TSHB Gene

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thyroid stimulating hormone beta

genes

1. Normal Function

The *TSHB* gene provides instructions for one piece (subunit) of a hormone called thyroid stimulating hormone (TSH). Thyroid stimulating hormone consists of two subunits called alpha and beta. The *TSHB* gene provides instructions for making the beta subunit. The alpha and beta subunits are bound together to produce the active form of the hormone. A particular segment of the beta subunit, known as the buckle or seatbelt, wraps around the alpha subunit to form the active hormone. This seatbelt region also helps stabilize the hormone's structure.

Thyroid stimulating hormone is made in the pituitary gland, a gland at the base of the brain. This hormone plays an important role in the growth and function of the thyroid gland, a butterfly-shaped tissue in the lower neck. It also stimulates the production of thyroid hormones, which play a critical role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism). The pituitary gland monitors levels of thyroid hormones. When thyroid hormone levels are too low, the pituitary gland releases thyroid stimulating hormone into the bloodstream. Thyroid stimulating hormone, in turn, signals increased thyroid gland growth and production of thyroid hormones.

2. Health Conditions Related to Genetic Changes

2.1. Congenital Hypothyroidism

Researchers have identified at least 10 *TSHB* gene mutations involved in congenital hypothyroidism, a condition characterized by abnormally low levels of thyroid hormones starting from birth. *TSHB* gene mutations are the primary cause of a form of the condition called central congenital hypothyroidism, which occurs when stimulation of thyroid hormone production by the pituitary gland is impaired.

TSHB gene mutations involved in congenital hypothyroidism alter the size or shape of the thyroid stimulating hormone beta subunit. Many of the mutations affect the beta subunit's seatbelt region. Some mutations severely shorten the beta subunit, eliminating the seatbelt region partially or entirely. Other mutations change the protein building blocks (amino acids) used to make the beta subunit. As a result, the seatbelt region cannot buckle around

the alpha subunit. *TSHB* gene mutations prevent the production of functional thyroid stimulating hormone or its release (secretion) from the pituitary gland. As a result, thyroid hormone production is not stimulated, leading to low hormone levels that are characteristic of congenital hypothyroidism. Additionally, the thyroid gland is reduced in size (hypoplastic) because its growth is not stimulated.

3. Other Names for This Gene

- thyroid stimulating hormone, beta
- thyrotropin beta chain precursor
- thyrotropin beta subunit
- TSH-BETA
- TSHB_HUMAN

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