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Congrnital Leptin Deficiency: Diagnosis and Therapy

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Editorial

Congenital leptin deficiency may be a condition that causes severe obesity beginning within the first few months of life. Affected individuals are of normal weight at birth, but they're constantly hungry and quickly gain weight. Without treatment, the acute hunger continues and results in chronic excessive eating (Hyperphagia) and obesity. Beginning in infancy, affected individuals develop abnormal eating behaviors like fighting with other children over food, hoarding food, and eating secretly.

People with congenital leptin deficiency even have hypogonadotropic hypogonadism, which may be a condition caused by reduced production of hormones that direct sexual development. Without treatment, affected individuals experience delayed puberty or don't undergo puberty, and should be unable to conceive children (infertile) [1,2].

Congenital leptin deficiency is caused by mutations within the LEP gene. This gene provides instructions for creating a hormone called leptin, which is involved within the regulation of weight. Normally, the body's fat cells release leptin in proportion to their size. As fat accumulates in cells, more leptin is produced. This rise in leptin indicates that fat stores are increasing [3].

Leptin attaches (binds) to and activates a protein called the leptin receptor, fitting into the receptor sort of a key into a lock. The leptin receptor protein is found on the surface of cells in many organs and tissues of the body including a neighborhood of the brain called the hypothalamus. The hypothalamus controls hunger and thirst also as other functions like sleep, moods, and blood heat. It also regulates the discharge of the many hormones that have functions throughout the body. Within the hypothalamus, the binding of leptin to its receptor triggers a series of chemical signals that affect hunger and help produce a sense of fullness (satiety).

LEP gene mutations that cause congenital leptin deficiency cause an absence of leptin. As a result, the signaling that triggers feelings of satiety doesn't occur, resulting in the excessive hunger and weight gain related to this disorder. Because hypogonadotropic hypogonadism occurs in congenital leptin deficiency, researchers suggest that leptin signaling is additionally involved in regulating the hormones that control sexual development. However, the specifics of this involvement and the way it's going to be altered in congenital leptin deficiency are unknown [3].

Congenital leptin deficiency may be a rare explanation for obesity. Researchers are studying the factors involved in additional common sorts of obesity.

Additionally, leptin remains a potentially forthcoming treatment for several other states of energy deprivation including anorexia or milder sorts of hypothalamic amenorrhea pending appropriate clinical trials.

Leptin replacement therapy reverses endocrine and metabolic alterations related to leptin deficiency. A number of these results could also be extrapolated to other diseases.

Leptin replacement therapy is currently available through a compassionate use program for congenital complete leptin deficiency and under an expanded access program to subjects with leptin deficiency related to congenital or acquired lipoatrophy. Additionally, leptin remains a potentially forthcoming treatment for several other states of energy deprivation including anorexia or milder sorts of hypothalamic amenorrhea pending appropriate clinical trials [4].

References

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