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TITLE: The Desmoid Tumor Research Foundation Registry and Natural History Study: 5 year status and update

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Abstract Purpose (95/200)

The Desmoid Tumor Research Foundation (DTRF) Registry and Natural History Study (NHS) are the first of its kind in that they are global and focused solely on the desmoid tumor (DT) patient population. The web-based model permits world-wide access, which is demonstrated by the diverse nationalities of the respondents. The overarching goal is that this study will be used to fill knowledge gaps in DT research and access to a patient population willing to donate their time and specimens for future studies. This abstract serves as a study update and guide to future data developments.

Abstract Background (193/200)

Desmoid tumors (also known as aggressive fibromatosis) are benign, locally invasive sarcoma or mesenchymal soft tissue tumors. While not colloquially considered cancer, they are most commonly treated with surgery, chemotherapy, and radiation, with varied success. Five to six out of a million people are diagnosed yearly with DT; while rare, there is a clear need to develop better interventions for DT, improve patient outcomes, and to better understand the DT patient experience from first symptoms through diagnosis, disease monitoring, and clinical treatment options.

The DTRF's mission is to aggressively fund research to accelerate the development of improved therapies, and ultimately find a cure for desmoid tumors. The DTRF launched the patient registry and natural history study (NHS) in 2017 and was one of the inaugural organizations in the I AM RARE project. Over the past 5 years, the study has enrolled participants to share their DT clinical journey. This abstract serves as an update to the NORD and entire rare disease community as to how the DTRF NHS has evolved over the first 5 years. The study is continually disseminating data, and study updates are on-going to improve the knowledge of this challenging rare disease.

Abstract Method(s) (199/200)

This registry and study has ethical approval for global data collection from the wcg IRB (Protocol Number: 120190346/ legacy IRB number: 2016-44).

The DTRF Patient Registry is designed to collect data for a prospective longitudinal web-based observational NHS. Participants with DTs will be followed throughout the course of their lives with either the participant or authorized respondents contributing data at varying intervals throughout the course of the study. Six of the surveys were developed from pre-existing, validated tools and included in a core data dictionary from NORD. These questionnaires are general to all rare diseases. The additional nine surveys were designed to capture information more specific to desmoid tumor patients.

All patients with a confirmed diagnosis of DTs are eligible for inclusion. The registry is open to recruiting patients of all ages who have ever had a diagnosis consistent with DTs. The study is continuing to evolve as the registry and NHS continue to collect information from the participants. Factors that contribute to this evolution include new scientific knowledge about DTs. As of July 23, 2021, 1188 DT patients or legally-authorized representatives (LARs) of DTs have consented to the DTRF NHS, and 729 participants have completed the Participant Profile.

Abstract Results (191/200)

Of those who have completed the Participant profile, 76.7% reported to be female, 22.9% reported to be male, and 86.7% reported to be white. While the participants are from 30 countries, 81.0% are from the United States. There are 27.4% who report abdominal DTs, whereas 13.6% have chest, 11.4% have lower extremity, and 9.5% have upper extremity DTs. At diagnosis 30.7% and 31.9% of the participants were 18-30 and 31-40 years of age, respectively. Of the 251 participants who responded, 59.0% disclosed having other medical conditions.

There is no standard of care of DTs, as demonstrated by the over 20 therapies, including clinical trials, that are reported. Additionally, 45.3% of participants (139/307) report taking non-steroidal anti-inflammatory medication for pain.

The study has been updated once to include changes to the Participant Profile (August 2018) and amendment 2 is currently being completed (July 2021) to address identified gaps.

De-identified data has been disseminated every year at the NORD Summit and the DTRF Research Workshop. Additionally, abstracts and posters have been presented at ASCO 2019, ASCO 2021, and submissions for CTOS 2021.

Abstract Conclusion (111/200)

In collaboration with the NORD, the DTRF NHS is built on the largest DT registry and has recruited more DT participants since launching in September 2017. It is serving to fill DT knowledge gaps and assist other researchers in their recruitment efforts for additional studies. As of July 23, 2021, 729 respondents had completed the Participant Profile. Abstracts and publications have been published and disseminated to share data in real time. On-going analysis is directing study amendments to fill identified gaps in knowledge, particularly in the DT-specific modules. Additionally, the study will be addressing the demographics biases

through targeted recruitment efforts throughout the United States with clinicians who treat sarcoma.