SOX10 Gene

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SRY-box 10

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1. Normal Function

The *SOX10* gene belongs to a family of genes that plays a critical role in the formation of tissues and organs during embryonic development. The SOX gene family also maintains the normal function of certain cells after birth. To carry out these roles, proteins made by genes in the SOX family bind to specific areas of DNA. By attaching to critical regions near genes, SOX proteins help control the activity of those genes. SOX proteins are called transcription factors on the basis of this action.

During embryonic development, the *SOX10* gene is active in cells called neural crest cells. These cells migrate from the developing spinal cord to specific regions in the embryo, where they give rise to many different types of cells. The protein made by the *SOX10* gene directs the activity of other genes (such as *MITF*) that signal neural crest cells to become more specific cell types. In particular, the SOX10 protein is essential for the formation of nerves in the intestine (enteric nerves) and for the production of specialized cells called melanocytes. Melanocytes produce melanin, a pigment that contributes to skin, hair, and eye color. Melanin is also involved in the normal function of the inner ear.

2. Health Conditions Related to Genetic Changes

2.1. Waardenburg syndrome

More than 30 mutations in the *SOX10* gene have been identified in people with Waardenburg syndrome type II and type IV (also known as Waardenburg-Shah syndrome). Both types of Waardenburg syndrome are characterized by changes in skin, hair, and eye coloring and hearing loss. People with type IV also have an intestinal disorder called Hirschsprung disease that causes severe constipation or intestinal blockage. Most *SOX10* mutations lead to the production of an abnormal version of the SOX10 protein or prevent the gene from making any protein. An abnormal or missing SOX10 protein cannot control genes that signal neural crest cells to become specific cell types. As a result, enteric nerves and melanocytes do not form normally during embryonic development. Missing enteric nerves in certain parts of the intestine cause the signs and symptoms of Hirschsprung disease. A lack of melanocytes affects the coloring of skin, hair, and eyes and causes the hearing loss characteristic of Waardenburg syndrome.

Researchers have found that mutations in the *SOX10* gene also cause a similar disorder known as peripheral demyelinating neuropathy, central demyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease (PCWH). This rare condition is a variant of Waardenburg syndrome type IV that also affects other parts of the nervous system. Like mutations that cause other types of Waardenburg syndrome, the mutations responsible for PCWH lead to the production of an abnormal version of the SOX10 protein that is unable to direct the activity of other genes.

3. Other Names for This Gene

- DOM
- dominant megacolon, mouse, human homolog of
- SOX10_HUMAN
- SRY (sex determining region Y)-box 10
- SRY box 10
- SRY-related HMG-box gene 10
- transcription factor SOX-10

- WS4

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