Bloom syndrome

Aflac Cancer & Blood Disorders Center
CHILDREN'S HEALTHCARE OF ATLANTA

Bloom syndrome is a genetic disorder characterized by short, but proportional, stature; hypersensitivity to sunlight, including a mnemonic facial butterfly rash; and a predisposition to leukemia, lymphoma and Wilms tumor during childhood.

Cause

Bloom syndrome is caused by genetic changes in the BLM gene, which encodes for a protein that acts as a "caregiver" of cell processes and helps repair cell damage. For an individual to be affected, he must inherit two genetic changes, or misspellings, one from each parent.

Diagnosis

Bloom syndrome can be diagnosed through genetic testing, including molecular and cytogenetic techniques. The diagnostic process is often facilitated by a genetic counselor and physician who specialize in hematology/oncology. The genetic counselor will help identify at-risk siblings and/or relatives in the family.

Additionally, genetic testing can be performed prior to pregnancy if one partner is a known carrier of genetic changes in the BLM gene. Carriers do not have Bloom syndrome.

Treatment

The two main strategies in regards to risk reduction and treatment of Bloom syndrome are to avoid exposure to UV light and radiation, and have a surveillance plan in place. A surveillance plan can be designed by the child's main healthcare provider, which is often a pediatric oncologist or other provider with expertise in children who have Bloom syndrome.

Cancer treatment can be complicated, as individuals with Bloom syndrome are extremely sensitive to radiation and chemotherapy.

Resource

Bloom Syndrome Foundation
bloomssyndrome.org

References

ncbi.nlm.nih.gov/books/nbk1398/
weill.cornell.edu/bsr/bloomsyndromeregistry