

Scans spot hidden tumors in rare cancer syndrome

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Researchers at Dana-Farber Cancer Institute report that full-body PET/CT scanning detected unsuspected, treatable tumors in 3 of 15 patients with Li-Fraumeni syndrome (LFS), a rare genetic cancer syndrome for which no screening tests have been recommended.

The results suggest that periodic scans in combination with physical exams might catch more tumors at a curable stage, the investigators report in the March 19 issue of the *Journal of the American Medical Association*. But they caution that further, larger studies are needed to determine whether PET/CT screening is beneficial in LFS patients, who are highly susceptible to a variety of cancers from an early age because of an inborn gene mutation.

"We need to be cautious, and we shouldn't say that every patient with the syndrome should have a PET/CT examination," said Annick Van Den Abbeele, MD, clinical director of radiology and director of nuclear medicine/positron emission tomography (PET) at Dana-Farber. "But the study showed some interesting findings that justify a larger, international study in these patients."

PET detects cancers by tracking their abnormal appetite for sugar (glucose) compared to normal tissues, while CT (computed tomography) uses X-rays to show anatomical and structural details. Combining the two modalities in a single machine allows a patient to undergo both exams in one session; the resulting images are superimposed to reveal the precise location of suspected tumors.

LFS is a rare hereditary cancer syndrome named for the researchers who first described it, Frederick Li, MD, of Dana-Farber and Joseph Fraumeni Jr., MD, of the National Cancer Institute. In most cases, the cause is a mutation in the TP53 tumor suppressor gene that can be inherited and creates a high risk of a variety of malignancies, including sarcomas, breast cancer, leukemia, brain tumors, and many more common cancers at unusually early ages, including childhood. Among individuals with LFS, the chance of developing any cancer has been estimated at 50 percent by age 30, and 90 percent by age 60. A survivor of one cancer has a high risk of developing a new malignancy over time.

The syndrome is usually diagnosed after several members of a family develop early-onset tumors, prompting a test for the mutated gene. At present, there is no specific screening test to detect cancers before they become symptomatic in people carrying the Li-Fraumeni mutation because they are prone to such a wide variety of cancer types.

"We decided to study this in the adult LFS population because PET/CT scanning is used in the care of many of the type cancers that occur in LFS. Both LFS families and physicians have been frustrated by the lack of information for families with a rare and burdensome condition," said Judy Garber, MD, MPH, senior author of the paper and director of Dana-Farber's Cancer Risk and Prevention Clinic.

The pilot study recruited 15 healthy members of Li-Fraumeni families carrying the TP53 mutation and who had not been diagnosed with cancer in the past five years. Many of the volunteers were members of Li-Fraumeni families who had participated in research through Dana-Farber and the NCI for many years.

The combined PET/CT images revealed thyroid cancer in a 31-year-old breast cancer survivor and in a 48-year-old survivor of breast cancer and a childhood sarcoma. In addition, a 36-year-old man with no cancer

history was found to have a tumor at the junction of his esophagus and stomach. All were given potentially curative treatments, the researchers said. Subjects are currently being followed one year later.

Whether earlier detection of tumors in LFS patients will translate into increased survival remains to be shown in longer-term studies, the scientists said. Another uncertainty is whether the potential benefit from periodic screening would outweigh the risks that the radiation exposure might trigger cancers in the cancer-prone LFS patients. Each PET/CT scan exposes the individual to an amount of radiation that is half of the allowable annual limit for a worker in the radiation industry, according to the paper. Still more questions exist about when to begin screening with imaging or colonoscopy in childhood, adolescence or adulthood.

"We will need to be cautious about the radiation issue, and to determine what is best for the patients in terms of a screening schedule," said Serena Masciari, MD, lead author. "We also need to know if there is a high rate of false-positive findings from screening that would have to be followed up," she added.

Source: Dana-Farber Cancer Institute

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