Commentary

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A Short Commentary on Tay-Sachs disease

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Introduction

Tay-Sachs disease is a disease of lysosomal retention acquired by the ubiquitous lysosomal acid hydrolase deficiency, hexosaminidase A (HexA). Enzyme deficiency primarily contributes to the accumulation of one of its substrates, GM2 ganglioside in neuronal cells leads to the death of apoptotic cells in the central nervous system. The disease varies clinically, with severe forms showing severe mental retardation, and death within 2-3 years of birth.

HexA is one of the three isozymees of β-hexosaminidase. Each isozyme effect results from one of the distinct interactions of α- and β-subunits. HexA, α-β heterodimer, and hexosaminidase B (HexB), β-β homodimer, are two major types of β-hexosaminidases. The α-α homodimer, hexosaminidase S, is a subgroup that appears to have a noninvasive pathogenesis. A- and β-subunits are encoded in two distinct genes, HEXA and HEXB, which are located on different chromosomes.

Tay-Sachs disease is caused by a genetic mutation involving HEXA that results in HexA deficiency. Like most lysosomal storage disorder, there is no cure for this deadly disease. Several aspects of lysosomal diseases suggest that these pathologies may not be funded by genetic or genetic therapy. First, the remaining enzymatic activity of only 10% of normal normal activity is sufficient to avoid the appearance of clinical symptoms as indicated by the so-called lysosomal enzyme pseudo deficiencies. Second, part of each lysosomal enzyme is secreted and can be absorbed by other cells through specific mannose-6-phosphate receptors and possibly by other unknown mechanisms. Therefore, we and others hypothesized that a group of modified cells that talk too much and produce large amounts of an enzyme can lead to insignificant activity in vivo cells that do not function well in vivo, using the hide-and-retrieve method.

The most serious neurodegenerative disease, Tays-Sachs syndrome, is caused by a deficiency of beta-hexosaminidase alpha-subunit that inhibits the formation of lysosomal heterodimeric alpha-beta enzyme, hexosaminidase A (HexA). There is no cure for this deadly disease; However, genetic therapy may represent a therapeutic approach. Previously we have also classified, in vitro, adenoviral and retroviral vectors coding the alpha- and beta-subunits of human beta-hexosaminidase. Here, we have determined the in vivo strategy that leads to the highest HexA activity in the highest amount of tissue in the hexA -dough available mouse. We have shown that intravenous co-administration of adenoviral vectors codes both alpha- and beta-subunits, which lead to special liver transmission, was essential for successful outcomes. Only the supply of both subunits approved by HexA overexpression leads to a significant secretion of the enzyme into serum, and the complete or partial reversal of enzymatic functions in all boundary tissues tested. Enzymatic alignment may be due to direct cellular transfer by adenoviral vectors and / or the detection of hidden HexA by different organs. These results confirmed that the liver was the preferred organ for delivering large amounts of hidden proteins. In addition, the need for overproduction of both heterodimeric protein subunits in order to achieve a high rate of extinction in defective animals in one place is emphasized. The endo native non-defected subunit otherwise becomes limited.

Here, we show that active release of hexosaminidase can be detected intravenous (i.v.), but not intramuscularly (i.m.), administration of adenoviral vectors. In addition, we show that co-transduction with two vectors with α- code and β-subunits, respectively, was required to achieve high HexA synthesis and secretion. After the α-chain vector-encoding transition was transmitted, only limited amounts of the active enzyme were produced, suggesting that β-chain was limited. This method has allowed us to obtain a complete HexA enzymatic modification using both direct transfer methods and secretory extraction methods. However, as reported, adenoviral vectors or hexosaminidases do not exceed the blood-brain barrier. Therefore, transplantation of gene therapy can be significantly altered in lysosome diseases without brain damage. Tay-Sachs and Sandhoff patients can benefit from this, however, as the efficiency of bone marrow transplantation in the Sandhoff mouse model suggests that the repair of non-neuronal cells can improve health despite brain lesions.

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None

Conflict of Interest

The author declared that there is no conflict of interest.