AMER1 Gene

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APC membrane recruitment protein 1

genes

1. Normal Function

The *AMER1* gene provides instructions for making a protein found in tissues throughout the body where it helps regulate the Wnt signaling pathway, which is a series of chemical signals that affect the way cells and tissues develop. Wnt signaling is important for cell division, attachment of cells to one another (adhesion), cell movement (migration), and many other cell activities. The AMER1 protein can promote cell growth by helping to turn on (activate) the Wnt pathway or prevent cell growth by helping to turn off (repress) the Wnt pathway. When repressing the pathway, the AMER1 protein is acting as a tumor suppressor, which means that it helps prevent cells from growing and dividing (proliferating) too rapidly or in an uncontrolled way.

2. Health Conditions Related to Genetic Changes

2.1. Wilms tumor

Mutations in the *AMER1* gene have been found in Wilms tumor, a rare form of kidney cancer that occurs almost exclusively in children. These mutations are somatic, meaning that they are acquired during a person's lifetime and are present only in kidney cells that give rise to the tumor. *AMER1* gene mutations result in a protein with a reduced ability to repress Wnt signaling. As a result, Wnt signaling is increased, which leads to the unchecked proliferation of kidney cells and tumor development.

2.2. Other disorders

Mutations in the *AMER1* gene that are present in cells throughout the body (called germline mutations) cause a bone disorder called osteopathia striata with cranial sclerosis. This condition occurs almost exclusively in females because males with the disorder usually die before or soon after birth. Affected females have excessive bone growth (hyperostosis), which leads to multiple skeletal abnormalities including an unusually large head (macrocephaly) and abnormal facial features. Males who survive infancy have bone abnormalities and heart, gastrointestinal, and genitourinary malformations. *AMER1* gene mutations that cause osteopathia striata with

cranial sclerosis lead to a lack of functional AMER1 protein, disrupting its role in regulating the Wnt signaling pathway. It is unclear why these mutations primarily affect skeletal development.

2.3. Other cancers

In addition to Wilms tumor (described above), changes in the *AMER1* gene have been reported to be associated with many other cancers. These mutations are somatic and are present only in cells that give rise to cancer. Studies have shown that *AMER1* gene changes are associated with stomach (gastric), breast, and colorectal cancers. It is likely that these gene changes impair the protein's tumor suppressor function, allowing cells to proliferate without control or order, which leads to cancer development.

3. Other Names for This Gene

- adenomatous polyposis coli membrane recruitment 1
- FAM123B
- family with sequence similarity 123B
- FLJ39827
- OSCS
- protein FAM123B
- RP11-403E24.2
- Wilms tumor gene on the X chromosome protein
- Wilms tumor on the X
- WTX

References

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