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Genomics Oncology

Unlock the Power of Genomics

CELL-FREE TUMOUR DNA TESTING IN CANCER

Cancer is a complex disease, caused by a wide range of genetic mutations. These mutations can be present in many combinations that are unique to each patient, making every case of cancer a rare disease.

Current treatment protocols rely on the assumption that patients with cancer of the same tissue type have the same underlying disease. However, each tumour has a unique genetic makeup, even amongst the same tissue type. Identification of these individual mutations is the key to developing personalised treatments for cancer patients.

What is ctDNA testing?

Cell-free DNA (cfDNA) are small fragments of DNA that are released from normal cells and tumour cells by programmed cell death (apoptosis) into the blood. Tumour cells may release circulating cell-free tumour DNA (ctDNA), which may contain identical mutations to those identified in the primary tumour. [1]

Release of ctDNA into the blood allows for a 'liquid biopsy' to be performed from a blood sample. This removes the need for invasive procedures, such as biopsies through fine needle aspiration or surgical excisions, which may be risky for the patient or not possible due to the size or location of the tumour.

ctDNA testing may give an indication of the size of a tumour or the amount of cancer cells in the body, monitor patient response to treatment and may alert doctors to early signs of recurrence or resistance.

Why Should I do ctDNA testing?

Check for Residual Cancer Following Treatment

- ctDNA is a potential marker of residual cancer cells following surgery, and may determine which patients will experience recurrence. [1]
- Radiology and imaging techniques can detect tumours approximately 7 – 10 mm in size. ctDNA is released by tumours around 20 times smaller than this, allowing for residual cancer cells to be detected much earlier. [5]
- The change of ctDNA levels in response to treatment has been observed across a range of tumour types, and some studies have found increased levels of ctDNA before or at the time of recurrence. [3]

Monitor Therapy and Check for Resistance

- ctDNA testing provides an overview of all the different cells within a tumour. Analysis can detect new molecular changes in the cells, which may provide an early indication of resistance to therapy.
- By understanding mutations and pathways in the tumour, personalised treatment approaches can be planned. This may help reduce the development of resistance in the tumour or improve response to treatment. [7]
- For patients currently undergoing treatment, testing may identify new treatment options.

Early Diagnosis and Detection

- Following further studies, ctDNA testing may potentially be used for cancer screening or early detection in the future.

Track the Number of Cancer Cells in Real-Time

- Levels of ctDNA increase with disease progression and decrease in response to treatment. [2,3]
- ctDNA can more accurately reflect the real-time tumour size and burden than other cancer markers. [4]
- Tracking ctDNA levels over time can be used to monitor response to treatment, as shown in Figure 1.

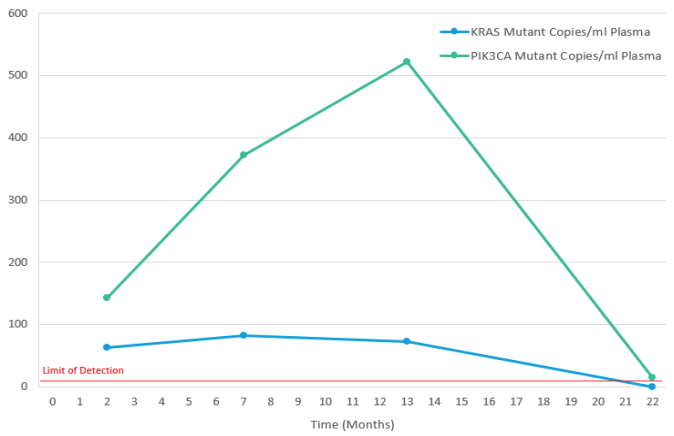

Figure 1: Tracking the amount of ctDNA in the blood for 2 specific mutations over a period of 22 months shows reduction in the tumour burden in this patient over time in response to treatment.

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What Might We Find?

We offer a range of assays that may detect mutations in the following genes. These genes may offer information on drug targets and resistance to current treatments.

LUNG

Oncomine Lung Cell-Free Total Nucleic Acid Assay

ALK	*MET*
BRAF	*NRAS*
EGFR	*PIK3CA*
ERBB2	*RET*
KRAS	*ROS1*
MAP2K1	*TP53*

BREAST

Oncomine Breast cfDNA Assay v2

ALT1	*FBXW7*
CCND1	*FGFR1*
EGFR	*KRAS*
ERBB2	*PIK3CA*
ERBB3	*SF3B1*
ESR1	*TP53*

COLON

Oncomine Colon cfDNA Assay

AKT1	*ERBB2*	*NRAS*
APC	*FBXW7*	*PIK3CA*
BRAF	*GNAS*	*SMAD4*
CTNNB1	*KRAS*	*TP53*
EGFR	*MAP2K1*	

PAN-CANCER

Oncomine Pan-Cancer Cell-Free Assay*

Hotspot Genes					Tumor Suppressor Genes	CNV Genes		Gene Fusions	
AKT1	*EGFR*	*FLT3*	*KRAS*	*PDGFRA*	*APC*	*CCND1*	*ERBB2*	*ALK*	*FGFR3*
ALK	*ERBB2*	*GNA11*	*MAP2K1*	*PIK3CA*	*FBXW7*	*CCND2*	*FGFR1*	*BRAF*	*MET*
AR	*ERBB3*	*GNAQ*	*MAP2K2*	*RAF1*	*PTEN*	*CCND3*	*FGFR2*	*ERG*	*NTRK1*
ARAF	*ESR1*	*GNAS*	*MET*	*RET*	*TP53*	*CDK4*	*FGFR3*	*ETV1*	*NTRK3*
BRAF	*FGFR1*	*HRAS*	*MTOR*	*ROS1*		*CDK6*	*MET*	*FGFR1*	*RET*
CHEK2	*FGFR2*	*IDH1*	*NRAS*	*SF3B1*		*EGFR*	*MYC*	*FGFR2*	*ROS1*
CTNNB1	*FGFR3*	*IDH2*	*NTRK1*	*SMAD4*					
DDR2	*FGFR4*	*KIT*	*NTRK3*	*SMO*					

*This assay can be used for most cancer types

Assay Sensitivity

Assays may not identify every mutation associated with malignancy. Whether a tumour has detectable levels of ctDNA in the blood varies depending on the tumour type and stage of disease. Some tumours, such as central nervous system metastases, are associated with lower levels of ctDNA.

The Pan-Cancer Cell-Free Assay has a flexible detection limit down to 0.1% for SNVs or 1 mutant copy in a background of 1,000 wild-type copies. To achieve 0.1% LOD, 20 ng of input cfTNA is required. %LOD is dependent on the cfTNA input amount.

How Do I Organise Testing?

Contact us for the test request form. When ready, return your completed form

→

We arrange for your blood or biopsy tissue sample to be sent to the laboratory.

→

We receive your tissue sample for testing.

→

Our testing partner completes the requested testing in their Brisbane laboratory.

→

Your personalised report is sent to nominated health care professional

References

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