Uncovering Genetic Foundations of Pediatric Liver Cancer

Researchers identify a number of genes that contribute to the development of liver cancer in Beckwith-Wiedemann Syndrome.

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Last fall, we published the story of Damon Runyon Clinical Investigator Jennifer M. Kalish, MD, PhD, a pediatric geneticist at the Children's Hospital of Philadelphia who has dedicated her career to the study of Beckwith-Wiedemann Syndrome (BWS), a rare genetic condition that causes overgrowth in certain parts of the body and predisposes children to cancers of the kidney and liver. As Founding Director of the hospital's Beckwith-Wiedemann Syndrome Clinic, Dr. Kalish established the country's first and only active BWS patient registry and biorepository storing blood and tissue samples necessary for research. In December 2020, her lab unveiled the first human cell-based model of the syndrome, developed using cells from patients in the registry.

Researchers know that BWS is caused by structural and epigenetic changes (how the DNA is "packaged") to chromosome 11. Dr. Kalish's research aims to uncover the genetic mechanisms by which BWS develops into pediatric cancers. "Once we figure out the pathways that lead to overgrowth and tumorigenesis, we can figure out how to intervene," she explained. "Then we can do screenings, but also provide a treatment to prevent the transition. That's the ultimate goal."

Now, a new paper from her lab represents an important step toward that goal. Using liver cancer samples collected from BWS patients, Dr. Kalish's team has performed the first multi-omic investigation (i.e., involving multiple data sets, including DNA sequencing, RNA sequencing, and epigenetic factors) of these tumors. By comparing BWS liver samples, both cancerous and not, to healthy liver samples, the researchers were able to identify a number of genes that contribute to the development of liver cancer in BWS patients. These genes, dubbed "the BWS oncogenesis network," can now be investigated as potential diagnostic or therapeutic targets.

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