Bloom syndrome

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