Rhabdoid Predisposition syndrome

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

Rhabdoid Predisposition syndrome is a genetic condition in which individuals are born with an increased likelihood of developing rhabdoid tumors that develop primarily in the kidneys and brain, but can also develop elsewhere in the body.

Cause

This syndrome is caused by germline changes in the SMARCA4 and SMARCB1 genes. Individuals with Rhabdoid Predisposition syndrome are born with these changes and in about 40 percent of cases are passed down from one parent. Both of these genes act as organizers of the genome, specifically chromatic structuring and packing, or chromatin remodeling.

Diagnosis

Rhabdoid Predisposition syndrome can be diagnosed through genetic testing, which is often facilitated by a genetic counselor or genetics services provider.

Surveillance

Guidelines exist for surveillance of children and adults with Rhabdoid Predisposition syndrome.

References

ncbi.nlm.nih.gov/pubmed/25494491