Mutation causing pre-cancerous condition found in Guj family

TIMES NEWS NETWORK

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Bengaluru: Researchers from a Bengaluru company and a Vadodara hospital have discovered a genetic mutation responsible for a rare pre-cancerous inherited condition in six members of a family.

Considered a breakthrough in cancer research, the study showed that six members of a Gujarati family were suffering from Familial Adenomatous Polyposis (FAP), a rare pre-cancerous condition which accounts for 1%-3% of colorectal cancer cases. The research began when a 52-year-old patient, Paresh (name changed), complained of weight loss and change in bowel movement in 2014. Diagnosis showed an abnormal growth in his colon, which was a benign polyp which could turn cancerous.

The research, conducted by Bengaluru-based MedGenome Labs and Vadodara-based Kailash Cancer Hospital and Research Centre, was possible because Paresh's family members agreed to share his clinical history and blood samples. A genetic analysis of 25 people from his family aged 6-60 was done. While the first patient was tested in October 2015, the entire family was tested in July 2016. MedGenome took about four weeks to identify the mutation in one female and five male members.

The analysis revealed mutation of Adenomatous Polyposis Coli (APC) gene present in six people who were diagnosed with FAP. It was also found in four other family members who didn't have FAP. However, no one in the family suffered from cancer, including Paresh who was screened first, but members were prone to it given their genetic disposition.

The research said in people with the gene mutation, 100-1000 benign growths appear in the colon and rectum in teenage years, and the condition could gradually turn malignant, leading to colorectal cancer by the age of 40-50.

Dr Rakshit Shah, surgical oncologist, KCHRC, Vadodara, said, "Detection of this genetic mutation in a patient will enable us to identify individuals most vulnerable to FAP and colorectal cancer. Preventive measures can be taken so that the incidence can be brought down," he said. According to him, every individual in the family of a patient suffering from colorectal cancer must undergo genetic analysis.

Under normal conditions, the APC gene is a tumour-suppressor one which prevents uncontrolled growth of cells. But mutations in same gene lead to malfunction, the scientists said.

"Our analysis revealed the presence of a mutation in the APC gene which hasn't been identified before. It underscored the power of genetic analysis in identifying individuals in the affected family at the risk of collateral cancer and hence saving lives through early detection and timely treatment," said Dr Arati Khanna Gupta, vice-president, MedGenome Labs.