

Kelly Mercier, PhD - DTRF 2022 Patient Meeting Webinar #1

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Jeanne Whiting: Another important way that we as patients can contribute to research that's so important is to share our stories through the Patient Registry. DTRF has had a Registry, which means you just logged in as a patient and you share your story the different phases you've gone through in your disease, in your treatment, in your response, in your lack of response, in your recurrence.

And this is called a Natural History Study just by answering surveys. So the Chair of our Registry or the PI of our Registry, which again you can get more information about on our website, is Dr. Kelly Mercier, and she has a recorded presentation here, and then we're gonna have questions after. But again, it is really life-affirming and invigorating as a patient turns you from being just a victim to being an advocate for a cure and for research by just sharing your story through this Patient Registry. We're gonna play a pre-recorded presentation from Dr. Mercier.

Kelly Mercier, PhD: Good morning. Thank you so much for that kind introduction. As was stated, my name is Kelly Mercier and I am the Principal Investigator for the DTRF Natural History Study and the DTRF Desmoid Tumor and Pregnancy Study. I'm here to talk to you today about how you can participate in desmoid tumor research today.

Now, there's all kinds of research that the Desmoid Tumor Research Foundation funds, so let's walk through some of these before we talk about the specific studies that I am leading. Now, there's basic, translational, and clinical research.

Basic research means that we are trying to uncover the basic knowledge of a disease.

So what's the underlying function of new molecules or cells strange or unknown phenomena with the disease. Translational research means that we want to move things from bench to bedside as soon as possible, and these techniques have been well investigated over the past 20 years to help us move things from a basic platform to the clinical platform.

Clinical research means that we are invested in human health. How can we improve the situation that you are in now, either with a diagnostic, a investigational therapy, or something that is primarily in-human at that moment? And an important aspect of human research is oversight. We always want to make sure that the participants of the studies, as you trust us with your body, with your data, that we take the utmost care in all of you.

And so anything that involves human subject research, involving a person, involving your data, has to be overseen by a committee. It's either called an investigational review board or an ethics committee. So you can see in the center here we have a flow chart. Does it involve living individuals? Do we need to get approval from you to ask you questions about your health, about your situation?

If both of those questions are answered, yes, it is human subject research. And for the studies that we run under the DTRF, they're considered human subject research, so the first thing that we ask you is for consent to collect your information. Another important consideration with your consent is that you can withdraw your consent at any time without any repercussion, so please keep that in mind as we talk about these projects.

So we have two ongoing studies with the DTRF for ways that you can contribute. The first is the DTRF Registry and Natural History Study. This is a collaboration that we have with the National Organization of Rare Disorders, otherwise known as NORD, and we launched the study all the way back in 2017. So we have been collecting data on desmoid tumor patients such as you since then. More recently we have started a collaboration with the Istituto Nazionale dei Tumori, a Italian organization for desmoid tumors, on desmoid tumor and pregnancy. I know that's a big concern for some of you, how we understand desmoid tumors in pregnancy, which led us to participate in this study.

The Natural History study has several components of this, and I want to walk you through this so you have an idea of the objectives of this study. So, as I've highlighted here, there are five. So we want to provide a convenient online platform for participants or their caregivers for legally authorized representatives of pediatric patients to self-report their information about their desmoid tumors.

It's also a platform for us to communicate within the desmoid tumor community about research opportunities, or clinical trials. We are able to further characterize and describe desmoid tumor experience through the information that you share with us in this platform, we can help assist the desmoid tumor patient community by understanding recommendations and standards of care.

Since this is a worldwide study, we have a pulse on what is happening throughout the world in terms of disease monitoring, diagnosis, and treatment. And then this is also a platform for us to collaborate with other researchers. So we have conversations with academics and pharmaceutical companies about de-identified data that is in the platform on how we can best explore additional studies that further investigate all of your situations that you could be encountering. But we do it in a de-identified manner, so none of your information is passed on to anyone else. But this is a way for us to further the information that we're gathering to something bigger.

Now, some of the data that we're collecting has to do with treatments that people are receiving in clinic today. We reported back in 2021 that the most common treatment initially is watch and wait or active surveillance. You will see that from the top of this graph where 69 patients reported that they did not receive an initial therapy.

And then we can see that other therapies that are listed include sorafenib, sulindac, anti hormonals, et cetera. So this is a way for us to understand how patients are being treated right now in clinic. While we are very interested in the therapies that participants are being given for their desmoid tumor, we are equally interested in additional therapies and are collecting that information as well.

You can see on the left hand side of this slide, 305 participants reported a sum of 1,118 therapies back in 2021. Now, there is a percentage of patients who reported that they do not receive any additional treatments. However, you will see that there is a percentage of 12% the received anti-inflammatories, anti-depressants, and opioids for pain.

So if you are receiving these medications, clearly you can see that you are not alone. Now, in addition to quality of life, treatment surveys, how people are diagnosed and monitored, we also have pregnancy questions in the Natural History Study, and these two graphs are meant to highlight that the participants who filled out the pregnancy survey match the tumor location in terms of the general population of the surveys.

And the reason why I point to this is because of the next slide, which will allow us to evaluate how the tumors respond to pregnancy. That being said, there is some concern by women who have desmoid tumors in the fertile age, how their tumors are going to respond during pregnancy. Now you can see from this set of bar graphs that the tumors seem to respond according to the participants similarly before or after pregnancy. And while this is a very small population, this helped drive us to participate in the desmoid tumor and pregnancy study.

Most recently, we have started recruiting for this study Desmoid Tumor and Pregnancy. You may have seen this on social media where we are recruiting for patients who are fertile diagnosed within 2000 and 2020. And there is a email address on here, pregnancystudy@dtrf.org if you wish to have more information. Now, this is a global endeavor. So there are many partners throughout the world led by the Fondazione IRCCS in Italy, and you can see that there are several collaborators throughout the world, Mount Sinai in Toronto, Dana-Farber, Institute Curie and a couple of other organizations. But we are one of the patient advocacy groups that is participating and recruiting participants into this study.

So who can participate in this study? Well, there's two different populations that we are investigating in this study. The first is trying to understand which women have desmoid tumors during fertile age to understand how many of you there are out there. The second thing we want to understand is how the desmoid tumors respond when you have a pregnancy. And so there's three different ways that we're going to investigate that. So, if you are diagnosed with a desmoid tumor during pregnancy if you have a tumor already known to you and being monitored, and then if you had one that was resected and you have minimal disease, if any, that's observed.

Much like the Natural History Study, these are all surveys that can be filled out online and through an interview. These are the kinds of things that we ask the participants to come with once we sit down for the interview. So, diagnosis date and tumor site, a pathology report, any radiology reports that you have, surgical type and treatment types, pregnancy information, and then the details about your labor and any complications.

Both of these studies highlight the kinds of research that we are doing as a foundation to better understand desmoid tumor in the population around the world. I hope that you take time to consider participating in these studies and please reach out to me if you have any questions. My contact information is below, and I really look forward to your questions during our session.

Jeanne Whiting: Well, hi, Dr. Mercier. You're here with us. I know you had some technical issues.

Kelly Mercier, PhD: I did, and I'm out in the world, so apologize if you can hear the wonderful music that's playing, but I'm really glad to be able to join you. Thank you.

Jeanne Whiting: We're so glad you're here. We have one really important question on the q and a for you.

Patient says, I joined the Patient Registry several years ago. I think I've only taken a repeat survey once or twice. Are we supposed to update it more frequently? If so, are you going to prompt us to do it with an email reminder or something else?

Kelly Mercier, PhD: Yeah, there should be email reminders going out for some of these surveys annually.

You are welcome to log in more frequently if you like to and update your results, but you should be receiving an email one year after you fill out some of the surveys, not all of 'em. So for instance, we are only gonna ask you about diagnosis once. What we want is our updates about your quality of life, and any outcomes or treatments that have changed over time. Additionally, we will be changing some parts of the Natural History Study in the next few months. We're in the process of making those changes and running them through our IRB to make sure that everything is kosher.

We've been working with the Patient Advocacy Committee with the DTRF to make sure that we're modifying the surveys to best capture what you all are interested in addition to researchers such as myself. So stay tuned for that. You'll be receiving an email from me if you are participant prompting you to go back and revisit the survey. So really appreciate your question. Thank you.

Jeanne Whiting: Okay, thank you. One other question. When will the DTRF release information related to the effectiveness of various treatments? Which is apparently something that we're studying, right?

Kelly Mercier, PhD: Right.

Jeanne Whiting: That is what most patients and caregivers are looking for as it may help them make decisions on future care.

Kelly Mercier, PhD: So we did release some information last year at ASCO, which is the premier oncology meeting so back in 2021 and you can find that poster on the DTRF website. There is also a new collaboration that we have that will be analyzing that data that we've collected all together. So that project is just kicking off and we should be able to have something in publication at another conference more thoroughly investigated sometime in 2023. I know y'all want this information as soon as possible. I'm trying as hard as I can. We're a lean staff, so thank you in advance for your patience.

Jeanne Whiting: I just also wanna emphasize that when you are, there are multiple surveys, right? So you can take your time, finish one survey, take a space, and then come back so you can get through them all.

Kelly Mercier, PhD: Yes.

Jeanne Whiting: But you get the benefit of once you complete one of these surveys, you get to see how it's been answered by other participants. So it informs you as you go along.

Kelly Mercier, PhD: Yes, please do not intend on finishing everything in one sitting. There are ways of saving as draft and you can go back to it. I highly recommend doing it. It can be pretty emotional, like going through all of these questions and really considering how you're feeling at a certain time. So please take it in bite size, go back to it. And if you don't go back to it, we will prompt you to go back to it. So thank you and yeah, thanks.

Jeanne Whiting: Yeah. Great. So, one question about the pregnancy study. I would love to participate in the pregnancy study. Yay. I have had two pregnancies since diagnosis in 2012. We do have a high level of interest in this of women of childbearing age, and we would love your participation. Do you wanna comment about that?

Kelly Mercier, PhD: Nope. I would go to our website and if you didn't grab the email address, pregnancystudy@DTRF.org just send an email and we will start the process of getting you consented to join the study.

Jeanne Whiting: Okay. Final question. Are you aware of any research into why there is an overwhelming correlation between pediatric desmoids in the head and neck and jaw, especially in under five?

Kelly Mercier, PhD: I personally do not, I will say that of the overall participation in the natural history study, pediatrics is by far the least. The last time I looked, there were less than 30 patients that were described in our pediatric setting. I would have to kick this question to Dr. Pollack if he has any information.

Dr. Seth Pollack: I will say that head and neck desmoids are more common in patients with APC gene, and that is a higher percentage of the pediatric patients that are getting desmoid tumors. So, that, that may be the phenomenon that you're seeing.

Jeanne Whiting: Kelly, thanks so much. You're such an important part of the DTRF team. Wherever you are in the world, we're so glad you could join us. Thanks so much.

Kelly Mercier, PhD: Absolutely. Have a good day, everyone.

Jeanne Whiting: Okay. That was great.