TITLE: A Retrospective Collection of Diagnostic Data from the Desmoid Tumor Research Foundation Natural History Study

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BACKGROUND AND OBJECTIVES: Desmoid tumors are a benign sarcoma diagnosed in 4-5 patients per million each year. The Desmoid Tumor Research Foundation (DTRF) launched the patient registry and natural history study (NHS) in 2017. This is a retrospective analysis of diagnostic data collected, tumor location, rates of misdiagnosis, how genetics testing is being incorporated into clinical practice, and additional clinical trial participation.

METHODS: The NHS launched September 2017 and contains 15 surveys covering diagnostics, disease, treatment, care management, and quality of life. Current reporting as of December 31, 2020, contains 619 participants or legally authorized representatives for which a subset have completed the surveys on desmoid tumor diagnoses.

RESULTS: Survey analysis documents that the most prevalent tumor locations were intra-abdominal 35.5% (220), joint / extremities 21.2% (131), and chest wall 14.7% (91). The majority of participants, 68.2%, reported that they had unifocal tumors (199/292), 19.5% reported that they had multifocal desmoid tumors (57). Biopsy procedures were the primary method of diagnosis according to 57.2% (167/292) of the participants, with needle biopsy comprising 19.8% (33/167). Biopsy as the primary method of diagnosis was most prevalent in tumors of the head and neck (18/26, 69.2%), chest wall (32/47, 68.1%) joint /extremities (50/90, 55.6%), and abdominal tumors (27/51, 52.9%). Additionally, imaging methods (CT, MRI) were the primary method of diagnosis in 22.6% (66/292) and surgical resection 14.4% (42/292).

Misdiagnosis is common for this tumor type, as 41.0% (119/290) participants reported an incorrect initial diagnosis. The reported incorrect diagnoses are described in the table. Genetic testing is not standard of care for desmoid tumors but is increasing in practice. A total of 78 participants (28%) of 282 participants report they had genetic testing (germline or somatic) of their tumor tissue. The majority of those participants, 65.4% (51/78), reported having Familial Adenomatous Polyposis (FAP). Of the participants that had genetic testing, 89.0% live in the United States. 10.5% of participants (37/353) have reported having participated in clinical trials. The majority of participants (317/368, 86.1%) are

willing to participate in other studies in the future, with 77.4% (285/368) willing to donate specimens for biomarker studies.

CONCLUSION: Participants 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 DTRF NHS is ongoing.

Misdiagnosis Prior to Desmoid Tumor	
Breast Cancer	2
Cancer (All Other)	16
Colon Cancer	1
Colon Polyps	3
Sarcoma (Not Specified)	19
Vascular tumor	2
Lymphoma	7
Scar (All)	11
Muscle Injury (All)	10
Hematoma (All)	6
Other	38