Study finds association between genetic mutation and age at diagnosis for common childhood cancer

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Certain mutations of the gene ATRX were associated with age at diagnosis in children and young adults with advanced-stage neuroblastoma, a cancer that grows in parts of the nervous system, according to a study in the March 14 issue of *JAMA*.

Neuroblastoma is the most common extracranial (outside the cranium) solid tumor of childhood and accounts for 15 percent of all cancer-related deaths in children. "Half of the patients (50 percent) with neuroblastoma present with metastatic disease; with current treatment approaches, the age at diagnosis has proven to be one of the most powerful predictors of outcome. The probability of overall survival is 88 percent in infants [age: less than 18 months at time of diagnosis], 49 percent in children [age: 18 months – less than 12 years], and only 10 percent in adolescents or young adults [age: 12 years or older]," according to background information in the article. "Genetic mutations associated with neuroblastoma and its clinical course are not completely understood."

Nai-Kong V. Cheung, M.D., Ph.D., of the Memorial Sloan-Kettering Cancer Center, New York, and colleagues conducted a study to identify genetic mutations that are associated with age at diagnosis in patients with metastatic neuroblastoma. Whole genome sequencing was performed of DNA from diagnostic tumors and their matched germlines (those cells of an individual that have genetic material that could be

passed to offspring) from 40 patients with metastatic neuroblastoma obtained between 1987 and 2009. Age groups at diagnosis included infants (0-