smear does not check for uterine cancer. The results of Lisa’s uterine biopsy at that time came back clear.

Just three years after Kim’s diagnosis, older sister Lisa, just shy of her 50th birthday, began experiencing heavy menstrual bleeding.

Thinking back to Kim’s symptoms, Lisa immediately made an appointment with her gynecologist, who sent her for an ultrasonic that indicated Lisa had a large cyst on her left ovary and a smaller one on her right side. Lisa also requested to have second uterine biopsy since three years had passed.

“This time, the results of my biopsy were not clear. There was uterine cancer,” says Lisa. “I decided to see Dr. Hackett, since he had treated Kim.”

Dr. Hackett performed a robotic hysterectomy on Lisa and she was diagnosed with stage 1 uterine cancer.

Based upon the diagnosis of uterine cancer in both Kim and Lisa at an early age, Dr. Hackett referred the sisters to the High Risk Cancer Assessment Program at the Leon Hess Cancer Center at Monmouth Medical Center for genetic testing.

“In the time since Kim had been diagnosed, recommendations had been made that patients diagnosed with colon or uterine cancer under the age of 50 should routinely have their tumor screened for an inherited condition called Lynch Syndrome,” says Sherry Grumet, M.A., M.S., C.G.C., board-certified genetic counselor, who leads the High Risk Cancer Assessment program.

Lynch Syndrome, occasionally known as hereditary nonpolyposis colorectal cancer (HNPCC), involves mutations in the genes that are normally responsible for correcting mistakes when DNA is copied. If one parent carries a gene mutation for the condition, there’s a 50 percent chance that the mutation will be passed along to each child. As a result, entire families, immediate and extended, can be affected, and those with the syndrome have an increased risk of certain cancers, including colon and uterine cancer.

“Because Lisa was under 50 when diagnosed, a tumor screening was automatically done by our lab to test for this condition,” continued Sherry. “Her initial results were based on immunohistochemistry (IHC) testing, which uses special dyes to stain tumor tissue samples and looks for the presence or absence of proteins produced by specific genes. In Lisa’s case, four proteins from the Lynch syndrome genes were tested and two of these proteins were absent, which was a big red flag.”

Lisa’s initial test results, coupled with family histories that suggested the possibility of Lynch Syndrome, were enough to convince Lisa and Kim to consider genetic testing through the High Risk Cancer Assessment Program.

Testing through the High Risk Cancer Assessment Program typically includes a pre-test counseling session, during which family medical histories are reviewed and a genetic counselor discusses the risk of developing certain types of cancer, including hereditary cancer. When appropriate, genetic tests are completed, either by a mouth swab or a blood test, to detect altered cancer genes.

“In this case, genetic testing revealed a mutation in the MSH6 gene, confirming the diagnosis of Lynch Syndrome in the family,” said Sherry. “Based on these results, the sisters will be screened frequently for colon and urinary tract cancers. At-risk family members will be notified of the discovery of the mutation in the family and the availability of genetic counseling and testing.”

Sherry continued, “Lisa and Kim, as well as their extended family, will benefit from knowing their risk and be able to take steps to prevent other related types of cancers from developing.”

“With uterine cancer, there is never a sigh of relief,” said Kim. “But, early detection, knowing your risk and preventative measures are key to being sure it doesn’t go any further.”

The goal of the High Risk Cancer Assessment Program of the Leon Hess Cancer Center at Monmouth Medical Center is to help patients and their families who may be confronting cancer risks make informed decisions by providing appropriate medical and genetic information, social support and resources. If you think you may be a candidate for high risk counseling and testing, visit www.mmemcancer.com/genetic.htm or call 732.923.6513.