That are at the better, but thank you so much for joining us today and Happy Friday. My name is Alex. I am a cancer genetic counselor here at Yale, and I’ll be acting as the host for today. So we have some amazing speakers who have worked really hard to put this all together for you. So we’re excited to be here and we certainly hope you find this helpful. But our goal today is to provide
a more 3D understanding of the genetic counseling field, and in reality this is a big ask because we’re a very diverse and multifaceted profession. So that’s why we have a very busy schedule. But we’ll start with the first half, which is an overview of genetic counseling, and we’ll have some genetic counselors speak. About different specialties within the genetic counseling field, that’s before we take a little break in the middle and moving on to the second half, which is more so about genetic counseling programs. Graduate School hearing from current
genetic counseling students, etcetera and there will be a final Q&A session at the very end. Over the course of our day, you will likely have some questions, so we do encourage you to utilize the Q&A function. But if you joined us last year, it’s going to look a little different. So if you ask that question, the questions will be picked out by the moderator to pose to the speaker at the very end of their talk, instead of addressing every question in the Q&A function just so
people can more so focus on what.

Is being presented.

This might mean that not all

However, our speakers have been

very gracious to share their

emails and I'll put that slide up

at the very end so if there isn’t

a time to address your question,

we’ll have the Q&A session at

the very end and as well as the

speakers emails as well.

So sometimes the speakers will

have a question for you guys,

so keep an eye out for the polls

and be ready to answer those.
And whether you’re able to attend for the full day today, whether you’re able to attend for maybe some of the day or not at all, all of the registrants who signed up for this event will be sent a short survey and maybe about a week’s time. And once you complete that survey that will take you to a recording of the event. So thank you in advance for answering those questions for us so we can help improve this event for future years.

A little bit about me now. I graduated back in 2017 with my Bachelor of Arts and Biology and Society.
I knew that I wanted to be a genetic counselor, so I felt that that really bridged both the science and people, which is what I liked about the genetic counseling profession. And then I went in to my Masters program at the Icahn School of Medicine at Mount Sinai in New York, and my first job and current job is working as a cancer genetic counselor here at Yale, New Haven. So when I tell people that I work as a genetic counselor, almost always they will ask me what
genetics has always been a hot topic.
Whether it's in the news or sci-fi.
So hopefully today over the course
of our session we'll be able
to take a better look at what a
genetic counselor is in reality.
And I always say that it's not.
It's not rocket science,
so if we think about genetic counselor
and break it down, we have genetics.
So thinking about DNA, genes,
chromosomes and then we have
counseling and planning.
Conversation, conveying information,
maybe eliciting feelings about
that information, etcetera.

Now at the most basic level, then a genetic counselor or genetic counselors or healthcare professionals with advanced training. In medical genetics and counseling, who educate, guide and support patients seeking information about inherited diseases and conditions in order to provide a better understanding of how genetic information impacts patients lives and the lives of their family members. So it’s a long winded explanation, but we wear a lot of different hats, so there’s no other way to put it.
But really this is a profession. That was born out of the understanding that genetic information can be sensitive. It can be confusing at times and personal always, so a genetic counselor can help patients navigate this path. And they might talk about whether genetic testing is right for a patient. That might mean explaining the genetic test results and what they mean for the patient and their families. Identifying resources for the patients afterwards, etcetera.
And it might also be helpful to think about what a genetic counselor is. Not so genetic counselors will typically have a masters degree and that focuses on both clinical genetics and counseling skills. Genetic counselors or GC’s can work with patient, or they can work with doctors, or they can work independently, and this is compared to a medical geneticist or clinical geneticist who have an MD or MD equivalent degree and specialized training in genetics. Meaning maybe they did an additional fellowship in genetics after completing their residency. They also have a specific board.
exam that they have to pass, and unlike genetic counselors. And they’re able to perform a physical exam. They’re able to perform procedures and diagnose diseases. A laboratory do not assist. On the other hand, is someone who has maybe an MD or MD equivalent degree. They might have a PhD and they’re working more so behind the scenes, so either with the testing technology itself or the interpretation of the findings but altogether genetic counselors, medical geneticists,
00:06:40.530 --> 00:06:41.616 laboratory geneticists,
NOTE Confidence: 0.885292538571428
00:06:41.616 --> 00:06:44.331 they work together to utilize
NOTE Confidence: 0.885292538571428
00:06:44.331 --> 00:06:47.753 genetic testing as part of someone’s
NOTE Confidence: 0.885292538571428
00:06:47.753 --> 00:06:50.037 overall comprehensive medical care.
NOTE Confidence: 0.8013637
00:06:53.260 --> 00:06:55.300 But back to genetic counselors.
NOTE Confidence: 0.8013637
00:06:55.300 --> 00:06:57.720 So we’re thinking about education,
NOTE Confidence: 0.8013637
00:06:57.720 --> 00:06:59.124 helping with diagnosis,
NOTE Confidence: 0.8013637
00:06:59.124 --> 00:07:02.400 and helping a families or individuals cope.
NOTE Confidence: 0.8013637
00:07:02.400 --> 00:07:04.680 Providing that soap psychosocial
NOTE Confidence: 0.8013637
00:07:04.680 --> 00:07:06.960 support advocating for patients,
NOTE Confidence: 0.8013637
00:07:06.960 --> 00:07:09.604 and contributing to risk
NOTE Confidence: 0.8013637
00:07:09.604 --> 00:07:12.248 estimation for inherited diseases.
NOTE Confidence: 0.904511643333333
00:07:16.770 --> 00:07:18.475 But if you’ve googled genetic
NOTE Confidence: 0.904511643333333
00:07:18.475 --> 00:07:21.334 counseling in the past, you were likely
NOTE Confidence: 0.904511643333333
00:07:21.334 --> 00:07:23.326 inundated with different acronyms,
NOTE Confidence: 0.904511643333333
00:07:23.330 --> 00:07:25.522 so I wanted to break that down a
little bit here in the ABC’s of the coming of genetic counselor.

As I mentioned earlier, genetic counselors obtain a Masters degree in genetic counseling that typically lasts for about two years and that is from a program that’s accredited by the Accreditation Council for Genetic Counseling a CGC. Currently there are 57 training programs within the United States and Canada, and as I mentioned earlier, it’s usually classroom based as well as clinical rotation. So going out and working with
genetic counselors in the field,
helping conduct sessions, etcetera.
Once someone graduates from one
of these accredited programs,
they sit for a board exam that’s
put out by the American Board
of Genetic Counseling or AB GC,
and that’s offered twice a year.
So once in February and once in August,
and once someone passes that
board examination,
they get the title of a certified
 genetic counselor or CGC.
About 90% of genetic counselors.
Hold that CGC certification.
And depending on where someone goes to work,
they might be required to obtain a state licensure as well. Here in Connecticut, that is something that we have to do, but it's just a matter of providing the appropriate documentation that you are legitimate genetic counselor. You have the appropriate training as well as paying a small fee. And about 62% of genetic counselors have one of these licenses. Compared to some other professions, the genetic counseling field is relatively new and the first genetic counseling program was founded at
Sarah Lawrence College in 1969. And since then, there have been many important milestones in the development of the genetic counseling profession. So our first professional organization was founded in 1979, that's the National Society of Genetic Counselors, or the NSGC, which you'll be hearing me reference multiple times throughout our talk and also of course, moving on from the first professional organization to obtaining state licensure in 2000, and To Utah was the first state to provide licensing for genetic counselors and in 2022,
NOTE Confidence: 0.95255442
00:10:06.101 --> 00:10:07.394 about 31 states.
NOTE Confidence: 0.95255442
00:10:07.394 --> 00:10:11.358 The last time I checked offer one of these
NOTE Confidence: 0.95255442
00:10:11.358 --> 00:10:13.618 offer licensing for genetic counselors.
NOTE Confidence: 0.80988526
00:10:16.800 --> 00:10:19.380 So first poll for today.
NOTE Confidence: 0.80988526
00:10:19.380 --> 00:10:22.334 Let’s see if I can access that.
NOTE Confidence: 0.9398842
00:10:31.580 --> 00:10:33.312 How many certified genetic counselors
NOTE Confidence: 0.9398842
00:10:33.312 --> 00:10:35.460 are there in the United States?
NOTE Confidence: 0.9398842
00:10:35.460 --> 00:10:39.380 So this is your closest estimate as of 2021.
NOTE Confidence: 0.838948271428571
00:10:45.240 --> 00:10:47.585 And now give maybe 10 more seconds
NOTE Confidence: 0.838948271428571
00:10:47.585 --> 00:10:50.227 or so to get your answers in.
NOTE Confidence: 0.6989328
00:11:00.630 --> 00:11:03.890 OK, so most people most
NOTE Confidence: 0.6989328
00:11:03.890 --> 00:11:07.338 people were on the nose.
NOTE Confidence: 0.6989328
00:11:07.340 --> 00:11:10.860 Most people said B are 5500.
NOTE Confidence: 0.6989328
00:11:10.860 --> 00:11:13.398 About 61% of you said that,
NOTE Confidence: 0.6989328
00:11:13.400 --> 00:11:16.640 and that’s correct.
There are currently a want to say

NOTE Confidence: 0.820902053333333

5629 CGC's within the United States.

NOTE Confidence: 0.820902053333333

There are maybe 2000 or so

NOTE Confidence: 0.820902053333333

practicing genetic counselors

NOTE Confidence: 0.820902053333333

outside of the United States.

NOTE Confidence: 0.820902053333333

According to a 2019 paper.

NOTE Confidence: 0.820902053333333

But yes, about 5500. So good job.

NOTE Confidence: 0.820902053333333

And this is a profession

NOTE Confidence: 0.820902053333333

that has continued to grow.

NOTE Confidence: 0.820902053333333

It’s growing 100% since 2010,

NOTE Confidence: 0.820902053333333

and the projected growth rate for

NOTE Confidence: 0.820902053333333

the genetic counseling profession

NOTE Confidence: 0.820902053333333

is about 21% through 2029.

NOTE Confidence: 0.820902053333333

That’s compared with a 7% average

NOTE Confidence: 0.820902053333333

growth rate for all occupations,

NOTE Confidence: 0.820902053333333

which is very reassuring to see.
No, this is a profession that has been named one of the best jobs by U.S. news for several years.

The NSGC puts out a professional status survey to their membership and nine out of 10 genetic counselors who responded to that survey reported that they are satisfied with their job, and this satisfaction can come from a variety of different reasons.

Whether it’s from intellectual stimulation or working with patients et cetera. And you can see the variety here.
different areas of practice as well.

So the traditional role is direct patient care.

I'm working in education and teaching of genetic counseling and about 51% of genetic counselors work in direct patient care roles.

That's compared to the 27% that are in non direct patient care.

Working in a laboratory as part of their variant curation team or even customer service and 22% of genetic counselors have the best of both worlds, so may be doing.
Working in the with the patients as well as research on this side. So if 50-50 split, most genetic counselors are working full time and the majority have worked remotely as part of or all of their position in 2021. Genetic counselors can also work in different specialties. The majority of genetic counselors are practicing in cancer genetics, followed by prenatal or reproductive genetics and pediatric genetics. They can also work in different settings, whether that’s in the...
laboratory or in the hospitals, and we're lucky to have a lot of speakers from these different specialty areas joining us today. Some people are practicing in multiple areas at the same time, all with the same genetic counseling degree. But I don’t know how these people here are doing for more practice areas at the same time. But there is variety and flexibility to a genetic counseling degree. And the good old fashioned way is working in person one, providing direct patient care. So meeting with someone in clinic nowadays,
especially with the onset of COVID-19, we’ve been utilizing telephone or web based service models, less so. Group counseling, understandably. But it makes it a little easier when we’re able to obtain DNA through a saliva sample, which can be collected at home. A blood drive, of course, is a little more difficult to collect from home, and genetic counselors are very flexible, so they’re typically utilizing...
multiple service delivery
models for their patient care.
Thinking about salaries, which is of course an important factor when deciding whether this is the right profession for you. The average salary according to the most recent professional status survey was just released to May 3rd this year, but the average salary is approximately $102 per month. But it’s important to note that this can vary significantly depending on your role. So whether you work in an indirect or non-direct patient care position and where you’re located.
NOTE Confidence: 0.868617618571429
00:15:47.730 --> 00:15:49.890 geographically speaking, as well as
NOTE Confidence: 0.868617618571429
00:15:49.890 --> 00:15:52.930 your number of years of experience.
NOTE Confidence: 0.868617618571429
00:15:52.930 --> 00:15:55.492 The average starting salary for a genetic
NOTE Confidence: 0.868617618571429
00:15:55.492 --> 00:15:58.090 counselor out of school was about 78.
NOTE Confidence: 0.83427527125
00:16:01.620 --> 00:16:03.455 And as part of that
NOTE Confidence: 0.83427527125
00:16:03.455 --> 00:16:04.556 professional status survey,
NOTE Confidence: 0.83427527125
00:16:04.560 --> 00:16:06.786 the NSGC also asks about different
NOTE Confidence: 0.83427527125
00:16:06.786 --> 00:16:09.756 demographics to get a better sense of
NOTE Confidence: 0.83427527125
00:16:09.756 --> 00:16:12.116 the representation within the profession.
NOTE Confidence: 0.83427527125
00:16:12.120 --> 00:16:14.856 At this time, the majority of
NOTE Confidence: 0.83427527125
00:16:14.856 --> 00:16:16.680 respondents identified as female,
NOTE Confidence: 0.83427527125
00:16:16.680 --> 00:16:18.664 about 10% of respondents,
NOTE Confidence: 0.83427527125
00:16:18.664 --> 00:16:20.648 identified as non white.
NOTE Confidence: 0.83427527125
00:16:20.650 --> 00:16:22.246 Almost all respondents
NOTE Confidence: 0.83427527125
00:16:22.246 --> 00:16:24.374 reported they speak English,
but a total of 51 spoken languages were noted by respondents as well. Recognizing that the diversity within the genetic counseling field can stand to improve and that as a profession we need to be supportive of existing minority genetic counselors and the NSC has prioritized justice equity, diversity and inclusion recently forming a JEDI Committee to address these issues. And you can see here the JDI committee has identified various strategies to support their goals, including working to recruit and retain diversity into the profession and the issue of increasing diversity within
the genetic counseling field has also been recognized outside of the NSGC. Somewhere recently, the Warren Alpert Foundation has given a five year, $9.5 million grant. To create the alliance to increase diversity in genetic counseling. And this is a program that will recruit and train 44 zero that is genetic counseling. Students providing full tuition, scholarships and stipends to cover living expenses during that time and this will be implemented through the five programs that you can see here. In addition,
the minority genetics professional

network or the MG PN was formed in November 2018 to provide a space for minority genetic counselors or diverse genetic counselors from diverse backgrounds to connect with one another, and they also have a prospective students slack channel, so feel free to join there. And finally, as far as diversity of nationalities, there is the International Special Interest Group or SIG that’s also available for prospective international students applying for graduate
training programs in the United States and so if you’re looking, or if you’re tuning in internationally from Canada or elsewhere, and this might be a great place to look for additional resources. So keep in mind that this can be very different depending on your role, but basically, or generally speaking, genetic counseling first starts with a detailed medical and family history,
so the pedigree or the family tree. That’s the bread and butter of genetic counseling. It’s used to document many features that are crucial to a genetic counseling session as far as different generations of individuals, how they’re related and maybe even traits of interest all in one neat, concise little picture. But instead of using dinosaurs and amoebas, we use shapes. So those who are assigned male at birth are designated by a square, while those who are assigned female at birth are designated by a circle.
and a diamond shape can be used for individuals who are gender nonconforming, or when the sex assigned at birth is not known. A line through the sheep is indicating as someone who has passed away. After obtaining that information, we’ll use that to perform a risk assessment or the likelihood of identifying a genetic mutation in a given patient. So genetic counselors are professionals trained in science communication, and we typically spend some time during a session to review genetics with the patient themselves.
If genetic testing is indicated, genetic counselors can communicate the risks, the benefits, the limitations of genetic testing in order to obtain informed consent before coordinating the testing needed, but frenetic testing is not always needed. And there are many different types of genetic tests, so on the left hand side those are medical or clinical genetic tests that are typically involving a genetic counselor to some capacity. So we have diagnostic testing which can be used to confirm a genetic disorder.
Carrier screening or carrier testing, and that is done prior to or during a pregnancy to see if the patient and/or partner are carrying a gene that might cause a congenital defect or disorder. And there’s also prenatal diagnosis testing, which is used to detect abnormalities in a fetus’s genes before birth to identify congenital disorders or birth defects. Newborn screening is a routine test mandated by law to screen for a set of inheritable diseases or disorders such as cystic fibrosis. This is something that can differ between states.
And then predictive diagnosis which is testing for known disorder in the family to understand risk for that disorder. So for example, if someone has a BRC A1 mutation in their mother and we’re doing targeted testing for that patient individually. On the other hand, on the right hand side there are also non medical or non clinical tests that do not typically involve the genetic counselor, but something that you might hear of or patients might bring bring to you. Obtaining a sample. It’s a bit of a pick your poison option or choose your own adventure here,
but usually it’s either done through a blood draw or a saliva sample. And finally, insurance companies are increasingly better about covering the top cost of genetic testing, especially when there’s an indication such as a personal and or family history that’s suggestive of an inherited disease. Of course, if you’re ordering testing for a patient, you then have to disclose the test results to the patient. We review information as far as screening and management.
recommendations or next steps based on those genetic test results.

You can help the patient by identifying research or resources for them and for their family members, as well as speaking through coordinating genetic testing and or counseling for at risk relatives who might also let’s say have that mutation or need genetic testing otherwise and at the very end you wrap it all up and a nice bow and summarize it for the patient so they can have it for their records.

So without further ado, now that you have a good
foundation of genetic counseling,

I’m going to pass it off to Maya.

She’ll be presenting on her role as a laboratory genetic counselor as the first of our many specialists,

so let me see.

Let me see if I can do this properly.

Sorry, just one second. Yeah.

It looks great.

OK. So are you saying the PowerPoint?

OK. So my name is Maya and I’m one of the genetic counselors at the Yale BNA Diagnostics lab and I’m going
to be speaking about what it’s like to work for a clinical laboratory in a university setting and then our other genetic counselor at the lab. Emily Voiceline, is going to be moderating in the chat, so. Please feel free to ask any questions that you’ve got there. So first we’ll talk about our own backgrounds. I’m a certified genetic counselor currently licensed in Connecticut. I graduated in 2021. From Long Island University Post and I did my undergrad degree at University of California,
Santa Barbara in biological anthropology at largely focusing on human evolution and human variation. So I was a non traditional student and took a five year gap between graduating from undergrad and then graduating from undergrad and then starting at my program in 2019. Out of undergrad, I shifted careers around a few times and then ended up becoming a copywriter for a marketing and public relations business for about five years worked there up until my last year of Graduate School. So I would be happy to speak about my experiences as a nontraditional student.
student later on in the Q&A.

Or, if you'd like to email me.

But this was my first job out of Graduate School, so I've been working here since August of last year.

So now I'm going to let Emily speak about herself and introduce herself.

Hi everybody, my name's Emily. I am also a certified genetic counselor in B DNA lab and licensed in Connecticut. I graduated from the Brandeis University program in 2021 and my undergrad I got my BS in Biology from Allegheny College in 2016, and so if you can do math,
you might have noticed.

I also had a few gap years in between undergrad and Graduate School. However, I took a little bit more of a traditional route and I actually worked as an accession or at a combined cytogenetics and molecular genetics lab in between that. And this is also my first job out of Graduate School, so I started at the DNA lab in September of 2021 and now we'll turn it back to Maya to talk about our daily duties.
the genetic counselors take on some pretty versatile roles. Umm? So my primary responsibility is being a liaison between the ordering providers at the billing and prior authorizations teams, and then the different members of the laboratory. Like the lab techs or our molecular genetics tests. So as part of my job, my offer post has support for owner ordering providers who have any sort of additional questions about the result or follow up. And finally we also do
00:28:34.520 --> 00:28:38.590 interpretation of the data for.

00:28:38.590 --> 00:28:39.420 Panel tests, tumor tests and then single site tests.

00:28:42.330 --> 00:28:44.145 So we work on variant interpretation for the lab also.

00:28:48.740 --> 00:28:51.491 So as I said before, assisting ordering providers as my primary role, I take questions about future orders, such as if the lab can offer a certain test. For example, we get a lot of questions about whether we can do testing for pseudogenes where next generation data might not be the best or for non sequencing tests. One well known example would be
fragile X testing where you’re not looking so much at single changes in the gene so much as these repeats. So that’s done with a different sort of test, and we do do that. It definitely helps to have a strong background and interest in molecular genetics to work at a lab. Then you definitely learn a lot more about molecular genetics as you work here. So I also assist in questions about insurance coverage and authorization. The lab has to review certain tests where we can’t get authorization from insurance and it’s my job to review the clinical notes from those orders.
to determine whether the patients meet criteria based on their payer as guidelines. So I do this with the assistance of somebody from the billing team or a coder who’s expertise is in actually reading and interpreting the policies themselves. Our lab offers something called virtual panels. So the entire exome is sequenced, then we only report on the genes or conditions that the providers specifically requests in the order. So I take calls from our providers or get emails.
Sometimes requesting customize Gene lists or we have set gene lists that we’re already using for some providers.

And then as a follow up to positive test results, all email providers to let them know about a pathogenic test result before it’s reported or directly to a patient’s chart and.

Umm, I get requests about all preconditions and maybe which other specialties they might want to refer their patients too.

And also take any sort of increase about existing orders and pending results.

Such as what the expected turn around time is,
or if the test is already processed.

So a laboratory stewardship is a really crucial part of working at a genetic testing laboratory. A lot of labs are hiring genetic counselors because our training is really focused on selecting the most appropriate tests based on individual or personal or family history of these individuals. So we know how significantly these results can affect patients' health and management if they do end up getting a pathogenic result. So we need to make sure that the...
00:32:32.192 --> 00:32:34.130 tests being ordered is appropriate,
NOTE Confidence: 0.891674482
00:32:34.130 --> 00:32:36.895 and then this also helps with insurance
NOTE Confidence: 0.891674482
00:32:36.895 --> 00:32:39.120 coverage because it can be very
NOTE Confidence: 0.891674482
00:32:39.120 --> 00:32:41.374 difficult to get some of these tests
NOTE Confidence: 0.891674482
00:32:41.441 --> 00:32:43.985 covered and we want to make sure that.
NOTE Confidence: 0.903106435714286
00:32:46.100 --> 00:32:48.284 Whatever is being ordered is going to
NOTE Confidence: 0.903106435714286
00:32:48.284 --> 00:32:51.008 be the most appropriate the first time.
NOTE Confidence: 0.903106435714286
00:32:51.010 --> 00:32:53.488 Because once you get a denial,
NOTE Confidence: 0.903106435714286
00:32:53.490 --> 00:32:56.832 resubmitting can be more and more
NOTE Confidence: 0.903106435714286
00:32:56.832 --> 00:32:59.060 difficult for additional tests.
NOTE Confidence: 0.903106435714286
00:32:59.060 --> 00:33:01.230 So I review every order
NOTE Confidence: 0.903106435714286
00:33:01.230 --> 00:33:03.400 that comes into the lab.
NOTE Confidence: 0.903106435714286
00:33:03.400 --> 00:33:06.487 Some of our ordering
NOTE Confidence: 0.903106435714286
00:33:04.772 --> 00:33:06.487 providers rely on the web,
NOTE Confidence: 0.903106435714286
00:33:06.490 --> 00:33:09.165 two select generalist based on
NOTE Confidence: 0.903106435714286
00:33:09.165 --> 00:33:11.840 the patients genotype or whatever
sort of condition or different symptoms they're experiencing.
And. So we are able to customize.
As I said before, and we accommodate those requests
and I work with our geneticist to determine what sort of gene list
would be appropriate for the order.
Another concern is that providers might simply place the wrong order because they can’t figure
out which one is the right option based on our lab test menu.
And we do it a little bit differently here than the commercial labs,
because we only have a set number of order tables and they order directly through our electronic medical system, which we use is EPIC. So maybe they want to place an order for a known familial variant only, but then they selected an extended gene panel. Because they didn’t know that they needed to search for something else in EPIC, so they might write a comment about a specific gene, so I would be the one reaching out to the provider who’s after reviewing the clinical information and presenting different test options.
and making sure that we actually do want to do an expanded test rather than that really targeted test. It’s very important that the patient does not receive information that they did not. And sent to when they were counseled about the results that they would be getting. So Emily is going to talk about one of her roles. Which is not directly related to genetic counseling, but it shows how our education and expertise can be applied to different areas.
And that’s data management and bioinformatics. I’m gonna let Emily take over again. So yeah, my position as mayor just mentioned. One part of it is the definitely not a traditional genetic counseling role, but that is something that I honestly really liked about the position when I was applying and it really just goes to show how versatile what we learn in genetic counseling school is and how that skill set can be used in a bunch of different ways other than just seeing patients or even the more traditional laboratory genetic counseling role so.
The biggest part of my job is bioinformatics and data management, so our lab does a lot of the lab work in house, we extract the DNA ourselves. We do the analysis ourselves. We do Sanger sequencing ourselves, but one thing we don’t do ourselves is next generation sequencing. So any sample that needs next generation sequencing is sent out to another part of the Yale and then when we get that next generation sequencing data back, it is my job to turn that sequencing data into data that is usable for our analysts.
00:36:23.230 --> 00:36:26.890 to be able to write the reports and.
NOTE Confidence: 0.874710332
00:36:26.890 --> 00:36:28.130 Get that.
NOTE Confidence: 0.874710332
00:36:28.130 --> 00:36:30.378 Was those results back to
NOTE Confidence: 0.874710332
00:36:30.378 --> 00:36:31.599 the ordering providers?
NOTE Confidence: 0.882301731666667
00:36:31.770 --> 00:36:34.500 And also when the data is
NOTE Confidence: 0.9326592175
00:36:35.070 --> 00:36:36.580 made usable, making sure that
NOTE Confidence: 0.9326592175
00:36:36.580 --> 00:36:38.519 it is in an easily accessible
NOTE Confidence: 0.9326592175
00:36:38.520 --> 00:36:40.490 location for everybody to find.
NOTE Confidence: 0.9326592175
00:36:40.490 --> 00:36:42.318 So I’m the person who if
NOTE Confidence: 0.9326592175
00:36:42.318 --> 00:36:43.566 somebody’s looking for something,
NOTE Confidence: 0.9326592175
00:36:43.570 --> 00:36:46.229 they come to me and at first glance
NOTE Confidence: 0.9326592175
00:36:46.229 --> 00:36:48.210 this might not seem like it is
NOTE Confidence: 0.9326592175
00:36:48.274 --> 00:36:50.489 super related to genetic counseling,
NOTE Confidence: 0.9326592175
00:36:50.490 --> 00:36:54.320 but in this process I’m also looking
NOTE Confidence: 0.9326592175
00:36:54.320 --> 00:36:56.850 over the orders for all the samples
NOTE Confidence: 0.9326592175
00:36:56.850 --> 00:36:58.500 that I’m working with that week.
And I’m kind of serving as yet another check after Maya and other people just to make sure that once again the orders are appropriate and there are no issues with insurance so.

My genetics knowledge that I gained in Graduate School is definitely very helpful for this and just allows me to serve as yet another checkpoint, because you can never have too many to make sure that everything looks good with these orders and we’re actually getting the providers the information that they want, and so I will turn it back to Maya for.
the other part of my job and something that we are both very involved in, which is very an interpretation and report writing.

So as Emily mentioned, both of us work on variant interpretation. So what we do is we analyze the raw data that comes into the lab and follow a CMG guidelines to classify variants and this is standardized across every genetic testing laboratory in the United States. So we use various databases to back our interpretation. Some of you might be familiar with them if you worked in the lab at all,
00:38:19.110 --> 00:38:21.906 or had any undergrad experience in genetics in a clinical setting.

00:38:21.906 --> 00:38:24.510 So we use glenvar genome for example,

00:38:24.510 --> 00:38:28.829 and then our own internal database.

00:38:28.830 --> 00:38:30.996 So the lab has our own database for we record the specific changes in

00:38:31.000 --> 00:38:34.152 the gene that we’ve seen before.

00:38:34.152 --> 00:38:36.987 And the phenotype or the symptoms that accompany those changes or

00:38:36.987 --> 00:38:39.927 what it’s been correlated with?

00:38:39.930 --> 00:38:42.672 Because they’re not always related, it is good to have that backup

00:38:42.672 --> 00:38:44.991 if it’s a very rare sort of change.

00:38:44.991 --> 00:38:47.076 So we also assist the lab directors

00:38:47.080 --> 00:39:00.679 in reviewing the reports that are
00:39:03.802 --> 00:39:07.187 ready for sign out. And which hack?
NOTE Confidence: 0.875489852222222
00:39:07.187 --> 00:39:10.400 Or we look at the clinical information
NOTE Confidence: 0.875489852222222
00:39:10.485 --> 00:39:13.579 and make sure that it’s clear and
NOTE Confidence: 0.875489852222222
00:39:13.579 --> 00:39:16.214 is appropriate for the condition
NOTE Confidence: 0.875489852222222
00:39:16.214 --> 00:39:18.050 that’s being reported,
NOTE Confidence: 0.875489852222222
00:39:18.050 --> 00:39:22.180 and that there are no major errors.
NOTE Confidence: 0.875489852222222
00:39:22.180 --> 00:39:24.020 For example, spelling errors,
NOTE Confidence: 0.875489852222222
00:39:24.020 --> 00:39:25.860 or if there’s any sort of error
NOTE Confidence: 0.875489852222222
00:39:25.860 --> 00:39:28.457 in the gene name like,
NOTE Confidence: 0.875489852222222
00:39:28.457 --> 00:39:30.519 because some of them,
NOTE Confidence: 0.875489852222222
00:39:30.520 --> 00:39:32.124 it’s just a series of letters and numbers.
NOTE Confidence: 0.875489852222222
00:39:35.560 --> 00:39:37.936 It can be really easy to miss those
NOTE Confidence: 0.875489852222222
00:39:37.936 --> 00:39:40.712 sort of typos if you’re staring at that
NOTE Confidence: 0.875489852222222
00:39:40.712 --> 00:39:43.230 report for hours as you’re working.
NOTE Confidence: 0.637223015
00:39:46.730 --> 00:39:49.354 So variant interpretation is
a role that a lot of labs are hiring genetic counselors for, and I personally was not aware of that prior to starting my graduate program and some of the programs do introduce very interpretation. So for example at Long Island University we had a course with one of the major commercial labs and we had weekly data interpretation assignments. But a lot of the labs are also training genetic counselors to analyze and write test reports because they have their own methods, their own data systems, so.
They really are just looking for this sort of clinical and molecular expertise that genetic counselor is doing up getting through our graduate degrees. So you’ll be hearing from some of our other genetic counselors about what their roles are like in a clinical setting and overall, laboratory genetic counseling is fairly different in a non patient facing role. And so I’m not in a patient facing role and my interaction with patients is very limited. I will sometimes take calls from anxious patients who are requesting updates on test results and my psychosocial skills that I’ve learned from genetic...
00:41:20.670 --> 00:41:24.600 counseling school come into play there.
00:41:24.600 --> 00:41:27.316 But I also need to redirect them
to contact their ordering.
00:41:27.316 --> 00:41:29.280 Provider or secret approval to genetics,
so I am not in the sort of role where you
would be counseling or speaking to patients.
00:41:32.130 --> 00:41:35.363 I need to review each test order.
00:41:35.363 --> 00:41:38.443 So I do get to see every test
order that comes into our lab.
00:41:40.540 --> 00:41:43.804 I need to review each test order.
00:41:43.804 --> 00:41:46.100 So I get to learn about a lot of
different conditions and what genes
might be associated with them,
because as I said before,
I'm reviewing those orders to find
appropriate gene lists that the
provider is not specific about what gene lists they do want included. It’s a great way to continue learning. In the jaw and I’m exposed to some really interesting and unique cases. So the skills that we learn in our graduate programs also get applied in new ways. Instead of reviewing clinical notes to prepare to counsel a patient, for example, I’m going to be reviewing those notes to determine what gene list can be applied to based on the differential diagnosis. Or, as I said before, seeing if they meet criteria for testing. If their insurance company
00:42:49.346 --> 00:42:52.160 is leaving it up to us.
00:42:52.160 --> 00:42:55.142 And in those cases where we can’t get prior authorization.
00:42:55.142 --> 00:42:57.130 So we also get to interact with a lot of different providers and a system as they’re following up with on results or want to learn more about what the result or the test is or what it means.
00:43:04.177 --> 00:43:07.330 With on results or want to learn more about what the result or the test is or what it means.
00:43:07.330 --> 00:43:10.309 For example, I once received a phone call from a primary care doctor who wanted me to help him come up with a strategy for explaining the results of his patient and advising on following.
up testing for family members.

Because he was an older doctor, he admitted to me that his education in genetics was very basic and he really didn’t know how to handle that status. So I really enjoy being able to collaborate with and educate some of the physicians or other providers like nurses or physicians assistants about genetic counseling. And helping them come up with strategies for speaking to their patients.

So Emily’s work in bioinformatics also showcases that we get the opportunity to learn new skills that would not necessarily be learned in a clinical
setting where we’re facing patients and also shows that genetic counseling degrees can be applied in some pretty interesting and diverse work settings. I think many of us pictured being in a clinical setting long term when we entered Graduate School. But sometimes you find that maybe that’s not what you want to do long term or you want to experience some other areas of genetics. And working in a genetics lab is really a great way to learn. To apply those skills that you’ve learned in a new and different way,
and it’s pretty rewarding.

And it’s very interesting.

So I’m going to stop sharing my screen, but I would be happy to take a look and see if there’s any questions.

Let’s see.

I think there’s one, and if maybe both of you would be able to comment on how did you decide or what was maybe the the precipitated the decision to move into laboratory genetic counselor after Graduate School?

So I can answer first,
since I’m already unmuted. But it wasn’t necessarily a plan for me. I applied to a lot of different roles and interviewed with a few different places, but I really liked just that. That sort of versatility of this role. I’ve always had an interest in molecular genetics, so more than just looking at the sort of symptoms and counseling people I really like knowing what goes into. What comes out of a test? Basically like how is that data being interpreted to say that? Because of this sort of
Genetic change, this is. This is what might happen with this person as far as their health. Goes. And something about my position is that I will be getting the opportunity to do more of a hybrid position. In that I'll be seeing patients once a week, so that was another thing that I really liked. I wasn't really drawn to a full time clinical position necessarily, because I do like being in the laboratory setting, but I just love the fact that I could really go in any direction that I wanted to.
By working at the lab, within reason, of course, but it’s just allowed me to. Really look how all this sort of different interests that I had.

Emily, do you have? Yeah, so I think for me a big part of it was actually my work that I did before Graduate School, so I was in a lab and they knew I wanted to be a genetic counselor so they were really great about keeping me in the loop.
00:47:55.686 --> 00:47:57.264 with everything that they were doing
NOTE Confidence: 0.880289061666667
00:47:57.264 --> 00:47:59.056 and kind of showing you the ropes.
NOTE Confidence: 0.880289061666667
00:47:59.060 --> 00:48:01.434 A little bit, and through that I came
NOTE Confidence: 0.880289061666667
00:48:01.434 --> 00:48:03.815 to really appreciate just how varied
NOTE Confidence: 0.880289061666667
00:48:03.815 --> 00:48:06.270 the genetic counselor role can be.
NOTE Confidence: 0.880289061666667
00:48:06.270 --> 00:48:08.318 Prior to that, I really only had
NOTE Confidence: 0.880289061666667
00:48:08.318 --> 00:48:10.436 exposure to clinical genetic counselors,
NOTE Confidence: 0.880289061666667
00:48:10.436 --> 00:48:12.690 so I really found it fascinating
NOTE Confidence: 0.880289061666667
00:48:12.690 --> 00:48:14.586 that I could be genetic counselor,
NOTE Confidence: 0.880289061666667
00:48:14.590 --> 00:48:17.276 but still kind of help behind the scenes,
NOTE Confidence: 0.880289061666667
00:48:17.276 --> 00:48:19.814 almost not necessarily seeing patients,
NOTE Confidence: 0.880289061666667
00:48:19.814 --> 00:48:22.403 but with the testing itself and
NOTE Confidence: 0.880289061666667
00:48:22.403 --> 00:48:24.825 that kind of drove me to think
NOTE Confidence: 0.880289061666667
00:48:24.825 --> 00:48:26.540 about maybe going into laboratory
NOTE Confidence: 0.880289061666667
NOTE Confidence: 0.94496872375
00:48:31.030 --> 00:48:34.230 And then I also see some other questions,
one specific to bioinformatics. So I actually worked with a bioinformatics expert and he kind of taught me the ropes to do what I need to do. I’m definitely not an expert whatsoever, but I can do the job and I think the reason that I was tired was they were more looking for somebody who could kind who had that genetic knowledge. And they figured it would be easier to have somebody who already had that, like I learned in genetic counseling school and teach me the little bit of bioinformatics that I needed to know, rather than getting a bioinformatics expert.
person and teaching them everything about genetics that is needed for my job.

Excellent and we do have some other great questions, but I have to be strict about our timing since we have such a busy schedule, so I'm pleased. Stay tuned afterwards and when we have the general Q&A we'll try to answer as many questions as we can, but I think we'll pass it over to Julie for our talk about reproductive genetics and thank you Maya and Emily. That was a great presentation, and we've been doing this for three years now and I feel like I
learned something new every time.

So thank you again.

Great hi everyone.

Julie, let me get you on the spotlight. There we go. Perfect.

So hello, my name is Julie. I’m a reproductive genetic counselor at Yale and I work in the Department of Obstetrics, Gynecology and Reproductive Sciences. Specifically in the section of maternal fetal medicine and I frankly could not do what I do without Maya and Emily.

And the work that they do because we may be the.
People in front of the patients, but they are doing the work as they said behind the scene that allows us to really provide answers for many of our patients.

So with regard to my professional background, I graduated from the Joan Marks graduate program in Human Genetics at Sarah Lawrence College. I am not going to say what year I have been working in maternal fetal medicine at Yale for over five years. And I would say that over 90%, probably more than 95% of the individuals that I see are considered high risk with regard to either their maternal or fetal
Concerns or a combination of the two. And I previously worked at two different medical centers and over the years have had the opportunity to specialize in not only prenatal or reproductive genetic counseling, but cancer, pediatric and adult genetic counseling services. So my jobs have predominantly involved direct patient care. I’ve also engaged in various clinical research studies and. Currently and with my last job, I would say at least 5% of my position involves teaching.
Currently I'm teaching the maternal fetal medicine fellows every two weeks. I provide a lecture so that they can be introduced to various genetic topics that will help them when they are out in the field, as well as help pass their board exams. But sometimes I'm called in to speak with nurses, social workers, sonographers, et cetera to really educate them about what I do and how I can support their job and their patient population. In the past I served as the Director of Clinical training and as a clinical rotation supervisor for students.
that were enrolled in the genetic counseling program at the Icahn School of Medicine at Mount Sinai, and to date I have had the pleasure of working with and supervising over 200 genetic counseling interns who were enrolled in various genetic counseling training programs across the US, so.

I would say they keep me on my toes and make sure that I’m staying on top of everything within my field, which is great.

So I wanted to step back and talk about the difference between when I say...
prenatal or reproductive genetic counselor,

NOTE Confidence: 0.7793304

since sometimes you’ll see

NOTE Confidence: 0.7793304

those used interchangeably,

NOTE Confidence: 0.7793304

but in if we start with prenatal,

NOTE Confidence: 0.7793304

that’s really talking about

NOTE Confidence: 0.7793304

what is occurring or existing

NOTE Confidence: 0.7793304

during pregnancy before birth.

NOTE Confidence: 0.7793304

So prenatal care is the health care

NOTE Confidence: 0.7793304

that women receive during pregnancy and.

NOTE Confidence: 0.7793304

Some genetic counselors refer to

NOTE Confidence: 0.7793304

themselves as prenatal genetic

NOTE Confidence: 0.7793304

counselors because they are either

NOTE Confidence: 0.7793304

predominantly or exclusively working

NOTE Confidence: 0.7793304

with individuals and their partners,

NOTE Confidence: 0.7793304

while a pregnancy is in progress.

NOTE Confidence: 0.7793304

Other genetic counselors refer to
themselves more broadly as reproductive genetic counselors because they are collectively working with. Individuals who are pregnant planning to become pregnant or interested in discussing concerns that arose during a previous pregnancy. And genetic counselors have filled an important role in supporting patients to make informed and value consistent reproductive decisions.

Since prenatal screening and diagnostic testing were first possible. So in my specialty, some common reasons for referral include
advanced maternal or paternal age,
which generally means that someone is 35 years old of age or older at the time of delivery.
If someone has a personal or family history of a known or suspected genetic condition, intellectual disability or congenital structural difference such as a congenital heart defect, Cleft lip or palate etc.
There can be atypical fetal ultrasound findings or abnormal prenatal screening or diagnostic results.
Concern about whether medications, drugs, alcohol,
environmental exposures that occurred prior to or during pregnancy may impact fertility or fetal development. Pregnancy outcome. Someone might be a carrier for an inherited condition or chromosome rearrangement and would like to discuss what this means for their family planning and another common reason is history of recurrent pregnancy loss or subfertility and infertility. And one question that I wanted to ask everyone is what percentage of pregnancies that result in first trimester miscarriage are found.
to have a chromosome disorder, which means either extra or missing chromosome material. And this is ranging from the low end of being five to six percent, 10 to 15 percent, 25 to 30, or 50 to 55%. Oh, we’re getting quite a range for our poll answers. We’ll give it maybe 10 more seconds. All right, 321. And can you see that I can? Right, that’s that’s very interesting to see. So the correct answer. Would be 50 to 55%, which I know is shocking to a lot of people and this was something that was really
flying under the radar years ago

before there was testing available on DNA that we received from cells.

From pregnancies that have miscarried.

So I think you know one thing that we try to tell patients when a chromosome problem is identified during pregnancy is not necessarily something that makes them feel better.

The fact that this is a common occurrence but can help them to feel that there’s not something wrong with them, and that there’s hope that they can go forward and have a successful pregnancy.

Other common reasons for referral
are individuals that are requiring assisted reproductive technologies to achieve pregnancy.

Individuals who are donating eggs or sperm for these purposes.

People who have multifetal pregnancies, including twins, triplets, quadruplets, and more.

Those with specific ethnic or racial groups, or geographic areas with a higher incidence of certain genetic conditions who are interested in having genetic carrier screening for those conditions, and just general interest in discussing test options that are available for individuals or
their reproductive partners prior to or during pregnancy. So this can include things such as.

Genetic carrier screening testing for chromosome conditions during pregnancy, etcetera.

So as Alex had mentioned, some things that happened during a reproductive genetic counseling session are very common in other settings as well, but typically we are obtaining medical, reproductive and environmental exposure histories. Taking a family history depending on the reason for referral,
we may go back multiple generations. Other times we may not have the time to allot to that and it may not be relevant to the reason for referral. So it may be a much smaller pedigree. We are explaining the risk for or the diagnosis of a genetic condition. Talk about inheritance recurrence risks, the benefits, limitations, and risks of test options that people have, prognosis management, current treatment options as well as prevention and current research options. We will interpret the results of this testing that they’ve elected to have. Discuss the implications for the
current fetus and future pregnancies, and talk about next steps.

We will talk to them about assisted reproductive technologies and we will also try our best to support them while they’re making these decisions. And trying to, by the way, forgive the sound in the background. That’s my dog growling.

So we’ll talk to them about decision making and try to take into account their personal, their religious, their ethical and moral values, et cetera. And I would say our biggest goal has to
be establishing rapport with patients.

The faster the better because we can’t do the other things on this list.

We can’t achieve the other goals unless we have established rapport.

We’ll try to assess their needs, exchange and discuss relevant information that’s specific to them and their situation.

We will try to elicit their thoughts and feelings, support their autonomy and their decision making, and we provide short term psychosocial support and patient advocacy.

And we know our limits as well, so that we identify situations
...where someone might need additional medical referrals or psychological referrals or support services.

You know whether that includes referring them to actual support groups, individual counselors referring them out to other medical specialists? And we try to serve as an ongoing resource as their needs and their desires evolve over time. And I selected a case that actually took place several years ago, but I thought that it is a really good example of someone’s needs and desires may evolve over time.
and how we have to pivot with them along that journey. So for this particular patient, she was 40 years old and referred for genetic counseling at approximately 12 weeks in pregnancy. Due to advanced maternal age. We know that this or we learn that this patient and her partner had one previous pregnancy that resulted in miscarriage at a that happened about six months prior to my meeting with them. And chromosome analysis that was performed on the products of conception from that pregnancy revealed that
NOTE Confidence: 0.757935746
01:02:19.573 --> 01:02:21.847 the fetus had a sporadic meaning,
NOTE Confidence: 0.757935746
01:02:21.850 --> 01:02:22.926 not inherited.
NOTE Confidence: 0.757935746
01:02:22.926 --> 01:02:25.078 Chromosome condition that is
NOTE Confidence: 0.757935746
NOTE Confidence: 0.900123333333333
01:02:29.000 --> 01:02:30.900 So you know just rapidly
NOTE Confidence: 0.900123333333333
01:02:30.900 --> 01:02:32.420 giving a little background.
NOTE Confidence: 0.900123333333333
01:02:32.420 --> 01:02:34.860 Individuals typically have 23 pairs
NOTE Confidence: 0.900123333333333
01:02:34.860 --> 01:02:37.859 of chromosomes for a total of 46.
NOTE Confidence: 0.900123333333333
01:02:37.860 --> 01:02:40.723 The 1st 22 pairs are numbered one
NOTE Confidence: 0.900123333333333
01:02:40.723 --> 01:02:43.579 through 22 and the 23rd pair are.
NOTE Confidence: 0.900123333333333
01:02:43.580 --> 01:02:45.980 Called the sex chromosomes,
NOTE Confidence: 0.900123333333333
01:02:45.980 --> 01:02:48.426 which are X&amp;amp;Y chromosomes.
NOTE Confidence: 0.900123333333333
01:02:48.426 --> 01:02:52.164 Most females have two X chromosomes
NOTE Confidence: 0.900123333333333
01:02:52.164 --> 01:02:55.687 and most males have 1X and Y1Y
NOTE Confidence: 0.900123333333333
01:02:55.687 --> 01:02:57.395 chromosome and chromosome aneuploidy
NOTE Confidence: 0.900123333333333
01:02:57.395 --> 01:03:00.824 is a term that’s given when there is
NOTE Confidence: 0.900123333333333
01:03:00.824 --> 01:03:02.859 an abnormal number of chromosomes.
NOTE Confidence: 0.900123333333333
01:03:02.860 --> 01:03:06.046 So for example there are 45
NOTE Confidence: 0.900123333333333
01:03:06.046 --> 01:03:08.660 or 47 instead of 46.
NOTE Confidence: 0.900123333333333
01:03:08.660 --> 01:03:10.345 Most of these chromosome conditions
NOTE Confidence: 0.900123333333333
01:03:10.345 --> 01:03:12.837 occur by chance as a result of an
NOTE Confidence: 0.900123333333333
01:03:12.837 --> 01:03:15.006 egg or a sperm cell that was created
NOTE Confidence: 0.900123333333333
01:03:15.006 --> 01:03:17.274 with an extra or missing chromosome.
NOTE Confidence: 0.900123333333333
01:03:17.280 --> 01:03:22.238 And as women get older,
NOTE Confidence: 0.900123333333333
01:03:22.238 --> 01:03:25.679 there is an increased chance to have
NOTE Confidence: 0.900123333333333
01:03:25.680 --> 01:03:27.941 a child with a chromosome abnormality.
NOTE Confidence: 0.900123333333333
01:03:27.941 --> 01:03:34.150 So for example, someone who is.
NOTE Confidence: 0.900123333333333
01:03:34.150 --> 01:03:36.398 20 years old has less than a one
NOTE Confidence: 0.900123333333333
01:03:36.398 --> 01:03:39.206 in 400 chance to have a child with
a chromosome disorder and at the age of 38 it has gone up to 1%. But I would like to view that as a 99% chance that there would not be a chromosome condition. So trisomy 13 is considered one of the more severe chromosome disorders, and it can result in miscarriage or the birth of a child who has severe intellectual disability and medical concerns and physical abnormalities. Years ago, they used to think that this condition was universally fatal within the first weeks of life, but now with...
Improved technology approximately 5 to 10% of children with this condition can survive past the first year. I am going to skip ahead. To say that during the counseling session we were talking about how this usually occurs by chance. This patient was 40 years old, so we talked about that her chance of having another pregnancy with a chromosome problem was not felt to be significantly different than anyone else. Her age in the general population, which is in a 1 to 2% range and we offered her some screening tests that can assess risk for some of the more
common chromosome conditions as well
diagnostic tests that permit us
to actually look at the chromosomes.
For the pregnancy under a microscope
with over 99% accuracy and we
talked about the benefits,
limitations and risks of both
diagnostic tests,
including the one in 400 risk
for miscarriage that these tests
are associated with.
So she expressed that although she
would love to have the information
that the diagnostic test can provide,
she did not want to have a test
that had a risk of miscarriage and elected to have cell free screening.

So by the way, the details that I've provided on these slides are really for later when you're looking through them in case you needed some background to understand what we're talking about. But I am just going to kind of cut to the chase for what happened with this couple. It turned out that she was not able to get a result from the cell free DNA screening, and that by itself indicates that the pregnancy could be at increased risk for certain. Chromosome disorders.

So we again offered her diagnostic testing and she elected to have an amniocentesis,
and when she arrived on the day of the test prior to performing the AMNIO, ultrasound revealed that the fetus had a brain abnormality that is called semi lobar lobar holoprosencephaly. So the counseling session involved talking to the couple about what holoprosencephaly is talking to them about the different outcomes that can occur depending upon the extent to which the abnormality exists and we talked about that this condition is not always genetic, it can be caused because of environmental problems or exposures it can occur.
Due to sporadic chromosomal or genetic conditions and it can be associated with some inherited conditions as well. They elected to proceed with the amniocentesis. The first thing that we found out is that the fetus had the correct number of chromosomes, which is 46. An additional test on that specimen revealed that there was no little pieces of chromosomes that are extra or missing, so that reduced the chance for over 150 different genetic syndromes. Only a small group of which could be linked to holoprosencephaly and then,
with the help of a laboratory

We were able to determine a panel of genes that would be appropriate to test. This fetus had one copy of a pathogenic variant in a specific gene that was called zic 2. And this disorders in one copy of this gene are associated with the condition that’s called holoprosencephaly type 5, and in this condition we commonly see semi lobar holoprosencephaly.
01:08:36.647 --> 01:08:39.414 variant was what caused the fetus to
01:08:39.414 --> 01:08:41.424 have holoprosencephaly and we each
01:08:41.424 --> 01:08:44.162 have two opportunities to have this
01:08:44.162 --> 01:08:47.508 gene working properly. If one is not.
01:08:47.508 --> 01:08:51.132 Then the person would be predicted to
01:08:51.132 --> 01:08:54.396 be at risk to have holoprosencephaly,
01:08:54.400 --> 01:08:56.479 but this gene is not fully penetrant,
01:08:56.480 --> 01:09:02.116 which means that some people with
01:09:02.116 --> 01:09:05.224 variants in this gene that are considered
01:09:05.224 --> 01:09:08.599 to have no symptoms or have such
01:09:08.599 --> 01:09:11.525 mild symptoms that they are never
01:09:11.525 --> 01:09:14.475 diagnosed as having this condition.
01:09:14.480 --> 01:09:17.344 So we recommended that the parents be tested.
01:09:17.350 --> 01:09:20.170 And it turned out that the
01:09:20.170 --> 01:09:22.960 partner had the same variant,
and although we certainly did not have information regarding his brain MRI, outwardly he had no signs of having holoprosencephaly. They were informed that with each pregnancy there’s a 50% chance that the fetus would inherit this pathogenic variant, but that not every fetus with the variant would have holoprosencephaly, and we also let them know that fetal ultrasound may not always show us that a fetus will have complications secondary to this variant. So we wanted to make sure they...
had the they were aware that they had the option of having. In vitro fertilization where the egg is fertilized outside of the body would then genetic testing on that would then genetic testing on that pre embryo to see if it was affected. Or they could have targeted prenatal testing during pregnancy to see whether or not the fetus inherited the variant and they elected to conceive their third pregnancy via IVF. There was targeted prenatal excuse me preimplantation genetic testing for this variant. And they were able to have a successful transfer and implantation of what was
01:10:39.588 --> 01:10:42.618 predicted to be an unaffected embryo.
01:10:42.620 --> 01:10:44.320 They elected to have prenatal testing early in pregnancy at around
01:10:44.320 --> 01:10:47.844 11 weeks to confirm the accuracy
01:10:47.844 --> 01:10:51.000 and it confirmed the fetus was.
01:10:51.000 --> 01:10:54.726 Not carrying this variant and
01:10:54.730 --> 01:10:56.840 they delivered a healthy baby boy,
01:10:56.840 --> 01:10:59.310 so this was really a very long haul
01:10:59.310 --> 01:11:02.518 for this couple to have a baby.
01:11:02.518 --> 01:11:05.430 It was over five years.
01:11:05.430 --> 01:11:08.710 But it was certainly a wonderful outcome.
01:11:08.710 --> 01:11:12.490 So in my last couple of slides I wanted to say that you know pregnancy
01:11:12.490 --> 01:11:14.578 can be a lot of things that can
01:11:14.578 --> 01:11:16.511 be planned and unplanned, desired,
not desired,

NOTE Confidence: 0.852805355833333

can be wonderful and exciting scary.

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Anxiety provoking etc.

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The list could go on and on and

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reproductive genetic counselors

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have both the responsibility and

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the privilege of educating,

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supporting and working with individuals

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who are faced with making really

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difficult decisions both prior to

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pregnancy and during pregnancy and

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working with this patient population,

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I can say has been extremely

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rewarding and challenging.

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And I have stayed consistent as

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a genetic counselor who does
01:12:00.838 --> 01:12:02.766 reproductive genetic counseling from
01:12:05.516 --> 01:12:09.332 that forces me to consistently be
01:12:13.090 --> 01:12:15.700 So sorry, I know I ran over by a few minutes.
01:12:17.090 --> 01:12:20.218 That was a great talk as
01:12:20.218 --> 01:12:22.626 always and I wanted to maybe pose a
01:12:22.626 --> 01:12:25.422 question to you before we hop over to
01:12:25.422 --> 01:12:27.287 Sarah’s talk about cardio genetics.
01:12:27.290 --> 01:12:29.830 Someone had asked about carrier
01:12:29.830 --> 01:12:31.862 testing specifically and whether
01:12:31.862 --> 01:12:34.088 that’s doing mostly sequencing
01:12:34.090 --> 01:12:35.959 of the genome or are there other
01:12:35.959 --> 01:12:37.650 ways to test for a carrier?
Could you just maybe expand upon that a little bit? The majority of genetic care carriers testing is being done through gene sequencing. Although for some of the more common genetic conditions that someone can be a carrier for other technology is actually better for identifying carriers than gene sequencing, so you know that is part of our responsibility is. Knowing what screening is appropriate for someone in general and how we might modify that based on personal history or family history or ethnicity so.
There’s not one straight answer, but yeah, overall gene sequencing is our go to for the vast majority of conditions. Thank you again and we’ll certainly tab back during the general Q&A, but let me see if I can get Sarah up and running. There we go. Yep, right there. OK, so let me just. we go. Yep, right there. OK, so let me just. we go. Yep, right there. OK, so let me just. we go. Yep, right there. OK, so let me just.
So first you know just a little bit about me. I am originally a jersey girl and from New Jersey I did my undergrad at Montclair State in New Jersey and then after two years I went back to grad school and I did the genetic counseling masters program at Rutgers, which was, you know, interesting to be a part of it. A new program. So I graduated from Rutgers in 2020, so working at Yale similar to some of the other presenters has been my first and only job since graduating, so they must have, you know,
good retention here, which is nice.

So I work more specifically in the heart and Vascular Center within the Yale New Haven Health System and to get more specifically than that I’m in the congestive heart failure program as well as the inherited hypertrophic cardiomyopathy program. That’s the program that my role is specifically built into, although I do see a lot of other indications outside of just HCM. So yeah, we do have an HCM that’s called the center of Excellence,
meaning that we have a clinic here that is, essentially a designated by the Hypertrophic Cardiomyopathy Association as being a multidisciplinary Center for patients. With this condition, including services such as genetic counseling and assistance with testing and family screening. OK so first just to kind of give an overview of what my role is like in the world of cardiology. Firstly, the people that I work with there is another genetic counselor
who specializes in cardiovascular genetics and if you attended this talk, you know a year or two years ago. I believe Arpita was the one, giving the talk about cardiac genetics. So if you were here at the year or two years ago you might remember some things from her. Call per and I have a pretty similar roles that overlap a lot, but aren’t entirely the same. So I work with a lot of different cardiologists. Actually, some cardiologists who are just general cardiologists and then also specialty cardiologists,
including electrophysiologists,

so doctors who work with us,

you know,

problems with the hearts electrical

specialists in cardiomyopathies,

you know, there are a lot of different types of
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how we build out our schedules. You know some programmatic concerns and things like that. You know there is a lot of collaboration with laboratory staff. Both. You know here at like the Yale DNA lab or you know, in some external genetics labs, you know we send some tests kind of through the L DNA lab as well as some. Uh, larger like commercial outside genetics labs. You know often just depends on things like insurance,
01:17:53.990 --> 01:17:56.530 turnaround time, things like that.
NOTE Confidence: 0.9532504
01:17:56.530 --> 01:17:58.966 And then finally I do work
NOTE Confidence: 0.9532504
01:17:58.966 --> 01:18:00.990 with some other research staff.
NOTE Confidence: 0.9532504
01:18:00.990 --> 01:18:03.790 There are a couple of research projects
NOTE Confidence: 0.9532504
01:18:03.790 --> 01:18:06.189 that I'm a little bit involved in,
NOTE Confidence: 0.9532504
01:18:06.190 --> 01:18:08.956 so I often work with, you, know,
NOTE Confidence: 0.9532504
01:18:08.956 --> 01:18:11.288 various researchers as well.
NOTE Confidence: 0.9532504
01:18:11.290 --> 01:18:13.778 And then I think a lot of people
NOTE Confidence: 0.9532504
01:18:13.778 --> 01:18:15.324 question how patients actually
NOTE Confidence: 0.9532504
01:18:15.324 --> 01:18:17.868 get to see a genetic counselor.
NOTE Confidence: 0.9532504
01:18:17.870 --> 01:18:20.026 So many of our patients will get
NOTE Confidence: 0.9532504
01:18:20.026 --> 01:18:22.850 referred to us, maybe through their PCP.
NOTE Confidence: 0.9532504
01:18:22.850 --> 01:18:23.800 For example,
NOTE Confidence: 0.9532504
01:18:23.800 --> 01:18:25.535 if their General practitioner happened
NOTE Confidence: 0.9532504
01:18:25.535 --> 01:18:27.706 to notice something like a cardiac
NOTE Confidence: 0.9532504
01:18:27.706 --> 01:18:29.616 murmur during a regular evaluation,
that’s something that might prompt further cardiac workup and potentially some genetic testing. These patients might get referred by a cardiologist. If they’re already seeing one for something, like high blood pressure, you know, high cholesterol if they identify some other problem that needs more specialized testing and discussion, they might refer to our specialty group. Some patients do self refer. We do get some self referrals. You know if you Google scale
cardiology genetic testing,

you know you’ll come to our page

and some patients contact us

that way also through the NSGC

find a genetic counselor tool.

There have been a handful of patients who

have utilized that to reach out to us.

You know, if they feel that there may be a need

for genetic testing within their families.

Additionally, you know we do a lot

of predictive testing for family

members when the proband tests positive for a genetic mutation,

so a lot of our patients do come

to us because they’re related to
someone else that I saw previously and provided testing for. And finally we do do some cross referring here between the different genetic counseling specialties. For example, a cancer genetic counselor might uncover a family history of cardiomyopathy or an arrhythmia? Something like that, and recommend that a patient sees you know a cardiac genetic counselor for more specialized testing and then vice versa.
we often find family history of something such as a cancer syndrome where they refer to another genetic counselor in a different specialty so we do have some collaboration here between all the disciplines. So and a question, I do get a lot from prospective students is just what does your week look like? You know, like what’s an average kind of week in the life for you as a cardiac GC. So obviously a large kind of chunk of my time gets taken up by the genetic counseling consults. And as mentioned earlier,
we do have, you know, kind of different ways we do the consoles. Currently we are doing a lot of virtual phone visits. Patients since the COVID pandemic, and then occasionally we do also have in person, consults with a cardiologist in the clinic potentially. In addition to that, you know a lot of my time taken up by charting. You know, putting in the visit nose, placing in the orders for genetic testing, and then of course everything you send to the lab will come back to you.
So a lot of time is taken up by reviewing patient results, and then of course calling those out and discussing those with the patient and their families to make sure that we have a plan for their management. We have a weekly cardiovascular genetics case conference with I and the other genetic counselor and a group of cardiologists where we discuss you know interesting or difficult cases. And then finally on Fridays I am always doing case Prep for Mondays so you can go and just start.
the cycle all over again.

And then outside of kind of the more

I do have some research duties as well.

I've helped out with a couple

projects here and there.

I have had the opportunity to supervise

some students which I love doing.

I've had some students shadow with me,

which was, you know,

a great opportunity for someone who

is not even too far out of school.

Myself, you know, we do have.

Within Yale,

multidisciplinary genetic counseling
01:22:28.090 --> 01:22:31.110 conferences and addition to that,
NOTE Confidence: 0.8024989945
01:22:31.110 --> 01:22:33.375 the National Society of Genetic
NOTE Confidence: 0.8024989945
01:22:33.375 --> 01:22:35.542 Counselors has a yearly conference,
NOTE Confidence: 0.8024989945
01:22:35.542 --> 01:22:38.110 so occasionally I may be working
NOTE Confidence: 0.8024989945
NOTE Confidence: 0.8024989945
01:22:40.590 --> 01:22:43.124 We we did mention special interest groups.
NOTE Confidence: 0.8024989945
01:22:43.130 --> 01:22:44.940 Earlier there is a cardiac
NOTE Confidence: 0.8024989945
01:22:44.940 --> 01:22:46.026 special interest group.
NOTE Confidence: 0.8024989945
01:22:46.030 --> 01:22:48.850 I am not personally super involved in it,
NOTE Confidence: 0.8024989945
01:22:48.850 --> 01:22:50.572 but it’s a great resource which
NOTE Confidence: 0.8024989945
01:22:50.572 --> 01:22:52.065 is something I really wanted
NOTE Confidence: 0.8024989945
01:22:52.065 --> 01:22:53.510 to include on this slide.
NOTE Confidence: 0.893053622
01:22:55.700 --> 01:22:56.522 Like I mentioned,
NOTE Confidence: 0.893053622
01:22:56.522 --> 01:22:58.440 a lot of our consoles are virtual,
NOTE Confidence: 0.893053622
01:22:58.440 --> 01:23:00.778 but we do also have inpatient consults.
NOTE Confidence: 0.893053622
01:23:00.780 --> 01:23:03.828 Sometimes with the patient in the
We might go to the cardiac intensive care unit. You know, we might be asked to go see them and speak with them about potentially doing genetic testing. And occasionally, I might have meetings with outside labs staff to discuss new testing panels that they are piloting and things like.

First of all, of course we have hypertrophic cardiomyopathy. It's really kind of my bread and butter. So first of all, of course we have hypertrophic cardiomyopathy.
I see a lot of this condition. Essentially it happens when the muscular walls of the heart become too thick, often because of, you know, a genetic mutation that someone carries. It’s actually a relatively common condition. Probably about one in every 250 people may have this condition according to you know, most recent estimates and then having this thickness this abnormal thickness in the heart muscle can cause a couple of different problems, including something that’s called diastolic dysfunction, which means that the heart can’t relax properly to fill with enough blood.
So sometimes there’s trouble with blood getting to the rest of the body. It can increase the risk for A-fib. In turn, increases the risk for stroke. And sometimes you know part of an HCM workup may involve differentiating what we call true genetic HTM from other conditions that can mimic genetic HCM. You know these are things such as hypertensive heart disease, something called athletes heart, which can happen when someone undergoes intense athletic training. For many years. It can kind of remodel the structure.
01:24:55.614 --> 01:24:58.710 of the heart and make it thicker.

NOTE Confidence: 0.90862958

01:24:58.710 --> 01:25:01.580 And often you know the types of

NOTE Confidence: 0.90862958

01:25:01.580 --> 01:25:05.010 HCM that we see are just isolated,

NOTE Confidence: 0.90862958

01:25:05.010 --> 01:25:08.706 but they can be present in in different

NOTE Confidence: 0.90862958

01:25:08.706 --> 01:25:11.030 syndromes as well as part of something.

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01:25:11.030 --> 01:25:11.702 For example,

NOTE Confidence: 0.90862958

01:25:11.702 --> 01:25:12.374 Noonan syndrome,

NOTE Confidence: 0.90862958

01:25:12.374 --> 01:25:15.350 HCM is just one feature of that condition,

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01:25:15.350 --> 01:25:17.810 but it may present with other

NOTE Confidence: 0.90862958


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01:25:19.040 --> 01:25:21.184 So in contrast to

NOTE Confidence: 0.90862958

01:25:21.184 --> 01:25:22.256 hypertrophic cardiomyopathy,

NOTE Confidence: 0.90862958

01:25:22.260 --> 01:25:23.772 we have dilated cardiomyopathy,

NOTE Confidence: 0.90862958

01:25:23.772 --> 01:25:26.040 as you can see in this

NOTE Confidence: 0.90862958


NOTE Confidence: 0.90862958

01:25:27.720 --> 01:25:29.743 The walls of the heart are kind
01:25:29.743 --> 01:25:31.380 of stretched down and thinner, so it makes it difficult for the heart to pump blood to the rest of the body.

01:25:37.180 --> 01:25:40.015 There is a risk of developing arrhythmias or other problems in the electrical system of the heart or risk of progression to end stage heart failure.

01:25:49.524 --> 01:25:51.930 Some of the genes that cause DCM may also present with muscular disease as well, so that’s something that we may want to pay attention to and additionally we do get some referrals for patients who have postpartum cardiomyopathy,
often from a provider who’s kind of an expert in cardio obstetrics. And it’s been found that about 15% of women who develop a postpartum or peripartum cardiomyopathy will actually carry a pathogenic mutation for a dilated cardiomyopathy. OK, and then there’s also something called arrhythmogenic cardiomyopathy, which has the name implies comes with an increased risk of ventricular arrhythmias. Given this sort of fibrofatty and scar tissue replacement of these normal healthy cells in the heart because of the gene mutation that someone may carry and this is
an interesting condition because exercise can actually make it worse. So these patients are often told to moderate their exercise and try, you know, more low impact things like walking or yoga as opposed to you know, high intense athletics and then this is just kind of a quick. Overview of how cardiomyopathies may be, you know there’s often a lot of imaging done. Genetic testing. Obviously blood work. Sometimes patients may present with symptoms such as shortness of breath, chest pain, or heart palpitations,
and sometimes people may have no symptoms at all, just those changes inside the heart. There are many different genes that can cause these problems. Some of them may only cause one specific type of cardiomyopathy and some can cause multiple different types. Most of the inheritance patterns that we tend to see with these conditions is mostly autosomal dominant, but there can be some other inheritance patterns as well. And finally, these conditions are managed through medications.
lifestyle changes, sometimes devices, surgery and then of course family screening is an important part of you know the management process. If an individual in the family is identified to carry a pathogenic mutation. We will discuss the option of genetic testing for that patients family members who may be at risk to also develop the condition. And then just briefly like I mentioned, we also see you know some inherited arrhythmias. We work with electrophysiologists for genetic testing.
For these patients, two of the most common ones that you may have heard of include Brugada syndrome and long QT syndrome. In essence, these are EKG changes that may increase the risk of a dangerous heart arrhythmia. So it’s something that has to be managed often with medications or also avoiding any triggers that can additionally cause these arrhythmias to appear. And then just a few more just kind of round out these slides. Sometimes we see patients sometimes we see patients for inherited aneurysms,
which is essentially a bulging of
the blood vessel that you see here.
This is also something that can
present as an isolated condition or
can be syndromic presenting with other
features of a connective tissue disease.
Another clinic that I work
in is an amyloidosis clinic.
You know, really,
until I started to get into cardiac genetics,
essentially it is this protein from the
gene called TR that becomes misfolded
and clumps up in different organs.
Primarily can cause heart failure neuropathy, but can also cause some kind of other unusual features such as carpal tunnel syndrome and gastrointestinal. It’s a really underrecognized condition definitely, so I kind of take every chance I get to spread awareness about it. And then finally we see a lot of dyslipidemias. One example of that is something that’s called familial hypercholesterolemia. Essentially, it’s you know a problem that causes an increased amount of LDL cholesterol,
01:30:31.930 --> 01:30:35.714 which is the bad cholesterol in the blood.

01:30:35.720 --> 01:30:36.947 And you know,

01:30:36.947 --> 01:30:39.401 exposure to these high levels of

01:30:39.401 --> 01:30:41.109 cholesterol overtime can increase

01:30:41.109 --> 01:30:43.563 the risk for heart disease and

01:30:43.563 --> 01:30:46.090 other cardiovascular complications.

01:30:46.090 --> 01:30:47.788 It can depend on the severity.

01:30:47.790 --> 01:30:51.774 If someone has one mutation or two mutations,

01:30:51.780 --> 01:30:54.462 and it is actually a condition

01:30:54.462 --> 01:30:57.030 that is pretty easily managed,

01:30:57.030 --> 01:30:58.766 these patients are encouraged

01:30:58.766 --> 01:31:00.068 to take statins,

01:31:00.070 --> 01:31:01.942 which is a medication that helps

01:31:01.942 --> 01:31:03.610 to lower the bad cholesterol

01:31:03.610 --> 01:31:05.740 in the blood and then also.
Modify their lifestyle.

You know proper diet and exercise to help decrease the risk of developing heart disease.

OK so I won’t, you know, belabor this slide too much because I think we already kind of know the basic components of a genetic counseling physic, because it tends to be, you know, fairly consistent between all the specialties, but something interesting that I’ve had come up in this setting is this question right here?

Can you tell me what I’ve been diagnosed with? And I can’t tell you how many...
times I have gotten this question. You know, from patients who recently were diagnosed with, for example, a cardiomyopathy. And they just have no idea what they’ve been diagnosed with. They’re very confused, and they’re very nervous about, you know the potential to say, pass it down to their children. So a big portion of the visit is me often explaining to the patient what they have, you know, and explaining kind of the basic genetics concepts of it, as well as the inheritance.
And then of course we want to work out you know what test is best? Should we order single gene? Should we order a panel? Should we order flexim sequencing?

That’s a big part of the conversation. And then finally, insurance concerns, you know, is something that’s probably, an issue for anyone in any specialty of genetic counseling. Patients are often concerned about things like cost, and you know, can these results be used against me? Will I have any any privacy
01:32:38.276 --> 01:32:39.440 with my genetic results?

01:32:39.440 --> 01:32:41.384 So that’s also something that we discussed with them.

01:32:41.384 --> 01:32:43.814 And then these are some concerns that are, you know,

01:32:46.530 --> 01:32:47.086 kind of come up often in cardiac genetic counseling.

01:32:48.754 --> 01:32:49.980 They may happen in other specialties as well, but this is just, you know,

01:33:01.741 --> 01:33:03.396 in the past two years here what I’ve noticed.

01:33:03.400 --> 01:33:05.746 which is something that can potentially indicate that there is a gene causing

01:33:05.746 --> 01:33:08.644 have a family history of sudden cardiac death,
arrhythmias in someone’s family.

So obviously you can expect that there is a psychological impact for the patients remaining relatives if they’re seemingly healthy family members suddenly died of an arrhythmia, so that’s something that you know.

I think a lot of my tough cases were these cases where there was a family history of just sudden death, with no apparent precipitating factors. In addition to that, something that comes up a lot in cardiology is uncertain.

Findings on genetic tests. You know,
01:33:43.100 --> 01:33:44.780 people may be aware that sometimes

01:33:44.780 --> 01:33:46.530 when we get genetic tests back,

01:33:46.530 --> 01:33:48.216 the results aren’t always clear cut.

01:33:48.220 --> 01:33:50.368 Sometimes we aren’t sure if the

01:33:50.368 --> 01:33:52.164 genetic change is actually causative

01:33:52.164 --> 01:33:54.580 of a disease and this makes it you

01:33:54.580 --> 01:33:56.796 know a little tricky to figure out.

01:33:56.800 --> 01:33:59.376 How do we best screen the family for

01:33:59.376 --> 01:34:01.344 this condition? Are they at risk?

01:34:01.344 --> 01:34:03.909 And also how do we communicate this

01:34:03.909 --> 01:34:06.439 uncertainty to the patients themselves?

01:34:06.440 --> 01:34:10.298 So the patient doesn’t feel overwhelmed.

01:34:10.300 --> 01:34:13.404 And then a type of testing that comes

01:34:13.404 --> 01:34:15.303 up occasionally in cardiovascular

01:34:15.303 --> 01:34:17.918 genetics is post mortem testing.
You know, often in these cases of a family history of sudden death they may pursue what’s called a molecular autopsy, which is genetic testing on the deceased to help check and see if there’s any potential mutations we can identify and can kind of help us figure out why did this person passed away suddenly, and you know what is the risk to the remaining? Family members should the remaining consider something such as getting a defibrillator, for example, to protect them from any dangerous heart rhythm. So super quick. This is just kind of
01:34:53.154 --> 01:34:55.137 an example pedigree of a patient that I had seen and it was interesting, not specifically because of the cardiac genetics of it, but kind of for some other factors.

You can see all these yellow sections here indicate patients affected with the condition called HT. Essentially it causes abnormal blood vessel formations in the body. It can cause problems with bleeding however these.

Individuals here also had a dilated cardiomyopathy and their deceased brother was thought to have heart failure.
and in rare cases, HT can cause basically a certain type of heart failure. But looking at all these cases of dilated cardiomyopathy and another relative on this side, who is thought to have some type of heart issues, we were having a discussion about, you know, is it possible that there are? Actually, two different genetic conditions running in this family. Or are these heart problems just related to the HT and these patients are just happening to
have some rare complications, so this was an interesting case, not even because of the genetics so much, but because of, you know, this patient had already gone through all this counseling and testing for this other condition and now we were coming in and talking to him about the potential there being a second condition in the family. You know he had. Already known that his children didn’t inherit the HHD from him, but now we had new concerns about,
you know,

the potential of dilated cardiomyopathy

gene running through the family as well.

So this was just kind of an interesting case.

Kind of, you know,

some interesting psychosocial aspects of this as well.

OK,

and that’s it,

I think I’m a little we’re we’re a little behind the Times Now,

a little behind the Times Now,

so maybe I will stop sharing and

maybe won’t take any questions

at this time and come back for

questions at the end.

And of course, here is my email address.
NOTE Confidence: 0.872706955
01:36:57.010 --> 01:36:59.302 If anyone has any interest in cardiovascular genetics, please email me.
NOTE Confidence: 0.872706955
01:37:01.270 --> 01:37:02.870 I love answering prospective student questions.
NOTE Confidence: 0.8997892
01:37:06.220 --> 01:37:08.271 Thanks, Sarah, just quickly we did get a question about shadowing and we’re not currently able to accommodate requests at this time. Is that right?
NOTE Confidence: 0.895826615
01:37:11.740 --> 01:37:14.150 shadowing and we’re not currently able to accommodate shadowing requests at this time. Is that right?
NOTE Confidence: 0.895826615
01:37:14.150 --> 01:37:16.078 able to accommodate shadowing requests at this time. Is that right?
NOTE Confidence: 0.895826615
01:37:16.078 --> 01:37:17.960 we did get a question about shadowing and we’re not currently able to accommodate shadowing requests at this time. Is that right?
NOTE Confidence: 0.898508115454545
01:37:19.130 --> 01:37:21.909 Yeah, so it, it depends if it’s someone already in a program or if it’s someone who is not in a program.
NOTE Confidence: 0.898508115454545
01:37:21.909 --> 01:37:24.513 someone already in a program or if it’s someone who is not in a program.
NOTE Confidence: 0.898508115454545
01:37:24.513 --> 01:37:27.390 it’s someone who is not in a program.
NOTE Confidence: 0.898508115454545
01:37:27.390 --> 01:37:31.006 At this point it is a little bit more more difficult just because of the regulations between like
the hospital and the university.

But if someone has specific questions about wanting, you know information about potentially shadowing here, I would encourage them to reach out to me. Just because there may be some more specific information that I could give. OK, perfect thank you. So I am going to try to share my screen again. Give me one second I'm going to.

Emily Chen, who spoke to us last year and wasn’t able to be here today, but she wanted me.
She wanted me to share her talk for her, so let’s get that going. Please let me know if you can’t see and or hear it, but here we go.

Hi everyone, my name’s Emily and I’m a genetic counselor here at Yale, working under the General Genetics Clinic. I’m here to talk today a little bit about pediatric and general genetics.

So to start off with, I’ll go over a brief biography, I actually graduated from UConn to the University of Connecticut, and I studied psychology and...
NOTE Confidence: 0.855333176363636
01:39:09.960 --> 01:39:11.600 Afterwards I went to the
NOTE Confidence: 0.855333176363636
01:39:11.600 --> 01:39:13.115 University of California, Irvine,
NOTE Confidence: 0.855333176363636
01:39:13.115 --> 01:39:15.974 where I did my masters in genetic
NOTE Confidence: 0.855333176363636
01:39:15.974 --> 01:39:18.621 counseling and the first job I took
NOTE Confidence: 0.855333176363636
01:39:18.621 --> 01:39:20.980 out of grad school was at Veritas.
NOTE Confidence: 0.855333176363636
01:39:20.980 --> 01:39:24.690 It was a genetic testing startup company.
NOTE Confidence: 0.855333176363636
01:39:24.690 --> 01:39:26.760 That was performing whole genome
NOTE Confidence: 0.855333176363636
NOTE Confidence: 0.855333176363636
01:39:28.902 --> 01:39:30.847 individuals who wanted to just
NOTE Confidence: 0.855333176363636
01:39:30.847 --> 01:39:32.792 learn about their disease risk.
NOTE Confidence: 0.855333176363636
01:39:32.800 --> 01:39:33.752 And then from there,
NOTE Confidence: 0.855333176363636
01:39:33.752 --> 01:39:35.860 after about 3 1/2 years at Veritas.
NOTE Confidence: 0.855333176363636
01:39:35.860 --> 01:39:38.868 I since then been here at Yale working
NOTE Confidence: 0.855333176363636
01:39:38.868 --> 01:39:40.982 in the general Genetics clinic
NOTE Confidence: 0.855333176363636
01:39:40.982 --> 01:39:44.300 for the past 2 1/2 years or so.
So what exactly is general Genetics? While it can be split into Pediatrics and adult genetics, and that’s mainly based on depending if the hospital you work at has a separate Children’s Hospital or not. If it does, then you might be only seeing pediatric patients here at Yale. We don’t have a separate Children’s Hospital, it’s all just under young New Haven, so that’s why our clinic is called General Genetics, which you might sometimes see interchangeably with adulters clinics.
But the key piece about our clinic is that we don’t specialize in a specific disease group or specific type of disease or indication. We really just see everything under the sun, so that includes cancer, cardio, and anything else that you might hear about today.

So some of the common reasons people are referred to us when they’re trying to better understand if there’s a genetic reason or genetic etiology for why
they’re experiencing their symptoms, or some kind of diagnosis that might tie multiple symptoms together, but there are other physicians haven’t been able to figure out. A lot of times you might hear about medical mysteries or diagnostic odysseys that’s often found in the general genetics clinic. We are trying to solve a lot of these cases. Some of my patients are over 40 years old and they have significant symptoms that they’ve just, you know, lived their entire life with, and doctors haven’t been able to
figure out before they finally come to the genetics clinic, we’re able to possibly give them the diagnosis in some cases. So I’ve listed some of these common indications here, so we see a lot of kids who have developmental delay or intellectual disabilities or autism spectrum disorder. Also see adults with these indications as well. We see people who have skeletal dysplasias, which means a problem with the bones. Sometimes they can be very fragile, sometimes they can be too hard and sometimes individuals can be short,
or they might have just proportionate bones or other types of problems with their bones.

We also see people who were born with what we call brick defects, so it can be a problem with their heart or a cleft lip or palate, or they can have extra fingers or toes or fused fingers and toes or defects just refers to anything that someone may be born with.

In addition to people who have seizures, there’s a genetic cause for the seizures, and if so, sometimes there’s a better
We also see individuals with what we call metabolic conditions, so those conditions are typically found on newborn screening, where baby has a heel prick and they’re tested for these disorders. There are other metabolic conditions that aren’t necessarily covered in newborn screening. Essentially, these are the types of conditions that sometimes can be treated with a diet adjustment and special formula added to their diet, and that’s kind of how we treat those.
Some of those conditions. Uh, we also see patients who have muscular weakness or atrophy in addition to connective tissue disorders of people who might have very stretchy skin in addition to you know, family history of strokes or aneurysms, for example. So you can see that we see a lot of complex conditions that might affect multiple systems. And I listed one example here. So one condition called Stickler syndrome patients can have.
They can have a cleft palate, which means the palette on the top part of the mouth inside didn’t close completely when forming mitral valve prolapse, which is a problem with the heart. Vision loss due to retinal detachments and the retinas. The back layer of the eye that you know takes in the light and sends the signals to the brain to process of your retinal. Then it can definitely result in vision loss and sometimes it can be reattached with surgery and then these individual can also have bone and joint problems even within the same family. Some people might have one symptom.
Some people might not have any symptoms of Stickler syndrome, or they’re very mild and hard to pick up. It can present very differently, but if we do diagnose someone stickers syndrome, You wanna check all of those different organ systems and make sure they’re working OK and check them overtime to make sure they don’t develop hearing loss later, or develop the vision loss due to the retinal detachment later. So usually after we find a diagnosis
01:44:36.480 --> 01:44:38.060 with diagnosis for someone,
NOTE Confidence: 0.889700138571429
01:44:38.060 --> 01:44:39.638 we have to coordinate their care.
NOTE Confidence: 0.889700138571429
01:44:39.640 --> 01:44:41.004 That includes helping testing
NOTE Confidence: 0.889700138571429
01:44:41.004 --> 01:44:42.970 any other relatives or providing
NOTE Confidence: 0.889700138571429
01:44:42.970 --> 01:44:44.394 recommendations for the condition.
NOTE Confidence: 0.889700138571429
01:44:44.394 --> 01:44:47.003 In some rare cases we actually might be
NOTE Confidence: 0.889700138571429
01:44:47.003 --> 01:44:49.278 able to direct them to curative treatment,
NOTE Confidence: 0.889700138571429
01:44:49.280 --> 01:44:52.088 whether it be an enzyme replacement
NOTE Confidence: 0.889700138571429
01:44:52.088 --> 01:44:54.120 therapy or you know some kind of
NOTE Confidence: 0.889700138571429
01:44:54.120 --> 01:44:55.919 gene therapy that might be available.
NOTE Confidence: 0.889700138571429
01:44:55.920 --> 01:44:57.770 So that’s not common,
NOTE Confidence: 0.889700138571429
01:44:57.770 --> 01:45:01.298 but it’s becoming more more well
NOTE Confidence: 0.889700138571429
01:45:01.298 --> 01:45:04.438 more studied and more medications.
NOTE Confidence: 0.889700138571429
01:45:04.440 --> 01:45:05.984 They’re starting to come
NOTE Confidence: 0.889700138571429
01:45:05.984 --> 01:45:07.930 out so definitely a hot
NOTE Confidence: 0.95306331
01:45:07.940 --> 01:45:08.990 area right now.
The team members that you'll probably be working with overall include administrative staff. You may be. If you’re lucky, you might have a genetic counseling assistant as well, or a nurse coordinator. Those individuals can have additional training in medicine, so they might be able to discuss negative results or may be able to. You know, determine whether additional records are needed for a visit before the patient comes to see us or gather family history.
information beforehand as well.

You may work with a social worker who

provides that additional

psychosocial support or identifies

resources for families that might not

necessarily be specific to traumatics.

The genetic counselor is probably a

little bit better suited to searching

for what kind of advocacy groups are

appropriate based on a diagnosis,

but in terms of daily types of difficulties that

families might be going through.

For example,

if they’re looking for disability services,

a social worker might be better well.
Equipped to identify those for the families.

Metabolic dietitian if you see metabolic patients. They’re registered dietitians who have specialized training for these taking care of these patients.

You may work with nurse practitioners or physician assistants, so these are advanced practice providers that typically have some additional genetics training, usually on the job training. So after some time they may, you know, specialize in certain type of disorder. It just depends where you work.
And then there’s also the medical.

Genesis, which is a key part of the general genetics team, so that is a physician who will be seeing the patients, and they typically have already done a residency in Pediatrics or internal medicine and then they do an additional residency in genetics. You might hear a called a fellowship or residency basically mean the same thing.

So what’s the role of a genetic counselor during these visits? Then we helped elicit the patients concerns. Excuse me excuse me, we also gather
their medical and family history

If it hasn't already been done.

If it has already been done,

we’ll probably go over it again and make

sure there’s nothing else that we’re missing.

A physical exam that’s usually

done by that advanced practice

provider or medical geneticist.

We also review the benefits, risks,

and limitations of genetic testing,

and we go over the different types

and we go over the different types

of results that are possible.

And then when the results do come back,

we help to interpret and return

those results to the family
and then digestible manner. Uh. Then afterwards will help provide continued support and identify any other resources for the patient and their family that they might need. Something that I didn’t necessarily list on here, but it is a part of the job as well, oftentimes the genetic counselor is the person who helps fill out the medical part of the paperwork that has to be done is we have to talk about the symptoms and why this individual needs genetic testing.
So I wanted to go for one case example here. What I may or may not see in the genetics clinic. So for example, let’s say a 9 month old male is coming in to see me and they have they were born with a congenital heart problem called pulmonary valve stenosis. And the baby was born with a normal weight and size. But then over time the pediatrician noticed that the weight gain started slowing down as well as the growth in general.
So nine months old, that’s pretty concerning. We see a lot of babies who are starting to follow up growth curves, so this is a pretty common reason to refer to us and then, together with both of the above symptoms, the parents you know after talking to them some more and going over the different systems parents have also. Explained to you that they feel like the baby has pretty easy bruising. Maybe they had a blood draw before and noticed bruising just from you know, the nurse trying to take the blood for example. Or maybe they had prolonged
bleeding after a cut or something. So putting those three together when you’re doing a family history, you start asking a little bit more about conditions that you might be thinking about. Are your differential list. So the different possible diagnosis. So going over their family history, find out pretty pretty much not too much going on on. The only thing is, let’s say mom is shorter than expected and she also had some kind of unknown heart problem when she was a baby. She doesn’t know what it was, but she was.
Otherwise healthy and didn’t need surgery.

So putting all this together, oftentimes families will ask you or the geneticist you know, what do you think my kid has?

And maybe 20 years ago, 30 years ago when we only knew about a handful of conditions, it might have been easier back then.

But now we know about, you know, over 7000 different conditions. In addition to that, we now understand that a lot of individuals can present very mildly, we expanded the spectrum of symptoms that someone can have in the spectrum.
01:50:42.235 --> 01:50:44.437 of severity that we can see.

01:50:44.440 --> 01:50:45.772 So now when people.

01:50:45.772 --> 01:50:46.438 Ask us,

01:50:46.440 --> 01:50:47.508 you know what condition

01:50:47.508 --> 01:50:48.843 do you think my child

01:50:48.860 --> 01:50:50.828 has or I have.

01:50:51.040 --> 01:50:53.272 Oftentimes, we just I would say

01:50:53.272 --> 01:50:55.487 I've never heard myself or the

01:50:55.487 --> 01:51:00.145 we are certain that the child has

01:51:00.145 --> 01:51:02.392 any Commission in particular because

01:51:02.392 --> 01:51:05.296 of the expansion of this knowledge,

01:51:05.300 --> 01:51:07.580 we can’t really pinpoint anything.

01:51:07.580 --> 01:51:09.204 But sometimes what we will say is,

01:51:09.210 --> 01:51:11.498 you know, based on what I’m seeing here,
the multiple symptoms I do think it is genetic. I just don’t know what exactly it is and we need to do testing. With that so Fast forward, let’s say we do testing for this individual and we find out that they have Union syndrome, so that’s a condition that it kids can be born with what we call pulmonary valve stenosis again. And it’s something we often see together and think of immediately when we see a baby born with that. Especially when they start to slow down when their weight gain later on,
and they can also have, you know, problems with the playlets and which leads to the easy bruising or prolonged bleeding times. And they can also have some other symptoms. Sometimes there can be some mild hearing loss as well, so developmental delays depending on the type of Noonan syndrome so you can see that even if I said Noonan syndrome, it really depends on which gene. So all of that happens after the visit. I would go over the results with the family. Let’s say it’s a dominant form of Noonan syndrome here or autosomal dominance,
and we go over that inheritance pattern. Go over who else might need to be tested or might want to consider testing. So in this case example, it’s the mother. These individuals can also be of short stature. So for the baby we might consider sending to endocrinology to monitor and also see if maybe growth hormone is something that might be given at some point in the future. For the mom, we’re more concerned about adult onset symptoms of Newman syndrome. For example, they can have something called hypertrophic cardiomyopathy.
which is a thickening of the heart.

Muscle makes it harder for it to pump, so that's definitely something that we want to keep an eye out for.

And then Mom might just want to know her future risks to other children that she might be having in down the line. So we’d go over all of that with the family, and again, we help to coordinate care between any other specialists. They need to be followed with and then we provide that psychosocial support and resources, especially when given the new diagnosis.
that someone might not be expecting.

I will say as part of my role as a general genetic counselor,

we also get called to the hospital sometimes.

So when we’re on call,

that means that anyone on the floor or the ICU the intensive care units may call us for a genetic consult.

So for example,

if you have a patient that’s hospitalized with the symptoms that we just mentioned,

maybe the baby is severely, in addition to not really gaining a lot of weight,

a lot of weight,

maybe they’re they have seizures.
and they're coming in. And I think that might be genetic. They will be addressed, and then we'll have to go see and evaluate. And do you know, talk to the family about whether or not testing is indicated and where to go from there. So in addition to just seeing patients outpatient setting, sometimes we see patients in inpatient as well.

So I just wanted to end with some other common conditions that you may see.
in a general genetics clinic, so you may have heard of Down syndrome neurofibromatosis type 121 to 22 Q 11.2 deletion syndrome, which is a microdeletion syndrome. That’s pretty common. Carter, Willie, we already talked about Noonan, EKU or phenylketonuria and it’s one of those metabolic conditions cystic fibrosis and then Duchenne or Becker muscular dystrophy is another common indication or common condition that you may see. I will say though, a lot of hospitals have specialized
01:54:52.474 --> 01:54:54.479 clinics for these common diseases,
01:54:54.480 --> 01:54:56.286 so in those cases you may not
01:54:56.286 --> 01:54:57.908 end up seeing those patients
01:54:57.908 --> 01:54:59.878 in the general genetics clinic.
01:54:59.880 --> 01:55:02.659 We may see the more complex cases
01:55:02.660 --> 01:55:04.184 or these medical mysteries.
01:55:04.184 --> 01:55:06.915 Versus the the ones that are more
01:55:06.915 --> 01:55:09.130 easily diagnosed or taken care
01:55:09.130 --> 01:55:10.970 of by other providers.
01:55:10.970 --> 01:55:12.728 So that’s kind of, I think,
01:55:12.730 --> 01:55:15.172 interesting that these are some
01:55:16.180 --> 01:55:17.758 I would say 1020 years ago
01:55:17.770 --> 01:55:19.780 that you might have seen nowadays
01:55:19.790 --> 01:55:23.238 I I almost rarely see some of these
01:55:23.238 --> 01:55:25.065 conditions because they’re followed
NOTE Confidence: 0.874343541111111
01:55:25.065 --> 01:55:27.390 in other specialty clinics instead,
NOTE Confidence: 0.874343541111111
01:55:27.390 --> 01:55:29.435 as other physicians become more
NOTE Confidence: 0.874343541111111
01:55:29.435 --> 01:55:32.010 well versed with how to order.
NOTE Confidence: 0.874343541111111
01:55:32.010 --> 01:55:33.665 You know simple genetic testing
NOTE Confidence: 0.874343541111111
01:55:33.665 --> 01:55:34.989 rather than these medical.
NOTE Confidence: 0.874343541111111
01:55:34.990 --> 01:55:36.925 District cases where it’s best
NOTE Confidence: 0.874343541111111
NOTE Confidence: 0.840143292
01:55:41.250 --> 01:55:44.740 Alright, so other than that.
NOTE Confidence: 0.840143292
01:55:44.740 --> 01:55:46.195 That’s pretty much all I
NOTE Confidence: 0.840143292
01:55:46.195 --> 01:55:47.861 have for you guys today.
NOTE Confidence: 0.840143292
01:55:47.861 --> 01:55:51.640 I’m sorry I couldn’t be there in person,
NOTE Confidence: 0.840143292
01:55:51.640 --> 01:55:53.536 but hopefully this was helpful and
NOTE Confidence: 0.840143292
01:55:53.536 --> 01:55:55.839 gave everyone a taste of what pediatric
NOTE Confidence: 0.840143292
01:55:55.839 --> 01:55:57.795 or general genetics may look like.
NOTE Confidence: 0.840143292
01:55:57.800 --> 01:55:59.100 Thanks again and bye.
01:56:03.430 --> 01:56:07.410 OK, so that was Emily with general and pediatric genetics.

01:56:09.170 --> 01:56:14.330 I’m going to. Let’s see.


01:56:21.450 --> 01:56:23.850 to come. But last but certainly not least, we’ll have a meet here.

01:56:23.850 --> 01:56:26.210 not least, we’ll have a meet here.

01:56:36.350 --> 01:56:38.950 OK, good afternoon everyone.

01:56:38.950 --> 01:56:40.900 Hope that the.

01:56:40.900 --> 01:56:42.820 Your day’s been going well.

01:56:42.820 --> 01:56:44.356 I’m going to share my screen.

01:56:48.880 --> 01:56:50.973 So today I am pleased to talk

01:56:50.973 --> 01:56:52.870 to you about about cancer,

01:56:52.870 --> 01:56:53.650 genetic counseling.
My name is Amy Kelly. I'm a genetic counselor at Smilow Cancer genetics. I work closely with Alex. And just some background about me. I graduated from State University of New York, Oswego, in 2014 with my Bachelors of Science in Zoology and then took one year off between graduating and going to my masters because I wanted time to apply for programs. Also wanted to get some volunteering experience. I graduated from the Icon School.
NOTE Confidence: 0.931722196666667
01:57:27.502 --> 01:57:29.976 of Medicine at Mount Sinai with my
NOTE Confidence: 0.931722196666667
NOTE Confidence: 0.931722196666667
01:57:32.150 --> 01:57:34.558 And I’m board certified as a 2017
NOTE Confidence: 0.931722196666667
01:57:34.558 --> 01:57:36.474 and actually just recertified this
NOTE Confidence: 0.931722196666667
01:57:36.474 --> 01:57:38.904 year because as a genetic counselor
NOTE Confidence: 0.931722196666667
01:57:38.904 --> 01:57:41.610 need to recertify every five years.
NOTE Confidence: 0.931722196666667
01:57:41.610 --> 01:57:44.116 And I’ve been with the Smilow cancer
NOTE Confidence: 0.931722196666667
01:57:44.116 --> 01:57:46.048 genetics program since June of 2017,
NOTE Confidence: 0.931722196666667
01:57:46.050 --> 01:57:48.024 so coming up on my 5 year
NOTE Confidence: 0.931722196666667
01:57:48.024 --> 01:57:49.877 anniversary here where I practice
NOTE Confidence: 0.931722196666667
01:57:49.877 --> 01:57:51.290 specifically clinical cancer,
NOTE Confidence: 0.931722196666667
01:57:51.290 --> 01:57:52.140 genetic counseling.
NOTE Confidence: 0.746508814545454
01:57:54.590 --> 01:57:56.984 So just says an overview overview
NOTE Confidence: 0.746508814545454
01:57:56.984 --> 01:57:59.180 about hereditary cancer in general,
NOTE Confidence: 0.746508814545454
01:57:59.180 --> 01:58:01.760 we like to refer to red
NOTE Confidence: 0.746508814545454
flags for hereditary cancer.

That just means that there’s specific findings in someone’s family that may be more suspicious that the cancers could be hereditary. That would be the big thing. Cancers at early ages, and that’s not all cancers. Some cancers may naturally occur in younger ages, but for example, breast cancer diagnosed under 50, and that is in and of itself suspicious of a hereditary predisposition to develop that type of cancer. Another thing that we may see is...
NOTE Confidence: 0.746508814545454
01:58:35.628 --> 01:58:37.303 multiple family members in the
NOTE Confidence: 0.746508814545454
01:58:37.303 --> 01:58:39.037 same family with the same type
NOTE Confidence: 0.746508814545454
01:58:39.037 --> 01:58:41.030 of cancer or associated cancers.
NOTE Confidence: 0.746508814545454
01:58:41.030 --> 01:58:43.232 So that would be multiple people
NOTE Confidence: 0.746508814545454
01:58:43.232 --> 01:58:45.044 and multiple generations on one
NOTE Confidence: 0.746508814545454
01:58:45.044 --> 01:58:46.168 side of the family,
NOTE Confidence: 0.746508814545454
01:58:46.170 --> 01:58:48.966 all with colon cancer for example.
NOTE Confidence: 0.746508814545454
01:58:48.970 --> 01:58:51.370 Or there are some cancers when
NOTE Confidence: 0.746508814545454
01:58:51.370 --> 01:58:52.570 they are hereditary,
NOTE Confidence: 0.746508814545454
01:58:52.570 --> 01:58:54.550 they can be associated with other
NOTE Confidence: 0.746508814545454
01:58:54.550 --> 01:58:56.920 risks of other cancers such as breast
NOTE Confidence: 0.746508814545454
01:58:56.920 --> 01:58:58.900 or ovarian cancer or pancreatic cancer
NOTE Confidence: 0.746508814545454
01:58:58.900 --> 01:59:00.939 in the same family we’re seeing
NOTE Confidence: 0.746508814545454
01:59:00.939 --> 01:59:03.304 colon and uterine cancer in the same.
NOTE Confidence: 0.746508814545454
01:59:03.304 --> 01:59:03.661 Family,
so it’s not necessarily seeing more cancer in general, but sometimes it can be a risk factor, particularly this the same type of cancer or known associated cancers that would be a red flag. Rare cancer, some cancers are very rare and may not be hereditary, but there are specific cancers that are rare that are more likely to be hereditary that includes ovarian cancer, pancreatic cancer, or male breast cancer. Those cancers, specifically, are more likely to be hereditary. Other cancers,
such as these tumors with very long names, paragangliomas and pheochromocytomas, are rare tumors. Paragangliomas are typically benign. They occur along this axis of the body. But they are very rare tumors that are actually have a high percentage of high likelihood of being hereditary. A few chroma cytoma is essentially a paraganglioma that sits on the adrenal gland, so right above the kidney and people with a feel chroma cytoma presence may develop symptoms of essentially overactive fight or
flight symptoms such as anxiety, sweating, flushing, etcetera.

So those tumors, specifically rare tumors, are also known to be more likely to be hereditary. Cancers that are unusually aggressive so those cancers could be things like. Prostate cancer is very common in the general population for men, however, it’s less common for prostate cancer to be aggressive or be a cause of the man’s death. So when we see prostate cancer, that is aggressive or spread to other parts of the body.
That is also a red flag for that cancer being hereditary. Or one person having multiple types of cancer is common in the general population. One in three people will develop cancer. However, seeing one person with multiple cancers does increase suspicion that that person may have a genetic predisposition to develop more than one type of cancer. So that includes women or men who have bilateral breast cancer. So cancer in both breasts.
or someone who’ve had colon.

Additionally, we also know that individuals who are Ashkenazi Jewish are more likely to specifically have hereditary breast and ovarian cancer, which I will talk about a little in a little bit, but there are two genes, specifically BRC one and BRC two. However, individuals who are Ashkenazi Jewish have a higher likelihood of having mutations in these two genes, specifically one in 40 people that are Ashkenazi Jewish.
Will have a BRCA one or BRCA 2 mutation compared to the non Ashkenazi population which is about one in 400.

Now for a quick poll. So, so I mentioned cancer is very common, but approximately what percentage of cancers are hereditary? Is it less than 5%? Is it between 5 to 10% and again popping average? Here is between 20 to 25%? Or is it around 40%? So take a couple seconds think.

Approximately what percentage of cancers are hereditary? No 46% great guys have. That’s
basically all you guys can all be.

Cancer genetic counselors now so that’s correct.

So most about 5 to 10% on average definitely not a small amount, but the grand majority are actually not hereditary.

And there’s little nice little pie chart here, So what causes cancer?

70% of cancer we consider to be sporadic, meaning that it’s due to things like the environment like asbestos exposure,
radiation exposure, things like lifestyle. We know that tobacco use can be a risk factor for certain types of cancers, including lung cancer. The natural aging process is also a risk factor for cancer. That's why we tend to see cancers diagnosed and diagnosed in older ages because as we age we have a higher likelihood of acquiring a random mutation that could then develop into a cancer. Also, sometimes cancer does just occur due to complete random chance. You see Hereditary familial and those two.
Those terms do sound very similar.

We distinguish them in cancer genetics a little bit.

20% of cancer is familial. Familial means.

You may see clusters of the same type of cancer in someone’s family. However,

we do not find one single genetic change. One single gene mutation that is causing those.

Answers so we do think that for familial cancer there may be small genetic factors, possibly in multiple genes, possibly polygenic that is working with shared environmental
factors and lifestyle factors, because families often live in the same locations, live have similar lifestyles, and this combination may create an overall higher risk of cancer in that one family that’s not caused by one single gene hereditary. Is a type of cancer that we can do genetic testing for that means someone is born with or they inherit one single genetic change. A harmful gene mutation or pathogenic variant that predisposes them over their lifetime to developing.
certain types of cancers.

And this is an overview of what we think about. You know why? 

How why cancer develops.

This is an oversimplification, but I think it kind of drives the point home of why there is this predisposition.

So on the top these are cells in the body, so everyone as we know has 2 copies of every gene, so with sporadic cancer at the top.

Overtime a gene could acquire a mutation again due to some sporadic factors such as the environment,
lifestyle, aging, random chance, however, that second copy of the gene is still working, so that cell continues to grow and act normally. It’s only when someone acquires a second hit, someone the gene requires a second hit that that cell essentially maybe nonfunctional and through other complicated processes can then go on to become a tumor. Where that cell growth is now not regulated with an inherited mutation, it’s different because someone’s
already born with a mutation

already in one copy of their genes,

and this is present in all

the cells of their body.

However, they have one copy of the
gene is still working normally.

So essentially though,

that one copy can work throughout

someone’s entire lifetime,

but since someone’s essentially
down a line of defense,

if someone needs to acquire.

Only a single mutation in that one copy

of the gene to then start the process,

potentially of a tumor developing,

so that is why with hereditary cancers we
NOTE Confidence: 0.900804462727273
02:06:43.072 --> 02:06:45.695 may just see more cancer in the family,
NOTE Confidence: 0.900804462727273
02:06:45.700 --> 02:06:48.720 younger cancers, more rare cancers,
NOTE Confidence: 0.900804462727273
02:06:48.720 --> 02:06:50.700 multiple cancers in one person.
NOTE Confidence: 0.900804462727273
02:06:50.700 --> 02:06:51.681 Things like that,
NOTE Confidence: 0.900804462727273
02:06:51.681 --> 02:06:53.316 and this is called this.
NOTE Confidence: 0.900804462727273
02:06:53.320 --> 02:06:55.635 This process is called knudsens
NOTE Confidence: 0.900804462727273
02:06:55.635 --> 02:07:00.623 2 hit hypothesis of why cancer
NOTE Confidence: 0.900804462727273
02:07:00.623 --> 02:07:02.416 develops and why specifically
NOTE Confidence: 0.900804462727273
02:07:02.416 --> 02:07:03.928 The way or present the way
NOTE Confidence: 0.900804462727273
02:07:03.928 --> 02:07:05.447 it does in certain families.
NOTE Confidence: 0.891500639666666
02:07:08.480 --> 02:07:10.664 So just want to talk about a typical
NOTE Confidence: 0.891500639666666
02:07:10.664 --> 02:07:12.856 day for a cancer genetic counselor and
NOTE Confidence: 0.891500639666666
02:07:12.856 --> 02:07:15.494 I’d like to start by talking about an
NOTE Confidence: 0.891500639666666
02:07:15.494 --> 02:07:17.797 example case so I know we’ve talked.
NOTE Confidence: 0.891500639666666
My other colleagues have talked about genetic counseling in general, so I won’t go into the details of exactly the genetic counseling process, but essentially the patients that I see are those who have who have cancer, who have had cancer or who have family history of cancer, and the goal through a pedigree is to look for those red flags that I mentioned earlier to determine what is the likelihood or the risk that there is a hereditary predisposition to cancer in someone’s family.

So for this case this is a 63 year old female.
She was diagnosed with breast cancer when she was 56.

She never had genetic testing previously, but she came in now to talk about genetic testing in her family. There’s even see a lot of cancer going on. All those little dark dark corners. And her maternal side of the family, there is a mutation in a specific gene called ATM, so her cousin had breast cancer and has reportedly an ATM mutation which I could not confirm with records and ATM. Which I will talk about in a little bit is a moderate risk breast cancer gene.
So the ATM mutation may be playing a role in her cousin’s breast cancer and that is a hereditary cancer gene. However, it appeared based on what? Patient reported that that ATM mutation was coming from her cousin’s father, which is not a blood relative to my patient, meaning that my patient would not have been at risk of inheriting that same mutation. So thinking about red flags in her family when we’re also seeing is apart from her history of breast cancer, which is not a very young age. It was after menopause,
which is less likely to be hereditary.

We do see an ovarian cancer in her maternal great aunt, but a little bit distant to her and related through her mother who is 83 with no cancer on her father’s side. However, her paternal uncle did die from prostate cancer, and if you remember the prostate cancer is common in men. One in nine men will develop prostate cancer. Metastatic prostate cancer is less common and more likely to be hereditary.
and also thinking about a pedigree. I think about limitations in family histories. Her father’s side is small with only men, which can limit an assessment. So this is I like this case because it shows kind of the importance of taking into account both sides of the family thinking about those associated cancers, thinking about what. What can we confirm with the records? Ideally we always want to confirm test results with records, but sometimes we can’t. So this is a great case to show that even though this risk factors on maybe her
mom’s side, there’s also maybe more.

Significant risk factors on her father’s side.

So for her we talked about hereditary cancer, specifically BRC one and BRC A2, which I will talk about and talked about other hereditary cancer genes as well nowadays with cancer genetic counseling. Of course we go through the benefits of doing genetic testing, which for cancer genetic counselors is really prevention, particularly for individuals who maybe do not have cancer. Or who possibly could have a
predisposition to another cancer.

The goal of knowing about hereditary cancer risk is that if we know someone’s at higher risk of certain cancers, there’s certain screening options that possibly could be condoned be done, and also possible surgical options that can actually prevent cancer. Additionally, for people who have cancer, it can be important for treatment decisions, meaning that there are some chemotherapies or treatments that may be more targeted. Individuals with certain gene mutations and it also may be helpful in planning surgery. So for her we did genetic testing.
02:11:20.440 --> 02:11:22.765 and nowadays genetic testing tends to be comprehensive and there’s a lot of genes listed here the about 1212 years ago.


02:11:26.943 --> 02:11:30.509 10 years ago.

02:11:30.510 --> 02:11:31.585 8 years ago.

02:11:31.585 --> 02:11:32.650 7 years ago.

02:11:32.650 --> 02:11:33.649 6 years ago.

02:11:33.650 --> 02:11:35.590 We really only doing testing for two genes when we’re talking about hereditary breast cancer, colloquially.

02:11:35.590 --> 02:11:37.290 Called the Braca genes.

02:11:37.290 --> 02:11:38.789 A lot of people have heard about these genes since Angelina Jolie went public with her own BRC 1 mutation and
her decision to have a prophylactic bilateral mastectomy to remove both breasts. Initially, these two genes we've been testing for over 20 years, so we have a lot of information and they were the only genes we were testing for quite some time, so a lot of people are referred to these two genes as the breast cancer gene. However, there are a number of other genes related to hereditary breast cancer, including one syndrome called Lee F ormini syndrome, related to mutations in TP 53.
02:12:22.200 --> 02:12:24.252 which is which is more rare

02:12:24.252 --> 02:12:25.278 leaf armeni syndrome.

02:12:25.280 --> 02:12:28.368 You would expect to see cancers in childhood,

02:12:28.370 --> 02:12:31.374 including leukemias and childhood

02:12:31.374 --> 02:12:33.398 brain tumors, osteosarcomas,

02:12:33.398 --> 02:12:36.000 cancers of the bone, sarcomas,

02:12:36.000 --> 02:12:38.650 cancers of the soft tissue,

02:12:38.650 --> 02:12:40.510 and a risk of breast cancer,

02:12:40.510 --> 02:12:42.346 usually before the age of 35,

02:12:42.350 --> 02:12:44.108 so it is a very significant.

02:12:44.110 --> 02:12:45.709 Territory cancer syndrome.

02:12:45.709 --> 02:12:48.374 Cowden syndrome is another hereditary

02:12:48.374 --> 02:12:50.900 breast cancer syndrome caused by related

02:12:50.900 --> 02:12:53.540 to a high risk of breast cancer.

02:12:53.540 --> 02:12:55.468 Individuals also can develop

NOTE Confidence: 0.8435201725
rare polyps of the colon.
Uterine cancer, kidney cancer.
Additionally, they have on average a larger head size and may have specific findings on the skin called Trichomonas.
so another rare hereditary cancer breast cancer syndrome.
Another one is called ADHD, one hereditary diffuse gastric cancer syndrome where specifically individuals are at risk to develop lobular type breast cancer,
a type of breast cancer and a rare stomach cancer called diffuse gastric cancer,
which is a type of gastric cancer that’s very hard to screen for. So for individuals with mutations in these genes and CDH, one that actually is a recommendation for a prophylactic gastrectomy to remove the stomach to be prevented in against the high risk. Gastric cancer, which is oftentimes not able to be screened for. Another syndrome is called puts STK 11. You can also see your risk of breast cancer with this syndrome. However, what you may can also see is
there at risk to develop polyps of the small bowel and they may cause they may develop something called inception where the small bowel collapses on itself. They also have distinctive, oftentimes distinctive lift markings. I'm almost like I've been told, almost like someone ate it like a bunch of Oreos, kind of like. Are freckling on the lips or on the fingers? And risk of other cancers as well tends to another rare syndrome. There are also other genes that are more of a moderate risk and are actually more common than we're finding a lot more often now that we're doing
more comprehensive genetic testing.

I mentioned ATM, but there's another

one called palb 2 and check two,

so those are the mainly hereditary

breast cancer genes.

Hereditary colon cancer, the most common one that we talk

which is mainly characterized by increased

risk of colon cancer and endometrial cancer.

That we may see risks of other

such as stomach cancer, ovarian cancer,

pancreatic or bile duct cancer.

There's even other genes related
02:15:02.830 --> 02:15:04.370 to risk of ovarian cancer,
NOTE Confidence: 0.885240066923077
02:15:04.370 --> 02:15:06.180 specifically that are more in
NOTE Confidence: 0.885240066923077
02:15:06.180 --> 02:15:07.990 a moderate risk called rat.
NOTE Confidence: 0.885240066923077
02:15:07.990 --> 02:15:10.550 51 CD rate, 51 D and brip one,
NOTE Confidence: 0.885240066923077
02:15:10.550 --> 02:15:14.006 and there’s a lot more so testing nowadays.
NOTE Confidence: 0.885240066923077
02:15:14.010 --> 02:15:15.670 How we lead with testing,
NOTE Confidence: 0.885240066923077
02:15:15.670 --> 02:15:17.278 at least in our program and
NOTE Confidence: 0.885240066923077
02:15:17.278 --> 02:15:18.350 other programs as well,
NOTE Confidence: 0.885240066923077
02:15:18.350 --> 02:15:21.694 is we tend to do more comprehensive testing
NOTE Confidence: 0.885240066923077
02:15:21.694 --> 02:15:24.148 because there are a lot more genes.
NOTE Confidence: 0.885240066923077
02:15:24.150 --> 02:15:26.654 Out there that we know of and their
NOTE Confidence: 0.885240066923077
NOTE Confidence: 0.885240066923077
02:15:29.480 --> 02:15:32.301 So doing bigger testing now is is
NOTE Confidence: 0.885240066923077
02:15:32.301 --> 02:15:34.602 a benefit because our technology
NOTE Confidence: 0.885240066923077
02:15:34.602 --> 02:15:36.120 has gone cheaper,
NOTE Confidence: 0.885240066923077
02:15:36.120 --> 02:15:38.248 faster and better and we can rule
02:15:38.248 --> 02:15:39.802 out multiple previous positions at

02:15:39.802 --> 02:15:41.853 the same exact time while about 10

02:15:41.853 --> 02:15:43.756 years ago we were limited to just

02:15:43.756 --> 02:15:46.078 testing for BRC one and BRC 2.

02:15:46.078 --> 02:15:49.060 So my patient did have a comprehensive

02:15:49.149 --> 02:15:52.560 panel testing and she had a BRC 1

02:15:52.560 --> 02:15:54.318 mutation which was actually it was

02:15:54.318 --> 02:15:55.816 was a little bit surprising because

02:15:55.816 --> 02:15:56.984 as we talked about,

02:15:56.990 --> 02:15:59.294 the only significant risk factor in

02:15:59.294 --> 02:16:01.675 her family apart from her history

02:16:01.675 --> 02:16:04.009 of breast cancer was her paternal

02:16:04.009 --> 02:16:05.579 uncles metastatic prostate cancer.

02:16:05.579 --> 02:16:07.937 What’s interesting with BRC one is

02:16:07.937 --> 02:16:10.005 there is a slightly increased risk

NOTE Confidence: 0.885240066923077
02:16:10.005 --> 02:16:12.017 for men to develop prostate cancer
NOTE Confidence: 0.885240066923077
02:16:12.017 --> 02:16:13.997 that tends to be more aggressive,
NOTE Confidence: 0.885240066923077
02:16:14.000 --> 02:16:16.338 so it’s based on the family history.
NOTE Confidence: 0.885240066923077
02:16:16.340 --> 02:16:18.230 It was most likely that Shane hitter.
NOTE Confidence: 0.885240066923077
02:16:18.230 --> 02:16:21.470 This from her father’s side of the family,
NOTE Confidence: 0.885240066923077
02:16:21.470 --> 02:16:22.370 and you can see there.
NOTE Confidence: 0.885240066923077
02:16:22.370 --> 02:16:24.926 There’s also,
NOTE Confidence: 0.885240066923077
02:16:24.926 --> 02:16:26.527 by my my other colleagues,
NOTE Confidence: 0.8000724714375
02:16:26.530 --> 02:16:28.708 but there wasn’t a variant of
NOTE Confidence: 0.8000724714375
02:16:28.708 --> 02:16:30.615 uncertain significance which are very
NOTE Confidence: 0.8000724714375
02:16:30.615 --> 02:16:32.570 common nowadays in cancer genetics.
NOTE Confidence: 0.8000724714375
02:16:32.570 --> 02:16:34.688 When we do these bigger panels,
NOTE Confidence: 0.8000724714375
02:16:34.690 --> 02:16:36.300 and especially with bigger tests,
NOTE Confidence: 0.8000724714375
02:16:36.300 --> 02:16:38.586 nowadays we do find uncertain results
NOTE Confidence: 0.8000724714375
02:16:38.586 --> 02:16:41.266 about 20 to 30% of the time with
02:16:41.266 --> 02:16:43.969 these panels and most of the time in cancer genetics uncertain results,
NOTE Confidence: 0.8000724714375
02:16:43.969 --> 02:16:45.769 variants of uncertain significance,
NOTE Confidence: 0.8000724714375
02:16:45.770 --> 02:16:47.362 or the US.
NOTE Confidence: 0.8000724714375
02:16:47.362 --> 02:16:48.556 End up being reclassified to benign so we act on and the laboratory in the future usually takes even a few years.
NOTE Confidence: 0.8000724714375
NOTE Confidence: 0.8000724714375
02:16:50.681 --> 02:16:52.937 Update us to either upgrade the result to a positive, which is less likely,
NOTE Confidence: 0.8000724714375
02:16:52.937 --> 02:16:55.518 or downgraded to a negative result.
NOTE Confidence: 0.8000724714375
02:16:55.518 --> 02:16:57.597 And that’s and that’s mutation for my patient.
NOTE Confidence: 0.716247819777778
02:17:00.001 --> 02:17:01.800 And that’s that
NOTE Confidence: 0.716247819777778
02:17:01.800 --> 02:17:03.092 mutation for my patient.
NOTE Confidence: 0.716247819777778
02:17:03.092 --> 02:17:05.010 And that’s that
NOTE Confidence: 0.716247819777778
02:17:05.010 --> 02:17:07.140 mutation for my patient.
NOTE Confidence: 0.716247819777778
02:17:07.140 --> 02:17:09.255 And that’s and that’s that
NOTE Confidence: 0.716247819777778
02:17:09.255 --> 02:17:10.547 mutation for my patient.
NOTE Confidence: 0.716247819777778
02:17:10.547 --> 02:17:12.660 And that’s that
NOTE Confidence: 0.716247819777778
NOTE Confidence: 0.716247819777778
02:17:14.847 --> 02:17:17.034 And that’s and that’s that
NOTE Confidence: 0.716247819777778
NOTE Confidence: 0.716247819777778
02:17:19.255 --> 02:17:21.449 And that’s and that’s that
NOTE Confidence: 0.716247819777778
NOTE Confidence: 0.716247819777778
02:17:23.646 --> 02:17:25.871 And that’s and that’s that
NOTE Confidence: 0.716247819777778
And she was. She was very surprised in a way, and as a I was a little bit surprised as well and she was someone who really struggled a little bit with it with these results because she was concerned about her risk of a second breast cancer. And is that something that she should have a prophylactic mastectomy for to be preventative at her age? You know she’s not. She’s not very young, but she’s definitely not not older. She has. She has many years to live.
So it was kind of thinking about how should I go about dealing with this risk.

She still had her ovaries, so something that she had to think about in terms of removal of the ovaries, which is recommended for women who are here say one or beer, say 2 positive because our screening for ovarian cancer is not as effective as our screening for breast cancer and so the goal, as I mentioned of doing this type of testing is prevention and increased screening when possible.
I did with this patient, but also with other cases in general. Again, if we’re talking about my typical day that I discussed results with the patient and then based on the results, I would refer to any appropriate providers for management such as a gynecologic oncologist for my patient to discuss removal of the ovaries and discuss the limitations of ovarian cancer screening, referral to a high risk breast oncologist to talk about. Screening for breast cancer. The risks and benefits of doing a
bilateral mastectomy to remove both breasts in terms of prevention and in terms of other types of results.
I would refer to other specialists familiar with the syndrome who can also provide guidance in terms of cancer screening.
And of course, for this patient and other patients, I provide resources for themselves and family members, including a letter for family members they can share with their family, describing their results and
recommendations for testing.

Also, there’s a lot of resources online in terms of support groups, information and directing patients to those support groups and possibly even referral to a psychiatrist to talk to just for more psychosocial counseling and dealing with finding out someone has a mutation. Every patient takes results a little bit differently. For some it’s it can be very difficult to hear these new news, but they find it important for other people.
It's almost a relief to have an answer and a plan going forward. So I would say the patients demanded what the results are. It's information and especially for hereditary cancer, it can be very powerful for themselves and information they can provide to relatives. A lot of what a lot of what Jane counselors do is document in the medical record, which is very important, so other providers know what was discussed. They can go back to my notes,
02:20:10.660 --> 02:20:13.240 see the cancer risks I mentioned,
NOTE Confidence: 0.837548962
02:20:13.240 --> 02:20:14.623 see screening recommendations,
NOTE Confidence: 0.837548962
02:20:14.623 --> 02:20:16.467 and any patients questions
NOTE Confidence: 0.837548962
02:20:16.467 --> 02:20:18.740 that I answered at that time.
NOTE Confidence: 0.837548962
02:20:18.740 --> 02:20:19.228 Of course,
NOTE Confidence: 0.837548962
02:20:19.228 --> 02:20:20.448 I notified that the referring
NOTE Confidence: 0.837548962
02:20:20.448 --> 02:20:21.580 provider of the results,
NOTE Confidence: 0.837548962
02:20:21.580 --> 02:20:23.967 so making sure the whole teams are
NOTE Confidence: 0.837548962
02:20:23.967 --> 02:20:26.512 aware of what the results are so
NOTE Confidence: 0.837548962
02:20:26.512 --> 02:20:28.684 the patient is fully plugged in.
NOTE Confidence: 0.837548962
02:20:28.690 --> 02:20:29.840 The results were scanned in
NOTE Confidence: 0.837548962
02:20:29.840 --> 02:20:30.530 the medical record.
NOTE Confidence: 0.837548962
02:20:30.530 --> 02:20:32.497 Again,
NOTE Confidence: 0.837548962
02:20:32.497 --> 02:20:34.300 that these results are clear that
NOTE Confidence: 0.837548962
02:20:34.300 --> 02:20:35.504 they’re easily accessible that
they're part of the patient's medical record and that the medical team is aware of writing a summary letter which summarizes the results in detail because the report can sometimes be really not clear sometimes to patients who may not have the medical terminology, so the letter is really helpful in providing those cancer risks recommendations. What we talked about, what that means for relatives. In detail, so they can also share that with their own with other providers.
or with their relatives.

For our program, here we present cases at Case conference, which is great, but essentially our team meeting once a week. We present positive results or difficult cases and we get the team's input. If there's anything additional that they should be tested for should be screened for, and it's it's nice to have that group consensus. And throughout the rest of the day, I'm doing other clinical and program tasks. One of my other roles here at the program is actually triaging
incoming referrals to be scheduled. So I look at every incoming referral, determine the indication, and determine how they should be scheduled and following up with providers looking at past test results to see if any testing is indicated, which I enjoy because it's another way to use my clinical brain and also helps the admin team in terms of their own scheduling process. Overall, overall cancer genetic counseling. It’s something that I’m very passionate about. I think it’s it’s very important.
It is information that can really change lives change outcomes for patients. Provide information in terms of cancer risk. Some days can be difficult and you know disclosing some information. Talking with patients, but overall I find it very rewarding to provide that information. If you will have, I will answer questions now and also at the end. But if there are any questions you have specifically about being cancer, genetic counselor or my background. Any more of my day to day task,
02:22:36.450 --> 02:22:38.370 please email me.

02:22:43.730 --> 02:22:46.234 Thanks Amy, that was a really great talk.

02:22:46.240 --> 02:22:48.417 I’m just going to share my screen

02:22:48.417 --> 02:22:50.806 because I put I started the

02:22:50.806 --> 02:22:54.194 countdown until we come back for

02:22:54.194 --> 02:22:55.490 the second half of our session.

02:22:55.490 --> 02:23:00.370 A little earlier just to get us back

02:23:00.370 --> 02:23:02.170 on track. But if you don’t mind,

02:23:02.170 --> 02:23:05.369 Claire did you happen to see regarding

02:23:05.370 --> 02:23:07.230 insurance and genetic testing?

02:23:07.230 --> 02:23:09.370 Yes, so there was one question Amy

02:23:09.370 --> 02:23:11.024 about whether there are any barriers

02:23:11.024 --> 02:23:13.720 that the patient or the genetic

02:23:13.730 --> 02:23:15.656 counselor might experience when

02:23:15.656 --> 02:23:16.940
it comes to getting genetic testing covered through insurance. Great question. So insurance is probably the bane of every genetic counselor's existence. I will say from what I've heard from my other colleagues and other specialties, cancer genetic testing is more easily covered than other specialties, but that we can still run into barriers. So insurance has specific guidelines mainly based kind of off the red flags I talked about, meaning that if a patient does not have this family history or this personal history, they will not cover.
Testing most insurance companies align with national guidelines recommendations, which can make it very easy. However, some insurance companies make up their own guidelines, meaning that someone could technically meet national guidelines and recommendations, but they will not meet their insurance guidelines. So sometimes the specific insurance is a barrier. Another barrier is that even though I would say we’re past that, we’re past the era of testing for just BR A1 and BRC A2.
A lot of insurance companies.

Aren’t in that area yet. They’re kind of living in the past. They will only want to cover testing for beer, say one and B RC2, which to us based on a person’s family history. If they’re his family, history is concerning. For hereditary breast cancer specifically doing just beer, say one and two, testing is not sufficient, so sometimes it can be difficult when we want to do a more expanded panel. A panel meeting again, looking at multiple types of genes in one, one test.
Some insurance companies do not want to cover. A larger panel, even though to us we feel it’s clinically indicated so that can sometimes be a barrier in terms of getting that covered, and every insurance policy is a little bit different. What I will say for cancer genetic testing is that the cost has gone down and there are some laboratories out there that will actually do even a full panel full comprehensive cancer panel for an out of pocket cost of $250.
which I will. Which is not a small amount of money, however. Compared to how the cost was even seven years ago, it has gone down, which is good because there are some patients who testing for them is indicated, but their insurance will not cover either panel testing or will not cover testing at all because of certain requirements, but they are able to get the testing they need for an out of pocket cost that may be reasonable to them. Alright, it looks like that was
all of the questions for right now, but I'm sure Amy would be happy to answer more if there are some later.

And just about a minute until we start the second half of our day today. Alex, do you want me to pull my slides up? Yeah, why don’t we do that? Since I have 400 things open. That was a good sign. Yeah, I guess except. Huh? Hold on one second. Let me try this again. Sorry my daughter was helping me do this last night and. Something
02:26:53.920 --> 02:26:54.898 got screwed up.
NOTE Confidence: 0.8261397
02:26:56.210 --> 02:26:57.510 That’s alright, you have some time.
NOTE Confidence: 0.53137577
02:26:58.700 --> 02:26:59.350 Excellent.
NOTE Confidence: 0.24556684
02:27:04.700 --> 02:27:05.680 Sue
NOTE Confidence: 0.878536135
02:27:19.320 --> 02:27:21.128 OK, so hopefully you can
NOTE Confidence: 0.91927697
02:27:21.370 --> 02:27:23.990 see my slides, yeah? Looks good
NOTE Confidence: 0.655074604
02:27:24.030 --> 02:27:27.164 and hopefully only my slides and not
NOTE Confidence: 0.655074604
NOTE Confidence: 0.827497305
NOTE Confidence: 0.899826294285714
02:27:35.250 --> 02:27:36.755 And I’m good to go whenever you
NOTE Confidence: 0.9488798775
02:27:36.770 --> 02:27:37.558 want me to start.
NOTE Confidence: 0.724136723333333
NOTE Confidence: 0.78868455
02:27:47.370 --> 02:27:48.500 OK so hi
NOTE Confidence: 0.796865546
NOTE Confidence: 0.796865546
02:27:50.050 --> 02:27:52.360 I’m the director of the Master
NOTE Confidence: 0.89528445375
02:27:52.370 --> 02:27:53.935 of Science and Genetic Counseling

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Training program at Bay Path University which is in Western Massachusetts and I thought before I tell you the things that you probably want to know about what to expect in Graduate School. I give you a 32nd bio of who I am and how I got here. So I've been the program director at Bay Path for four years at which I was a clinical genetic counselor for 29 years. Nine and a half of which was in prenatal genetics with a little bit of Pediatrics thrown in.
and then for 20 years I worked in cancer centers seeing patients for cancer risk assessment and genetic testing. Much like you’ve just heard about over those years, our fields changed a lot and grown a lot, and I learned a lot along the way as I’m sure you will when you begin your journey to become a genetic counselor. I had always wanted to be a program director and four years ago I finally made it work for myself, so I thought we would talk about some of the basics of what to expect. In a graduate program and genetic counselor. As you may know,
There is an accreditation organization called the Accreditation Council for Genetic Counseling or a CGC. This organization was formed to accredit genetic counseling training programs and it sets standards for all programs that they need to follow in order to be accredited. At the beginning and then subsequently throughout the years, they need to be reaccredited so they need to continue to prove over and over that they’re meeting the requirements. Naturally, when you’re applying to programs, you’re going to want to be sure.
that the programs you’re looking at are accredited by a CGC, and it will say that on their website. So if it doesn’t say it on the website, you know to be a little bit cautious of that. I think it’s pretty unusual for you to find anything like that because you know we all know what genetic counseling training programs are out there, and if there’s something that’s not accredited, it’s not going to stick around. But due to these accreditation standards, all programs must provide a certain things and make sure that their students graduate.
02:29:50.378 --> 02:29:51.824 with specific competencies.
02:29:51.830 --> 02:29:54.210 The three legs of the stool, so to speak,
02:29:54.210 --> 02:29:56.050 for all programs include coursework,
02:29:56.050 --> 02:29:58.684 field work and research in the form
02:29:58.684 --> 02:30:00.800 of a thesis or Capstone project.
02:30:02.090 --> 02:30:04.240 So let’s start with the coursework. I realize
02:30:04.250 --> 02:30:05.786 that Bay path is not necessarily
02:30:05.786 --> 02:30:07.589 the same as every other program,
02:30:07.590 --> 02:30:09.610 and I can only speak to how we do things,
02:30:09.610 --> 02:30:11.682 but I’d be really surprised if they
02:30:11.682 --> 02:30:13.969 aren’t all very similar in this regard.
02:30:13.970 --> 02:30:15.836 The main difference between our program and
02:30:15.836 --> 02:30:18.238 most others is the fact that ours is online,
02:30:18.238 --> 02:30:20.110 which means our lectures are mostly
02:30:20.110 --> 02:30:22.480 prerecorded and not presented synchronously.
with everyone in class together.

Some of them are, but it’s not the norm,

and as I mentioned,

each program must be accredited by a CGC,

which means that.

There are rules we must all follow and core competencies.

We must make sure that our students achieve so every program will provide

for you medical genetics courses, reproductive and cancer genetics

Research courses that lead you through the process of writing a thesis or capstone project,

and of course,
clinical coursework that goes along with your field work. Rotations that will prepare you for and allow you to process and present cases to your supervisors and classmates. And become increasingly capable of seeing patients on your own. All of this is typically done within a framework of medical ethics, equity, diversity, inclusion, justice, and belonging in each course. I would expect that you would have a lecture accompanied by readings or videos and an assignment. Some
Assignments will involve role plays, standardized patients, or other ways to interact with their classmates, genetic counselors and other healthcare professionals to learn the skills you need to counsel appropriately and effectively. Other assignments may help you learn how to write a patient chart note or a summary letter, or research a specific disease to present to a patient or create educational materials for patients or healthcare professionals. Of course, you'll also be tested on your knowledge,
and many programs will use test questions in the style of the board certification exam to get you used to the format and the pacing needed to pass the exam. Second, there's your clinical work again. Every program works differently, although there are core fundamentals that you must have. In general, you'll start out observing a genetic counselor, or several of them. You will likely be asked not to say anything during the session since your skills are not developed yet,
but you will have the opportunity to learn not only the scientific material, but the nuances that the genetic counselors use in assessing the patients knowledge, interest, and receptivity to the information we find that. Not every patient is willing to hear that much and some are in a fragile emotional state if say they or their child or fetus was recently diagnosed with a serious condition. So determining our patients medical literacy and ability to handle the information we’re presenting may be the most important aspect of our jobs as GC’s and that’s one of the
fundamental things you’ll begin to learn in your observations, and they may ask you to draw a shadow pedigree while they’re doing their cases. To see how your pedigree compares with theirs and that helps in your learning too. And then of course, as time goes by, you take on increasingly greater responsibility in cases until the final semester of your training, when you’d likely perform the whole session by yourself. With supervision in terms of
content, every student must have what we call the big three rotations in prenatal Pediatrics and cancer. In a prenatal setting, you’ll see patients who are pregnant or would like to be and have concerns about their ability to have healthy children. This may be related to things like maternal age exposures to toxic substances, family histories of genetic conditions, abnormalities identified on ultrasound, or even a blood relationship between the patient and her partner. In a pediatric setting, you’ll see children who have features
that may be consistent with a genetic condition and need to be diagnosed. If that’s possible, or you may see children who are diagnosed previously but are coming back for periodic follow up, you may even see newborns in the neonatal ICU who are suspected to have a condition that needs to be diagnosed so that treatment or surgery can be initiated in a cancer setting which you just heard all about. You now know that you’ll see patients who have a personal and or family history of cancer who are hoping to
find out if there’s a hereditary.

Component that that information of course can both guide treatment options and provide risk assessment for family members. It could even inform the patient if there are increased risks for more cancers than they’ve already had. Some programs like ours will not have you doing any rotations in your first semester, so that you can get used to Graduate School and knock out more of the academics. In the beginning, we start in the second semester, while some don’t start until the summer.
But in any program, these three clinic types are required to collect the cases you need to be eligible to sit for the board certification exam. But there are many other types of settings that you may have opportunities to rotate through, like cardio, genetics clinics or ophthalmology, Neurology or psychiatry you may even wish to do a rotation in a laboratory or other industry type setting or public health department or newborn screening lab.
The Sky’s the limit, really.

If you have the time and if your program allows it, so again when you’re researching programs, think about what might be important and ask the questions that will help you figure out what it is that you want to do and where would be the best place for you to do it.

The third leg of the stool is your research project, and because I’ve always wondered this and thought you might too, I looked up the difference between a capstone project and a thesis.
A capstone project attempts to address an issue in the field by applying existing knowledge towards a real life problem, whereas a thesis seeks to create new knowledge through student research, trying to prove or argue a hypothesis rather than just investigate a topic. So each program has a research component that is required, but which type of component varies and of course is something else you may wish to research ahead of time. Most programs, I believe,
02:36:10.206 --> 02:36:12.710 are like ours with a research course first
NOTE Confidence: 0.9487990225
02:36:12.773 --> 02:36:15.017 that helps you understand the process.
NOTE Confidence: 0.9487990225
02:36:15.020 --> 02:36:16.572 The difference between qualitative
NOTE Confidence: 0.9487990225
02:36:16.572 --> 02:36:17.736 and quantitative research,
NOTE Confidence: 0.9487990225
02:36:17.740 --> 02:36:19.798 and how to write a proposal.
NOTE Confidence: 0.9487990225
02:36:19.800 --> 02:36:21.828 Then there are the capstone courses
NOTE Confidence: 0.9487990225
02:36:21.828 --> 02:36:24.172 during which you apply for Institutional
NOTE Confidence: 0.9487990225
02:36:24.172 --> 02:36:26.676 Review Board approval and do the
NOTE Confidence: 0.9487990225
02:36:26.676 --> 02:36:28.236 data collection and write up.
NOTE Confidence: 0.9487990225
02:36:28.240 --> 02:36:29.840 Most programs will encourage you,
NOTE Confidence: 0.9487990225
02:36:29.840 --> 02:36:31.744 though they may not require you to
NOTE Confidence: 0.9487990225
02:36:31.744 --> 02:36:33.388 present your research on campus
NOTE Confidence: 0.9487990225
02:36:33.390 --> 02:36:35.382 and or submit it for presentation
NOTE Confidence: 0.9487990225
02:36:35.382 --> 02:36:37.256 or publication to a professional
NOTE Confidence: 0.9487990225
02:36:37.256 --> 02:36:39.666 organization such as the National
NOTE Confidence: 0.9487990225
02:36:39.666 --> 02:36:41.594 Society of Genetic Counselors,
the American Society of Human Genetics,
American College of Medical Genetics,
American Society of Clinical Oncology,
or American College of Obstetrics and Gynecology. I could go on all day.
But it’s a really good opportunity to show the research that you’ve done and to kind of show the world what you’re what kinds of research your program allows. So those are the required components, but most programs, if not all of them, also have supplemental activities that you can potentially engage in, as there are so many other areas from which students can learn,
they may be required in some programs and voluntary or absent in others, so these may include things like book groups, journal clubs, guest speakers, webinars provided by outside organizations like commercial Genetics Labs. You could join some special interest groups by becoming a student member of an SGC. You can also request extra or different field work rotations to expand your knowledge and experience. Most program leadership members are very open to suggestions for these kinds of supplemental activities, so you want to be creative and
ask for what you want.
You will likely be pleasantly surprised.
I'll give you an example.
We've had a few students over the years who said, you know, I really want to do psychiatric genetic counseling, and that's a really difficult thing to find.
There are just not a lot of psychiatric genetics clinics in the country.
And that's probably largely because the genes for psychiatric illnesses haven't been discovered cloned.
You know, tests made available for them,
and so it’s all counseling.

It’s no genetic testing, but we’ve had students who’ve gone out to Vancouver for a few weeks to the 1st and to date one of very few psychiatric genetic counseling clinics in the world. So you know there are other kinds of things you can apply for lab rotations, and you know, I know in our program our students are required to do 30 day rotations in a semester, but if they really want to, let’s say, cardiac genetics,
02:38:42.122 --> 02:38:45.007 then we can shave it down a little bit
02:38:45.007 --> 02:38:47.549 so they can do 20 minute to 20 minutes.
02:38:47.550 --> 02:38:50.115 20 days in a Cancer Center and then the
02:38:50.115 --> 02:38:52.189 other 10 days in the cardiac clinic.
02:38:52.190 --> 02:38:53.186 Things like that.
02:38:53.186 --> 02:38:53.850 So again,
02:38:53.850 --> 02:38:55.890 if you want something, ask for it.
02:38:55.890 --> 02:38:57.210 They may very well be able
02:38:57.210 --> 02:38:58.828 to accommodate your request.
02:39:01.190 --> 02:39:02.260 And then the last thing I
02:39:02.270 --> 02:39:04.898 really wanted to touch on is the very deep
02:39:04.898 --> 02:39:06.573 emotional components of our profession
02:39:06.573 --> 02:39:08.790 and the rigors of training for it.
02:39:08.790 --> 02:39:09.840 And because of all of that,
02:39:09.840 --> 02:39:11.681 we feel it’s really important to care
02:39:11.681 --> 02:39:13.812 and
for yourself as it always is in life.

So as part of a training program, whether it’s actually embedded in the program or something that you do on your own, we feel that self care is very important. It’s easy to get kind of pulled under, sometimes by again the emotions of our sessions with patients. You’re stress regarding coursework or rotations. Interactions with classmates and colleagues so. Of course, maybe your thing.
02:39:41.260 --> 02:39:43.120 is to meditate or do yoga,
NOTE Confidence: 0.939172687
02:39:43.120 --> 02:39:45.416 cook or be with family and friends.
NOTE Confidence: 0.939172687
02:39:45.420 --> 02:39:47.256 Some programs build it in and
NOTE Confidence: 0.939172687
02:39:47.256 --> 02:39:48.699 some will expect you to care
NOTE Confidence: 0.939172687
NOTE Confidence: 0.939172687
02:39:49.800 --> 02:39:51.640 And in your own time.
NOTE Confidence: 0.939172687
02:39:51.640 --> 02:39:52.900 I know in our program,
NOTE Confidence: 0.939172687
02:39:52.900 --> 02:39:54.628 especially because it’s online and our
NOTE Confidence: 0.939172687
02:39:54.628 --> 02:39:56.220 students are not physically together,
NOTE Confidence: 0.939172687
02:39:56.220 --> 02:39:58.236 they have created movie nights at game
NOTE Confidence: 0.939172687
02:39:58.236 --> 02:40:00.359 nights and other kinds of social events.
NOTE Confidence: 0.939172687
02:40:00.360 --> 02:40:01.613 And when we do have our students
NOTE Confidence: 0.939172687
02:40:01.613 --> 02:40:03.797 together on campus once or twice a year,
NOTE Confidence: 0.939172687
02:40:03.800 --> 02:40:06.488 we always build in some time for
NOTE Confidence: 0.939172687
02:40:06.488 --> 02:40:07.640 socializing and decompression.
NOTE Confidence: 0.939172687

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But of course that involves the students being with each other and sometimes self care requires you to be alone or to be with people who are completely disconnected from the pieces of your lives that are creating the stress. What you do is not as important as the fact that you’re doing it, and we feel strongly that this should be part of everyone’s Graduate School Experience and in fact when we interview students or applicants, we ask them what kinds of self care they engage in. So something to think about. I’m not saying that other
02:40:36.910 --> 02:40:38.299 programs ask that question,
02:40:38.300 --> 02:40:39.784 but it’s probably a good thing to
02:40:39.784 --> 02:40:41.462 have in your pocket if they ask you
02:40:41.462 --> 02:40:43.059 how do you take care of yourself?
02:40:43.060 --> 02:40:46.120 You want to have some kind of answer and
02:40:46.120 --> 02:40:47.150 hopefully it’ll be actually something
02:40:47.150 --> 02:40:48.700 that you practice on a regular basis.
02:40:50.300 --> 02:40:51.260 So that’s what I wanted you
02:40:51.270 --> 02:40:52.320 to know about what to
02:40:52.320 --> 02:40:53.160 expect in Graduate School.
02:40:53.160 --> 02:40:54.616 I hope you will feel free to
02:40:54.616 --> 02:40:56.667 reach out to me at anytime with
02:40:56.667 --> 02:40:58.039 questions about genetic counseling,
02:40:58.040 --> 02:40:59.912 training programs in general,
02:40:59.912 --> 02:41:02.720 or Bay Pass program in particular.
02:41:02.720 --> 02:41:05.166
And I welcome your questions.

Janice thought was great and I love the self care bingo to feel that for me. You find any graphic online.

But I know that there were a couple of questions that have popped up in the first half of the session. I think I'll wait to ask them until Maria has given her talk and then have you 2 can address them in tandem.

Maria, let’s see here. Perfect.
Great. Alright. So hi everyone, thanks for coming and sticking with us through the afternoon. My name is Maria Geyer, I am the program director for the UConn genetic counseling program as well as some other graduate programs within the UConn family. So I wanted to chat with you a little bit today about applying to programs and how to strengthen your application. It can be quite an intimidating process, as some of you may have already experienced, so I kind of wanted to go through a few steps and how to start that and
some of the questions that you might want to ask yourself as you’re thinking about what programs to apply to. So step one of this process is to know yourself. You know genetic counseling has lots to offer in terms of a career, but you want to make sure that choice is right for you. So really take some time to reflect on what’s important to you, what you know, what makes you tick, and what do you think will be fulfilling in a career in general. And sometimes the best way to do
02:42:51.270 --> 02:42:53.390 this is to talk to people you know.

02:42:53.390 --> 02:42:55.806 Being here today is a great is

02:42:55.806 --> 02:42:58.280 a great start and I think you.

02:42:58.280 --> 02:43:00.098 Probably now met some folks that

02:43:00.098 --> 02:43:02.365 you could reach out to to question

02:43:02.365 --> 02:43:04.357 and ask and maybe have conversations

02:43:04.357 --> 02:43:05.389 about the career,

02:43:05.390 --> 02:43:07.052 but that’s really a good starting

02:43:07.052 --> 02:43:09.224 point is to kind of do your due

02:43:09.224 --> 02:43:11.446 diligence in terms of what you as a

02:43:11.446 --> 02:43:14.990 potential applicant are looking for.

02:43:14.990 --> 02:43:15.726 So Step 2,

02:43:15.726 --> 02:43:17.390 so once you kind of determine that and

02:43:17.439 --> 02:43:19.063 that this is the right step for you,

02:43:19.070 --> 02:43:20.430 is to know the programs.

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So as you’ve heard, there are many programs throughout the country and Canada, and although they’re very similar in the fact that they’re held to specific standards by our accreditation agency, you know many programs have their own strengths or niches. Some really stress psychosocial skills. For example, you know some are really big on research or technology, or you know it might be important to think about the types and amounts of clinical exposure that.
That each program has to offer you know what are the patient populations based on where you are or where you would be. So I encourage you to do your homework around that. There's a lot of different ways to choose programs and and we're going to talk about that, but just know that not all programs are created equal in terms of what they want to emphasize to their students. So when you're at the phase of wanting to select a program, what do you do? So?
different things that are important and things that go into the formula of what makes somebody want to apply to a particular program. So these are just some things on the left that could be questions or checkboxes for you to think about, or things that could be important to someone. So first is education delivery. You know there are different types of programs in terms of whether they’re face to face, whether they’re completely online, do they have a hybrid modality? So that might mean you go to
campus sometimes, but not all the time. And how does that play into class size? Like? Are you someone who likes to be part of a larger group, or do you like a smaller class size? I mean, I had three classmates when I went to Graduate School, so it’s very different than some of the class sizes that are out there today with 2224 upwards of 30 students per cohort. So is that? Is that a deal breaker for you, or is that something that makes you gravitate toward? Away from a program.
Cost is important, so tuition and fees obviously play a big role into which programs you might want to select. Are there scholarships that are available to help alleviate some of those costs? What is the cost of living within a program and where it’s located? You know this can be a huge deterrent and attraction based on whatever your particular financial situation is, but it’s important and tuition and fees potential. Scholarship availability are things that a program might have online so that you can kind of do your
your homework and your research by going to each of the websites of the programs that are of interest to you. You should be able to glean some really good information regarding cost straight from their website. Location is important, so similar to cost it tends to be a biggie in terms of why applicants choose particular programs. You know, maybe you're someone who you like a more urban setting or more suburban setting or very rural. There are programs that kind of check

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all of those boxes in different places you know. Do you need a program that’s close to family or friends for whether it be support systems or for living arrangements? Do you have to travel for field? Or, you know, is that kind of within the bounds of public transportation? Or do you need to have a car for campus? So these are all questions that are logistically important and necessary to consider when you’re thinking about a program.
location is important, but if a program Member asks, you know why you chose them as a program over others like please, please, please, don’t ever say us because my aunt lives here and I want to be close to family or I just always wanted to move to Boston or things like that. So while they’re important, they tend to be your personal reasons, so make sure that that you can make your choice accordingly, but also relay that appropriately.
Faculty are important, so you know who’s going to be your instructors and mentors and advisors are very important, and while you might not know who they specifically, before you get there, you know it can be important to research. What is the faculty to student ratio you know is the class so big that you don’t get a lot of engagement from your faculty members? Or is the class you know so small that you’re hearing from them all the time? It’s all this comes kind of down to personal preference and what you feel like. You need to be successful in your graduate education.
Affiliations are also important, so these are collaborators that your GC programs will have made relationships with, and these can include other universities or colleges. Are they part of a medical school? Are they part of the university based hospital? Are there international partnerships so there may be some places that you’re hoping to train or hoping to get some exposure to particular industry partners or laboratories. So it’s important to look at.
have and the different affiliations

and where you’ll where you rotate.

So that kind of brings me
to training opportunities.

You know, while while Janice
talked about having you know,
kind of the big three rotation of PEDs,
prenatal and cancer.

What does that look like within each program?

Are you going to get all of your education?

Kind of within one university based hospital that they have their tentacles
outside where you can experience different patient populations.

There are different hospitals and specific
clinics and there are specialties.

Do you get exposure to something like advocacy or community outreach or?

Maybe public health is really important to you, so there’s there could be a lot to unpack in the training opportunities.

And again, this is. This all comes down to what you want out of a program, so as much as they would be.

You know, interviewing you and asking you questions. These are the questions you want to ask of a program and kind of to
02:49:15.563 --> 02:49:17.125 yourself before you get there to make
NOTE Confidence: 0.88896096
02:49:17.125 --> 02:49:18.511 sure that that program would be a
NOTE Confidence: 0.88896096
02:49:18.511 --> 02:49:20.378 good fit should you match with them.
NOTE Confidence: 0.8376192
02:49:22.850 --> 02:49:24.020 So once you kind of have your
NOTE Confidence: 0.941018385555556
02:49:24.030 --> 02:49:28.494 list of programs that may fit your needs and.
NOTE Confidence: 0.941018385555556
NOTE Confidence: 0.941018385555556
NOTE Confidence: 0.941018385555556
02:49:31.724 --> 02:49:33.710 They can look like very elaborate
NOTE Confidence: 0.941018385555556
02:49:33.724 --> 02:49:38.034 spreadsheets, sometimes in terms of.
NOTE Confidence: 0.941018385555556
02:49:38.034 --> 02:49:39.548 Prerequisites, and then where
NOTE Confidence: 0.941018385555556
02:49:39.548 --> 02:49:40.628 they’re located and how much
NOTE Confidence: 0.941018385555556
02:49:40.628 --> 02:49:41.970 they cost and things like that.
NOTE Confidence: 0.941018385555556
02:49:41.970 --> 02:49:43.200 So whatever is important to you,
NOTE Confidence: 0.941018385555556
02:49:43.200 --> 02:49:44.904 make sure to put that on your list.
NOTE Confidence: 0.941018385555556
02:49:44.910 --> 02:49:46.468 But the ways that you can
NOTE Confidence: 0.941018385555556
02:49:46.468 --> 02:49:47.776 explore programs can vary,
so you can first review websites, and I think that’s a great place for everybody to start. I put the website here for the accreditation agency that lists the program directory, so you can kind of look by state for who is currently accredited and accepting students. You can visit campuses and I understand that during COVID obviously sometimes this makes it difficult, but you can always have a virtual tour or virtual visit emails and reaching out to program directors and other faculty. So if you can explore the area if it’s a
new town just to kind of get a feel for whether you like that type of a setting.

I completely understand that this could be very cost prohibitive,

but I think once you narrow your list down to to just a few.

Visiting and exploring a campus in itself is worthwhile.

Engaging students and alumni.

So again, some of the most important information you can glean from this process could be to speak to current students,

and many programs will have a student representative.

Generally a second year student who
you know has already gone through the program for the first year who has been able to develop an opinion on some of the pros and the cons, and what would they do differently and how happy they are. And generally these conversations could be very, very helpful, so I encourage you to reach out to students or.

Folks who have graduated from a program asked them their experience. So the last little bucket here is to ask questions, and you know these are some of
the questions that you might want to ask a program to help give you the information that might help you make a more informed decision. So for example, what do programs emphasize in terms of educational content and delivery? So this is kind of like what makes your program special, and then how do you disseminate that information to students? Is it all online? Is it a kind of hand holding? Is it throw you in the deep end? You know you could get a variety of answers here,
but I think all of it. Be very, very important and you know obviously you want to know what the strengths of the program are. What are their kind of accolades and accomplishments? And I think that’s important to be able to consider in making your decision of how what a program’s been able to do with their time. So step three is to know the process of the application and there’s quite a process. So while most programs are similar
in terms of their requirements, they’re not all the same. Some require one semester of organic chemistry, and some might require 2, and you know, so this is again where your spreadsheet might come in handy. If you have created one to kind of list out what the prereqs are for coursework. Timing for applications is important because the deadline for submission for applications is not the same throughout each program, so some may have a deadline of November 30th or December 15th or January 1st or February 1st. So we really can go throughout that whole winter time,
so make sure you are well aware of what your timelines are.

So applying to more than one program, I put that here feels ironic to say because I only applied to 1 program and I got into my program, but that is not the norm. I think some of the studies show that people who apply to four or more programs are statistically significantly more likely to gain entrance into a program than if you applied to one or two, which makes sense, but so I think in terms of how many programs to apply to there’s.
You know, there's things to consider in terms of cost and time and effort and letters of recommendation and all of that goes into it. But I think the typical applicant will apply to a few at least a few programs. And now with interviews being virtual, that does help with cost. It used to be that you'd have to fly everywhere to go to your interview and or find a hotel or stay with a student, make arrangements, take time off of work, things like that. So I will say that you know one of the only benefits to COVID is that it has
made interview the interview process
more accessible to more applicants.
So we chatted about cost a little
more bit so there are costs associated
simply with applying to programs.
You have the match fee,
so I don’t think we really
touched on the match yet,
but it is something that all genetic
counseling applicants will have
do is register with the match,
and I think the match is about $100.
Some programs do have waivers
for fees for applications.
I think that the match system has an Ms,
02:54:30.040 --> 02:54:32.684 has potential waivers for fees for the match as well, so.
02:54:32.684 --> 02:54:38.694 Be able to investigate where those incentives or alleviations might be.
02:54:42.210 --> 02:54:46.209 interview expenses can be costly if you have to travel there, so it’s a process, and do have empathy for that.
02:54:47.810 --> 02:55:00.150 It has lots and lots of words on it about application requirements.
02:55:00.208 --> 02:55:01.480 The important thing to know is that this is not an exhaustive list,
so there is lots and lots and lots to do when it comes to an actual application and things to consider. So do you have all of your prereqs and order by the time that you apply, know that for some programs you have to have all of your prereqs on a transcript prior to submitting your application or it has to be prior to the interview process. So there are different timelines. One caveat to note is AP courses, and in general if you took, you know a psychology and high school
and got credit for it and didn’t

then it generally won’t fulfill the

So I get this question from students a lot.

And again,

every program is different in terms

so this is all general information.

So typically AP courses won’t be accepted.

Fulfilled prereqs for GC school.

The GRE is becoming a little bit

less and less required from programs,

So what I’ve seen in the past few years

is a shift where in the past every
single program requires the GRE’s to, you know, just a couple were outliers where they didn’t require the GRE. So if you were going to apply to multiple programs, you kind of had to take the GRE because chances are you know at least one or two of your your programs would require that’s not becoming the case, so I definitely encourage you to look at that piece of the application process. The requirement within the programs does vary quite widely in the
terms of the numbers of programs that are no longer requiring the GRE. Language requirements are are still fairly standard, so English is your second language. There are going to be requirements based on school in terms of what they will accept for, you know, sample total scores or or writing assignment things like that. They may differ from university to university, but they generally will have some type of language requirement. You’re going to have to submit transcripts.
I’m going to have to submit your GPA.

I have some shameless plugs in here of the clinical genetics and genomics certificate that I run at UConn, as well as the clinical communication and counseling certificate that I run at UConn as well. These are a set of of courses which can gain you a graduate certificate in these areas, which a lot of students join and apply to these programs in order to boost their GPA so.

I tend to get the question of like what is the golden number for GPA
and what do I have to do and this

varies widely for programs as

well some will come out and say

what they will and will not accept for GPA.

Some are a little more ambiguous

and some programs are moving to a

more holistic space of not relying

as much on GPA as they are trying to

look at the applicant as a whole.

So, but for people who are interested in,

you know taking more graduate level.

Courses which are related to

clinical genetics specifically or

the psychosocial piece which is

the communication certificate.

You know, these two certificate programs
are available through UConn and and many students go on to genetic counseling. Graduate schools from these programs. Letters of recommendation. Usually it’s about 3. You want to kind of have the trifecta of letters of recommendation so you know you want to have someone from academics who can speak to your academic prowess and your potential for success in a graduate program. You know if you’ve done a research in undergrad or as a position that you hold right now. Research is a good letter of
02:58:44.054 --> 02:58:46.010 recommendation from a PI.
NOTE Confidence: 0.82073437631579
02:58:46.010 --> 02:58:48.831 I have folks who have advocacy and
NOTE Confidence: 0.82073437631579
02:58:48.831 --> 02:58:51.394 outreach experience so kind of the
NOTE Confidence: 0.82073437631579
02:58:51.394 --> 02:58:53.519 counseling portion of the genetic
NOTE Confidence: 0.82073437631579
02:58:53.519 --> 02:58:55.626 counseling hat that’s a good another
NOTE Confidence: 0.82073437631579
02:58:55.626 --> 02:58:58.195 base for the triangle there to have you
NOTE Confidence: 0.82073437631579
02:58:58.195 --> 02:59:00.358 want to have basically a well rounded
NOTE Confidence: 0.82073437631579
02:59:00.360 --> 02:59:02.710 list of letters of recommendation.
NOTE Confidence: 0.82073437631579
02:59:02.710 --> 02:59:04.110 Do not ask family.
NOTE Confidence: 0.82073437631579
02:59:04.110 --> 02:59:05.510 Do not ask friends.
NOTE Confidence: 0.82073437631579
02:59:05.510 --> 02:59:08.302 Do not ask peers really to assess you
NOTE Confidence: 0.82073437631579
02:59:08.302 --> 02:59:11.427 and give you a letter of recommendation.
NOTE Confidence: 0.82073437631579
02:59:11.430 --> 02:59:13.030 It’s not as professional looking.
NOTE Confidence: 0.82073437631579
02:59:13.030 --> 02:59:14.486 It doesn’t carry as much weight and
NOTE Confidence: 0.82073437631579
02:59:14.486 --> 02:59:16.195 you want to make sure you really only
NOTE Confidence: 0.82073437631579
02:59:16.195 --> 02:59:17.714 get like those two or three letters
you want to make sure they are from folks who can really speak the language that graduate programs need to hear and say the things that they need to say.

A personal statement.

So I get a lot of questions about personal statements and a lot of students who are looking for assistance with this. You know this can be your time to shine. This can be your opportunity to be able to say what it is that makes you different from other applicants.

Don’t challenge yourself to write
02:59:46.560 --> 02:59:47.456 it in a weekend.
NOTE Confidence: 0.82073437631579
02:59:47.460 --> 02:59:49.145 You should be going through
NOTE Confidence: 0.82073437631579
02:59:49.145 --> 02:59:50.493 multiple drafts of this.
NOTE Confidence: 0.82073437631579
02:59:50.500 --> 02:59:52.530 You should be going to a writing
NOTE Confidence: 0.82073437631579
02:59:52.530 --> 02:59:55.277 center if you have access to one at
NOTE Confidence: 0.82073437631579
02:59:55.277 --> 02:59:57.954 your current university, if not there.
NOTE Confidence: 0.82073437631579
02:59:57.954 --> 03:00:00.000 Are actually places online that
NOTE Confidence: 0.82073437631579
03:00:00.000 --> 03:00:02.310 you can send your statement to to
NOTE Confidence: 0.82073437631579
03:00:02.376 --> 03:00:04.314 get edited to have questions asked
NOTE Confidence: 0.82073437631579
03:00:04.314 --> 03:00:06.250 to help you with rewrites,
NOTE Confidence: 0.82073437631579
03:00:06.250 --> 03:00:08.440 so I encourage you to really,
NOTE Confidence: 0.82073437631579
03:00:08.440 --> 03:00:10.305 really really take the personal
NOTE Confidence: 0.82073437631579
03:00:10.305 --> 03:00:11.797 statement piece pretty seriously
NOTE Confidence: 0.82073437631579
03:00:11.797 --> 03:00:13.521 because that can be what sets
NOTE Confidence: 0.82073437631579
03:00:13.521 --> 03:00:15.190 you apart from somebody else’s
NOTE Confidence: 0.82073437631579
03:00:15.190 --> 03:00:17.710 volunteer experience is important.
They want to see that you’ve had experience putting on that psychosocial hat.

Crisis counseling is very common for applicants to have for bereavement counseling.

Support groups working with the disability community.

All types of these things that could be local to you or they could be virtual now in times of COVID. Basically what you want in your application to show is that you’ve done your due diligence in terms of investigating the profession and knowing that this is the right fit for you.
So everything that you do you do with a purpose and you know shadowing or interviewing genetic counselors is wonderful. It’s not required. Shadowing is becoming increasingly impossible to find in times of COVID, so just please don’t be discouraged if you don’t have a shadowing. Experience prior to application. We as programs understand that it’s very difficult to get that type of experience. There are other ways of kind seeking that information. You could call a genetic counselor and try to interview them or talk to them. There is a master genetic counselor.
series which is a set of videos that are free to watch through the National Society of Genetic Counselors and SGC. Which shows examples of different genetic counseling settings, it’s role play, you know, through with actors and actual genetic counselors so you can see what an actual session looks like. They have prenatal and cancer and PEDs I think, but or can’t. Yeah they have all three. But they’re about 1/2 an hour piece and they get digested afterwards. For for questions and things like that.
And it’s really a great. Another great way to kind of get some of that experience all right. So one of the last steps here you’re going to listen to my daughter. There’s a picture of my dad. He’s holding my son that’s Nicholas and my dad always said if the jobs worth doing, it’s worth doing right. So that means if you’re going to go through this process and it is a process, it’s big and it’s overwhelming at times that you want to put your whole everything into it, OK? So you’re going to do your best when it comes to that personal statement.
You’re going to make sure you check off the boxes for prereqs well before you have that application, you’re going to make sure that if you don’t have volunteer experience that you’re not doing it just a month or two right before your application process, you have to show them that you’re really invested in this career path, so no half assing everything. Do everything to your best of your ability and it will show in your application. Those are words from my dad. So lastly, I’ll leave you with some helpful information this relates to.
I think I saw a couple of questions about this, potentially taking a gap year. So for some students they feel like it’s looked upon negatively to have a gap year. I’m here to tell you that it is absolutely not looked down upon to have a gap year. You know, for students who take that gap year or longer, it took longer. You know that means you might actually be in a professional setting, so you’re gaining professionalism. You’re gaining experience in the field if it’s related. Hopefully it’s related. You know you’re saving money so that finances don’t become such a burden.
You know you’re doing what you need to do to prepare for grad school, so taking a gap year not a bad thing at all. If you go through this process and you don’t match, it is disheartening, obviously, but you are not alone. This is very common. There are many, many, many applicants for not a lot of spots. So if you’re not accepted, please please, please reach out to the programs that you wanted to match with and get feedback. You may think I didn’t get in because I didn’t have a great GPA,
but maybe it had nothing to do with it

and it was the fact that you didn’t have enough volunteer experience.

Or your professional or your personal statement just wasn’t up to par, you know?

So just please seek feedback so you can work and make the right strides to get in.

Contact programs to make sure you’re fulfilling their requirements, so if there’s ever a question when you’re going through the application process of like ooh, I don’t know if this is going to count or not.
Don’t just wing it because it might not count. Please feel free to contact programs. There are folks who can answer those types of questions very quickly, so you’d have to kick yourself later for being like, oh, I just didn’t know that. And again, familiarize yourself with the profession. So this is you doing your homework and doing your due diligence. You know, review the NSGC code of ethics, the position statements that they put out, the policy statements that are written,
be able to have an intelligent conversation about some of those things, should they come up in an interview, or when you’re talking to a genetic counselor while you’re interviewing them. Read the genetic counseling literature so lots of things come out about genetics right now, and it’s very important to kind of stay abreast of that and that actually shows that you’re very passionate and invested in the profession. Again, the master genetic counselor videos at nsgc.org are free, and they’re available, and they’re wonderful,
so I encourage you to do that. You can do this. It is hard. I know it’s hard, it’s a lot. There are a lot of pieces that go into this and it’s a long process but stay positive, keep working hard and you’ll make it happen eventually. So if you have questions, I’m available, that’s my email. Feel free to reach out. I’m always happy to chat with students and potential students, applicants, whatever,
03:05:35.820 --> 03:05:38.070 and answer any questions that you
NOTE Confidence: 0.81817406
03:05:38.070 --> 03:05:39.953 might have about the process or
NOTE Confidence: 0.81817406
03:05:39.953 --> 03:05:42.130 programs and and so on and so forth.
NOTE Confidence: 0.81817406
03:05:42.130 --> 03:05:43.678 So so thanks for your time.
NOTE Confidence: 0.81817406
03:05:43.680 --> 03:05:45.080 I hope I didn’t go too too long.
NOTE Confidence: 0.81817406
03:05:45.080 --> 03:05:46.574 I tried to speak very very
NOTE Confidence: 0.81817406
03:05:46.574 --> 03:05:47.570 fast and I’m happy
NOTE Confidence: 0.89725314375
03:05:47.625 --> 03:05:49.335 to answer questions at the end.
NOTE Confidence: 0.811920988
03:05:51.310 --> 03:05:53.950 Thanks Maria, that was really inspirational.
NOTE Confidence: 0.811920988
03:05:53.950 --> 03:05:55.672 I feel like I’m ready to
NOTE Confidence: 0.811920988
03:05:55.672 --> 03:05:58.652 apply again round two,
NOTE Confidence: 0.811920988
03:05:58.652 --> 03:06:01.058 but I feel like your presentation
NOTE Confidence: 0.811920988
03:06:01.058 --> 03:06:03.547 was almost like an FAQ in itself,
NOTE Confidence: 0.811920988
03:06:03.550 --> 03:06:05.590 so I’m going to just switch
NOTE Confidence: 0.811920988
03:06:05.590 --> 03:06:07.630 it over to Olivia and Kim.
NOTE Confidence: 0.811920988
03:06:07.630 --> 03:06:08.790 I appreciate if you could
03:06:08.790 --> 03:06:09.950 stay around till the end,
03:06:09.950 --> 03:06:12.950 but of course you did provide your email,
03:06:12.950 --> 03:06:16.889 so let’s get this show on the road.
03:06:16.890 --> 03:06:17.679 There we go.
03:06:29.220 --> 03:06:31.420 Let me just share my screen here.
03:06:38.100 --> 03:06:39.790 OK, can everyone see this?
03:06:41.590 --> 03:06:42.790 I think we’re good.
03:06:42.790 --> 03:06:46.370 OK, great OK so I know we’re
03:06:46.370 --> 03:06:49.658 short on time so I am Kim Freya.
03:06:49.658 --> 03:06:52.682 I am just recently graduated bapak
03:06:52.682 --> 03:06:54.986 the university this week so we’re
03:06:54.986 --> 03:06:57.237 kind of here to talk to you a little
03:06:57.237 --> 03:06:59.403 bit about the the Graduate School
03:06:59.403 --> 03:07:01.650 experience and kind of what we went
03:07:01.650 --> 03:07:03.460 through in the last couple of years.
And I'm Olivia, I just became a second year at Bay Path program. And like Kim said, we're just going to go over a little bit about our backgrounds. My education background I completed my Bachelors of Science.
At University of Oklahoma, I worked on my bachelors for over a decade because I was in the Air Force for 10 years. Active duty and I was taking classes. Kind of one or two at a time while I was also working full time in the military, I transitioned over into the reserve so that I could finish my bachelor’s degree and then also apply and go through Graduate School so that particular piece I would say is. I’m very different than most students that I’ve met and experienced.
Not many have gone through a military background as follows. As far as volunteer goes in my work as a Air Force member, I did a lot of crisis intervention counseling as a part of my job, but I also did suicide intervention training, so I was. I'm actually a trainer that does that program to teach others about suicide intervention. So that’s another. Different kind of volunteer experience, so I don’t really have a very typical background as a student, that’s you know,
applying to a genetic counseling program,
but.
So, like Kim was saying, is actually a great example.
My backgrounds are very different from Kim,
so I have a Bachelors of Science and a Masters of Science and infectious disease from the University of Saint Joseph’s kind of in between getting those degrees,
I worked as a cancer clinical research coordinator, so I was enrolling patients on to clinical trials.
I kind of focused on the Memorial Sloan Kettering impact study and the GRAIL.
Money which developed the new cancer
screening blood test called Gallery and

after that I did a medical scribe position

at an asthma allergy place when I was there.

I actually got accepted into PA school

and did go but I found out while I

was there it really wasn’t for me.

So that was like a big kind of

twist in my journey that I would say

that it wasn’t expecting.

So I actually took a gap year and.

Kind of wanted to just really look

over my experiences and kind of figure

out what I wanted as a career who I

was just kind of about my future.

So I kind of was looking back when

I was a researcher and I remembered
my interactions with the genetic counselor and I really didn’t know anything about genetic counseling. I never even heard about it, so I kind of did a deep dive into the profession and I just kind of fell in love with it.

So I decided to volunteer as a genetic counseling assistant at a maternal fetal Medicine and kind of really got to learn the role of the genetic counselor and really just could see myself doing this for the rest of my career. So I decided to kind of build up my resume so that I could apply and it was like I said,
my gap year.

So I kind of wanted to do something that was of interest of me.

Something that I've always wanted to do and that I had the time.

I decided to volunteer at a equine assisted therapy program.

And I'm a lifetime horseback rider,

so it was really just a passion of mine and I decided to really help those we were doing frontline workers at the
time during the pandemic and we also did veterans and children with autism or other disabilities so they would.

take us on trail rides and we would just teach them about horsemanship.
03:11:11.150 --> 03:11:13.030 So it was just something that I really.

03:11:13.030 --> 03:11:15.890 Was passionate about and UM,

03:11:15.890 --> 03:11:17.102 while during the pandemic.

03:11:17.102 --> 03:11:19.349 I also decided to do like a

03:11:19.349 --> 03:11:20.336 virtual teaching program,

03:11:20.336 --> 03:11:23.530 so I taught immigrants and refugees.

03:11:23.530 --> 03:11:25.310 English as a second language,

03:11:25.310 --> 03:11:26.870 which was brand new to me.

03:11:26.870 --> 03:11:28.262 It was very challenging,

03:11:28.262 --> 03:11:30.756 but I really loved it and kind

03:11:30.756 --> 03:11:32.742 of just what Kim was saying.

03:11:32.730 --> 03:11:34.320 These were things that I was

03:11:34.320 --> 03:11:35.834 passionate about and I like thought

03:11:35.834 --> 03:11:37.542 they would look good on my resume,

03:11:37.542 --> 03:11:40.519 but that really wasn’t why I was doing it.

321
So I would just kind of encourage you guys to do things that you really passionate about. Have like a really good interest in and that kind of makes you unique in your application. So that's just what I wanted to say about that. And that's pretty much my background.

So Kim and me wanted to just do another slide about you guys getting into the program. And during the program, so I'll let Kim take over.

Yeah so.
not for the faint of heart.

I don’t think it really matters.

Kind of what degree you end up going into with like getting into higher levels of education Graduate School PHD’s,

it’s going to be a lot of work.

It’s going to take a lot from you.

And so like Olivia said, doing things that help set you up for that kind of thing that you’re passionate about.

Really makes a difference going into those Graduate School interviews.

You know, saying I did these things because I really like them.
And I know a lot about myself and
I know that this career is going to be for me because I know that it’s going to be something that I am passionate about and why that is. Can really help. Kind of key into the you know to those that are interviewing you, those that are interviewing you, that you’ve really thought about it and you really kind of know a little bit more about. And then maybe when you started. I would also say that highlighting kind of what makes you unique is a really good aspect in.
and also going through your clinical rotations and those kinds of things. Being able to again know yourself, make help yourself stand out a little bit from what other people make them unique. You know, so you can really stand out in the minds of those that are interviewing you. Those that you’re working with. It it definitely is a asset and not something that’s a hindrance and then also Graduate School like I
03:13:38.108 --> 03:13:39.900 said it's a lot of work.
NOTE Confidence: 0.824636757272727
03:13:39.900 --> 03:13:42.136 It's a lot of time management
NOTE Confidence: 0.824636757272727
03:13:42.136 --> 03:13:43.931 skills going between didactic
NOTE Confidence: 0.824636757272727
03:13:43.931 --> 03:13:45.632 work and your clinical rotations,
NOTE Confidence: 0.824636757272727
03:13:45.632 --> 03:13:48.710 and being able to do your capstone or thesis
NOTE Confidence: 0.851004264
03:13:49.400 --> 03:13:50.860 all at the same time.
NOTE Confidence: 0.88671245625
03:13:51.200 --> 03:13:52.875 Sometimes it's a little disheartening
NOTE Confidence: 0.88671245625
03:13:52.875 --> 03:13:55.162 where you feel like you know,
NOTE Confidence: 0.88671245625
03:13:55.162 --> 03:13:56.999 why did I get into this in the 1st place?
NOTE Confidence: 0.88671245625
03:13:57.000 --> 03:13:57.900 Can I really do this?
NOTE Confidence: 0.88671245625
03:13:57.900 --> 03:14:01.440 Trying to have those.
NOTE Confidence: 0.88671245625
03:14:01.440 --> 03:14:03.520 The reminders about why you decided
NOTE Confidence: 0.88671245625
03:14:03.520 --> 03:14:05.110 to do this in the 1st place can
NOTE Confidence: 0.88671245625
03:14:05.110 --> 03:14:06.960 really help push you through some
NOTE Confidence: 0.88671245625
03:14:06.960 --> 03:14:08.664 of those really rough days where
you're really like tasked to the Max with case Prep and also getting an assignment done or an oral exam. By the way, your thesis professor is going to say hey, have you done this part for your thesis yet? You should probably really be thinking about that. You can have a lot of things going on at one time, so being cognizant of why you really want to do this can really bolster your energy and your motivation to keep going and to keep trying.
and and to

NOTE Confidence: 0.845946166666667

give yourself confidence that you

NOTE Confidence: 0.845946166666667

really can do it because you can.

NOTE Confidence: 0.845946166666667

Everybody can they.

NOTE Confidence: 0.845946166666667

You know if you get to the place

NOTE Confidence: 0.845946166666667

where you’ve applied and they’ve

NOTE Confidence: 0.845946166666667

accepted you into a program,

NOTE Confidence: 0.845946166666667

can see that you can do

NOTE Confidence: 0.795178388

this and they have faith

NOTE Confidence: 0.824639572

in you and that’s why

NOTE Confidence: 0.923178063333333

accept you into the programs

NOTE Confidence: 0.923178063333333

because they feel like you’re ready

NOTE Confidence: 0.923178063333333

and they know that you’re you

NOTE Confidence: 0.9252614675

can do it so. Keep that confidence up.

NOTE Confidence: 0.832892882

So for mine I just. I guess because

NOTE Confidence: 0.832892882

I’m a second year, I still remember
applying and being actually doing this program last year as an applicant. So I just want to say, don’t compare yourself to anyone else. I think a lot of times when we do these webinars and the students talk about their backgrounds, a lot of applicants tend to think, well, I don’t have this or I didn’t do this. Or should I do this and it’s? It’s just everyone has their own journey like me and Kim like are a good example like we are very different in our backgrounds, but we both made it into the program and she graduated.
I'm a second year so we're definitely doing it.

So just don't compare yourself. We all have our own strengths and weaknesses. Our own unique abilities and experiences.

So really, like I said, just you know, do things that you really enjoy and shows who you are as a person.

And I think that will make you a really strong applicant.

So just don’t compare yourself to anyone. I think that goes along with a really strong applicant. even in the program.

I think a lot of times we have imposter syndrome like did they really choose me and I think I
03:16:09.128 --> 03:16:10.820 still have that a little bit.
03:16:10.820 --> 03:16:12.962 So just remember that if you do get in you are there for a purpose
03:16:12.962 --> 03:16:15.112 and if you don’t it’s really a numbers game like there is limited spaces and limited programs.
03:16:19.105 --> 03:16:20.719 We’re all really qualified.
03:16:20.720 --> 03:16:23.072 Sometimes you just have to up a little bit and like I said before, like talk to your program director or any interviewers and see what you can do to improve your application. Because most of the time you are qualified, it’s just a numbers game and then my
second one and Janice already touched upon.

This is just self care and Kim, like Kim said, it can be a really stressful program, but it’s worth it in the end and I think to able to get through the program in one piece. Home is really self care. You gotta have to remember why you went into it in the self care of just doing anything that you enjoy. Whether that’s going for a walk. Reaching out to friends taking 20 minutes a day, maybe taking a couple hours off during your program.
03:17:11.750 --> 03:17:13.205 Just anything that will keep
you a little bit like yourself.

03:17:13.205 --> 03:17:15.250 Because like Janice was saying,
you can really get in the undertow
and kind of forget about that.

03:17:15.250 --> 03:17:16.540 So just self care is really
important during the program.

03:17:16.540 --> 03:17:17.870 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:17.870 --> 03:17:19.229 So just self care is really
important during the program.

03:17:19.230 --> 03:17:20.682 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:20.682 --> 03:17:21.650 And while we’re teaching right now,
they’re teaching us about compassion,
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03:17:21.650 --> 03:17:23.606 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:23.610 --> 03:17:24.990 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:24.990 --> 03:17:25.918 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:25.918 --> 03:17:27.474 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:27.474 --> 03:17:29.224 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:29.224 --> 03:17:30.629 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:30.630 --> 03:17:32.486 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

03:17:32.490 --> 03:17:34.714 And while we’re teaching right now,
they’re teaching us about compassion,
fatigue for genetic counselors,
so I think that’s really important.

And while we’re teaching right now,
We’re super happy.

To have you guys contact us.

That’s why we’re giving you her email.

Please use it if you have other questions.

It’s is a very short amount

do to explain

so please feel free to

reach out to us

definitely, and I’ll stop sharing now.

Thank you both and Kim,

congratulations on graduating

making it to the promised land

and hang in there. I love you you.

You’ll be there in no time.

But let me I might share my screen
03:18:08.930 --> 03:18:11.798 to go over. Where did that go?
03:18:11.798 --> 03:18:15.110 The emails for our great panelists?
03:18:15.110 --> 03:18:16.358 Thank you again.
03:18:16.358 --> 03:18:19.710 So much for taking the time out of
03:18:19.710 --> 03:18:22.833 our day out of your day to share your
03:18:26.480 --> 03:18:29.574 Let’s see now I can see that
03:18:29.580 --> 03:18:31.326 there were a couple of questions.
03:18:31.330 --> 03:18:33.465 I wonder if there was anyone on
03:18:33.465 --> 03:18:36.460 the panel who had last minute
03:18:36.460 --> 03:18:38.994 remarks to make to our group here
03:18:38.994 --> 03:18:41.030 before we closed for the day.
03:18:43.390 --> 03:18:45.654 As I kind of look through these questions.
03:18:58.440 --> 03:19:01.024 OK, Maya or Emily?
03:19:01.024 --> 03:19:05.910 Are you guys still on? Yeah.
OK, and there was a question from earlier about deciding whether you wanted to work. Continue working in the lab or exploring multiple specialties, or if there is anyone here who has kind of transitioned from 1 specialty to the other, and if you could just comment on that. Yeah, so for the time being, I really don’t know if this is something that I wanna stay in long term. I like it right now. But I also like the fact that I’ve been offered the opportunity to start seeing some patients.
03:19:45.900 --> 03:19:48.696 It will only be, I believe.

03:19:51.180 --> 03:19:55.689 Two days per month so it will be limited,

03:19:55.690 --> 03:19:59.254 but it will be nice to actually get back

03:19:59.254 --> 03:20:04.680 into the clinic and see people. But, uh.

03:20:04.680 --> 03:20:07.305 As far as long term, I don’t know.

03:20:07.305 --> 03:20:10.064 I do find it rewarding in its own

03:20:10.064 --> 03:20:12.248 sort of ways, but it is different.

03:20:18.590 --> 03:20:21.100 And I probably

03:20:21.150 --> 03:20:24.126 will stay in lab for my whole career,

03:20:24.130 --> 03:20:25.870 but one of my favorite things

03:20:25.870 --> 03:20:28.240 about GC is if that does change. I

03:20:28.250 --> 03:20:30.180 do have the opportunity to

03:20:30.270 --> 03:20:32.200 go to a different specialty.

03:20:35.520 --> 03:20:37.908 And anyone who might have taken

03:20:37.908 --> 03:20:40.100 some time between their undergrad

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between that and Graduate School, and how did those jobs or your experience during that time that prepare you? I can maybe provide an answer to that. That’s OK, so personally I had taken two years off in between undergrad and grad school. I did not know that I wanted to be a genetic counselor when I was in undergrad. I think I had heard of the profession, but not known enough about it to consider it seriously as a career. I personally majored in psychology and had a minor in biology, so in between, you know, going back to school,
there were like two other classes.

I think that I had to take and I did those at like a local Community College because of money of course.

And in the time between in those two years, I worked primarily in mental health, so I worked with individuals in a mental Health Center and also with some individuals with disabilities such as cerebral palsy. And I actually volunteered to help teach a kind of an art class for these individuals with disabilities at a day. Center kind of an art class you know modified to their levels, and I definitely think that working
with individuals with you know various mental health issues. You know, even though it wasn’t specifically related to genetics, was actually enormously helpful for me. I think just moving into those, kind of needing to put on the Counseling hacked so they say when working with patients. Excellent, there was a question for Emily and Maya. What do you find and? Or a difference between pursuing a masters in genetic counseling and not a PhD in let’s say,
genetics? Or lab work.

So I find that they’re pretty different.
Pursuing a PhD in genetics depending on if it’s molecular genetics, which I think is most likely what you’re asking about.

It’s a very different sort.

Of course, genetic counseling is much more focused on.

The interaction with people and looking at their different sort

If you’re going into molecular genetics, it’s much more about the DNA itself.

And I find that they they are very different.
Jose because. They really focus on different aspects of genetics. They do have a lot of crossover, but they're fairly different fields. Great.

OK, well as people are dropping off ready for the weekend, any last comments? Thank you all again for attending and speaking. I was really, really really informative and helpful.

OK great, well this recording will be available after that short survey, so I'll send that out in the coming weeks. I put in the contact information.
03:24:46.930 --> 03:24:48.900 for our great panelists today,

03:24:48.900 --> 03:24:52.876 but you can always reach me afterwards.

03:24:52.880 --> 03:24:54.637 And if you have a more specific

03:24:54.637 --> 03:24:55.940 question for a panelist,

03:24:55.940 --> 03:24:58.558 I encourage you to contact them directly.

03:24:58.560 --> 03:25:00.522 But thank you all so much

03:25:00.522 --> 03:25:02.999 again and have a great weekend.