GenomeConnect Newsletter

Fall 2018

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Share Your Story With Us!

Have you received an update about your results from GenomeConnect, your doctor, or the genetic testing lab?

Email us if you would be willing to share your story and experience with us!

Share Your Story With Us!

Your doctor likely ordered genetic testing for you (or one of your family members) in order to find a cause for your particular health problem(s) or to find out if you were at risk for future health problems. Your results may have listed one or more genetic changes, each with an "interpretation" (what the laboratory thought each change meant in terms of your health). They may have used terms like "Pathogenic," "Variant of Uncertain Significance," etc. Over time, as we learn more and more about genetic changes, interpretations may change.

Have you received an update about your results from GenomeConnect, your doctor, or the genetic testing lab? When your results were updated, did this impact your medical care? When initially having testing were you aware that genetic test results may change over time?

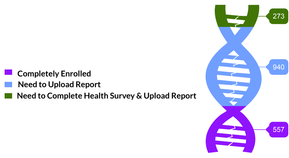
Email us if you would be willing to share your story and experience with us!

Click here to email us

Participation Update

As of August 2018, GenomeConnect has over 1770 participants. Of participants, 557 have completed all steps to enroll! This means that they have enrolled, completed their health survey, and uploaded a copy of their genetic testing report!

In total, 1213 participants are enrolled, but need to take action to completely participate! 940 participants have enrolled and completed their health survey, but have not uploaded a copy of their report, and 273 participants need to complete their health survey and upload their report!



If you haven't complete your health survey or haven't uploaded your report, do so today! To complete the health survey, login to your GenomeConnect account and click "GenomeConnect Survey" under "Incomplete Surveys."

If you haven't uploaded your report, check out this short video that shows you how - http://bit.ly/UploadGC

Working with Other Patient Groups

Geisinger is a ClinGen grantee that coordinates GenomeConnect. To help more patients share their genetic and health data, the team at Geisinger plans to help rare disease patient advocacy groups and other rare disease registries collect and share genetic and health information.

Data sharing can ...

Help us better understand genetic changes and can help others get clearer genetic test results

Increase our understanding of a condition's features, which may help identify possible interventions and treatments

Give you the option to receive updates about your genetic test results

To learn more about the Patient Data Sharing Program, visit the ClinGen website: http://bit.ly/ClinGenData

Ask our Genetic Counselors - What are Inheritance Patterns?

We all have two copies of almost every gene in our body, one copy from our mother and one copy from our father. Whether or not we show features of a particular genetic condition depends on many factors, including the condition's inheritance:

Autosomal Dominant

If a variant in only one copy of the gene is enough to cause symptoms or features of the condition, the condition's inheritance is autosomal dominant.

Autosomal Recessive

If variants in both copies of the gene are necessary to cause symptoms or features of the condition, the condition's inheritance is autosomal recessive.

X-Linked

If the gene that causes the condition is located on the X chromosome, the condition is called X-linked.

X-Linked Dominant

Females have 2 X chromosomes, and if a variant in one copy of the gene causes the condition, the condition's inheritance is X-linked dominant.
Males only have one X chromosome (and a Y chromosome). If a condition is X-linked dominant, a variant in the only copy of the gene causes the condition in males.

X-Linked Recessive

If variants in both copies of the gene cause the condition in a female, the condition's inheritance is X-linked recessive.
A variant in a male's only copy of the gene causes the condition. Males are more frequently affected with these conditions since they only have one copy of the X chromosome.

Raw Data from At-Home Genetic Testing

Direct to consumer or at-home genetic testing is testing that you order online, by phone, or in some retail stores. The types of results returned through these at-home tests vary. Some give you information about traits (like your wine preferences), others provide ancestry information, and some return health information and carrier status. Some at-home genetic testing companies also offer the option to download your raw data from testing. These files include hundreds of lines of your genetic information in the letters A, T, G, and C.

This raw data is very difficult to interpret on your own. There are several online "third-party interpretation" services that offer analysis of the raw data from at-home genetic testing. These services can possibly provide you with more information, but it is important to understand that there are risks and limitations:

- Raw data has not been validated meaning that there are often errors. Because of this reports from these services can include false positives. This means that the service indicates an increased risk of disease when your risk is not actually higher than that of the general population. In many cases, it is important that results from a third party service are reviewed and validated.
- The results may include unexpected or upsetting information about your disease risk or family relationships. If this is the case, find a genetic counselor to help you understand and process these results.
- Privacy measures from these third party services vary. Be sure to read their privacy policy to know how your data is stored and used.

If you have questions about your raw data or reports from a third party service, find a genetic counselor in your area: www.nsgc.org/findageneticcounselor