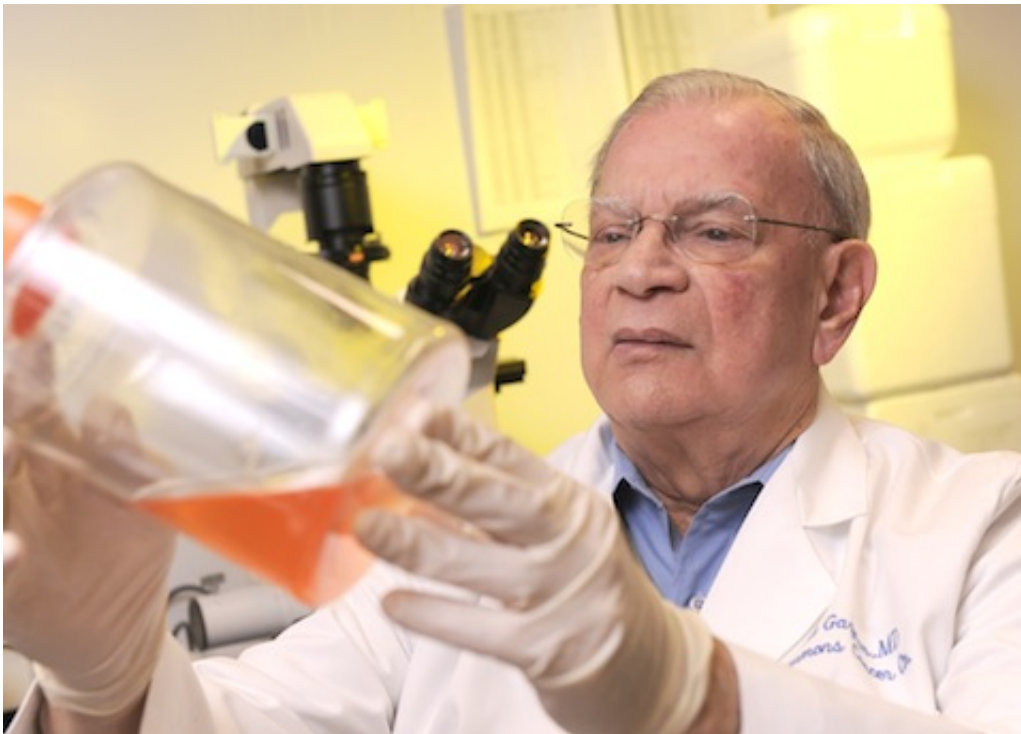


Inherited mutated gene raises lung cancer risk for women, those who never smoked

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This is Dr. Adi Gazdar, Professor of Pathology and Deputy Director for the Nancy B. and Jake L. Hamon Center for Therapeutic Oncology Research.
Credit: UT Southwestern Medical Center

People who have an inherited mutation of a certain gene have a high chance of getting lung cancer—higher, even, than heavy smokers with or without the inherited mutation, according to new findings by cancer researchers at UT Southwestern Medical Center. Although both genders

have an equal risk of inheriting the mutation, those who develop lung cancer are mostly women and have never smoked, the researchers found.

People with the rare inherited T790M mutation of the epidermal growth factor receptor (EGFR) gene who have never smoked have a one-in-three chance of developing lung cancer, researchers found. This risk is considerably greater than that of the average heavy smoker, who has about a one-in-eight chance of developing lung cancer – about 40- fold greater than people who have never smoked and do not have the mutation.

The likelihood of developing lung cancer is so strong for women with the [mutated gene](#) and people with the mutated gene who have never smoked that they may need to get screened for lung cancer at regular intervals, according to Dr. Adi Gazdar, Professor of Pathology and Deputy Director for the Nancy B. and Jake L. Hamon Center for Therapeutic Oncology Research at UT Southwestern.

"Fortunately the mutation, which is extremely rare, can be detected by a blood test. Only people suspected of having the mutation and their [family members](#) need to be tested for the mutation," said Dr. Gazdar.

"This is a very rare inherited mutation in the general population, but because it confers a greatly increased risk of developing lung cancer, it is present in about one in every hundred lung cancer cases."

UT Southwestern investigators said the link with women and those who had never smoked was completely unexpected.

"I even had to convince my coworkers that this finding was correct, but the data were overwhelming," he said.

Dr. Gazdar made the discovery with Dr. Joan Schiller, Professor and Chief of the Division of Hematology-Oncology in Internal Medicine and

Deputy Director of UT Southwestern's Harold C. Simmons Comprehensive Cancer Center, by studying a young woman with lung cancer and her family for five generations, along with a review of data in the literature.

The findings appear online today and in the April print edition of *Journal of Thoracic Oncology*. An accompanying article in the same issue of the *Journal* provides additional data confirming the findings of the UT Southwestern investigators. An accompanying editorial in the same issue states "These studies now solidify the fact that routine clinical management of lung cancer now has to include the awareness of this inherited cancer syndrome."

"It's rare, but you're still faced with families like this. As people get more attuned to recognizing [lung cancer patients](#) with this mutation, we're going to be identifying more family members with this inherited risk, and we will have to develop guidelines on how to manage them," Dr. Gazdar said.

The rare mutation was discovered first in the patient's lung cancer and then in the bloodstream. That prompted a more intensive investigation that involved blood tests of multiple family members and piecing together medical and smoking histories of family members for five generations, some deceased, by Linda Robinson, Assistant Director of Cancer Genetics.

In doing so, researchers found that the mutated gene was passed down on the mother's side in this family.

"It took a huge amount of detective work and involved a lot of leg work for Linda Robinson, and we were fortunate to get great co-operation from the family."

Lung cancer is the leading cause of cancer death in the United States. More than 159,000 Americans died from [lung cancer](#) in 2013, accounting for more deaths than breast, colorectal and prostate cancer combined, according to the National Cancer Institute.

Provided by UT Southwestern Medical Center

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