New genetic signatures in childhood leukemia create paths for precision medicine

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Erin Crowgey, PhD. Credit: Nemours Children's Health System

Researchers with Nemours Children's Health System utilized Next Generation Sequencing (NGS) to more precisely identify genomic characteristics of leukemias in children, the most common childhood cancer. The study, published today in *BMC Medical Genomics*, identified new genetic structural variants that could be used to assess the presence of minimal residual disease during the course of chemotherapy and help determine response to various therapies.

"Progress in the field of genomic research and advanced technology has allowed us to find new variants that can better target treatments for kids with cancer," said Erin Crowgey, Ph.D., lead author of the study and Director of Clinical Bioinformatics at Nemours. "Pediatric leukemias have a diverse and complex genomic structure, and older sequencing techniques were missing a lot of the important information that guides our clinical evaluation, risk identification, and therapeutic strategy for patients."

Researchers analyzed DNA and RNA from bone marrow, cell lines, and umbilical cord samples from 32 pediatric leukemia patients and five adult leukemia patients. Patient samples were collected at diagnosis, end of the first treatment, and relapse. These samples were sequenced using molecular barcoding with targeted DNA and RNA library enrichment techniques. The sequencing identified multiple novel gene fusions and previously unknown copy number losses in leukemia genes. This approach enabled more sensitive detection of these genetic variants.

"Hearing that your child has cancer is scary; particularly that the clinical presentation of his disease is unique and there is no other research that matches his case," said Sophie Hayes, mother of Eliot Hayes, a patient at Nemours. "So, when the genomic results came back and showed that Eliot's cancer was not unique at the genetic level, we were very grateful to the scientific community and to Nemours. This early knowledge enabled us to incorporate an additional line of attack into his treatment

and offered us hope."

Researchers note that the ultimate goal is to bring the genomic testing into routine clinical practice so that it can be used regularly, leading to a pediatric onco-specific genomic treatment program at Nemours. "Dr. Crowgey's work brings complex genomic testing closer to the bedside to improve the treatment of children with cancer," said E. Anders Kolb, MD, Director of Nemours Center for Cancer and Blood Disorders and a co-author of the paper.

More information: Erin L. Crowgey et al, Error-corrected sequencing strategies enable comprehensive detection of leukemic mutations relevant for diagnosis and minimal residual disease monitoring, *BMC Medical Genomics* (2020). DOI: 10.1186/s12920-020-0671-8

Provided by Nemours Children's Health System

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