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Gene therapy resolves ocular problems linked to ANT1 gene mutation

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"Mitochondrial myopathy, associated with muscle weakness and progressive external ophthalmoplegia, is caused by mutations in mitochondria oxidative phosphorylation genes including the heart-muscle isoform of the mitochondrial adenine nucleotide translocator (ANT1)," scientists in the United States report.

"To develop therapies for mitochondrial disease, we have prepared a recombinant adeno-associated viral vector (rAAV) carrying the mouse Ant1 cDNA. This vector has been used to transduce muscle cells and muscle from Ant1 mutant mice, which manifest mitochondrial myopathy," according to A. Flierl and colleagues, University of California Irvine, MAMMAG.

"AAV-ANT1 transduction resulted in long-term, stable expression of the Ant1 transgene in muscle precursor cells as well as differentiated muscle fibers. The transgene ANT1 protein was targeted to the mitochondrion, was inserted into the mitochondrial inner membrane, formed a functional ADP/ATP carrier, increased the mitochondrial export of ATP and reversed the histopathological changes associated with the mitochondrial

myopathy."

"Thus, AAV transduction has the potential of providing symptomatic relief for the ophthalmoplegia and ptosis resulting from paralysis of the extraocular eye muscles cause by mutations in the Ant1 gene," study authors suggested.

Flierl and colleagues published their study in *Gene Therapy* (Adeno-associated virus-mediated gene transfer of the heart/muscle adenine nucleotide translocator (ANT) in mouse. *Gene Therapy*, 2005;12(7):570-578).

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