YY1AP1 Gene

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YY1 associated protein 1: the YY1AP1 gene provides instructions for making part of a group of associated proteins known as the INO80 chromatin remodeling complex.

Keywords: genes

1. Normal Function

In the cell nucleus, this complex attaches (binds) to chromatin, which is the network of DNA and proteins that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling by the INO80 chromatin remodeling complex allows cells to control the activity (expression) of certain genes. This complex also helps regulate several other critical cell functions, including repair of damaged DNA, cell specialization (differentiation), and cell growth and division (proliferation). Activity of the INO80 chromatin remodeling complex appears to be particularly important in smooth muscle cells, which line the walls of blood vessels.

2. Health Conditions Related to Genetic Changes

2.1. Grange syndrome

At least five mutations in the *YY1AP1* gene have been identified in people with Grange syndrome. This rare condition causes narrowing (stenosis) and blockage (occlusion) of blood vessels that supply blood to many organs and tissues, including the kidneys, brain, and heart. Grange syndrome can also be associated with short fingers and toes (brachydactyly), fusion of some of the fingers or toes (syndactyly), fragile bones that are prone to breakage, learning disabilities, and heart defects.

Mutations in the *YY1AP1* gene are described as "loss-of-function" because they prevent the production of any functional YY1AP1 protein. A loss of this protein disrupts the function of the INO80 chromatin remodeling complex, which alters the activity of multiple genes in smooth muscle cells. Researchers suspect that these changes in gene expression lead to reduced cell proliferation and differentiation. However, it is unclear how these changes cause narrowing and blockage of arteries. It is also unknown how *YY1AP1* gene mutations are related to other features of Grange syndrome, such as bone abnormalities and learning disabilities.

3. Other Names for This Gene

- GRNG
- HCCA1
- HCCA2
- hepatocellular carcinoma susceptibility protein
- hepatocellular carcinoma-associated protein 2
- YAP
- YY1AP

References

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