

# One single genetic test for all children with cancer improves diagnosis and treatment

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Analysis of the entire tumor RNA picks up more clinically relevant genetic changes in children with cancer than traditional diagnostic methods, new research has shown.

A piece of [tumor](#) tissue is taken from all [children](#) with cancer to determine the exact form of their disease. The piece of tissue is looked at under the microscope, but also analyzed for genetic faults. Changes in both DNA and RNA—a translation of the DNA code—can provide important clues about the exact cancer type, the aggressiveness of the tumor, and the possible benefit of targeted drugs.

## Replacing classical tests

All available RNA in a piece of tumor tissue from every child with cancer in the Netherlands is studied using so-called "RNA sequencing" – currently in addition to traditional diagnostics. This was made possible by the opening of the Princess Máxima Center for [pediatric oncology](#) in 2018, the research hospital where all children with cancer in the Netherlands are treated.

In some children, RNA sequencing has already led to improved diagnosis and adapted treatment. For a large number of cancer types, the Diagnostic Lab in the Princess Máxima Center has already replaced the traditional tests with broad RNA analyses. The researchers expect that they will also be able to switch completely quickly for the other cancer types—especially forms of leukemia.

## Fusion genes

In a new study, scientists at the Princess Máxima Center compared the effectiveness of RNA sequencing with traditional methods that allow you to specifically search the DNA and RNA for known gene changes. The study was published today (Thursday) in the journal *JCO Precision Oncology*, and was funded by NWO, KiKa and the Adessium Foundation.

The team analyzed tissue samples from 244 children referred to the Princess Máxima Center between late 2018 and mid-2019 after suspected cancer. They focused on picking up so-called fusion [genes](#)—a kind of genetic fault where two separate genes together form one new, faulty gene. Such fusion genes are found in many cancers and can often influence treatment decisions.

Using RNA sequencing, the team picked out a total of 78 fusion genes. That was 23—or 40%—more than they found using traditional techniques. Traditional methods did find the other 55 [fusion genes](#)—but often only one of the two genes was picked up from the fusion, while information from both genes can be important for diagnosis or treatment.

## **More accurate diagnosis**

In almost a third of the 23 RNA sequencing specific gene fusions, the finding led to a more accurate diagnosis or possible treatment. In the case of one girl, the pathologist's diagnosis was modified on the basis of RNA sequencing to infantile fibrosarcoma, a soft tissue tumor, with a so-called NTRK fusion. This meant she was able to receive a new targeted drug as part of a clinical trial.

The diagnosis of a one-year-old boy with a brain tumor was changed from glioblastoma to hemispheric glioma, a tumor with a less bad outcome. When standard treatment stopped working, he was given a

precision medicine that kept him stable for another year.

For a third of the 23 abnormalities 'missed' by traditional diagnostics, no specific tests currently exists. For the other missed gene changes, technical reasons played a role—or the relevant test was simply not requested because it did not seem relevant for the tumor type.

Dr. Bastiaan Tops, head of the Diagnostic Lab at the Princess Máxima Center for pediatric oncology, and co-leader of the study, says that "RNA sequencing was already used before, but only in children who were very ill, and for whom standard treatment had stopped working. In our research hospital setting at the Princess Máxima Center, we have implemented RNA sequencing into standard diagnostics. Our new study shows that this approach is paying off."

"Because we can look at the full genetic landscape of a child's tumor at diagnosis, we can discuss possible consequences for treatment with the child's doctor right away. That means we can offer children with cancer the very best opportunities, based on the latest scientific insights."

Dr. Patrick Kemmeren, group leader and head of the Big Data Core at the Princess Máxima Center for Pediatric Oncology, and co-leader of the study, says that "in this study, we show that a single test that searches the entire tumor RNA is almost one and a half times more sensitive to genetic faults in childhood cancer. I expect that the test we have developed will replace the various traditional methods in the foreseeable future."

"In my group, we conduct much broader research into DNA and RNA abnormalities in childhood cancer. If we discover new abnormalities, they can also be immediately included as part of new diagnoses—and even tested retrospectively in children who are already under treatment. In this way, children with [cancer](#) benefit as quickly as possible from new

findings within fundamental research."

**More information:** Improved Gene Fusion Detection in Childhood Cancer Diagnostics Using RNA Sequencing, *JCO Precision Oncology*, DOI: [10.1200/PO.20.00504](https://doi.org/10.1200/PO.20.00504)

Provided by Princess Máxima Center for Pediatric Oncology

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