutgeneticcounselors>

Meet the genetic counselors on the GenomeConnect Team!



[Juliann Savatt, MS, LGC](#)

Juliann Savatt graduated from Allegheny college and went on to receive her Master's of Science in Genetic Counseling from the University of North Carolina at Greensboro in 2015. Juliann started her career at Geisinger and currently works at Geisinger's Autism and Developmental Medicine Institute as a genetic counselor and research coordinator. In terms of her research interests, she is motivated to empower patients to be active partners in genomic discovery and is working towards that goal by being a part of the GenomeConnect team.



[Erin Rooney Riggs, MS, LGC](#)

Erin Rooney Riggs is a certified genetic counselor. She received her Master's degree in Genetic Counseling at Northwestern University in 2007, and started her career as a clinical genetic counselor and research coordinator at Emory University. She worked at Geisinger from 2013-2017 as a coordinator for the Clinical Genome Resource ([www.clinicalgenome.org](http://www.clinicalgenome.org)). Erin continues her work with ClinGen and now is an assistant professor at Geisinger's Autism and Developmental Medicine Institute.



[Danielle Azzariti, MS, CGC](#)

Danielle Azzariti is a certified and licensed genetic counselor. She trained at Brandeis University and has been working as a genetic counselor in the Boston area since 2009. She started her career as a clinical genetic counselor and research coordinator for the Massachusetts General Hospital Neurogenetics Program. She has been working at the Laboratory for Molecular Medicine at Partners HealthCare Personalized Medicine since 2013 as a coordinator for the Clinical Genome Resource and Matchmaker Exchange.



[Emily Palen, MS, LGC](#)

Emily Palen graduated from Hamilton College with her Bachelor of Arts in Neuroscience and went on to Boston University's School of Medicine for her Master's in Genetic Counseling. Upon graduating in 2016, Emily moved back to her home state of Pennsylvania to work at Geisinger's Autism and Developmental Medicine Institute as a genetic counselor and research coordinator. Emily is particularly interested in the study of developmental brain disorders, the promotion of health literacy and health empowerment, and the creation of opportunities to exchange data and information to improve patient care.



[W. Andy Faucett MS, LGC](#)

Andy Faucett directs community engagement and education for Geisinger's biobank, the MyCode® Community Health Initiative. His research focuses on genetic testing, ethical and consent issues for genetic research, genetic counseling, and patient engagement for research. He serves on the Geisinger IRB and the IRB leadership team. He helped develop and provides oversight for GenomeConnect. He also is involved in the ethical, legal and social issues aspect of the ClinGen with the CADRe project. Faucett has a B.S. from the Baptist College at Charleston, M.S. in Human Genetics from Sarah Lawrence College, certification from the American Board of Genetic Counseling (ABGC) and is licensed as a Genetic Counselor in Pennsylvania. He previously served on the NSGC, ABGC and National Coalition for Health Professional Education in Genetics Boards of Directors, and the American Society of Human Genetics Information and Education Committee. He held positions at Emory University School of Medicine; Baylor College of Medicine; Memorial Medical Center, Savannah, GA; and the Center for Disease Control and Prevention (CDC) prior to Geisinger.