

INFORMATION CARD

McArdle Disease

A rare genetic disorder of muscle metabolism

Patients appear healthy but activity results in premature fatigue, exaggerated heart rate, pain and muscle spasm.

Cases of muscle breakdown (rhabdomyolysis) can lead to life-threatening kidney failure.



AGSD

Association for Glycogen
Storage Disease (UK) Ltd

Registered charity
no. 1132271

For more information:

www.agsd.org.uk

Guidance in event of rhabdomyolysis

If after strenuous or unusual exercise/activity† you have one or more of the following signs:

Dark coloured urine - This is called myoglobinuria or proteinuria and appears as reddish tea to cola coloured urine. (However, if you have eaten strongly coloured food such as beetroot/beets there is probably no need for concern.)

Feeling very unwell after activity - perhaps with 'flu-like symptoms, can be a sign of rhabdomyolysis (muscle breakdown).

Low volume of urine - Producing a very low volume or no urine at all, constitutes a medical emergency (unless simply caused by dehydration) as the complications can become life threatening.

†A few people have experienced these symptoms without activity.

You should:

- Drink plenty of water to help clear your urine.
- Go to hospital promptly for medical assessment.
- Take a urine sample with you, if possible.

If in doubt, telephone your McArdle Disease consultant or specialist nurse.

To the emergency room doctor:

Treatment guidelines:
www.agsd.org.uk
and follow: Type V and McArdle Emergencies

McArdle's physician:

People with McArdle Disease may require assistance with:

- A place to sit down for 10 minutes rest.
- The use of a courtesy wheelchair (e.g. at airports)
- In the event of a rhabdomyolysis crisis, to be taken to hospital.

Thanks for your help.

McArdle Disease is also known as Glycogen Storage Disease Type V (GSDV). It is an inherited condition which results in the absence of an enzyme (myophosphorylase) in the muscle cells. This enzyme is needed to convert glycogen (fuel stored in the muscle) into energy.

The result is a serious energy deficit during the first 10 minutes of any activity, and throughout all intensive activity.