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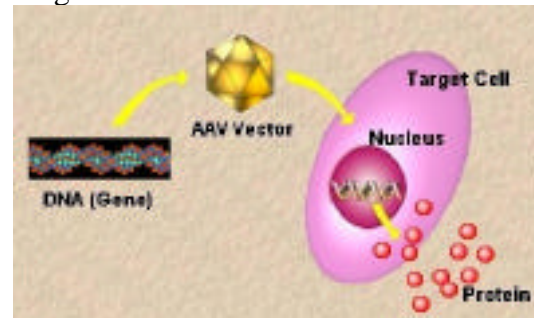
### Reversing the Genetic Mutation in Muscular Dystrophy

Muscular dystrophy is a genetic disease caused by mutations in the dystrophin gene. It weakens the skeletal muscles, causes difficulties in breathing, skeletal deformities and in some cases the heart problems. In most cases its X-chromosome recessive, thus affecting more males than females. One type is limb-girdle muscular dystrophy, which generally affects the areas of the hips and shoulders. LGMD is not a fatal disease but it does cause weakness to the lung and heart muscle, which may lead to death. Some require respiratory or physical therapy or even pacemakers.



A new study at the University of Pittsburgh used gene therapy to successfully treat heart failure in an animal model of a human with LGMD. The past attempts failed because the virus was not small enough to reach muscle cells. Dr. Xiao and his team found a virus-adeno-associated virus, or AAV- which was small enough to deliver the gene to the cells. They encoded the virus with a normal copy of delta-sarcoglycan, the defected gene which causes sever muscle weakness. This virus was injected into both young

(10 day old) and adult LGMD hamsters. This study showed that the normal gene soon appeared in the skeletal and cardiac muscle cells. When examined after 8 months the young hamsters had developed normal hearts, while the muscle in the adults' hearts improved greatly. The group of hamsters that were not given the virus had developed abnormalities of the heart tissue, liver problems, fluid in the chest, and swollen lungs.



The greatest result was the improvement in the lifespan of the treated versus untreated. Also the treated hamsters had the normal proteins a year later. The untreated hamsters ended up dying of heart failure and muscle complications. The treated group all lived past 48 weeks.

This study has made great progress, but this is only the beginning. Now that this virus works for hamsters, it is only time before it will be adapted for humans. The major obstacle that needs to be overcome is how to make the virus be undetected to an immune system. Also a large amount of the treatment is needed to target every muscle cell. This study has created a remarkable breakthrough for treating heart failure in muscular dystrophy.

#### References

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