

Physiotherapy exercise advice for McArdle disease

McArdle disease and related disorders service

Scope of guidance

This information leaflet has been developed by the Highly Specialist Physiotherapist based in the National McArdle disease and related disorders service in London. This document should be used as a reference guide only. For further advice and information, please contact 02034488132 or email enquiries.mcardle@uclh.nhs.uk.

What is McArdle disease?

McArdle disease is a rare metabolic muscle disorder also called Glycogen Storage Disease Type V (GSD V). It is caused by the lack of an enzyme called muscle glycogen phosphorylase. This enzyme is important in producing one of the fuel sources required by skeletal muscle for energy through conversion of glycogen into glucose.

This means that after using the initial stored energy in the muscles, the muscles are unable to generate further energy through anaerobic metabolism and have to wait until an alternative muscle energy metabolic pathway becomes active (aerobic metabolism), usually 10 minutes after starting any physical activity or exercise. During this time people with McArdle disease develop symptoms of pain, muscle weakness and increased heart rate. If people with McArdle disease continue to exercise despite developing pain or weakness, a severe muscle spasm or contracture will occur and they are at risk of a very severe episode of muscle damage called rhabdomyolysis. Therefore, people with McArdle disease are advised to either slow down or stop and rest for a minute if their symptoms start to increase beyond "moderate".

Second wind

The “second wind” is a phenomenon whereby after 6-10 minutes, exercise induced pain and fatigue begins to subside and the person with McArdle disease can exercise more freely. This is probably due to a switch in muscle energy metabolism from glycolysis to oxidative phosphorylation which utilises an increased reliance of fatty acids as the energy source; thereby providing energy for more normal sub maximal muscle functioning. During the pre-second wind period, sympathetic activity is abnormally high, and manifests itself by a high heart rate relative to the level of exercise intensity. The onset of a significant decline in heart rate and discomfort is an easily measured key marker of the “second wind”.

It is important to remember that the “second wind” is specific to the muscles being used. For example, if walking for 15 minutes, leg muscles will be in second wind, but the upper limb muscles will not be in second wind and will therefore not be warmed up.

Aerobic exercise

Aerobic exercise is the safest form of exercise for people with McArdle disease. It is also the only known management strategy for this population currently (Quinlivan et al, 2011). Conditioning as a result of exercise training and/or controlled physical activity strategies may improve performance by increasing the ability to achieve a “second wind”.

For people with McArdle disease to get aerobic training effects they should be worked at 60 – 70% of their heart rate reserve. The recommendation for people with McArdle disease is to initially start on improving their walking. However it is important to note they will probably need to build up to this and focus should be on starting off slowly and gently and working on achieving a second wind first and build up exercise training slowly as they will be de-conditioned. They should ultimately aim to achieve the government recommendations of 150 minutes of moderate intensity aerobic exercise per week, with bouts of at least 30 minutes to allow exercise training once in “second wind”.

Strengthening exercises

There is emerging evidence that strengthening protocols can be used with patients with McArdle Disease; however these should only be done with close supervision by a physiotherapist. The following information provides basic advice on strength training and is based on work done by Santalla et al (2014). **However please contact the specialist McArdle physiotherapist on 02034488132 or enquiries.mcardle@uclh.nhs.uk PRIOR to starting any strengthening programmes.**

- **All muscle groups** to be involved must be in second wind prior to strength training. Therefore if the upper limbs are to be strengthened, they should be

warmed up for 15 minutes using, for example, an arm ergometer or cross trainer.

- Low number of repetitions (**4-5 repetitions maximum**).
- **No/low resistance** should be used.
- **Isometric contractions should be avoided** to prevent muscle damage and therefore all movements should be dynamic.
- A rest period of 2-3 minutes between each set of repetitions **is essential** to allow muscle energy to be restored.
- There should be a period of **at least 48 hours** between any muscle strengthening activities/sessions.
- All exercises should be guided by pain (**less than 4 on the RPP scale**).

Other activities to avoid are squatting, sustained positions such as stretching and more vigorous exercise such as rushing up stairs for example.

Borg's RPP Scale:

0	Nothing at all	"No P"
0.3		
0.5	Extremely weak	Just noticeable
1	Very weak	
1.5		
2	Weak	Light
2.5		
3	Moderate	
4		
5	Strong	Heavy
6		
7	Very strong	
8		
9		
10	Extremely strong "Max P"	
11		
↵		
●	Absolute maximum	Highest possible

Borg CR10 scale
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Back care advice

A recent audit of patients attending The National McArdle Service demonstrated a high proportion of people with McArdle disease experience low back or neck pain. There may be a number of reasons for this, including reduced physical activity

levels, increased weight and the need for strengthening exercises to be modified to avoid muscle damage.

People with McArdle disease should be encouraged to carry out gentle aerobic exercise as described above. Core stability exercises can be prescribed as long as the strengthening principles stated above are followed.

Outcome measures

A twelve-minute self-paced shuttle walk is used instead of a six minute walk test as the onset of the “second wind” is likely to occur after five to eight minutes of exercise (Buckley et al, 2014). It accommodates the specific limitations of the anaerobic glycolytic function in these patients and their potential inability to maintain a steady or incremented pace in periods less than five minutes. During the test, heart rate and rate of perceived pain (see scale above) are measured each minute in order to identify the “second wind” and reduce the risk of muscle damage.

Contact details

For further information and advice, please contact:

Specialist Neuromuscular Physiotherapist (McArdle disease)

Telephone: 0203 448 8132

E-mail: enquiries.mcardle@uclh.nhs.uk

References

Buckley JP, Quinlivan RM, Sim J, Eston RG & Short DS. (2014) Heart rate and perceived muscle pain responses to a functional walking test in McArdle disease. *Journal of Sports Science*, 32(16), pp1561-1569.

Quinlivan R, Vissing J, Hilton-Jones D & Buckley J (2011) Physical training for McArdle disease. *Cochrane Database of Systematic Reviews*, doi: 10.1002/14651858.CD007931.pub2

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