

Well, here we are again. Last year in our IMPACT we recapped all that had changed in the world, all we had collectively lost and our enduring hope that when we sent our next IMPACT, the world would look remarkably different. Although some parts of our country are finally seeing sustained declines in infection rates due to vaccination, far too many people are still suffering the effects of our nation's worst pandemic. Inside this edition will be a message from our Executive Committee recapping this past year and our plans for the new year. However, with the holiday season approaching and the continuing popularity of at home DNA kits, we thought we would take this opportunity to reprint our article from 2018 on genetic privacy and protecting your DNA.

GENETIC PRIVACY – WHAT AM I SHARING?

Have you seen the commercials offering you the chance to find out if you might be related to a great warrior or perhaps royalty? Maybe your love of fishing comes from your great-great grandfather who was an Irish fisherman. Companies like Ancestry.com and 23andMe have built substantial businesses offering information about ethnicity based on your genetic sample.

In addition to ancestry, 23andMe has a far more extensive array of tests available directly to the consumer. They were one of the first to enter the direct to consumer genetic testing market. They have approval from the FDA to offer testing for three BRCA ½ variants, genetic health risk reports and carrier screening reports. In October 2018, the FDA authorized 23andMe to sell tests for 33 pharmacogenetic variants directly to consumers. These types of variants are often used to determine if a specific medicine, such as a chemotherapy regime, will be more or less effective based on genetic variants present. As with all direct to consumer genetic testing, the FDA warns that results should be used for informational purposes only and consumers should consult with healthcare professionals for any treatment decisions. The FDA also acknowledges that these tests do not provide definitive information about an individual's variants and the effectiveness of any specific drug.



In 2008, landmark legislation was passed protecting an individual's genetic information. The Genetic Information Non-Discrimination Act (GINA) protects people from discrimination by health insurers and employers on the basis of DNA information. The law stipulates that an employer or an insurer cannot deny an individual coverage on the basis of genetic testing results. It also mandates that an individual cannot be forced to undergo genetic testing by an employer or an insurer. There are some exceptions. The law does not cover life insurance, disability insurance and long-term care insurance. This is an important piece for people to consider before undergoing predictive tests such as the BRCA gene. Unfortunately, many individuals have learned they are unable to obtain life or disability insurance after testing positive for the BRCA gene. This is where meeting with a genetic counselor before undergoing screening can be so beneficial. Most individuals are counseled to consider obtaining coverage before undergoing any type of predictive screening.

In past issues of the *IMPACT* we've discussed the overwhelming amount of information that is available through genetic testing. The decision to undergo screening is a personal decision that should be made with the support of a genetic counselor to understand the risks, benefits and limitations. Interpretation of results is something a qualified medical professional should handle. However, with the exploding direct to consumer genetic testing market offering individuals the opportunity to learn everything from their heritage to their risk for Alzheimer's or Parkinson's disease, all with a cheek swab, privacy is a concern that is often overlooked.

Before GINA, many people declined testing or paid for the screening out of pocket for fear of discrimination. As new tests and therapies become available, the need for protection is even greater. In March 2017, medical, genetic and privacy advocacy groups became alarmed by the introduction of H.R. 1313: Preserving Employee Wellness Programs Act. H.R.1313 was seen as an attempt to circumvent the protections offered by GINA. The bill would allow employers to see their employees' genetic information and impose financial penalties on employees who wanted to opt out. Proponents argued

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**"TAY-SACHS
AND CANAVAN
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TO COME."**

Impact is a
publication of
National Tay-Sachs
& Allied Diseases
Association of
Delaware Valley

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GENETIC PRIVACY – WHAT AM I SHARING? *Continued from page 1*

that it was not a violation of GINA because it was part of an employee wellness package, allowing employers to better manage their employee's health. Amid fierce opposition, HR 1313 did not move out of the House. Yet with healthcare costs dominating spending in the US, the potential to use genetic information to manage those costs will remain a target for employers and insurers.

So, what does all this have to do with if I send a cheek swab in the mail to find out if I am royalty? The answer is everything. For many, ordering an ancestry kit is a fun family surprise with few risks beyond an unknown ancestor discovered. However, when you give someone a sample of your DNA, you are not just giving them your heritage, you are supplying your entire genetic make-up. You may only pay to get the ancestry results, but that doesn't mean an entire array of tests were not performed. The terms and conditions for many of these companies include language where the consumer agrees the company may use their genetic material for research.

In 2018, GSK and 23andMe announced a four-year collaboration focused on developing new medicines and potential cures, using human genetics as the basis for discovery. 23andMe is providing the genetic data from five million deidentified customers to GSK researchers. Many 23andMe consumers were shocked to hear that the company had sold their genetic data to another company, even though they consented to the data sharing when they purchased their kit. The company also has agreements with Genentech and the Michael J. Fox Foundation.

Genetic research is vital. The discoveries made in the last 10 years alone have changed the course of medicine forever. Companies such as Spark Therapeutics have developed treatments that cure rare forms of blindness through genetics. Cancer treatments have become more targeted and effective. Closer to home, recent trials for a cure for Tay-Sachs disease are moving into the human subjects phase. The mapping of the human genome holds the promise for so many treatments and cures.



Informed consent is the foundation for all research. The single largest funder of all research, the National Institutes of Health requires the investigators they fund to obtain broad consent for genomic data sharing. Sometimes though giving consent can seem a little like accepting the 32 pages of terms of conditions every time you update the software on your phone or tablet.

Researchers at Vanderbilt University recently conducted a literature review to examine the issue of genetic privacy. They analyzed 53 studies that touched on genetic privacy. Responses from more than 47,000 participants were included. There was a mix of participants included – 18 of the studies were from participants in the general public or direct to consumer testing users and 31 studies focused on patients and families directly involved as research participants. The remaining studies were more general. For the most part, investigators asked participants if they were concerned about privacy but not why they were concerned. The review also found that participants tended to view privacy with related factors such as confidentiality, control and security. A huge gap identified was that few studies included participants thoughts on the Genetic Information Non-Discrimination Act or other laws that provide protections.

Vanderbilt researcher Ellen Clayton said in a statement, “The picture of genetic privacy that emerges from this systematic literature review is complex and riddled with gaps. Much more needs to be learned about which concerns about genetic privacy matter most to people as well as the social forces that influence their views in order to develop policies that create trust necessary to allow people to make optimal use of advances in genomics.”

Does this mean I should just listen to Aunt Gert at Thanksgiving when she tells me I'm related to a Duke and not send in my test? That is completely up to you. Like any other genetic test, the decision to participate is entirely yours to make, but we want our readers to understand their rights and protections.

HERE ARE A FEW THINGS TO KEEP IN MIND FOR ALL GENETIC TESTING:

1 By agreeing to the terms and conditions for these kits you agree that your data may be shared (often for a price) for research. Your data is deidentified, meaning that your name and specifics are not attached to it, just your genetic markers. Directions for opting-out of this option can be found on each company's website if you do not want to participate in research.

2 If you choose to have any type of genetic health report or carrier screening done through a direct to consumer service, please meet with a genetic counselor or physician to determine what those results actually mean. There are significant limitations and variations in the reports which require proper interpretation. The results should be for informational purposes only. All medical treatment decisions should be made with your healthcare team.

3 GINA protects your genetic information from being used by your employer or health insurer. It also protects your right to

decline to undergo genetic testing by an employer or insurer. These protections do not apply to life insurance or long-term disability. Before undergoing any type of predictive screening, please meet with a genetic counselor to determine what these results could mean. You should also consider obtaining these coverages before you undergo screening.

4 Finally, although H.R.1313 did not move forward, it does not mean it won't come back. As we continue to unlock the potential of genetics to predict and treat disease, the rising costs of these treatments will only inflate healthcare spending. Employers and insurers will continue to argue that they have a right to control those costs. If the protections afforded to you under GINA matter to you, please be sure you stay engaged with your government officials on a state and national level. Your voice and your vote count. ✨

EXECUTIVE COMMITTEE MESSAGE

Dear Friends:

For NTSAD-DV this past year has been a year of ensuring that anyone who needed to be screened could be screened. Unfortunately, none of our normal education and outreach activities were able to resume during the past year. The schools we normally visit were still virtual or not allowing speakers, the events we host with our Young Leadership Council remained cancelled, the health fairs and festivals we participate in never happened. And for the second year in a row, our largest fundraising event did not happen. Yet, our doors stayed open (virtually) and we continued to financially support carrier screening and genetic counseling. We were able to do that with your continued support. Grassroots organizations can only continue if they have a base of support to help them carry out their mission. Your generosity and support ensure that we can continue in this most worthy cause- the birth of healthy children in this generation and for generations to come. Our organization has operated on a shoestring budget these past 20 months. We know that many of you continue to be impacted negatively from the pandemic, but if you can make a donation, please do. We are making plans to resume as many of our pre-pandemic activities as possible and your support will ensure our mission continues for generations to come.

Thank you for your generosity.

NANCY DONEGAN MICHAEL J. OSSIP LAURI SUSSMAN SIEGEL



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STAY HOME, GET SCREENED!

If you have not been screened for Tay-Sachs, Canavan and the other genetic diseases and are considering starting or expanding your family – screening and virtual counseling is safe and accessible through our partnership with Einstein's Prevention of Jewish Genetic Diseases Program (formerly the Victor Center).

- There is no preparation for the testing and usually results are available in four to six weeks.
- You do not need to fast, and blood can be drawn at any time of day.
- If you have a partner, it is recommended that your partner be screened at the same time.
- It is recommended that you share your results with your PCP and OB/GYN.

With our changing population, the pan-ethnic panel is recommended. Patients can be screened for more than 280 diseases in the extended carrier screening or pan-ethnic panel which covers 101 Jewish genetic diseases. The pan-ethnic carrier screening panel is not based on specific ancestry. If you are unsure of your heritage, the pan-ethnic panel is recommended. Each year the testing panels expand and it is recommended to learn about diseases added to the panel before each pregnancy. American College of Medical Genetics (ACMG) supports extended carrier screening in all populations. ❄️

For more information, please contact info@tay-sachs.org

THANK YOU FOR YOUR GENEROSITY in returning the enclosed envelope.

You can also make a secure donation online at <http://www.tay-sachs.org/donations.php>

LET'S GET FIT FOR A GREAT CAUSE!

*This spring we are planning to run
a virtual fitness challenge.*



Help us take 1,000,000 steps for Tay-Sachs and Canavan prevention! Participants in our challenge will be able to track their activity (walk, run, bike, swim etc.) and record their steps completing those activities for a month. Just like in a 5K or a Walk, you will register, form a team and have the opportunity to have friends support your success with a donation to our prevention efforts.

Ready to get moving for a great cause?

Contact Rebecca at info@tay-sachs.org for more information. ❄️

WAYS TO GIVE

*There are several options to support NTSAD-DV.
We are grateful for your support.*

■
You can make a secure donation online at
<http://www.tay-sachs.org/donations.php>

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Return the enclosed envelope

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Designate NTSAD-DV as the recipient of your
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■
National Tay-Sachs and Allied Diseases
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National Tay-Sachs & Allied Diseases Association of Delaware Valley

PO Box 441
Jenkintown, PA 19046

215-887-0877
www.tay-sachs.org
info@tay-sachs.org

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