This project is funded by the Duchenne forum - a collaboration established to accelerate progress in the search for treatments and eventually cures.

Genome surgery for Duchenne muscular dystrophy

Professor George Dickson and his team have developed an innovative technique with the potential to repair the genetic mutation that causes Duchenne muscular dystrophy. The ground-breaking technique, described as genome surgery, could be the first therapy that offers permanent correction of the genetic mutation in a person’s own DNA. The technique is relevant to all boys and men with Duchenne muscular dystrophy and could also be used to treat people with Becker muscular dystrophy.

What are the researchers aiming to do?

Duchenne muscular dystrophy is caused by mutations in the dystrophin gene. This gene contains the instructions for making dystrophin protein which acts as a shock absorber to prevent damage when the muscle contracts. The loss of dystrophin in Duchenne muscular dystrophy leads to wasting of the muscle, with the muscle fibres gradually being replaced by fat and scar tissue. Repairing the mutations in the dystrophin gene could restore production of dystrophin to the muscles which could be a viable therapeutic approach to prevent muscle damage and slow the decline in muscle function.

Professor Dickson and his team have been working on a strategy described as genome surgery, which has the potential to permanently correct a mutation in a person’s own DNA. This is done by using enzymes called endonucleases that act like molecular scissors to cut the DNA. These scissors are designed to cut out the precise part of the gene containing the mutation. Other molecular tools are used to add in the correct DNA sequence and join the cut ends together. This would correct the genetic mutation and allow production of the full-size dystrophin protein.

In this PhD studentship, the molecular tools required for the gene surgery technique will be generated and then tested in cell culture models of Duchenne muscular dystrophy and in a mouse model of the condition. If proven to be effective, this therapy has the potential to be beneficial for all boys with Duchenne muscular dystrophy regardless of their mutation. The technique may also be able to treat people with Becker muscular dystrophy and has the potential to be adapted for the treatment of other muscular dystrophies or related neuromuscular conditions.
How will the outcomes of the research benefit patients?

The gene surgery approach has the potential to permanently correct the genetic alteration causing Duchenne muscular dystrophy, something that has not been achieved before. The approach is applicable to all the mutations which cause Duchenne muscular dystrophy and Becker muscular dystrophy and could also be adapted for the treatment of other neuromuscular conditions.

What do the researchers say?

“Recent astonishing advances in science have yielded ‘molecular scalpels’ and ‘repair patches’ to specifically cut open and correct the damaged X-chromosome in Duchenne muscular dystrophy patients’ cells. With this new funding from the Muscular Dystrophy Campaign, the exciting and pioneering technology of genome surgery will produce a universal gene repair system that could be applied to all types of Duchenne and Becker muscular dystrophy mutations.”

Grant information

Project leader: Professor George Dickson
Location: Royal Holloway (University of London)
Conditions: Duchenne muscular dystrophy; Becker muscular dystrophy
Duration: Three years, starting 2013
Total project cost: £ 79,704Official title: Correction of Duchenne Muscular Dystrophy Mutations using Endonuclease-Mediated Genome Surgery

For further information

If you would like further details about this research project, please contact:

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