

Epilepsy treatment could yield cure for McArdle disease

January 15 2015, by Narelle Towie



Prof Howell's team injected or administered tablets of sodium valproate into 17 merino sheep carrying a mutation for McArdle Disease and then biopsied their limb muscles. Credit: Steven Depolo

WA scientists may have found a cure for the crippling muscle condition McArdle disease, using a drug already approved for use in humans.

McArdle [disease](#), also called glycogen storage disease V, is a [rare genetic condition](#) that affects one in every 100,000 people.

Patients with the disease have muscles that lack a key enzyme that converts stored sugar into glucose, meaning their cells are unable to generate enough energy.

Sufferers are unable to do simple physical activities, such as walking, without experiencing cramping and exhaustion.

In serious cases, the disease causes muscle tissue to break down, leading to [acute renal failure](#).

Until now, there was no [treatment](#) for McArdle disease.

But Murdoch University veterinarian Professor John Howell says his team have switched on the missing enzyme in [sheep](#) and made their symptoms disappear using an anticonvulsant [drug](#) called sodium valproate.

"We have been working on it for years and we used a number of compounds and when we gave the sheep sodium valproate we found that it stimulated the missing enzyme to be activated and so it worked as a cure for the disease in sheep," Prof Howell says.

Sheep used in initial experiments

"We developed a treatment that works in sheep and that treatment is now being used to treat humans."

Murdoch University veterinarian Associate Professor Kate Creed says the drug had to be continuously administered over four months.

She says sheep with McArdle disease exhibit similar clinical effects to humans and the work was a significant breakthrough in the treatment of the disorder.

"It's very nice to feel that we might be able to benefit people with McArdle disease," A/Prof Creed says.

Prof Howell's team injected or administered tablets of sodium valproate into 17 merino sheep carrying a mutation for McArdle disease and then biopsied their limb muscles.

Sodium valproate is currently used as a treatment for epilepsy and the compound belongs to a family of drugs, called Histone deacetylase inhibitors, that can switch genes on and off.

The Murdoch researchers found the missing enzyme, called glycogen phosphorylase, was produced in much greater amounts in the skeletal muscle fibres of sheep given the drug compared to control animals administered saline.

Prof Howell says the drug was effective within days of being injected into the animals, but it is too early to say if sodium valproate will work in humans.

The researchers say the drug is currently being trialled in treating humans with McArdle Disease in London.

More information: "Investigating sodium valproate as a treatment for McArdle disease in sheep." DOI: [dx.doi.org/10.1016/j.nmd.2014.10.002](https://doi.org/10.1016/j.nmd.2014.10.002)

Provided by Science Network WA

Citation: Epilepsy treatment could yield cure for McArdle disease (2015, January 15) retrieved 20 March 2024 from <https://medicalxpress.com/news/2015-01-epilepsy-treatment-yield-mcardle-disease.html>

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