Characterizing the Patient Journey to Diagnosis of Desmoid Tumor: Desmoid Tumor Research Foundation Natural History Study

ABSTRACT #1574376

Shengfan Zhou,[1*] Amanda Lucas,[2] Lynne Hernandez,[3] Timothy Bell,[1] Sandra Goble,[4] Kelly Mercier[2,5]

[1]SpringWorks Therapeutics, Inc., Durham, NC, USA; [2]Desmoid Tumor Research Foundation, Jacksonville Beach, FL, USA; [3]Desmoid Tumor Research Foundation, Philadelphia, PA, USA; [4]Study conducted while at: SpringWorks Therapeutics, Inc., Durham, NC, USA; currently at: Medidata AI, a Dassault Systèmes company, Statistical Innovation Group, New York, NY, USA; [5]Duke University, Durham, NC, USA

*Presenting author. Email: sophia.zhou@springworkstx.com

Copies of this poster obtained through a Quick Response (QR) code are for personal use only and may not be reproduced without permission from CTOS or the author of this poster.

INTRODUCTION

- Desmoid tumors (DT; aggressive fibromatosis) are rare, locally invasive, infiltrative, and often aggressive soft-tissue tumors that do not metastasize and have an unpredictable disease course[1–3]
- Depending on tumor location, signs and symptoms of DT can include pain and impaired quality of life that can impact activities of daily living[3]
- Because of the rarity of DT and their histologic and clinical similarities to other diseases, DT are commonly misdiagnosed initially[1,4,5]

OBJECTIVE

- To characterize the self-reported patient journey from signs and symptoms to diagnosis of DT using data from the largest international DT registry and natural history study to date

METHODS

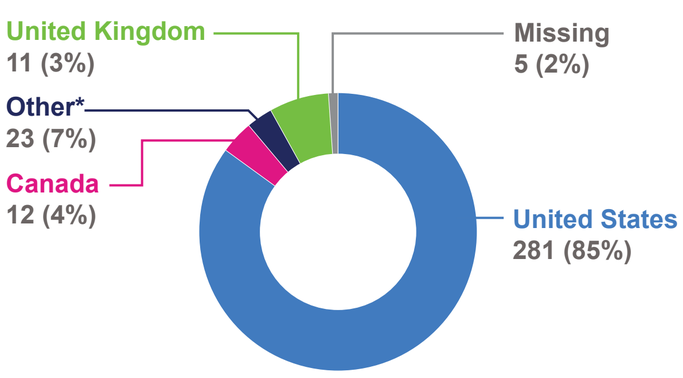
- The Desmoid Tumor Research Foundation (DTRF) Natural History Study includes surveys based on templates developed by the National Organization for Rare Disorders (NORD) for patient organizations that describe the course of rare diseases from initial symptoms through different clinical stages
- Additionally, investigators from the DTRF Natural History Study designed several surveys to ascertain information specific to DT and the typically chronic nature of this disease
- The present analysis is based on data from September 2017 to the cutoff date of October 2022
- The DT diagnosis was self-reported by the study participants and/or reported by their caregivers
- In this analysis, continuous variables were described using standard summary statistics and categorical variables using proportions; chi-square tests were used to examine the association between categorical variables

RESULTS

PARTICIPANT CHARACTERISTICS

- A total of 332 participants (93% adult [≥18 years old], 5% pediatric [<18 years old], 2% missing data) completed the DT diagnosis survey, with a mean age of 39 years (standard deviation [SD], 13.76) and median age of 38 years (interquartile range [IQR], 31–48 years; range, 2–78 years)
- Participants were mainly female (70%), White (86%), and located in North America (88%) **(Figure 1)**

Figure 1. Participant country (N=332)

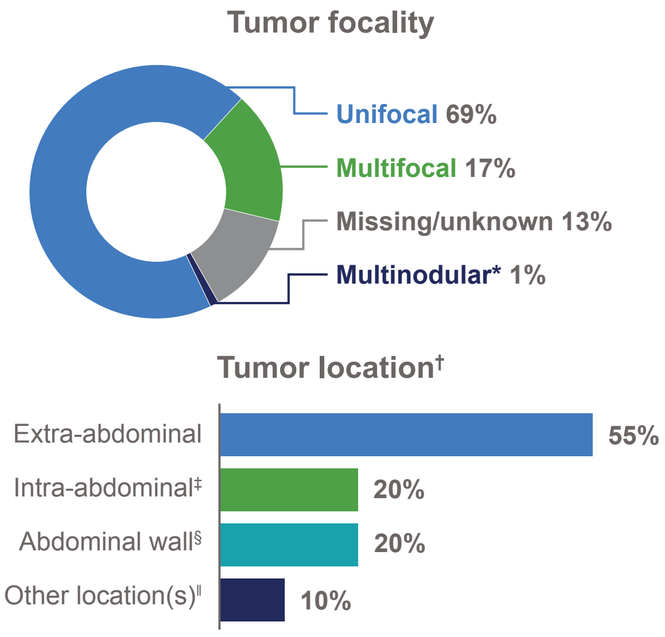


Percentages total more than 100% due to rounding.
*16 countries with <10 participants: Argentina (1), Australia (7), Belgium (1), Brazil (1), France (1), Germany (1), India (1), Ireland (2), Italy (1), Japan (1), Jersey (1), Jordan (1), Norway (1), Poland (1), Romania (1), and United Arab Emirates (1).

DIAGNOSIS

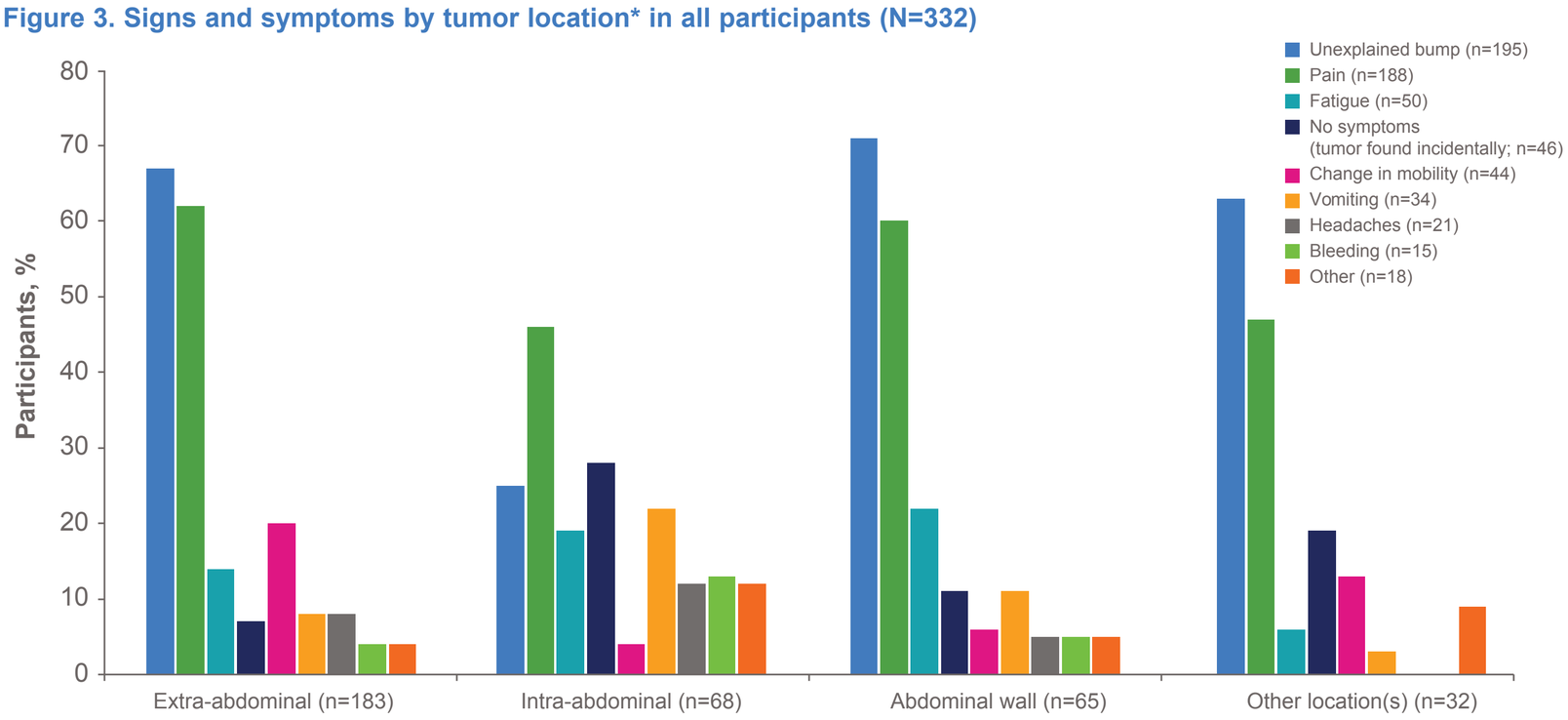
- Most participants (79%) were diagnosed between 2011 and 2022, with the first sign or symptom of DT experienced at a median age of 32 years (IQR, 21–41 years)
  - The median time from experiencing the first sign or symptom to diagnosis was 1 year (n=281)
  - Most participants (250/281; 89%) were diagnosed within 5 years of sign or symptom onset
- The majority of participants (69%) had unifocal disease at diagnosis **(Figure 2)**
  - Unifocal disease is one or multiple tumors in one part of the body
  - Multifocal disease is tumors in different locations, eg, one in the extremity and one in the abdomen
- The most common tumor location was extra-abdominal (55%), including head, neck, joint/extremity (eg, hip, knees, shoulders, arms, hands, feet, legs), and chest wall **(Figure 2)**
- The median maximum tumor length at diagnosis was 5.0 cm (IQR, 2.7–10 cm; range, 0.5–28 cm; n=157)
- The most common signs and symptoms at diagnosis were unexplained bump (59%), pain (57%), and fatigue (15%)
- **Figure 3** shows the full list of signs and symptoms by tumor location

Figure 2. Tumor characteristics at diagnosis (N=332)



*Multiple tumors in the same area of the body.
†Some participants reported more than one option; therefore, percentages do not total 100%.
‡Intra-abdominal: eg, deep in stomach/pelvis, wrapping up bowels, kidney, pelvis.
§Abdominal wall: eg, quite superficial on the stomach muscle.
‖Other location(s): eg, axilla and surrounding muscles and underarm tendon, back, brachial plexus, breast, left underarm, left breast, left-flank iliac crest, left side of spine at waist level and in left glute, lower back, lower lumbar spine, lower right back in paraspinal muscle.

Figure 3. Signs and symptoms by tumor location* in all participants (N=332)

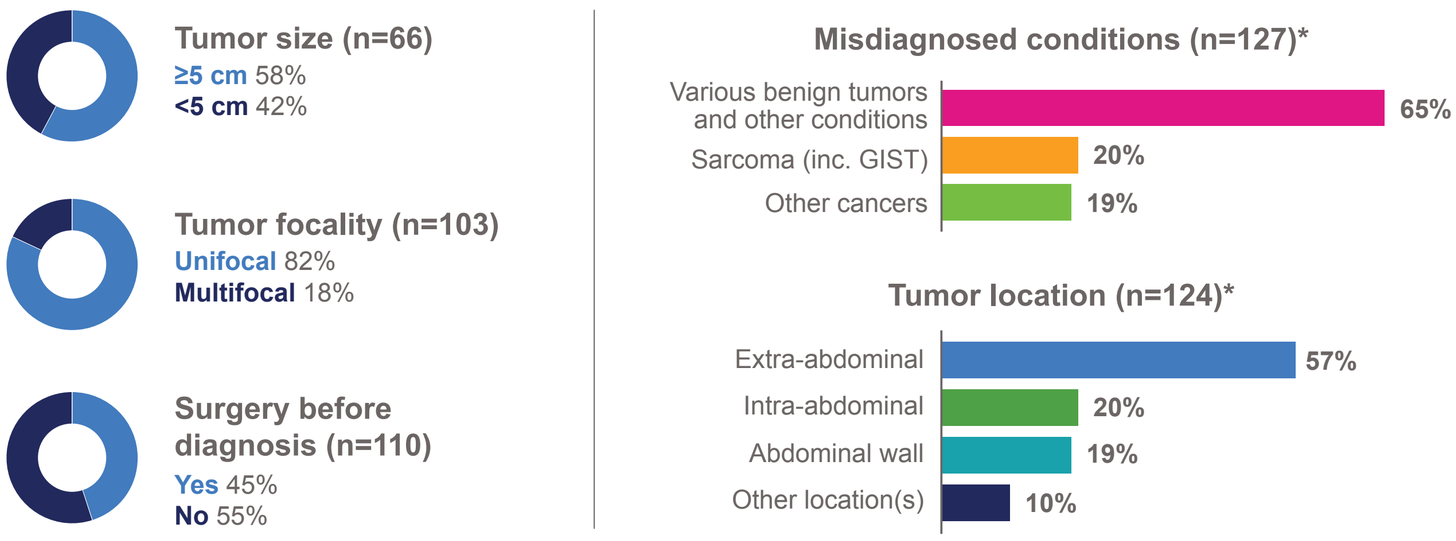


*Participants could have multiple symptoms and tumor locations at diagnosis; therefore, percentages do not total 100%.

MISDIAGNOSIS

- There were high rates of misdiagnosis among participants (127/329; 39%), with significantly higher rates observed in females (99/228; 43%) than males (26/94; 28%; P=0.008) and in participants with any other medical conditions (63/146; 43%) than those without any other medical conditions (28/100; 28%; P=0.016)
- Of the 127 participants who were misdiagnosed, 20% were diagnosed with sarcoma, including gastrointestinal stromal tumor (GIST); 19% were diagnosed with other cancers, including lymphoma, vascular tumor, and breast cancer; and 65% were diagnosed with other benign tumors and conditions, including muscle injury, lipoma, colon polyps, postoperative scars, early-onset arthritis, hernia, and Baker's cyst **(Figure 4)**
- Among participants who were misdiagnosed, 45% (49/110) had surgery before diagnosis, 58% (38/66) had a tumor ≥5 cm in diameter, 82% (84/103) had unifocal tumors at diagnosis, and 57% (71/124) had extra-abdominal tumors **(Figure 4)**

Figure 4. Clinical characteristics of participants who were misdiagnosed



Note: N values vary for these analyses due to missing data for some participants.
*Some participants reported more than one option; therefore, percentages do not total 100%.

CONCLUSION

- On their journey to a diagnosis of DT, patients may experience burdensome symptoms, such as pain and decreased physical functioning, that can adversely impact their quality of life
- Most participants in this study had unifocal disease at diagnosis, with an unexplained bump as the most common sign and pain as the most common symptom
- The median time to diagnosis for participants in this study was 1 year
- More than one-third of participants reported a prior misdiagnosis, and nearly half of these participants underwent surgery before DT diagnosis
  - Thus, a timely and correct diagnosis is critical to ensure that patients receive the most appropriate care and disease management
- Misdiagnosis was more likely to occur in women (than men) or in those with any existing medical condition (than those without)
- Raising DT disease awareness would help achieve an early and accurate diagnosis for people with DT, and thereby facilitate appropriate DT management according to established guidelines

ACKNOWLEDGMENTS: *This presentation was supported by SpringWorks Therapeutics, Inc. Writing and editing support was provided by Rebekka Harding-Smith and Daria Renshaw of IQVIA with funding from SpringWorks Therapeutics, Inc.*

REFERENCES: 1. Kasper B, et al. *Ann Oncol*. 2017;28:2399–2408. **2.** Riedel RF, Agulnik M. *Cancer*. 2022;128:3027–3040. **3.** Gounder MM, et al. *Cancer*. 2020;126:531–539. **4.** Penel N, et al. *Eur J Cancer*. 2016;58:90–96. **5.** Huss S, et al. *Histopathology*. 2013;62:294–304.