Evidence BasedRecommendations for Treating Desmoid Tumors: Consensus of the Desmoid Tumor Working Group

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INTRODUCTION:

Desmoid tumors are locally invasive soft tissue lesions that frequently occur in the extremities and are treated by orthopaedic surgeons. There have been substantial advances in diagnosis, natural history, and treatments in the past five years that have altered management recoomendations. An international team used a formal process to develop evidence based treatment recommendation for desmoid tumors with a sub-focus on those seen by orthopaedic surgeons.

METHODS:

89 clinicians or researchers with expertise in desmoid tumors and leaders of patient advocacy groups were invited to participate. Data from a prior consensus paper was used as a baseline. New articles were assessed in several subtopics, and a meta-analysis was undertaken when appropriate data was available. A modified Delphi process was used with over 60 experts actively participating to develop the recommendations.

RESULTS:

The diagnosis can be reliably by needle biopsy. Mutational analysis of the CTNNB1 gene, which encodes beta-catenin, should be undertaken to confirm the diagnosis. In mutation negative cases, consideration should be given to genetic counseling or germ line APC mutational analysis, as these patients may have a form of familial adenomatous polyposis and routine colonoscopy is needed for surveillance. Almost a quarter of tumors will regress over time, and as such, active surveillance should be the first line of therapy unless a tumor is causing significant morbidity or has the potential for mortality. New MR imaging analysis approaches can be used to suggest the treatment aproach. Surgery should be avoided, even in the smallest tumors, as recurrence rates are quite high, and overall outcome, as shown in meta-analysis, is worse than without surgery.

In case of progressive or life-threatening situations, first line therapy should be a methotrexate vinblastine regimen. Tyrosine kinase inhibitors can be considered in the case of progression under this treatment. Gamma secretase inhibitors show promise as a second line of treatment, However, this is associated with ovarian dysfunction, and should this be used with caution in girls. Radiation therapy is effective, but associated with side effects and should be avoided, especially in children . The role of local therapy with other modalities, such as cryotherapy, is not well defined but could be considered in small progressive lesion and/or refractory disease after several lines of therapy.

DISCUSSION AND CONCLUSION:

A Delphi process resulted in treatment recommendations for patients with desmoid tumors.

There was consensus around a shift away from surgery and towards active surveillance as a first line treatment. Given the significant regression rate, other theories should be instituted after demonstrated progression or in cases of significant symptoms on prevention. New drug therapies hold promise, but should be used with caution in certain populations due to the side effect profile, such as in pediatric patients.

FIG 1.
Management of Patients with Desmoids from diagnosis to treatment.

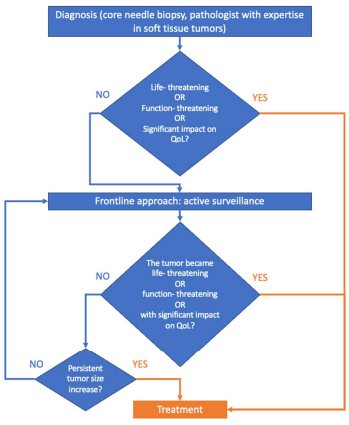

FIG 2.
Management of Patients with Desmoids of the extremities requiring treatment