

Cancer Panel Analysis of Circulating Tumor Cells in Patients with Breast Cancer

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Study Aim: To isolate circulating tumor cells (CTCs) from breast cancer patients using a novel size-based filtration method and perform comprehensive cancer gene panel analysis on the isolated CTCs.

Methods:

- Blood samples were collected from 8 breast cancer patients
- CTCs were isolated using a size-based filtration method developed by the authors
- Isolated CTCs were counted via immunofluorescent staining for EpCAM and CD45
- Genomic DNA from CTCs was amplified and analyzed using the Ion AmpliSeq Comprehensive Cancer Panel, which covers 409 cancer-related genes
- White blood cells (WBCs) from the same patients were used as controls
- CTC-specific mutations were identified by comparing CTC and WBC mutation profiles

Key Findings:

- EpCAM+ CTCs were detected in 7/8 patients, with an average of 8.6 CTCs per 5 mL blood
- The isolation method yielded sufficient DNA quantity and quality for genomic analysis
- CTC-specific mutations were detected in 62.5% of patients
- Mutations were found in genes including EZH2, NOTCH1, ARID1A, STK11, FLT3, MYCN, APC, and PTEN
- Some mutations were detected even in samples without EpCAM+ cells, suggesting the method can isolate EpCAM- CTCs

Conclusions:

- The size-based CTC isolation technique was effective at obtaining CTCs at sufficient purity for genomic analysis
- Comprehensive cancer panel analysis of CTCs is feasible and may have potential applications in precision medicine
- This approach allows detection of CTC-specific mutations by comparison to WBC controls from the same patient

This study shows that isolating and genetically analyzing circulating tumor cells from breast cancer patients is feasible and potentially useful for personalized treatment.