

INFORMATION CARD

GSD Type VII

**is 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 exercise - 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 exercise.

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 GSD Type VII doctor or specialist nurse.

To the emergency room doctor:

For further information:
www.agsd.org.uk
and follow: GSD Type VII

GSD Type VII doctor:

People with GSD Type VII may need help with:

- Use of a courtesy wheelchair (e.g. at airports)
- In the event of serious rhabdomyolysis, to be taken to hospital.

If unwell, please provide:

- A quiet place to sit down for 10 minutes.
- A glass of water.
- Access to a toilet.
- Use of a telephone.

Glycogen Storage Disease Type VII (also known as phosphofructokinase deficiency or Tarui Disease) is an inherited condition which results in the deficiency of an enzyme in the muscle cells. This enzyme is needed in order to use glucose to fuel the muscle.

The result is a serious energy deficit at the start of any activity, and throughout all intensive activity.