

**Docket #:** S24-017

# **A Liquid Biopsy Approach to Distinguish Leiomyosarcoma from Leiomyoma in the Uterus**

Stanford scientists have discovered a DNA methylation signature on circulating tumor DNA (ctDNA) that can distinguish between the aggressive Leiomyosarcoma (LMS) from its benign counterpart leiomyoma (LM) in the uterus.

A large number of women annually present with uterine masses that require surgery. The majority of these tumors are benign leiomyomas that can be removed with simple surgical methods. A small subset however is malignant, and these will spread through the peritoneal cavity when not resected with an appropriate oncological techniques. Biopsies of uterine masses are not performed and imaging methods often fail to be conclusive about this differential diagnosis. As a result, patients are often assumed to have the more frequently occurring Leiomyoma and undergo surgery of their tumor without definitive diagnosis, which can lead to dissemination of an unexpected malignancy in women with LMS. This leads to significantly poorer clinical outcomes for patients. In the previous published work, Stanford researchers have demonstrated that it is possible to detect genomic markers of LM and LMS in ctDNA (see Publications). In new unpublished work, Stanford researchers have now identified key epigenomic markers in the ctDNA of LM and LMS patient plasma samples which allows for the enhanced diagnosis through DNA sequencing. This finding addresses a critical unmet need for the diagnosis of LMS to improve patient outcomes.

## **Stage of Development**

Proof of Concept - verified from patient samples.

## **Applications**

- Blood test for diagnosis of leiomyosarcoma (LMS)/leiomyoma (LM)
- Detection of disease progression in LMS
- Monitoring treatment effectiveness for LMS
- Companion diagnostic

## Advantages

- First in class LMS diagnostic
- Non-invasive
- Cheaper and faster than current diagnostic methods

## Publications

- Przybyl, J., Chabon, J. J., Spans, L., Ganjoo, K. N., Vennam, S., Newman, A. M., ... & van de Rijn, M. (2018). [Combination approach for detecting different types of alterations in circulating tumor DNA in leiomyosarcoma](#). *Clinical Cancer Research*, 24(11), 2688-2699.
- Przybyl, J., Spans, L., Lum, D. A., Zhu, S., Vennam, S., Forgó, E., ... & van de Rijn, M. (2019). [Detection of circulating tumor DNA in patients with uterine leiomyomas](#). *JCO precision oncology*, 3, 1-9.

## Patents

- Published Application: [WO2026015497](#)

## Innovators

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- Joanna Przybyl

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