

Children genetically prone to cancer benefit from early standardized surveillance

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Recent advances in genomic sequencing have revealed that 5–15% of children with cancer have an underlying genetic predisposition. Although genetic predispositions increase the risk for new tumors, it has been unclear when to begin surveillance for these tumors.

St. Jude Children's Research Hospital scientists showed that beginning

[surveillance](#) soon after recognizing a patient's predisposition often led to discovering one or more early-stage asymptomatic tumors. About half of these tumors could be completely removed by surgery alone, thus avoiding the need for toxic chemotherapies or radiation therapies.

The findings inform [clinical practice](#) after [genetic testing](#) and were [published](#) in *JAMA Oncology*.

"We've shown that following standardized surveillance protocols provides a very effective way to detect new tumors at their earliest and most treatable stages," said corresponding author Kim Nichols, MD, St. Jude Division of Cancer Predisposition director, Department of Oncology.

"Understanding whether children have an underlying genetic risk for cancer can greatly impact their clinical care, and proactive surveillance for new tumors is an important component of this care."

Findings showed the benefit of early surveillance applied across multiple cancer types and predisposing conditions. The researchers examined 274 St. Jude [pediatric patients](#) with 35 different cancer-predisposing conditions over a median of three years. Surveillance revealed tumors in 27 patients, representing a broad array of solid and central nervous system neoplasms.

Early detection leads to better treatment

Conventional practice typically results in surveillance that begins months to years after detecting a genetic predisposition. Further, data on the effectiveness and outcomes of surveillance are sparse. Strikingly, most of the new cancers detected in the study were found with early surveillance right after the genetic predisposition was discovered.

Nearly one out of every three tumors were found at the first surveillance intervention soon after diagnosis, and two out of three were found within two years of that initial surveillance visit. Surprisingly, this included a small subset of children already being treated for a different cancer.

"About 17% of the children had a new tumor identified while they were still undergoing treatment for a prior cancer," Nichols said.

"Often, providers wait until a child has finished treatment for their first [cancer](#) before they start screening for other potential malignancies. If you do that, it is possible you're going to miss a new tumor. Providers should now recognize the importance of starting the recommended surveillance tests the moment they discover an underlying predisposition."

Beginning surveillance right away also resulted in finding cancers in earlier stages. Without the early screening, many of the tumors would have remained unnoticed until much later because almost all were asymptomatic at the time of discovery.

By finding these tumors at an early stage, oncologists could remove most of them by surgery alone. In addition, findings showed that most tumors (around 70%) were entirely removed without leaving even microscopic traces behind.

"We were able to remove these tumors without any evidence of disease left behind," Nichols said. "These patients will have better outcomes and may need less or even no chemotherapy or radiation therapy." Avoiding chemotherapy and radiation can prevent long-term treatment-related side effects, a poignant consideration when treating children.



(L to R) Co-first authors Alise Blake and Melissa Perrino, MD, St. Jude Department of Oncology and corresponding author Kim Nichols, MD, St. Jude Division of Cancer Predisposition director, Department of Oncology. Credit: St. Jude Children's Research Hospital

Modern cancer surveillance methods worked, when available

To find potential cancers and guide treatment, clinicians used various surveillance methods. For example, a predisposition for a solid [tumor](#) may lead to annual full-body magnetic resonance imaging (MRI). Standard surveillance methods had minuscule false positive and false negative rates.

"Overall, across these different reports and reviews of the relevant

images, there was an excellent performance of the different modalities," Nichols said.

"We had very few false positive or negative findings, meaning we did not need to pursue unnecessary interventions or miss any incipient tumors. Both factors are essential in optimizing the care for our patients with underlying predispositions."

The study may enable more providers to order these effective, yet sometimes expensive, tests. It can be challenging for providers to get insurance companies to pay for early screening.

Historically, this has created a difficult cycle where providers could not order the tests, which meant there was no strong evidence of their value, preventing their use. Patients at St. Jude are treated regardless of insurance coverage, thus enabling physicians to order the appropriate tests.

The excellent performance of the standard screening methods is evidence that early testing can improve detection and outcomes. This may give insurance companies the evidence needed to cover the tests for children with a predisposition.

"I hope that providers will initiate recommended surveillance as soon as they learn that someone has an underlying predisposition," Nichols said.

"They shouldn't hesitate to start looking as soon as they learn about the [predisposition](#), and it's imperative to follow through with that surveillance. Then, we can find these tumors early, while they are potentially easier to treat."

More information: Alise Blake et al, Performance of Tumor Surveillance for Children With Cancer Predisposition, *JAMA Oncology* (2024). [DOI: 10.1001/jamaoncol.2024.1878](https://doi.org/10.1001/jamaoncol.2024.1878)

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