



THE DESMOID TUMOR
RESEARCH FOUNDATION

Desmoid Tumors

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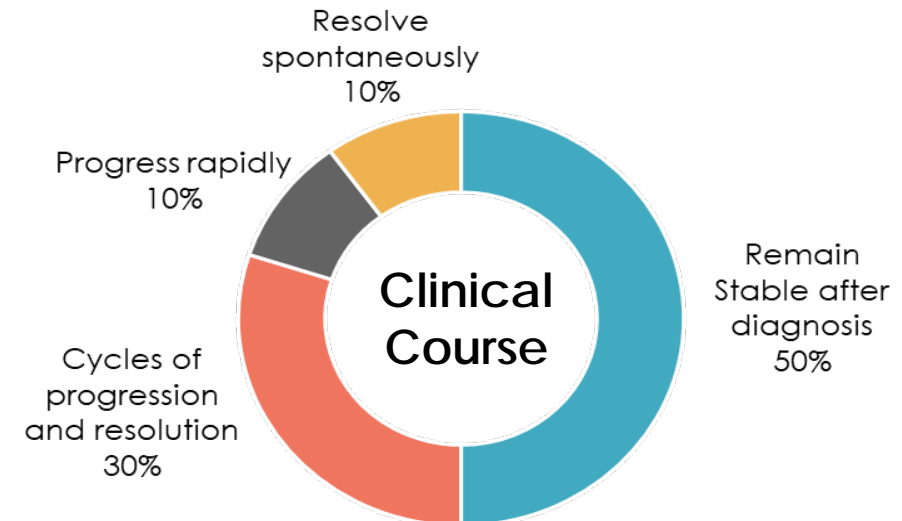
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Background



- Desmoid tumors are a deep connective and soft tissue neoplasm.
- Desmoid tumors are locally aggressive, infiltrative, and destructive.
- They are not classified as a malignant cancer because they do not metastasize.
- Desmoid tumors have a highly variable and unpredictable course.
- Desmoid tumors are also known as aggressive fibromatosis.



Incidence

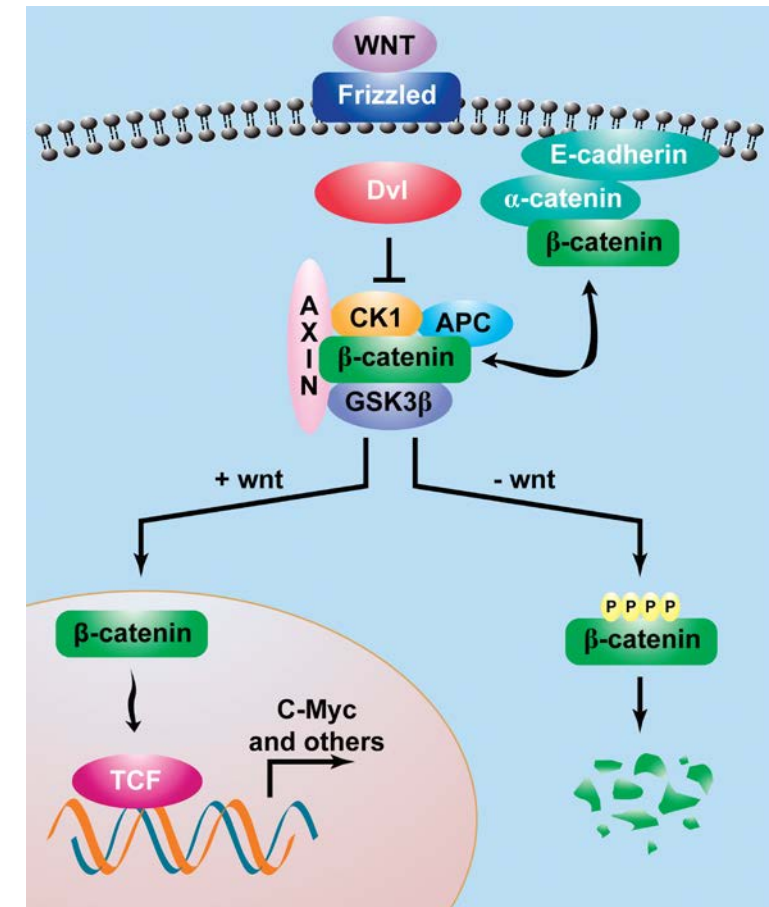


- Desmoid tumors are rare and incidence varies but is estimated at 2-4 per million per year
- In the US, it is estimated that 900 to 1,500 people are diagnosed with desmoid tumors each year.
- Desmoid tumors are about twice as common in women than in men.
- Desmoid tumors are most often diagnosed in people aged between 10 and 40 years, but occur in all age groups.
- The true burden of desmoid tumors is likely understated due to challenges in diagnosis and reporting.

Diagnosis and Cause



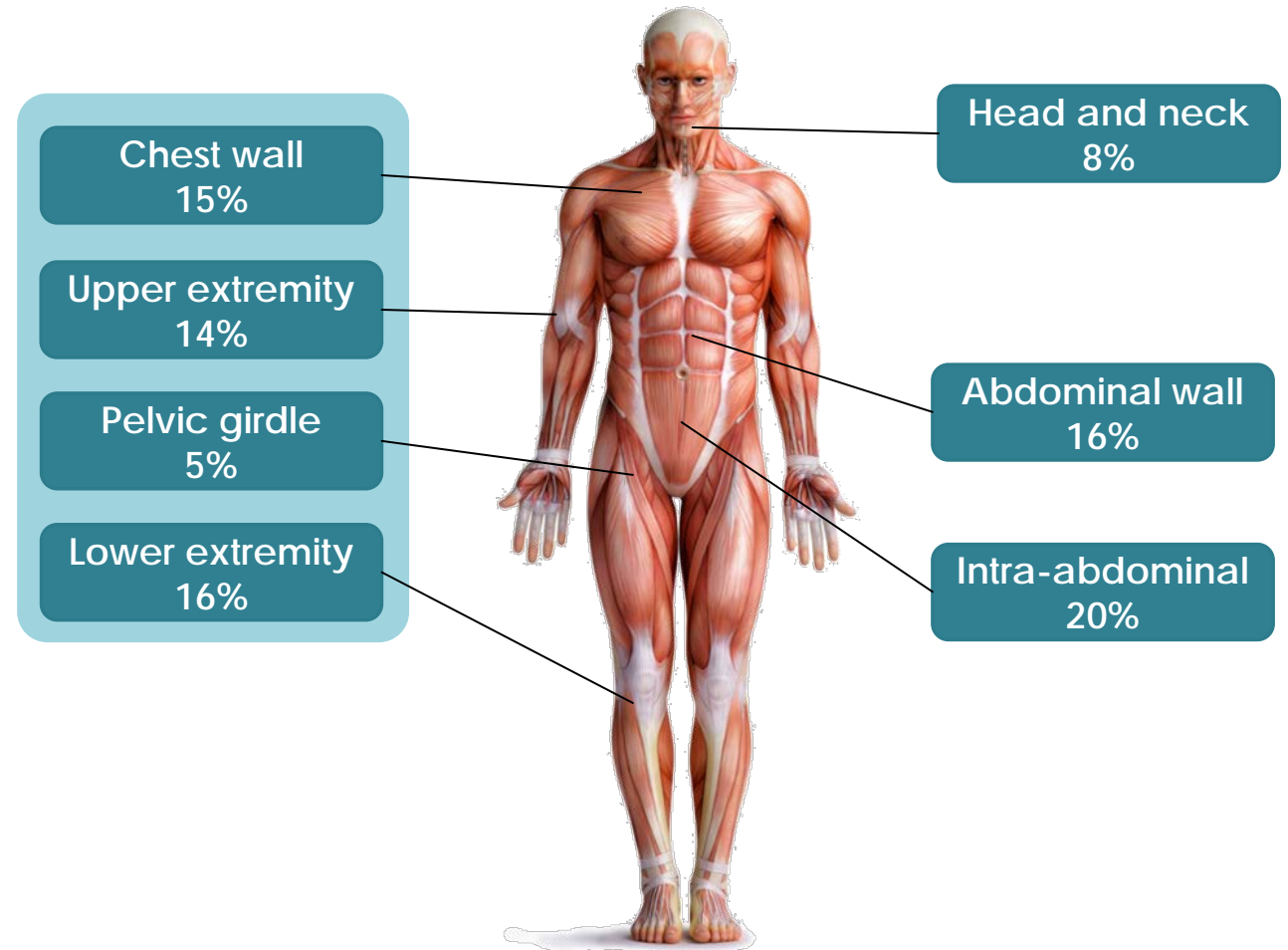
- Diagnosis of desmoid tumor is usually made by clinical exam and imaging and confirmed by biopsy
- The cause of desmoid tumors remains unknown but is thought to be related to wound healing
 - There is high rate of recurrence, especially after surgery
 - Desmoid tumors are associated with trauma
 - Up to 90% of desmoid tumors have a mutation in the β -catenin gene
 - APC Mutations (Familial Adenomatous Polyposis) is associated with increased likelihood of desmoid tumors



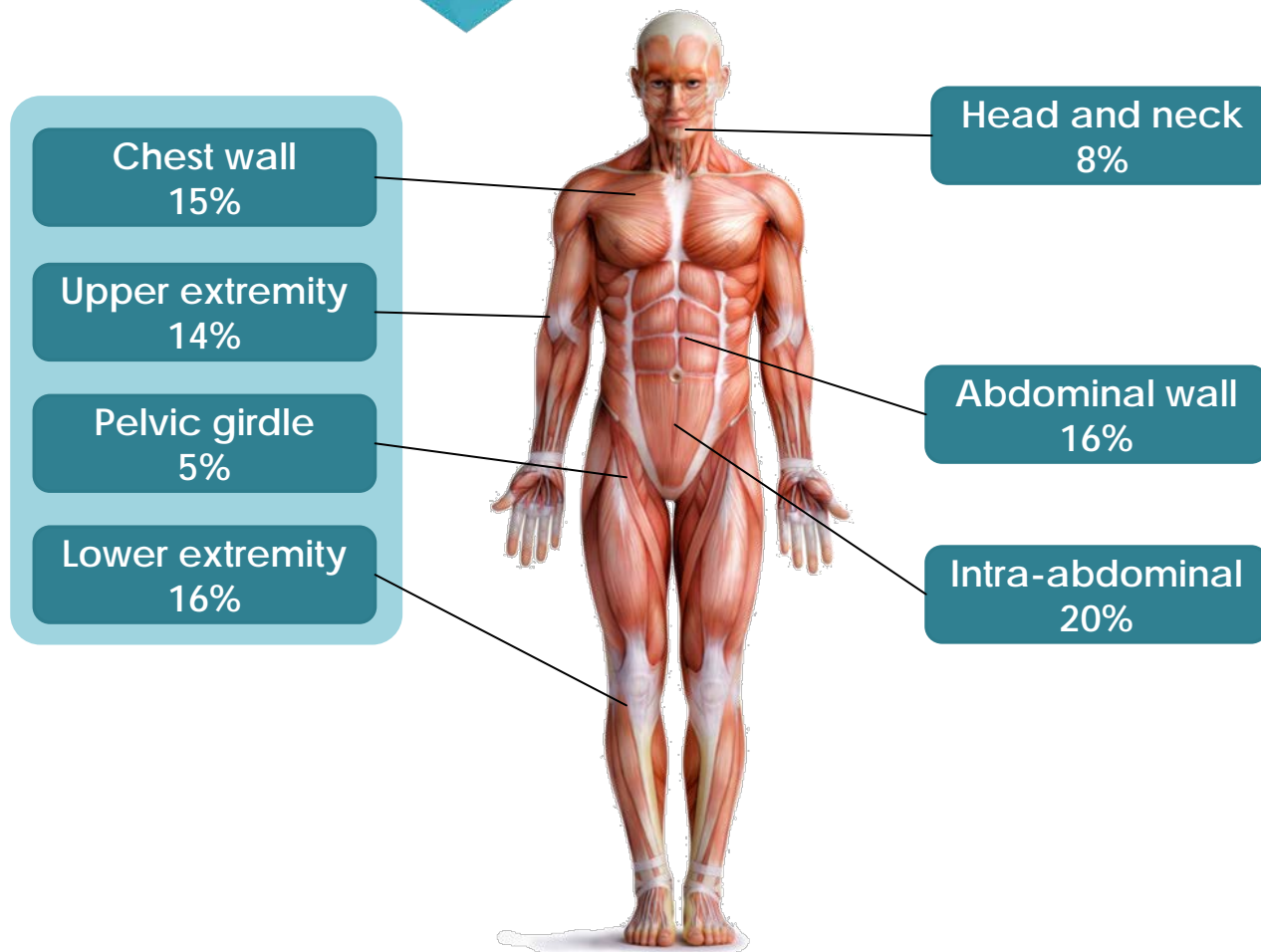
Common sites of disease



- Connective tissue is found everywhere in the body, so desmoid tumors may occur at any site^{1,2}
- In practice, the locations can be grouped into 4 general areas³
 - Extremities, girdles, chest wall
 - Head and neck, intrathoracic
 - Abdominal wall
 - Intra-abdominal, retroperitoneal



Symptoms and Presentation

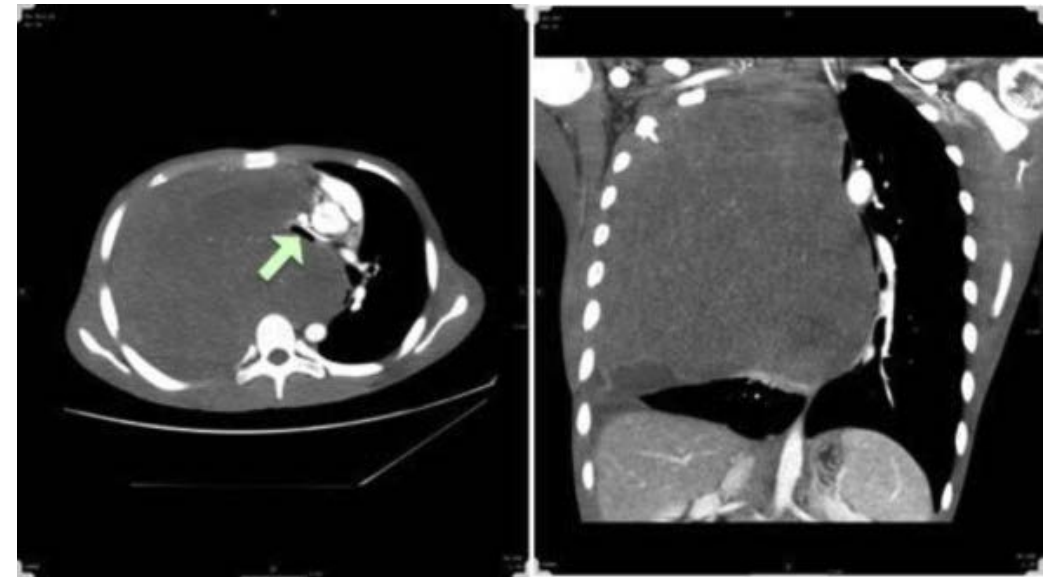


- **Extremities, girdles, chest wall:** pain, restricted mobility, palpable mass
- **Head and neck, intrathoracic:** dysphagia, shortness of breath
- **Abdominal Wall:** palpable mass, may be revealed during pregnancy
- **Intra-abdominal, retroperitoneal:** may be asymptomatic (silently enlarging), weight loss, organ involvement (e.g., bowel obstruction, compression of ureters)

Severity



- The more serious desmoid tumors are often found in:
 - Head and neck/intrathoracic
 - Intra-abdominal, retroperitoneal, or pelvic cavity
- This is usually because of the vital organs in these areas including:
 - Intestines
 - Kidneys
 - Lungs
 - Vena cava
 - Mesenteric arteries
 - Carotid arteries



Robinson et al. J of Ped Surg Case Rep. 2016 (5):15-18

Treatment Options



Desmoid tumors are often cared for by oncologists, in particular sarcoma specialists, and generally have the same treatment options as for malignancy

Active Surveillance

- “Watch and wait” is often the front line approach.
- Exception is head and neck tumors which are often treated more aggressively because of the proximity to important structures

Surgical

- Original thinking was to just remove them and be done.
- This is falling out of favor because of the high rate of recurrence at the same site.

Ablative

- Radiation is commonly used
- New methods being studied: High Intensity Focused Ultrasound and Cryoablation

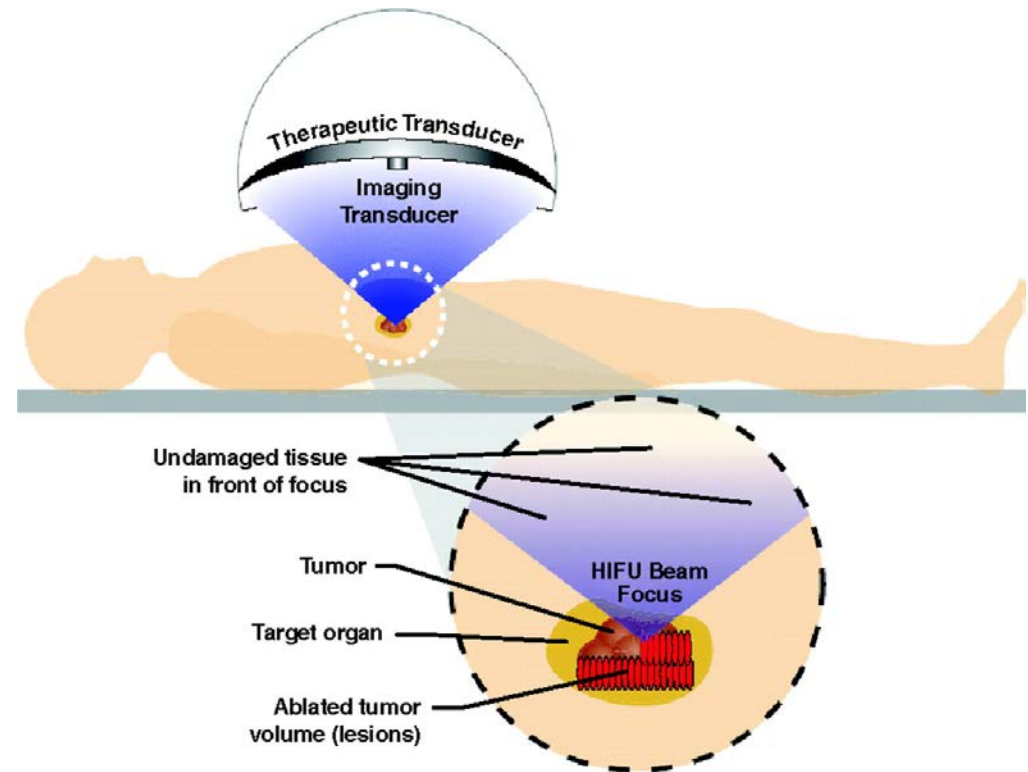
Medical

- Hormone-blocking agents and NSAIDs
- Chemotherapies
- Kinase inhibitors/ gamma secretase inhibitors/ other targeted therapies

Treatment (cont.)



- At progression:
 - Abdominal wall – surgery; medical treatments
 - Extremities – medical treatments; surgery
 - Intra-abdominal – medical treatments; surgery and radiation



Dubinsky, et al. *Am J of Roentgenology* (2008) 190: 191-199

Thank you for your
attention and
consideration.

Questions?