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Cancer Genes

Genetic testing is now available for dozens of hereditary cancer syndromes, most of which are caused by alterations in genes that suppress tumor growth.

To date, the most commonly requested test is for BRCA1 and BRCA2. Alterations in these two genes predispose individuals to breast, ovarian, and other cancers.

Although these mutations can affect people of any race or ethnicity, they are found more frequently in families of Ashkenazi Jewish descent. Among female carriers, BRCA alterations may confer a lifetime breast cancer risk of up to 85 percent (though new research suggests breast cancer risk varies among families and may only be as high as about 50 percent) and an ovarian cancer risk of up to 50 percent. Male carriers also have increased risk for breast, prostate, and other cancers.

Other frequently used genetic tests are for two colon cancer syndromes: hereditary nonpolyposis colon cancer (HNPCC) and familial adenomatous polyposis (FAP). Both syndromes are associated with a high risk for colon cancer (80 percent and virtually 100 percent, respectively) and other cancers.

Recently, several familial syndromes have been linked to kidney cancer. In von Hippel-Lindau disease, patients inherit a defect in a specific gene called the von Hippel-Lindau gene, though people without the inherited disease can also have problems with the VHL gene. The VHL gene is a tumor suppressor gene, so inactivation of the gene can cause up to 80 percent of all sporadic (non-hereditary) clear-cell renal cell carcinomas, the most common type of kidney cancer. These discoveries provided researchers with a specific target for developing therapies to treat kidney cancer.

For those who test positive for a genetic mutation, increased screening is needed. Some patients may also consider chemoprevention strategies to help lower their risk of second cancers or recurrence. In other cases, cancer may be prevented altogether using prophylactic (preventive) surgeries.