# Study examines genetic defects linked to body abnormalities in patients with childhood cancer

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Children with cancer have a higher prevalence of body abnormalities, such as asymmetric lower limbs and curvature of the spine, suggesting that the genetic defect responsible for the abnormality may play a role in the development of cancer, according to a study in the January 2 issue of JAMA.

Certain genetic syndromes can be associated with an increased risk for tumor and cancer development in children. Several studies have shown that developmental genes, which play a role in body plan formation during embryogenesis, are also involved in the development of cancer, according to background information in the article.

Johannes H. M. Merks, M.D., Ph.D., of Emma Children's Hospital, Academic Medical Center, Amsterdam, the Netherlands, and colleagues conducted a study to assess the prevalence of morphological (body structure) abnormalities in a large group of childhood cancer patients. The study, conducted between January 2000 and March 2003, included 1,073 patients who underwent a physical examination for morphological abnormalities (such as differing length in limbs; broad hands or feet; prominent ears; curvature of the spine). The patient group consisted of 898 long-term survivors of childhood cancer and 175 newly diagnosed pediatric patients with cancer. The control group consisted of 1,007 schoolchildren examined in an identical way. The average ages of patients and controls were 21.2 and 10.4 years, respectively.

The researchers found that both major abnormalities and minor anomalies were significantly more prevalent in the pediatric cancer group (per 1,000 cases, patients had 268 major abnormalities and controls had 155 abnormalities). One or more major abnormalities were present in 26.8 percent of individual patients (15.5 percent in controls), two or more abnormalities in 5.1 percent of patients (1.6 percent in controls), and three or more abnormalities were found in 0.9 percent compared with none in controls.

One or more minor anomalies were found in 65.1 percent of individual patients (56.2 percent in controls), two or more minor anomalies in 32.8 percent of patients (22.1 percent in controls), and three or more minor anomalies were found in 15.2 percent of patients compared with 8.3 percent in controls.

In 42 patients (3.9 percent), an established clinical genetic syndrome was diagnosed. Analysis showed 14 age-independent morphological abnormalities that were independently and significantly associated with childhood cancer. For two of these (blepharophimosis [eyelid abnormalities] and asymmetric lower limbs), the researchers identified statistically significant patterns of co-occurring morphological abnormalities suggestive of new tumor predisposition syndromes. Thirty-four patients fit one of the two tumor predisposition patterns.

Source: JAMA and Archives Journals