Inherited Genetic Mutations Are Common in Young Adults With Early-Onset Cancer

These cancers include those that are rare among adults under 40, such as breast, colon, pancreas, kidney, prostate and ovarian cancer.

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Adults under 40 who develop cancers that are rare for their age bracket have a high rate of inherited gene mutations associated with cancer.

People with inherited genetic mutations who develop such cancers—known as early-onset cancers—are more likely to develop new malignancies unrelated to their first cancer and to experience other health complications.

The most common early-onset cancers, which by definition are rare malignancies among younger adults, are breast, colon, pancreas kidney, prostate and ovarian cancer. These are distinct from other types of cancer that are more commonly seen among young adults, such as sarcoma and brain cancer.

Thus, the new findings from a recent study, presented this week at the American Association for Cancer Research (AACR) Virtual Annual Meeting II, have important implications for how younger adults with cancer might receive genetic counseling that could inform their treatment as well as their ongoing monitoring as they progress through life.

“This study supports a role for genetic testing irrespective of tumor types,” Zsofia Stadler, MD, a medical oncologist at Memorial Sloan Kettering Cancer Center in New York, said at an AACR press briefing.

Stadler and her colleagues assessed inherited genetic mutations among 1,201 young adults diagnosed with cancer at Sloan Kettering between 2015 and 2019, looking specifically for up to 88 genes known to be linked to a higher risk of cancer.

Eight hundred seventy-seven of the participants had early-onset cancer—most commonly colorectal, breast, kidney, pancreatic and ovarian cancer; 324 had young-adult cancers that are

common in this age group—most commonly sarcoma and brain, testicular and thyroid cancer.

The researchers found that 21% of those with early-onset cancers and 13% of those with young-adult cancers had inherited genetic mutations (also known as germline mutations).

Among people with early-onset cancers, BRCA gene mutations and those associated with Lynch syndrome (the most common cause of hereditary colon cancer) were seen most often. Among those with young-adult cancers, inherited TP53 mutations were more common; these are characteristic of Li-Fraumeni syndrome, which is associated with childhood cancers.

“Although they only represent about 4% of all cancers, young adults with cancer, defined as those diagnosed with cancer between the ages of 18 and 39, face unique challenges,” said Stadler. “Identifying whether a young patient’s cancer occurred in the setting of an inherited cancer predisposition syndrome is important as it can result in a substantial change in clinical management, such as increased cancer surveillance aimed at early detection and risk-reducing surgery to prevent new cancers and may even have reproductive implications for young families.”

To read the study abstract, click here.

To read a press release about the study, click here.
