The Desmoid Tumor Research Foundation (DTRF) Natural History Study includes surveys based on data from September 2017 to the cutoff date of October 2022. Participants were mainly female (70%), White (86%), and of median age of 32 years (IQR, 21–41 years) at diagnosis. Among participants who were misdiagnosed, 45% (49/110) had surgery before diagnosis, 58% (38/66) had a tumor ≥5 cm in diameter, and 82% had unifocal disease. Due to the rarity of DT and their histologic and clinical similarities to other diseases, 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. MISDIAGNOSIS: There were high rates of misdiagnosis among participants (127/332; 38%), with significantly higher rates observed in females (95/228; 42%) than males (30/104; 29%; P=0.018) and in participants with 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 (cy)4–6. Among participants who were misdiagnosed, 45% (49/110) had surgery before diagnosis, 58% (38/66) had a tumor 25 cm in diameter, and 82% (94/113) had unifocal tumors at diagnosis, and 57% (71/124) had extra-abdominal tumors. 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. The majority of participants (250/281; 89%) were diagnosed within 5 years of sign or symptom onset. The median time from experiencing the first sign or symptom to diagnosis was 1 year (IQR 281). Most participants (250/281; 89%) were diagnosed within 5 years of sign or symptom onset. The majority of participants (89%) 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, 2–10 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) Tumor focality Tumor location1

Unifocal 69%
Multifocal 17%
Missing/unknown 13%
Multinodular* 1%

1Multiple tumors in the same area of the body.
*Some participants reported more than one option; therefore, percentages do not total 100%.

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

Radiation (n=1)
Surgery (n=1)
Irritation (n=1)
Neck (n=1)
Hair loss (n=1)
Numbness (n=1)
Other symptoms (n=1)

Other location(s) (n=52)

Fatigue (n=50)
Change in mobility (n=44)
Vomiting (n=23)
Headaches (n=21)
Bleeding (n=15)
No symptoms

Abdominal wall§

United States
281 (85%)

Canada
12 (4%)

United Kingdom
11 (3%)

Other*
23 (7%)

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

Several 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 25 cm in diameter, and 82% (94/113) had unifocal tumors at diagnosis, and 57% (71/124) had extra-abdominal tumors (Figure 4) Figure 4. Clinical characteristics of patients who were misdiagnosed Tumor size (n=66)

35 cm 58%
<5 cm 42%

Miskdiagnosed conditions (n=127)*

Various benign tumors and other conditions
Sarcoma (inc. GIST)
Other cancers

Tumor focality (n=103)

Unifocal 82%
Multifocal 18%

Tumor location (n=124)*

Extra-abdominal
20%
Intra-abdominal
20%
Abdominal wall
19%
Other location(s)
10%

No symptoms

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 Misdagnosis 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.