What is McArdle Disease?

McArdle Disease is a rare metabolic muscle disorder also called Glycogen Storage Disease Type V (GSD V). People born with McArdle Disease are unable to produce an enzyme called muscle phosphorylase. This enzyme is important in producing one of the fuel sources required by skeletal muscle for energy.

The means the body is unable to use stored glycogen for muscle energy and until an alternative muscle energy metabolic pathway becomes active, which normally takes up to 10 minutes, people with McArdle Disease develop symptoms of pain and muscle weakness with fatigue. 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”.

People with McArdle Disease have a maximal work capacity of about one half that of the normal population and this, together with a fear of intense exercise and its consequences, can be debilitating for affected individuals.

Second Wind

The “second wind” is a phenomenon whereby after about seven minutes exercise induced pain and fatigue begins to subside and the patient can exercise more freely. This is probably due to a switch in muscle energy metabolism from glycolysis to oxidative phosphorylation that utilises an increased reliance of fatty acids as the energy source. Thus providing energy for more normal sub maximal muscle functioning. During the pre-second wind period, sympathetic activity is abnormally high, and is 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”.

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”.

Outcome Measures:

The outcome measures which are useful and also used in our clinic are:
• **SF36** – to monitor how their health impacts on quality of life
• **12 minute walk test** – to assess functional capacity and provide a baseline for physiotherapy intervention.

**How to complete a 12 minute walk test:**

A twelve-minute self-paced shuttle walk is used instead of a six minute walk test, because the onset of the second wind is likely to occur after five to eight minutes of exercise. 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.

**Equipment required:**

- 10 metre length measured on the floor or a treadmill which records distance.
- Heart rate monitor
- CR10 scale (Borg Rating of perceived pain scale- RPP)– please see appendix 1

**Instructions:**

1. Ideally they should be seated for 30 minutes prior to the assessment.

2. The patient should be familiarised with the Borg Rating of perceived pain scale prior to commencing the assessment. A rating of “0” equated to no muscle pain at all and “10” was the most pain the patient can ever recall suffering. The scale has points in excess of 10, which should be described as being potential levels of pain beyond those previously experienced.

3. Attach the heart monitor to the patient.

4. Give the patient instruction to complete as many 10 m shuttles as possible in 12 min, but not to exceed a walking speed that would elicit a leg RPP of greater than level “4” (between moderate and strong). If RPP exceeds 4, instruct the patient to slow down or stop and only to continue when the RPP was ≤ 4. If shuttle walking you can walk next to the patient to ensure you are monitoring their heart rate.

5. This 12 minute walking assessment can be completed on a treadmill and the speed slowly increased but do not exceed a speed which elicits a leg RPP of greater than level 4.

6. At the end of each minute the number of shuttles/ distance walked on the treadmill completed, heart rate and RPP is recorded.

7. Patients are given the opportunity to immediately stop and sit down if required if in pain or they have other symptoms arising such as dyspnoea, dizziness, or nausea.

8. Even if the patient stops and rests the clock remains running.
How to interpret the results of a 12 minute walk

**Aim:** Over approximately 500 metres

For those with an inability to walk 500 m in 12-minutes equates to an inability to sustain activity at levels greater than 3 to 4 metabolic equivalents a level associated with activities of daily living. It is these patients, for whom exercise therapy may be the most beneficial. Low levels of aerobic fitness and inactivity are also significant risk factors of premature mortality as a result of developing more prevalent chronic conditions such as diabetes and cardiovascular disease.

We have a number of patients who peak between 800-900 metres in 12 minutes indicating a good level of aerobic fitness.

**Exercise Advice**

**The use of sugar pre exercise**

Many therapeutic studies have been undertaken in McArdle disease, most of which have employed small numbers of patients. The only intervention shown to improve symptoms of McArdle disease is oral sucrose which, when ingested immediately prior to exercise, facilitated the “second wind”. This is helpful in some circumstances and patients do this in the form of a sports or sugary drink which they sip. However this should not be used instead of aerobic conditioning and it is a cause of significant weight gain.

**Strengthening and stretching**

Isometric contractions and lifting of heavy weights should be avoided to prevent muscle damage. At present we have no evidence for safe strengthening protocols in patients with McArdle Disease. Focus should be directed towards aerobic exercise.

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

**Aerobic exercise - How hard should they exercise?**

For patients to get aerobic training effects they should be worked at 60 – 70 % of their heart rate reserve. The recommendation is for patients 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.

**Minimum Heart Rate (Min HR):** Ideally this should be taken first thing in the morning

**Maximum Heart Rate (Max HR):** 220 – age
**Heart Rate Reserve (HRR):** Maximum Heart Rate – Minimum Heart Rate

50% of Max HR = (0.5 x HRR) + Min HR = _____ bpm (beats per minute)
60% of Max HR = (0.6 x HRR) + Min HR = _____ bpm
70% of Max HR = (0.7 x HRR) + Min HR = _____ bpm

**IMPORTANT:** Please note all patients need to start slowly so they achieve their second wind as reported above and then gradually increase their speed. All aerobic exercise should be guided by the RRP scale and if the patient experiences any symptoms over 4, the speed should be slowed or stopped as guided by the patient. They should eventually aim for 30-40 minutes of exercise. It is important to warm up and cool down.

People with McArdle Disease can also improve their fitness using an exercise bike or cross trainer. The principles remain the same ensuring they maintain their pain levels below 4 and heart rate is monitored.

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APPENDIX 1: Borg CR10 Scale

0  Nothing at all          No pain
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        Maximum pain
11

● Absolute maximum         Highest possible