Single-cell analysis and deep learning reveal a novel diagnostic biomarker for endometriosis

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Innosuisse

The Problem

Endometriosis patients face a delay in diagnosis of up to 7.5 years on average. This is associated with a significant impact on the quality of life for patients, and a conspicuous economic burden ($70 billion dollars per year, in the United States alone) (1). New diagnostic solutions are urgently needed to allow prompt referral and appropriate treatments to prevent disease progression and to address possible infertility early on, as well as to significantly reduce the socio-economic burden. Scailyte and Inselspital Bern aimed at identifying novel endometriosis-specific biomarkers for the development of a non-invasive, in vitro diagnostic tool to aid in the diagnosis of Endometriosis.
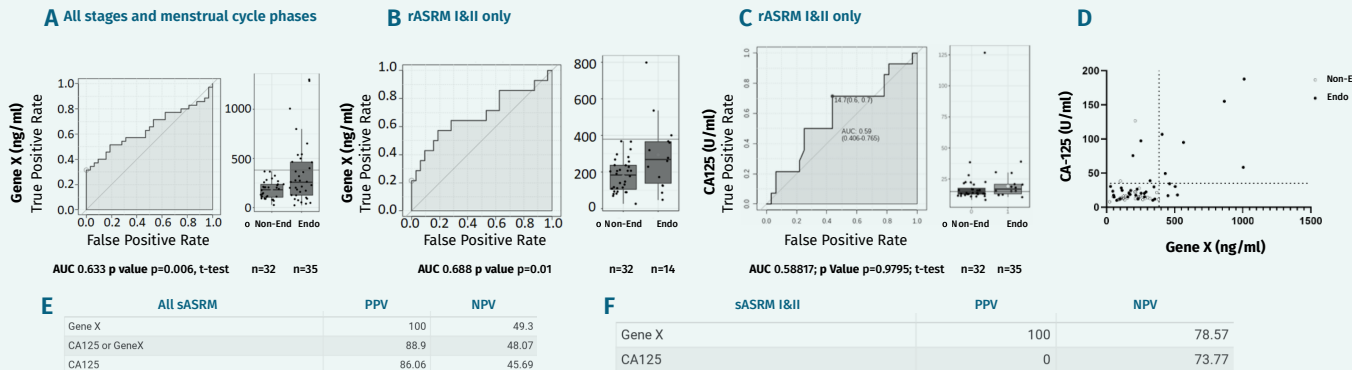
PBMCs and endometrial biopsies were collected during diagnostic laparoscopy from 32 patients suspected of suffering from endometriosis. Single-cell RNA-sequencing and a machine learning algorithm, ScaiVisionTM, was performed to investigate the presence of biomarker signatures present specifically in endometriosis patients.

In this poster, the blood-based biomarker (Gene X) is presented for the first time. Gene X was quantified and validated using ELISA method in an independent cohort of 67 human serum samples confirmed endometriosis or non-endometriosis by laparoscopic surgery.

The Science

Endometriosis prediction performance of Gene X in serum

Inclusion criteria:
- Laparoscopy / pathology report
- Signed Informed Consent Form

Exclusion criteria:
- Hormonal treatment
- Adenomyosis (in non-endometriosis group)
- Unclear menstrual phase
- Unclear diagnostic

A All stages and menstrual cycle phases
AUC 0.633 p value p=0.006, t-test n=32 n=35

B rASRM I&II only
AUC 0.688 p value p=0.01 n=32 n=14

C rASRM I&II only
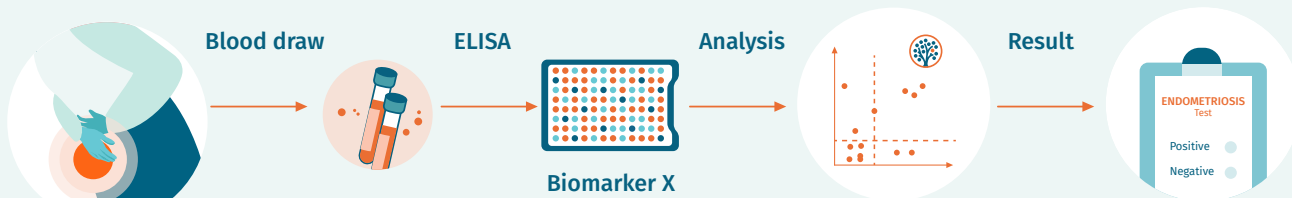AUC 0.58817 p Value p=0.9795; t-test n=32 n=35

D

Gene X accurately predicts Endometriosis in all stages and cycle phases (A). Accuracy further increases in rASRM I and II (or moderate Enzian score) (B). Gene X accuracy is positively affected in the secretory phase (data not shown). Gene X outperforms the prediction accuracy of CA125 in all patients and in rASRM I and II patients (C). CA125 complements Gene X and increases the sensitivity in detection of patients with endometrioma and high grade endometriosis (D). Gene X Positive Predictive Value exceeds CA125 and their combination is all stages and further more, in rASRM I and II

Our results show a novel and promising biomarker for the diagnosis of early stage endometriosis patients directly from serum.

E

All sASRM	PPV	NPV
Gene X	100	49.3
CA125 or GeneX	88.9	48.07
CA125	86.06	45.69

F

sASRM I&II	PPV	NPV
Gene X	100	78.57
CA125	0	73.77

The Solution

First-in-class Endometriosis Diagnostic Assay

Blood draw → ELISA → **Biomarker X** → Analysis → Result

Patient suspected of having Endometriosis

ENDOMETRIOSIS Test — Positive / Negative

Our product comprises of Gene X detection via ELISA from patient serum, and integrated results on our platform to give an accurate diagnosis of Endometriosis at early stages.

We are seeking advisors and collaborators to expand our network and bring our product to your clinic by 2024.

References:
(1): Wang Y, Nicholes K, Shih IM. The Origin and Pathogenesis of Endometriosis. Annu Rev Pathol. 2020;15:71-95. doi:10.1146/annurev-pathmechdis-012419-032654

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