

## **Introduction**

You are being asked to participate in GenomeConnect because you (or one of your family members) have had genetic testing, are considering genetic testing, and/or have been diagnosed with a genetic condition. The purpose of this registry is to collect information about genetic test results and health from people that have had or are considering the testing.

If you choose to participate, you may be providing information about yourself, a family member, or someone you have legal guardianship over. Keep in mind that, throughout this document, “you” will refer to the person whose information is being shared in this registry. For example, if you are enrolling your child for GenomeConnect, every time it says “you” we mean “your child.”

The ClinGen Resource Project, which includes the GenomeConnect registry, is funded by the National Institutes of Health (NIH). The registry system has been developed and is hosted by PatientCrossroads, a company specializing in patient registries. There is no cost to you to participate.

### **What is the purpose of this registry?**

Though everyone’s DNA code is similar, there will always be changes from person to person. We can find these changes by doing genetic testing. New methods of genetic testing have allowed doctors and scientists to find more genetic changes than ever before, but we still do not understand how all of these changes affect our health. GenomeConnect is one way the information can be gathered for doctors and researchers to understand more. We will share the genetic and health information you provide to us, separated from your personal identifying information, in databases approved by GenomeConnect staff, so that further studies and research can be done to improve health care.

### **Who will be in the registry?**

GenomeConnect is open to adults (age 18 and older) and children (under age 18) who have the consent of a parent or legal guardian meeting at least one of the following criteria. Please check the box (or boxes) from the criteria list that describe you:

- Have already had genetic testing
- Are considering genetic testing
- Are family members of someone who has had genetic testing

Adults who are able to consent for themselves must do so, and a parent/guardian must consent for a child. Children ages 10-17 also have to provide assent to participate. We will contact people who joined GenomeConnect as children when they turn 18-years old, to get consent for them to continue participating. A child or adult who is not able to consent (for reasons like a cognitive disability) can still participate but must get help from their parent or guardian.

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## **What will I be asked to do?**

All information you provide will initially be stored on the secure PatientCrossroads computer systems.

1. You will fill out an initial online questionnaire about your health. It will ask about the type(s) of genetic testing you may have had, the results, and questions about your health and development.
2. You will upload a copy of your genetic testing report(s), if applicable. This allows us to learn important information about the particular genetic change(s) listed on a test report. If you do not have a copy of your report, registry personnel will help you try to get one. Once we receive your report, we will contact your testing lab and let them know you are participating in our registry.
3. We will stay in touch with you about new surveys and research studies that may apply to you. Once a year, we'll contact you to ask you to update your profile information so that it is up-to-date.

## **What information will be shared, and with whom?**

- The GENETIC TEST RESULTS you provide
- The HEALTH INFORMATION you provide

This information will be separated, or deidentified, from your personal identifying information like your name and contact information. We will NEVER share your personal contact information outside of GenomeConnect without your permission.

Deidentified information will be shared with the following groups so that it can be studied to improve health care:

- *The Clinical Genome Resource (ClinGen)*: ClinGen is a project funded by the NIH that is focused on understanding the role genetic changes play in human health and development. GenomeConnect is part of this project. ClinGen is the guardian of the information contained within the registry and is responsible for collecting, storing and sharing the data, but only GenomeConnect staff will have access to your personal identifying information, like contact information, for purposes of recontacting you.
- *Partner databases outside of GenomeConnect that have GenomeConnect approval*: These partner databases are accessible via the Internet, and they include both public access databases (that anyone can view) and controlled-access databases (only approved users can view). The public access databases will have limited information (like single genetic variants listed by themselves, not connected to other genetic variants). It would be unlikely that this general "one single variant" information could be tracked back and identified as you. To

learn more about the databases we have partnered with, please click on this link: [Partner Databases](#).

- *Other registered users of GenomeConnect*: This could include other participants (patients, family members, etc.) and members of the scientific community (doctors, researchers, etc. who want to learn from information in the registry).
- *Geisinger Institutional Review Board* (the committee that oversees this research)

I understand this section about who has access and what deidentified information is shared.

### **What are the risks to my privacy?**

The risks of participating in GenomeConnect, which we feel are minimal, involve the security of your information and your privacy. Because many genetic changes are rare, there is always the chance that you could be identified based on your genetic information alone. Other possible risks include potential breaches in the computer security system. We believe that the risk for these is low.

GenomeConnect takes these risks very seriously, and we work hard to protect the information we collect. Partnering with PatientCrossroads, a company specializing in registries, has helped us to lower risks. PatientCrossroads will store information we receive from you on its private computer systems, which have tight security measures in place to protect your information. You can learn more about PatientCrossroads security measures here:

<http://www.patientcrossroads.com/securityandprivacy.html>.

Before any of the information you provide is shared outside of GenomeConnect, we will remove your personal information such as name, date of birth, and address, and replace it with a unique number code only known/seen by approved staff involved with GenomeConnect. This code is stored in a secure place on the PatientCrossroads server and protected with a password.

Your personal information may be disclosed if we are required by law. Federal Privacy Regulations provide safeguards for privacy, security, and authorized access. If there are any breaches, a Federal law, called the Genetic Information Nondiscrimination Act (GINA), has been put in place to protect you against discrimination by health insurance companies, group health plans, and most employers. If we think you may have been affected by a breach in the registry's computer system or identified by any of your information, we will notify you.

### **What are the benefits of being part of this registry?**

You might not see any immediate personal benefits; however, your participation may help you, members of your family, and others with the same genetic change or disease in the future by

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improving understanding of genetics and health. By participating, there is a possibility you may be able to connect with other participants with similar conditions or genetic changes. We will notify you if someone, like another person in the registry or a health researcher, wants to get in contact with you. It is up to you whether you respond or not. If we receive information about research, medical advances, or other news that applies to you, we will make an effort to let you know about it.

At this early stage of genetic research, we may not find meaningful genetic information of use to you.

I understand this information about the risks and benefits of participating in GenomeConnect.

### **What are my rights?**

You do not have to join this registry. Participation is voluntary. Your decision to participate in this registry will not affect your health care.

You can also choose to stop participating in the registry at any time, and for any reason. We will stop all invitations for additional surveys and requests for patient or research contact. We will not be able to remove any information submitted before you withdrew.

### **Where can I get more information?**

If you have any questions about GenomeConnect, or want to withdraw your participation, please contact the registry coordinator at 570-214-1721 or email [info@genomeconnect.org](mailto:info@genomeconnect.org). To report concerns that result from your participation in the registry, you may use the contact information above, or contact study investigator, Andy Faucett, at 570-214-4862.

For questions about your rights as a research participant, contact the Human Research Protection Program staff of the Geisinger Institutional Review Board (the group of people who review this research to protect your rights) at (570) 271-8663.

I understand that I can choose to stop participating in the registry at any time and have been told how to contact study staff for more information.

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### ***Assent for Children Ages 10-17***

***If the person you are enrolling in GenomeConnect right now falls into the 10- to 17-year old age group, please read and complete this page. Otherwise, check this box to move ahead.***

### **Explanation of assent for 10-17 year olds:**

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Children ages ten through 17-years old need to agree to participate in the GenomeConnect registry to be enrolled. This is called “assent.” If the child is not able to understand the details of the study for reasons like a cognitive impairment, the child’s assent is not needed (i.e. it is “waived”).

Please have your child read (or listen to you read) the following information. He or she needs to type in his or her name or initials and date of birth to move forward with participating in GenomeConnect, but you can help them do it if needed.

**Please have the child (ages 10-17) read or listen to the following:**

You are being asked to take part in the GenomeConnect registry because you or a family member has had genetic testing, is thinking about having the testing, or has a genetic condition. The reason for GenomeConnect is for doctors to learn more about genetic reasons for health issues. This study may not benefit you personally, but doctors might find out something from your genetic and health information that may help other patients in the future or one day in the future, help you. You might be able to connect with someone who is similar to you.

Your parent or another adult who cares for you will be sharing your health information and genetic test results with us. You can help them answer the questions if you want. The people who run the study will see your name, your age, and some other details about you. When we share your health and genetic information with researchers and doctors, we won’t give out those personal details about you.

If you feel uncomfortable with a question we ask, you do not have to answer it. There is a chance someone could get your information out of our computer system or use your genetic information to figure out who you are, but the GenomeConnect staff does everything they can to make sure this does not happen.

You do not have to be in the study, and it is okay if you don’t want to be or if you change your mind in the future. When you turn 18, we’ll ask you again if you want to be involved. You and your parent/guardian need to let us know if you want to stop participating. By checking the box below, you are agreeing to participate in GenomeConnect.

I understand the contents of this form. I have had the opportunity to ask any questions, and if I had questions, they have been answered. I understand that I can print a copy of this form.

**Final page of consent for participating in GenomeConnect**

You must check the box below to give your consent to participate. Parents enrolling a child aged 10-17 also MUST check this box, even if child provided his/her assent on the page before this one.

**If you do not want to participate**, please exit this form without checking the box below. There will be no effect on your regular health care, medical treatment, or insurance benefits. Contact the GenomeConnect coordinator at 570-214-1721 or send an email to [info@genomeconnect.org](mailto:info@genomeconnect.org) if you are not sure and would like to ask questions before you decide.

**If you want to participate**, please check the box below. By doing this, you are consenting to participate in the registry. You will be able to change your mind and withdraw at any time by contacting the coordinator at the phone number listed above.

I understand the contents of this form. I have had the opportunity to ask any questions, and if I had questions, they have been answered. I understand that I can print a copy of this form.

**To contact Study Staff:**

Study Phone Number: 570-214-1721

Study Email: [info@genomeconnect.org](mailto:info@genomeconnect.org)

Study Address/Team:

Principal Investigator -- W. Andrew Faucett, MS, LGC

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Danville, PA 17822-2620

Ph: 570-214-4862

Study Coordinator -- Brianne E. Kirkpatrick, MS, LGC

Co-Investigators -- David H. Ledbetter, PhD, FACMG

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