

Cancer researchers link DICER1 gene mutation to rare childhood cancer

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Research published today in *Science Express* from the journal *Science* demonstrates the first definitive link between mutations in the gene DICER1 and cancer. By studying the patterns of DNA from 11 families with an unusual predisposition to the rare childhood lung cancer pleuropulmonary blastoma (PPB) investigators found that children with the cancer carried a mutation in one of their two DICER1 gene copies.

DICER1 makes an important protein that works to suppress other genes through intermediary molecules known as microRNAs. Scientists have learned that microRNAs can fine-tune the expression of many other genes, which is particularly important in normal human development. Recent research has also focused on DICER1 as having a potential role in [cancer](#) because the micro-RNA molecules it produces appear vastly different from normal when found in cancer [cells](#); some suggest that the pattern of microRNAs in cancers resembles an embryonic stage.

"When we realized that DICER1 was in the segment of chromosome that was shared among children with PPB we were very excited," said D. Ashley Hill, MD, lead author and chief of Pathology at Children's National Medical Center.

"PPB is a tumor that appears to arise out of a localized area of abnormal lung development. The implications of a defect in a master controller gene for normal organ development would be significant."

Hill says not everyone who inherits a mutation develops PPB and

children with PPB are typically normal in every other way. The team theorizes that something else must happen to the normal copy of DICER1 in [lung cells](#) for a tumor to develop. When the research team looked at PPB tumors to see if there is any DICER1 protein being made from the remaining normal copy of the gene, they were surprised by the results: "We expected to see that the tumor cells had no DICER1 protein giving us a nice explanation for why the tumor cells had gone haywire." But that wasn't the case.

They found that the benign cells that grew on the surface of the tumor had lost the DICER1 protein. "During [lung development](#) the cells that line the airways and the cells that make up the support structures have to communicate." Hill explained. "Loss of DICER1 in the airway lining cells could disrupt this communication, possibly setting the stage for abnormal growth."

Studying [tumor cells](#) for genetic [mutations](#) has led to many advances in the understanding of cancer. This discovery may represent a first step in understanding a new mechanism for how cancer begins. Conceivably, the mutated cells do not turn into tumors themselves. Instead, these cells influence surrounding cells to grow quickly, setting the stage for additional genetic mutations in cells that then become cancerous. Hill's team will now focus on demonstrating the sequence of events that result from loss of DICER1.

Only 50 or 60 cases of PPB are diagnosed worldwide each year. The cancer presents as cysts in early stages and progresses to solid lung tumors over time. If detected in the earliest stages, 90 percent of patients appear to be cured when treated with a surgical intervention and sometimes chemotherapy.

Source: Children's National Medical Center ([news](#) : [web](#))

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