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(54) Title: THERAPEUTIC ADENO-ASSOCIATED VIRUS USING NUCLEIC ACID ENCODING ALPHA-GLUCOSIDASE (GAA) FOR TREATING POMPE DISEASE, WITH HETEROLOGOUS SIGNAL PEPTIDE



Fig. 1

(57) Abstract: Disclosed herein is a method for the treatment of Pompe Disease comprising administering a recombinant AAV (rAAV) vector comprising a rAAV genome comprising a heterologous nucleic acid encoding a heterologous BM40 signal peptide and an acid alpha-glucosidase (GAA) polypeptide, or N-terminal truncation thereof, where the heterologous nucleic acid is operatively linked to a liver-specific promoter, where the nucleic acid encoding GAA polypeptide can be wild type nucleic acid sequence, or modified nucleic acid sequence, or a codon optimized nucleic acid sequence, and can optionally be modified to reduce or completely eliminate CG and CpG dinucleotides and, optionally eliminated alternative reading frames (ARF) content.

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