

# Major birth defects associated with moderately increased cancer risk in children

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A multistate study led by researchers at the University of Utah has revealed that the risk for childhood cancer is moderately increased among children and young adolescents with certain types of major birth defects. Children born with non-chromosomal birth defects have a twofold higher risk of cancer before age 15, compared to children born without birth defects, according to this study published in July in *PLOS ONE*. However, cancer risk varies by the specific type of birth defect, and is not significantly increased in many of the more common birth defects.

An estimated 3 percent of U.S. babies, approximately 120,000, are born with major birth defects each year. In many countries, including the United States, birth defects are the leading cause of infant death and a major contributor to disability and pediatric hospitalizations. Birth defects are an increasing health concern worldwide and, in 2010, the World Health Organization identified birth defect prevention and care as a global priority.

"There is a large body of evidence for increased [cancer risk](#) in children with Down's syndrome, a genetic birth defect caused by the presence of an extra copy of [chromosome 21](#)," says Lorenzo Botto, M.D., professor of pediatrics at the University of Utah School of Medicine and first author on the study. "However, studies to date have provided inconsistent findings on cancer risk in children with structural birth defects that are not caused by [chromosome abnormalities](#)."

In this study, funded by the U.S. Centers for Disease Control and Prevention (CDC), Botto and his colleagues analyzed information from birth defect and cancer surveillance programs in Utah, Arizona and Iowa and observed that cancer risk was increased in children born with [eye defects](#), [cleft palate](#), some heart and kidney defects, and microcephaly, a condition where the head is smaller than normal. The types of cancer for which risk was increased included neuroblastoma, retinoblastoma, and hepatoblastoma, all cancers that arise from immature cells and typically develop in early childhood.

"While there is an increased risk for cancer in young people with certain types of birth defects compared to children without birth defects, the overall cancer risk for a child with a birth defect is still relatively low, so it is important for health care providers to be careful not to produce unnecessary concern among parents and families," says Botto. "In addition, we found that the incidence of cancer was highest in the first three to five years of life, so clinical surveillance can be focused by age, as well as by birth defect."

The scientists also observed that cancer risk was 14 times higher among children with Down syndrome, mainly due to leukemias. However, cancer risk was not increased with many common birth defects, including cleft lip, hydrocephalus, and hypospadias. Hydrocephalus is an abnormal buildup of fluid in the brain and hypospadias is a birth defect in which the opening of the tube that drains urine from the bladder is on the underside, rather than at the tip, of the penis.

"It's reassuring that many of the common major birth defects are not associated with any increase in cancer risk," says Botto. "Our study helps to identify who is, and who is not, at increased risk for cancer, and this information can be used to focus future research on potential genetic or environmental factors that contribute to cancer risk."

This study was conducted in collaboration with Timothy Flood, M.D., from the Arizona Department of Health Services; Julian Little, Ph.D., from the University of Ottawa; and Paul Romitti, Ph.D., from The University of Iowa College of Public Health.

"A large, population-based study like this one would not have been possible without access to the high-quality birth defect and cancer [surveillance programs](#) in the states of Utah, Arizona, and Iowa," says Botto. "Thanks to the support of the CDC and the work of generations of people who recognized the importance of collecting health data, we were able to conduct a rigorous study with findings that could potentially translate into better care and long-term outcomes for young people with birth defects."

Provided by University of Utah Health Sciences

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