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Circulating tumor cells in desmoid tumors

Alexcia C. Braun², Fernando Campos¹, Emne A. Abdallah², Anna P. C. Ruano², Milena Tariki¹, Fabio Pinto¹, Celso A. L. Mello¹, Vilma R. Martins², Ludmilla T. D. Chinen².

1 Department of Medical Oncology, AC Camargo Cancer Center, São Paulo, Brazil; 2 International Research Center, AC Camargo Cancer Center, São Paulo, Brazil

Background: Desmoid tumor (DT) is a rare neoplasm with high local recurrence rates. CTNNB1 mutations are found in most sporadic cases and are located in exon 3, usually in codons 41 or 45 (p.T41A, p.S45F and p.S45P). Circulating tumor cells (CTCs) isolated from the peripheral blood of patients with sarcomas and other neoplasms can be early biomarkers of tumor invasion and dissemination. Moreover, CTCs can also re-colonize their tumors of origin through a process of “tumor self-seeding.” Objectives: To identify CTCs in the peripheral blood of patients with DT; to evaluate their expressions of β -catenin and vimentin proteins.

Materials and methods: We conducted a prospective study of patients with initially diagnosed and relapsed DT with measurable disease. Blood samples from each patient were processed and filtered by ISET® (*Rarecells, France*) for the isolation and quantification of CTCs. The expressions of β -catenin and vimentin in CTCs were analyzed by immunocytochemistry (ICC).

Results: A total of 18 patients were included, and all had detectable CTCs. We found a concordance of β -catenin expression in CTCs and in primary tumors in 42.85% of cases by using ICC and immunohistochemistry respectively.

Conclusions: Our study identified a high prevalence of CTCs in DT patients. Concordance of β -catenin expression between primary tumor and CTCs bring new perspectives to assess the dynamics of CTCs in the blood microenvironment, opening new avenues for studying the biology and behavior of DT. In addition, these results open the possibility of using CTCs to predict DT dynamics at the time of progression and during treatment. Further studies with larger sample sizes are needed to validate our findings.