



Impact of *HSD17B3* Mutations on Hormone Balance in Genetic Insights

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DESCRIPTION

The *HSD17B3* gene is a critically important gene within the human body, playing a key role in the metabolism of hormones and cholesterol. It encodes an enzyme known as 3-beta hydroxysteroid dehydrogenase type 2, which is responsible for converting certain hormones into their active forms. This process is essential for normal sex hormone balance, fertility, and cholesterol regulation. Mutations in the *HSD17B3* gene may lead to developmental disorders or diseases related to steroid metabolism. These mutations have been associated with infertility and metabolic disorders due to changes in hormone levels caused by alterations in enzyme activity. By exploring the molecular pathways of these two novel mutations, we can gain valuable insight into how they disrupt normal hormone metabolism and contribute to disease development.

To determine two novel mutations of the *HSD17B3* gene, this is responsible for encoding an enzyme responsible for fatty acid metabolism. This new information has provided insight into a possible link between genetic mutation and fatty acid metabolism-related illnesses. In order to understand this connection and uncover the molecular pathways to exploring the impact of these mutations on cell functioning. The first mutation, termed R160X, is a result of a frame shift caused by a single base pair deletion in the coding sequence of *HSD17B3*. This results in an early termination codon that causes any translation initiated at the site to halt prematurely and eliminates the functional protein from being expressed. Similarly, the second mutation, termed 519A>G, is also a frame shift but is caused by a single base pair substitution instead from adenine to guanine. As with its predecessor, this alteration also produces an early termination codon leading to failed protein expression. To assess how these mutations influence cellular activity, we are utilizing yeast cells as their model system. They will then be able to characterize any changes in enzyme function or production as well as compare transcript levels associated with this gene and its related enzymes before and after introduction of either mutation. Recently it has identified two novel mutations

in the *HSD17B3* gene. These mutations have been associated with certain forms of male-pattern baldness and alopecia, as well as a higher risk of developing cancer due to changes in metabolism. In order to better understand their impact on cellular processes, we must explore their molecular pathways in detail. The *HSD17B3* gene provides instructions for making an enzyme called 3β-hydroxysteroid dehydrogenase type 3 (*HSD17B3*). This enzyme is involved in many different metabolic pathways within cells, such as those related to steroid hormone synthesis and breakdown. The two novel mutations of this gene are classified as missense mutations, meaning they cause a change in one amino acid along the entire protein chain. In terms of their effects on metabolic pathways, one mutation leads to an increase in fatty acid metabolism by activating the expression of genes related to fatty acid oxidation. The other mutation increases glycogenolysis, which is the breakdown of glycogen into glucose for energy production. Both mutations can potentially lead to increased DNA damage and cancer susceptibility due to changes in metabolic pathways within cells. In addition, these mutations can also have indirect effects on other cellular processes. For example, they can alter the levels of certain hormones like testosterone or estrogen which could impact hair growth or development of certain cancers associated with hormone levels. Overall, further exploration into the molecular pathways associated with these two novel mutations will help us gain a better understanding of their impact on cellular processes and provide insight into potential treatments for male-pattern baldness and alopecia. The molecular pathways of two novel mutations in the *HSD17B3* gene has been a valuable exercise and is an important step in understanding the genetic basis for diseases related to *HSD17B3*. Through examining *HSD17B3*'s structure, its interactions with other molecules, and its effect on protein levels throughout the body, we can gain a better understanding of how these mutations could affect human health. This information is invaluable when it comes to developing potential treatments or therapies that might be used to address any negative health effects associated with *HSD17B3* mutations.

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