Dear Reader,

Welcome to the fourth issue of Genetic Counseling: Undergrad Edition! Members of the Genetic Counseling Club at the University of Pittsburgh established this newsletter firstly as a resource for students who are interested in genetic counseling but are unable to attend meetings. However, we hope it will also serve as a guide to genetic counseling for everyone, even those with just a passing interest in the field. We hope to advise, inspire and, most of all, educate.

With the COVID-19 pandemic shifting most of our classes and activities online this year, we thought it would be interesting to see how genetic counseling operates virtually by taking an in-depth look at telegenetics. If you enjoyed reading or have thoughts on what we should explore in our next issue, please reach out to us at pittgeneticcounselingclub@gmail.com. Otherwise, look for our next publication near the end of the spring 2021 semester.

Gopika Rajanikanth, Editor-in-Chief

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The current COVID-19 pandemic has continued to ripple the normalcy in society, impacting how people communicate and interact within all environments. In medicine, particularly, the unexpected is expected, and as seen with rising positive cases and upwards of 200,000 deaths in the United States alone, COVID-19 remains an unforeseen yet consistent threat. Fortunately, the existence of modern technology in health care operations such as telehealth—and more specifically, telegenetics—medical professionals have been able to combat the discrepancies in patient communication imposed by COVID-19.

Telehealth practices provide a different yet intimate health care experience for patients in need of medical assistance. Usually conducted through various modes of technology, patients are able to connect and ask questions with their health care providers in a virtual setting. The demand for medical care is a vital, ongoing commitment, which telehealth technology maintains in everyday life and under disease-outbreak conditions. Amid a global pandemic especially, this virtual connectivity allows patients to inquire about their health, and health care providers to facilitate the appropriate care while preserving the safety of both parties.

Within the broad range of telehealth consultations, telegenetic communication remains a possibility. Telegenetic services have allowed genetic counselors to continue to discuss genetic risks, treatment and testing options with patients. Rather than in-person appointments, telegenetic services allow patients and genetic counselors to interact virtually or over the phone in the event that in-person contact is not possible or safe. One of the major components in a genetic counseling appointment is the counseling aspect, which is why implementing telegenetic services is especially important for the interaction between patient and professional; while in-person communication may not be feasible, virtual discussion allows patients to experience the full support and acknowledgement of concern by the genetic counselor.

Another important consideration is the cost of such telegenetic medical care: according to a study comparing in-person and virtual telegenetic services, there was an observed decrease by about $150.00 for telehealth appointments versus in-person consultations. In this specific research, telehealth proved to be a more affordable option. By the same study surrounding patient experience in telegenetic services (cancer genetic specific), patients provided a substantial amount of positive feedback regarding their genetic counseling experience. These results indicate that despite the notable difference in health care settings, the quality and commitment to care remains unchanged. It is understood that the digital environment cannot fully replace in-person interaction, however, professionals recognize the importance of connecting with patients and the use of telegenetic services extends the opportunity for patients and their desired care.

Despite the supportive feedback from patient surveys, challenges with technology are an inevitable component in the health care industry. The first major constraint in telegenetic services are the technological inconsistencies including access to the technology itself, and connection quality; furthermore, imminent skepticism about patient privacy in a virtual environment continues to question the efficacy of such telemedical advancements. In all aspects of health care, it is important to consider the diversity in patient experience within a medical setting, especially with regard to personal preference and concerns. Overall, however, telehealth remains a viable option to conduct medical protocol and aims to follow the same professional standards required in medical practice.

Telegenetics falls within the broad spectrum of telehealth programs and serves as another opportunity for interaction between patients and genetic counselors, aside from the typical in-person appointments. Telehealth services continue to revolutionize the dynamics of healthcare and have served as promising tools for providing efficient and effective health care under pandemic conditions and beyond.
The genetic counseling program at Bay Path University is one of the few programs with a pre-eminent online curriculum. The university, located in Longmeadow, Massachusetts, contains a graduate college with 34 distinct programs. Their genetic counseling program has options for many students of many different backgrounds, and it is certified by the Accreditation Council for Genetic Counseling, Inc. With an average class size of 15 students, the program provides a personalized, engaging learning experience. The highly accessible genetic counseling program encourages students to participate in-person and online in a diverse, inclusive environment.

While many of Bay Path’s programs have components of tele-education, their genetic counseling program has a unique hybrid curriculum, consisting of online courses, campus sessions, and clinical experiences. Currently, like many other programs throughout the country, courses are exclusively online due to the COVID-19 pandemic. However, Bay Path University has years of experience in tele-education and creating communities through computer screens. Alumni of the program describe having a strong sense of community despite the online learning conditions. With telehealth increasing throughout the world, partially due to new technologies and the current pandemic, the program’s online courses can increase students’ telehealth skills. In past years, to supplement their online education, the students would attend 2-3 day sessions at the Longmeadow campus twice a year. Whether a student’s future career involves in-person care or telehealth, Bay Path’s genetic counseling program prepares them for both.

The genetic counseling curriculum is mainly divided among first year and second year students. In general, first year students focus on the necessary online courses, and they begin clinical, laboratory, and community experiences. Second year students focus more on clinical experience, while participating in online courses as well. Over the two years in the program, students individually complete a capstone project, beginning their research in the first year. The capstone projects are completed during the second year, and each student presents their research results at the annual Bay Path University Academic Achievement Day.

Outside the online classroom, fieldwork and clinical experiences are an extremely important part of the genetic counseling program. Students are expected to complete 840 hours of fieldwork, through 4 clinical rotations. During the spring of the first year, students should participate in 80 hours of fieldwork. Through the summer, fall, and spring of the second year, each semester should include 240 hours of fieldwork each. Since the program mainly consists of online education, students are physically located throughout the country. Therefore, Bay Path has partnered with clinical genetics centers in many different regions. Most partners are located in Western Massachusetts, Connecticut, and New York, but there are fieldwork opportunities in farther states like California and Texas as well. Clinical rotation sites include pediatric, prenatal, cancer, and laboratory genetics, so students can engage in different areas of research and fieldwork.

Bay Path University’s genetic counseling program is a balanced mix of online courses, in-person sessions, and fieldwork experience. With a hybrid online curriculum, the program prepares students for future careers in the genetic counseling field. Telehealth and online communication are skills that could help all future genetic counselors.

Learn more about Bay Path University’s genetic counseling program here:

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What is the Match? By Gretchen Murphy-Zug

The Match is a program designed to pair prospective genetic counseling students with genetic counseling programs. It can often seem mysterious but it is the same program that medical schools and various other health programs use to pair students and programs. In the 2020 admissions cycle, 50% of the participants were matched with a school.

The Match came into existence as a matter of organization and convenience. The Match allows everyone to find out on the same day if they got into a genetic counseling program. Before the Match it was a chaotic process where different programs would put in offers at different times. This led to a lot of confusion and waiting games for both students and the programs. This prompted a shift to the Match as a way of standardizing the results even if the applications are all different.

So, how exactly does the Match work? First any applicant to any school must register because you are required to submit your match number with your application. Then, if the program is interested, they will invite you to interview at the program. After applying and interviewing, you then must rank the schools that you would be willing to attend in order from most to least. This is very important because whichever school you are matched with you must attend. An example of this would be if you were applying to Pitt, Baylor, and the University of Maryland (UMD) and ranked them in this same order from most to least. Then the genetic counseling programs will rank the students that they would like to see in their program. Keeping with our example, Pitt ranks you 8th, Baylor ranks you 7th and UMD ranks you 2nd. So which matters more, your ranking or the programs rankings? Your rankings matter more! You would go to Pitt as you had them ranked higher than the UMD even though UMD ranked you higher.

There are also multiple tracks listed for the same school. This is due to scholarships and possible job positions. If you want to apply for a scholarship then you select the scholarship track. This means you can actually rank the same school twice, once for a regular track and once for a scholarship track. However, these vary from school to school in exactly what the scholarship or position constitutes and how many are available. There is also the matter of how much it costs to register for the match. It costs $100 regardless of whether or not you end up participating in the match. The match results this year will be announced April 23, 2021. Last year there were 5 unfilled positions which could be applied for separately from the match after the results are announced.

The Match can seem daunting at first but it truly boils down to a few key dates and steps. The key is to stay on top of the dates that rankings are due and to make sure that you are completely satisfied with going to any of the schools on your list. It seems scary to find out in just one day but it is much better than the previous drawn out process. Good luck with your Matches!

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2020-2021 Admission Cycle Important Dates

**December 15, 2020-** Deadline to register for Match

**April 14, 2021-** Rank Order List Deadline

**April 23, 2021-** Match Day
Haley Kulas is a second-year genetic counseling student at the University of Pittsburgh. Since high school, Kulas has known she has wanted to be a genetic counselor. She has muscular dystrophy and had a genetic counselor herself. Originally from Pittsburgh, PA, she attended Washington & Jefferson College in Washington, PA where she majored in general biology and minored in psychology. During her undergraduate career, she was a member of the fraternity Kappa Alpha Theta. After graduating, she was accepted into the Masters of Science in Public Health program at Pitt. She wasn’t sure if this was the correct career path for her, so the next year she applied for the Masters of Science in Genetic Counseling at Pitt and was accepted.

Q: Why did you choose Pitt’s Genetic Counseling program?
I like the organization of the program; I like how all of the coursework is taken in the first year and the second year is focused on clinics. Pitt has amazing program directors and department staff. They are supportive, understanding, and want you to succeed. It is such a warm environment. It is also very wheelchair accessible which is important to me.

Q: What was the most difficult part of the transition from undergrad to grad school?
It was actually a really easy transition for me. I came from a school with super high expectations and was well prepared for the workload. It was just a different environment; the faculty really wants us to succeed. It is a very supportive environment. I am also much more interested in what I am studying now, so that eased the transition.

Q: What were the most challenging aspects of the application process for you?
I think the most challenging part is writing the personal statement. It can be hard to get your clear thoughts down on paper and connect your experiences to what the job entails. Your personal statement can take a while to perfect. It is important to be able to really think about what roles genetic counselors play and see how your experiences can show that you understand the roles. Additionally, the anxiety of waiting for the match process is difficult; but it is important to have methods to deal with this anxiety. It is normal and everything will be worth it in the end. I would also suggest for applicants to do a mock interview; it is important to figure out how you want to answer certain questions before the interview.

Q: What are some of your favorite classes you’ve taken at Pitt?
I really liked the molecular basis of human inherited disease course and the principles of genetic counseling course taught by genetic counselors. I also enjoyed my intervention skills course; it was more focused on the psychosocial aspect of genetic counseling, which I value.

Q: Since you had a normal in-person start to your graduate studies, how has COVID-19 affected your studies and how the program is running?
It’s definitely weird, but the program directors are handling it well. Since we are in our second year, our main worry was whether we were going to be able to do clinics. Clinics are going well now though. Although clinics are mostly in person, we’ve had some neat remote opportunities. I think it’s important to just go with the flow.

Q: What advice do you have for undergraduates interested in genetic counseling?
I would suggest spending time really understanding why you want to go into genetic counseling. I would also suggest for undergraduates to spend enough time gaining experience in the field. Experience can include informational interviews or shadowing a genetic counselor. I think it is important to get experience in all three core areas: pediatric, prenatal, and cancer genetic counseling.
Genetic testing companies like 23andMe have had a recent surge in popularity due to rapid advances in technology and the convenience of direct-to-consumer testing. Direct-to-consumer testing involves customers completing their genetic test at home without help or supervision from a health care provider. The customer needs to simply provide a saliva sample and send it in the mail for analysis. This quick and efficient form of testing is appealing to many people. Through these tests, customers are able to test their DNA and receive reports about specific traits their genes may carry, wellness factors influenced by genes, and health predisposition and carrier status on certain genetic diseases.

23andMe tests for over ten health predispositions including the BRCA1/BRCA2 genes. Currently, 23andMe provides the only FDA authorized direct-to-consumer test for BRCA mutations. BRCA mutations are associated with higher risks of breast, ovarian, and prostate cancer. Two BRCA genes are found in each individual and play an important role in the prevention of breast cancer by acting as tumor suppressors. However, when mutated, the gene’s ability to suppress tumors is altered, potentially leading to uncontrolled cancerous tumor growth.

There are thousands of variants in the DNA code that result in cancerous genes. Only about 0.25% of the population carry a mutation in BRCA1 or BRCA 2. These individuals are both at a higher risk for developing breast cancer and developing cancer at a younger age. For example, 55 to 60% of women with BRCA1 mutations and 45% of women with BRCA2 mutations will develop breast cancer before they are 70 years old. Additionally, having these mutations lead to an increased risk of developing recurrent breast cancer and other types of cancers, such as fallopian tube, perinatal, prostate, and pancreatic cancer.

Although the percentage of the population with these mutations is small, certain populations are most at risk for these variants. For example, 1 in forty people of Ashkenazi Jewish descent have one of the three variants tested through 23andMe. Despite 23andMe testing for these three variants of the BRCA mutations, that are most prevalent in the Ashkenazi Jew population, the test is not comprehensive as there are over a thousand additional variants of the gene.

Understanding the limitations of these tests is necessary for consumers to accurately comprehend their results. According to 23andMe, 1 in four hundred people in the general population has a BRCA variant, yet many of these variants are not one of the three variants that they specifically test for. Therefore, people might misinterpret their results as no or low risk of developing breast cancer, when in reality they carry an alternative variant that may put them at risk. In addition to BRCA mutations, lifestyle choice, environmental factors, and other genetic variants can play a role in the development of cancer. Furthermore, inheriting a BRCA mutation does not necessarily mean you will develop cancer; it just means your risk for developing cancer increases. Nonetheless, testing negative for these mutations is not a reliable way to measure your risk of developing cancer since this test is not comprehensive.

To learn more about your risks it may be helpful to speak with a healthcare professional, such as a genetic counselor or a physician. A health care provider can determine if more comprehensive tests are required and provide higher insight into 23andMe results.

Many men and women are unaware that they carry a mutation that companies like 23andMe test for. It is important to have the knowledge to accurately assess risks to your future health. Overall, 23andMe is a good source for individuals interested in learning more about their genetics. People who get a 23andMe test should be sure to utilize various resources, such as genetic counselors, to gain a more accurate understanding of their results.
What is McArdle Disease?

McArdle disease is a rare muscle disorder. This disease occurs because a sugar called glycogen cannot be broken down. Glycogen is very important because your body gets a simple sugar called glucose from the food you eat, leading to an elevation in blood glucose levels. It’s important for the body to remove extra glucose from your blood and convert it into glycogen to balance your blood glucose levels. Your body stores glycogen in many parts of your body, such as your liver and muscles. This is because glycogen is one of the molecules your body uses to store energy. In order to work effectively, muscles continually need glucose. The stored glycogen can be broken down to glucose and used by the muscles.

If you are diagnosed with McArdle disease, your body is unable to break down glycogen. The enzyme missing in the muscles of people diagnosed with McArdle’s disease is myophosphorylase. Myophosphorylase is an enzyme that degrades glycogen. With McArdle’s disease, this missing enzyme leads to fatigue during exercise, cramps, muscle pain and other symptoms of the disease.

What Causes McArdle disease?

McArdle disease is an inherited disease with an autosomal recessive pattern. In order for this disease to occur, both copies of the PYGM gene (one from the mother, one from the father) must be mutated. The PYGM gene codes for the enzyme myophosphorylase which cleaves glycogen into glucose-1-phosphate, which will later be transformed into glucose. If both the PYGM genes have been mutated, there won’t be any functional myophosphorylase, which prevents glycogen from being broken down. Consequently, muscle cells won’t have enough energy to function properly.

What are the symptoms?

This disease causes weakness in the muscles. It makes it challenging for the person diagnosed to exercise without getting exhausted too quickly. Difficult exercise is nearly impossible after a couple minutes. A few other symptoms include brownish red urine, fatigue during or after exercising, muscle cramping and muscle pain. In addition, some people may experience long-lasting weakness in their muscles but this is rare. People diagnosed with McArdle’s disease usually experience these symptoms by the age of 15.

How is it diagnosed?

One must get tested before they are diagnosed with McArdle’s disease. A typical test is a physical exam and a muscle strength test. Some other tests include blood tests, a forearm exercise test, an MRI to scan your muscles, a muscle biopsy to see if glycogen has accumulated in the cells and a urine test.

How do you treat McArdle disease?

There is currently no cure for the disease. However, particular diets and several exercises can help with the disease. Moderate exercise can allow your body to use glucose most effectively. Other treatments include being cautious about the amount of carbohydrates consumed, taking creatine or Vitamin B-6 supplements, and ingesting a specific amount of sugar prior to exercising.
Dawn Nicotra, MS, CGC

Genetic Counselor Spotlight: Dawn Nicotra, MS, CGC

By Gopika Rajanikanth

Dawn Nicotra is a genetic counselor at MyGeneTeam, where she provides telegenetic counseling to inherited cancer families, while working remotely. After she graduated from the genetic counseling program at the University of Pittsburgh, Nicotra originally worked in pediatric genetics at Children’s Hospital of Pittsburgh for a few years before transitioning to her current position.

Nicotra had not heard about genetic counseling while she was studying biology as an undergraduate student, but she knew she enjoyed science, and specifically genetics. She had originally planned to pursue physical therapy, but she enrolled in an advanced genetics course towards the end of her degree, which made her reconsider that choice. “I decided not to go to grad school right away because I knew that sort of changed things for me,” she said. “I really wanted to go more in that direction, but I didn’t know how or in what way.” Nicotra knew she didn’t want to work in a lab, so she spent a few years testing out different careers and fields before she discovered genetic counseling.

Nicotra never expected to be working in telemedicine because when she finished graduate school, most genetic counselors worked in hospital settings. “I really didn’t see telegenetics coming back when I was graduating in 2005. And I feel like it happened so rapidly, but it seems so seamless and appropriate for the day and age that we live in,” she said. As a result of the COVID-19 pandemic, Nicotra believes that people are more willing to utilize telegenetics and talk to genetic counselors about their medical history and genetic testing options over the phone or by video. But there are some differences between telegenetic counseling and counseling in-person in a clinic or hospital setting. “As expected, you can’t read their body language or pick up other non-social cues as easily over the phone,” she said. Instead, Nicotra says that telegenetic counselors have to work harder at the beginning of a conversation to establish a warm connection with a patient before discussing anything else. She was surprised that forming this connection wasn’t as challenging as she anticipated when she first joined MyGeneTeam. “But I also love my job and love talking to people and I think most people and patients just pick up on that and respond to that. So, I think just making that connection a little more personal at the beginning helps,” she said.

As a genetic counselor at MyGeneTeam, Nicotra had the opportunity to work remotely. For Nicotra, this meant that she was able to eliminate some commute time as well as gain more flexibility with balancing her work life and family life. Telemedicine allows Nicotra to see patients from across the country from various time zones, which also adds flexibility to her work schedule. “If you want to work evenings, then there’s plenty of people on the west coast that are still in the midst of their workday. And vice versa. If you live on the west coast and you want to see early patients,” she said. Nicotra thought that working remotely would mean she wouldn’t be able to interact with her coworkers as often,
but she’s able to more than she thought she would. “Even though there’s a large team of us, and we’re scattered about the country, there is a strong sense of teamwork and it’s been easy to make connections. We have a lot of team meetings and I have certainly been able to form relationships with my colleagues,” she said.

Nicotra starts off her workdays by logging in and reviewing the cases that she has scheduled for that day. If there is time before her first call with a patient, she takes care of tasks such as answering emails from patients, coworkers or laboratory staff and following up on results or other lab related items.

Then Nicotra starts meeting with her patients that are scheduled for that day. “I schedule my patients fairly close, back to back. I finish a call, write my note, fax it all out and document everything that needs to be documented for insurance. And then I jump right into my next call,” she said. Nicotra sees around 4 patients a day. A call with a new patient usually lasts forty-five minutes, and then she spends around thirty minutes afterward filling out paperwork and writing the clinic note. A call with a follow-up patient usually takes fifteen to twenty minutes and she only needs ten minutes afterwards to fill out the paperwork. As a cancer genetic counselor, a lot of diagnoses that Nicotra sees are pretty similar, including family history of breast cancer, colon cancer and personal history of metastatic prostate cancer. But for Nicotra, every individual patient brings some unique aspect to the case, including their personal or family history, which keeps everyday interesting for her.

As genetic counseling graduate school programs continue becoming more competitive, Nicotra says that prospective students should try to be well rounded and gain exposure to areas including science, counseling, lab research, as well as shadowing genetic counselors. Exposure to writing and communication, even if it is not related to science, is helpful as well. She also thinks that real life work experience can be beneficial to a lot of prospective students to learn real life skills that come with having a job, such as teamwork and responsibility. “Don’t be discouraged if you aren’t accepted to a program the first year or the first time around. There’s always another year, and it’s never too late to do something that you’re passionate about. I speak from experience with regards to those things.”

Thank you for reading Issue IV! We hope you’ll check back next semester for Issue V!