TITLE: An Update on Diagnostic Data from the Desmoid Tumor Research Foundation Natural History Study

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BACKGROUND AND OBJECTIVES: The Desmoid Tumor Research Foundation (DTRF) launched the patient registry and natural history study in 2017. Understanding the diagnostic process is of the upmost important for rare diseases. Here, we describe the longitudinal data, to include patient demographics and diagnostic data.

METHODS: The registry launched September 2017 and contains 15 surveys covering diagnostics, disease, treatment, care management, and quality of life. As of May 1 2020, 1076 patients or legally authorized representatives have consented to participate in the study, completing a total of 4705 surveys.

RESULTS: Registry participants are mostly white (87%, 553/645) and female (75%, 481/641). The average age at diagnosis is 33. Desmoid tumor location was reported for 342 respondents with tumors at time of data collection. Most prevalent tumor locations were joint /extremities (25%, 86/342), intra-abdominal (15.8%, 54/342), and chest wall (16%, 55/342). Multiple tumor locations were indicated for 10.8% (37/342) of patients. The most frequently reported methods of diagnosis included biopsy (77%, 235/305), CT (73%, 223/305), MRI (69.5%, 212/305), and Surgical Resection (38.4%, 117/305), where multiple responses were permitted. Participants reported that they were misdiagnosed 39% of the time (117/300). Desmoid tumors are caused by two genetic entities: mutations in either the CTNNB1 or APC genes, for which 33% of the respondents (109/338) reported that they were tested for these mutations.

CONCLUSION: Patients with desmoid tumors report many methods of diagnosis for their diverse tumor locations, high rates of misdiagnosis, and increased rates of genetic mutation testing. Data collection through the NHS is ongoing.