HSD30

Characterization Of Patients With Desmoid Tumors And Their Treatment Patterns Within An Academic Health System In The United States

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Background and objective

- Desmoid tumors (DT) are rare, locally aggressive tumors of soft tissues. While they do not metastasize, DT tend to infiltrate the surrounding structures and have a high rate of local recurrence.[1]
- DT can lead to substantial morbidity. In some cases, DT are associated with debilitating pain that is non-responsive to analgesics and result in functional limitations such as restricted limb movement.[2]
- DT management recommendations, such as those by the National Comprehensive Cancer Network and Desmoid Tumor Working Group, have evolved from a primarily first-line surgical approach to encouraging active surveillance and considering systemic treatment first if active interventions are needed.[3,4]
- Study objective: To assess the patient journey of DT, including patient characteristics, misdiagnosis and treatment patterns

Methods

- Study overview: Retrospective cohort study at the University of Utah Health system, which includes Huntsman Cancer Institute
- Inclusion criteria: Diagnosed with DT between 1/1/2011 and 7/31/2023 and had ≥2 DT-related clinical encounters
- Data collection: Data was extracted from electronic health records and administrative claims (**Table 1**). Patients were followed from diagnosis of DT to the last encounter in the University of Utah Health system.
- Line of therapy (LOT) was defined as a distinct treatment regimen given until completion or discontinuation due to progression, side effects or patient/clinician preference.
- Misdiagnoses were identified based on billing codes of conditions commonly misdiagnosed in place of DT in the 2 years prior to DT diagnosis.

Table 1. Data elements extracted from electronic health records

Study variables	Data source
Demographic characteristics	Patient history
Tumor characteristics	Pathology/imaging reports, clinical notes
Treatment patterns	Pharmacy records, clinical notes
DT-related symptoms	Clinical notes
Disease progression	Imaging reports, clinical notes
Misdiagnosis prior to DT diagnosis	Billing data
Healthcare resource utilization	Billing data

Results

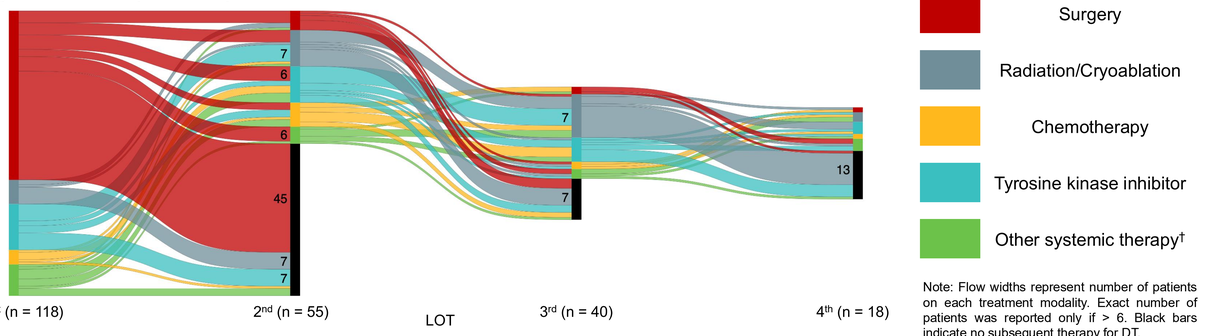
- A total of 148 patients met eligibility criteria with a median follow-up after diagnosis of 27 months. Tumor characteristics are shown in **Table 2.**
- The median age at diagnosis was 36 years (interquartile range: 27 to 46) and the majority were female (68.2%).
- Most patients (79.7%) received treatment, among whom 33.9% received ≥3 LOTs and the maximum LOT received was 10.
- Surgery was the most common treatment modality, primarily as the 1st LOT, while tyrosine kinase inhibitors were the most widely used systemic therapy in most LOTs (**Figure 1**).
- Use of surgery as 1st LOT decreased from 74.2% (46/62) in 2011–2017 to 44.6% (25/56) in 2018–2023, accompanied by an increase in the types of treatment modalities used (**Figure 2**).

Table 2. Clinical characteristics of patients with DT

Characteristics	N = 148
Primary DT size in mm, median (IQR)	60 (40-92)
Primary DT location, n (%): Abdominal wall	41 (27.7%)
Intra-abdominal	29 (19.6%)
Pelvic	7 (4.7%)
Chest wall	42 (28.4%)
Intrathoracic	9 (6.1%)
Head and neck	6 (4.1%)
Upper and lower limb	14 (9.5%)
Tumor focality, n (%): Single	128 (86.5%)
Multifocal	20 (13.5%)
Documented diagnosis of clinical FAP, n (%)	21 (14.2%)
Tested for *APC* mutation, n (%)	56 (37.8%)
Documented *APC* pathogenic mutation, n (%)	18 (12.1%)
Tested for *CTNNB1* mutation, n (%)	10 (6.8%)
Documented *CTNNB1* mutation, n (%)	7 (4.7%)

Abbreviations: DT, desmoid tumor; FAP, familial adenomatous polyposis; IQR, interquartile range

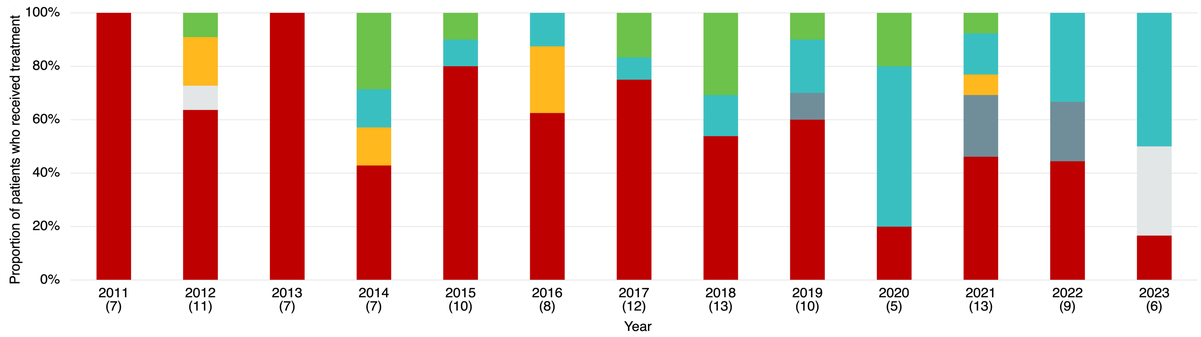
Figure 1. Treatment patterns of patients with DT from 1st to 4th LOT



Note: Flow widths represent number of patients on each treatment modality. Exact number of patients was reported only if > 6. Black bars indicate no subsequent therapy for DT.

†Other systemic therapy refers to tamoxifen with or without non-steroidal anti-inflammatory.

Figure 2. Treatment modalities received by patients with DT at 1st LOT across the study period



†Other systemic therapy refers to tamoxifen with or without non-steroidal anti-inflammatory.

- A total of 43 patients (29.1%) potentially had a misdiagnosis prior to a confirmed diagnosis of DT with a median misdiagnosis to DT diagnosis time of 0.8 years (interquartile range: 0.1 to 1.7).
- The most common misdiagnosis was non-DT benign tumor (20.3%); other conditions misdiagnosed as DT included low grade sarcoma (7.2%), nodular fasciitis (2.6%) and procedure-related scar (2.0%).
- In the year prior to DT diagnosis, a higher proportion of patients with misdiagnosis required outpatient visits, emergency room visits, and hospitalizations compared to those without misdiagnosis (**Table 3**).

Table 3. Healthcare resource utilization in the year prior to DT diagnosis

Healthcare resource utilization	Misdx (n = 43)	No misdx (n = 76)
Patients with outpatient visits, n (%)	28 (65.1%)	23 (30.3%)
Outpatient visits PPPY, median (IQR)	3 (0-14)	0 (0-4.3)
Patients with emergency room visits, n (%)	6 (14.0%)	2 (2.6%)
Patients with hospitalizations, n (%)	11 (25.6%)	2 (2.6%)
Length of hospitalization (days), median (IQR)	4 (3-13)	13.5 (11-16)

Abbreviations: IQR, interquartile range; misdx, misdiagnosis; PPPY, per patient per year

Conclusion

- The varied DT treatment approaches and multiple LOT support the use of better therapeutic agents and evidence-based guidelines.
- Patients receiving surgery for DT as 1st LOT decreased from 2011–2017 to 2018–2023, reflecting guideline updates in 2015 and 2017 suggesting that surgery is rarely an appropriate 1st LOT option for most patients; however, nearly 50% of patients still received surgery as 1st LOT after 2017 despite the associated morbidity.
- The occurrence of misdiagnosis and increased healthcare utilization due to a longer diagnostic journey and potentially worsening symptom burden suggest the need for timely and accurate DT diagnosis.

Acknowledgments

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Disclosures

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