

CASE STUDY

Genetic Analysis Identifies the Right Drug for Treatment of Bladder Cancer

Quick Summary

- o Amandeep Malhotra* was diagnosed with bladder cancer at the age of 45 years
- o Genetic analysis of the tumor was advised to him in order to choose optimal therapeutic options
- o A mutation in the *PIK3CA* gene was identified in his tumor biopsy
- o A targeted therapy drug, Everolimus, was prescribed to Amandeep, as a result of genetic testing
- o The choice of an accurate and optimal therapeutic, right at the initiation of cancer therapy eliminated trial-and-error, resulting in savings of cost and time for the patient



Patient Profile

Amandeep Malhotra was enjoying the prime of his life with a successful career in a leading pharmaceutical company, a content private life and good health. At 45, he exercised regularly and was careful with his health regimen. He believed in prevention rather than cure and was very vigilant about signs of health problems. In the summer of 2016, he had episodes of pain in the lower abdomen on one side of his body. He also had the urge to pass urine more frequently than before. However, when passing urine became painful and traces of blood were evident in urine, he consulted a leading oncologist in Delhi. The doctor advised a biopsy of bladder tissue in order to conduct a histopathological examination. Additionally, the doctor also recommended a genetic analysis of the tumor biopsy. This was done in order to determine the most suitable therapy for Amandeep.

Histopathological Report

Examination of biopsy tissue showed that Amandeep had a high grade papillary transitional cell carcinoma. The tumor had invaded the inner lining of the urinary bladder but had not yet reached the muscle layer outside it. Invasion of cancer cells into the bloodstream and lymphatic system were not evident in the histopathology exam.

Results of Genetic Testing

The StrandAdvantage 48-Gene Test was prescribed for analysis of Amandeep’s bladder cancer biopsy. This is a pan-cancer test that is designed to assay for the most commonly found mutations from many cancer types.

Therapy	Relevant Markers	Approved Indications	Trials
Everolimus	<i>PIK3CA</i> ^{E542K}	Pancreatic Neuroendocrine Tumor, Gastrointestinal Neuroendocrine Tumor, Lung Neuroendocrine Tumor, Kidney Cancer, Astrocytoma, Hormone receptor-positive HER2-negative Breast Cancer	NCT02449538 NCT02029001

*Name changed to protect patient privacy

- A mutation in the *PIK3CA* gene- *PIK3CA*^{E542K}- was identified in the tumor tissue.
- This gene is engaged in regulating cell proliferation. The mutant gene is constantly active. Treatment with Everolimus, a drug that acts on proteins that work downstream of the *PIK3CA* protein, can help to stop the growth of cells that have this mutant gene.
- Everolimus is being used in several clinical trials wherein the same gene is mutated, in solid tumors (NCT02449538; NCT02029001).
- Hence, a recommendation to use Everolimus to treat Amandeep's cancer was provided.

Conclusions

- Amandeep Malhotra, a 45- year old person was diagnosed with bladder cancer.
- Genetic analysis of the cancer revealed that a mutation in the *PIK3CA* gene was present in the bladder cancer.
- Everolimus, a targeted therapeutic, was recommended to the patient right at the initiation of cancer therapy.
- Genetic analyses can eliminate guesswork and trial-and error runs with chemotherapy drugs and enable the practice of precision medicine.

StrandAdvantage 48-Gene Test

The StrandAdvantage 48-Gene Test is a pan-cancer test designed to identify mutations in genes involved in the most common cancers. In addition to providing on-label drug recommendations, this test can also provide off-label recommendations where the drugs are undergoing trials for therapeutic efficacy in other solid tumors (besides the approved ones).

References

1. <https://clinicaltrials.gov/ct2/show/NCT02449538>;
2. <https://clinicaltrials.gov/ct2/show/NCT02029001>