

## Keeping Tabs on Cancer: The Liquid Biopsy Way

### Quick Summary

- o Ms. Preet Jhurani\*, a 65-year-old homemaker, was diagnosed with breast cancer.
- o Genetic analysis of the tumor biopsy revealed the presence of a mutation in the *TP53* gene.
- o The identification of the genetic profile of the tumor is crucial for tracking the progression of the disease in response to administered therapy.
- o Surgical excision of the tumor was performed and adjuvant chemotherapy was provided to the patient.
- o The tumor marker was not detected in a liquid biopsy sample, drawn at three months post-surgery.
- o The patient is well as of September 2017 and is under medical observation.
- o Additional liquid biopsies can be performed at 3-4 month intervals to ascertain recurrence of breast cancer, prior to the emergence of other clinical symptoms.



### Introduction

Breast cancer is now the number one lethal cancer in women, in India. It has overtaken cervical cancer to be the most frequent cancer in India (1–3). Treatment of metastatic breast cancer with chemotherapy is the standard of care, in practice.

The results of chemotherapy are assessed by clinical symptoms and imaging techniques such as PET-CT scans, ultrasound imaging and mammograms (4). Another useful technique that can be leveraged for monitoring the response of cancer to given therapies, is Liquid Biopsy.

The significant advantage of liquid biopsy tests is that there is no exposure to radioactive probes (PET-CT scan) and the results are obtained within 5 working days. A blood sample for liquid biopsy can be obtained in settings where a patient is comfortable and the frequency of these tests can be set by the doctor.

One can perform two liquid biopsy tests in the time interval that is mandatory between two PET-CT scans.

### Liquid Biopsy

Liquid Biopsy is essentially a blood sample from a cancer patient. Blood naturally contains DNA shed by cancer cells as well as normal cells when they die. Scientists can then extract tumor-specific DNA from this blood sample and understand the genetic profile of the cancer as well as estimate tumor burden.

\*Name changed to protect patient privacy

## Patient Profile

Preet Jhurani, age 65 years, was looking forward to visiting her son in Australia and spending 6 months with him. She was also a bit apprehensive about the upcoming trip because some nagging health issues bothered her. She hoped that her breast pain and discharge would not bother her during her stay with her son. She had also recently noticed a small lump on her right breast and dimpling in the areola.

Eventually, she decided to consult a physician and lay her doubts to rest before departing for Australia.

Preet's gynecologist suspected the incidence of breast cancer and referred her to a prominent oncologist in Bangalore, India. Medical investigations, including a mammogram and a tissue biopsy, led to the diagnosis of breast cancer.

## Results of Genetic Testing

Preet's oncologist recommended genetic testing of the breast biopsy sample, with two objectives in mind:

- A. Choice of targeted therapies
- B. Tracking the progression of cancer using liquid biopsies The StrandAdvantage 56-gene test was prescribed for the molecular diagnosis of Preet's breast cancer, in January 2017.

Sample Collection Date	Test	Gene	Mutation	Result	Details
Solid Tumor Result (DNA Source:FFPE)					Supporting Reads
19-Jan-2017	Strand Advantage Test Swift 56G Solid	TP53	R273H	Detected	9.80%

A mutation in the *TP53* gene - *TP53*<sup>R273H</sup> - was detected in the breast tissue biopsy.

## Treatment Options

A lumpectomy with wide margins was performed on Preet to curtail the growth and spread of cancer, in March 2017. Adjuvant chemotherapy was prescribed to her to ensure that remnant cancer cells, if any, would be eliminated.

## Follow-up Liquid Biopsy

A liquid biopsy sample was drawn from Preet to understand the progression of her breast cancer, in June 2017.

A highly sensitive detection assay was used on cell-free DNA (cfDNA) isolated from the blood sample. The detection test is designed to identify the precise gene variant - *TP53*<sup>R273H</sup> - in cfDNA.

Sample Collection Date	Test	Gene	Mutation	Result	Copies/ml plasma
27-Jun-2017	Strand Liquid Biopsy*	<i>TP53</i>	R273H	Not detected	N/A

The tumor marker mutation - *TP53*<sup>R273H</sup> - was not detected in the blood sample within 3 months of the surgical excision of the parent tumor.

## Conclusions

- Preet Jhurani, age 65 years, was diagnosed with breast cancer.
- A mutation in the *TP53* gene was identified from the breast tissue biopsy.
- Surgery and adjuvant chemotherapy were administered to Preet.
- A liquid biopsy was performed, 3 months post-surgery, to ascertain the tumor burden in the patient.
- The tumor marker mutation was not present in cell-free DNA isolated from blood plasma.
- At this time-point, the combinatorial therapy of surgery and chemotherapy seems to have limited the growth of breast cancer cells. The patient is in remission and is under medical surveillance as of September 2017.
- Follow-up liquid biopsies can be performed at 3-4 month intervals to gain insights about the progression of cancer before clinical symptoms of recurrence are evident.
- Detection of the tumor-specific TP53 mutation in cfDNA from subsequent liquid biopsies, will indicate proliferation of remnant breast cancer cells.

## StrandAdvantage 56-gene Test

StrandAdvantage 56-gene test is a pan-cancer test designed to identify mutations that are frequently found in most solid tumors. The 56-gene test has the advantage of being applicable for analysis of solid tumors as well as NGS analysis of cell-free DNA obtained from a liquid biopsy.



## References

1. Malvia S, Bagadi SA, Dubey US, Saxena S. Epidemiology of breast cancer in Indian women. *Asia Pac J Clin Oncol* [Internet]. 2017 Aug [cited 2017 Aug 16];13(4):289–95. Available from: <http://doi.wiley.com/10.1111/ajco.12661>
2. Badwe RA, Dikshit R, Laversanne M, Bray F. Cancer incidence trends in India. *Jpn J Clin Oncol* [Internet]. 2014 May [cited 2016 Dec 14];44(5):401–7. Available from: <http://www.ncbi.nlm.nih.gov/pubmed/24755545>
3. Mallath MK, Taylor DG, Badwe RA, Rath GK, Shanta V, Pramesh CS, et al. The growing burden of cancer in India: epidemiology and social context. *Lancet Oncol* [Internet]. 2014 May [cited 2017 Jul 31];15(6):e205–12. Available from: <http://linkinghub.elsevier.com/retrieve/pii/S1470204514701159>
4. Chu AJ, Chang JM, Cho N, Moon WK. Imaging Surveillance for Survivors of Breast Cancer: Correlation between Cancer Characteristics and Method of Detection. *J Breast Cancer* [Internet]. 2017 Jun [cited 2017 Sep 6];20(2):192–7. Available from: <http://www.ncbi.nlm.nih.gov/pubmed/28690656>



Strand Life Sciences Pvt. Ltd.

5th Floor, Kirloskar Business Park, Bellary Road, Hebbal, Bangalore - 560 024  
Phone: 1800-1022-695, support.strandx@strandls.com, www.strandls.com

#StayAheadOfCancer  

Strand is accredited by



Strand ID: LB-153527102017