

## **Type V Workshop Report for the 2007 Annual Conference**

### **Dr Ros Quinlivan reported on the work of the McArdle Disease Service at the Wolfson Centre for Inherited Neuromuscular Disease, Oswestry.**

We are very grateful to the Association for Glycogen Storage Disease (UK) for their continuing support. This year has been a very exciting one. The TORCH appeal has raised £3.55 million and building work for our new research facility is well under way. Dr Quinlivan co-hosted the first European Neuromuscular Disease Centre (ENMC) workshop in McArdle's disease and participated in the first International Glycogenoses workshop in Genoa in November 2006. Following these two meetings a collaborative network of European clinicians interested in McArdle disease was established (MADEN), the first steering group committee meeting was held in March 2007 in Conegliano, Italy. MADEN submitted a large (3 million Euro) grant to The European Union for a collaborative epidemiological study but sadly we were not successful in obtaining funding. We plan to continue to try and raise funds for a large International study and will be meeting in Sicily in October 2007.

The clinic at Oswestry has now seen 50 people confirmed to have McArdle disease. We are accumulating evidence to show that our management strategy is beneficial with our patients demonstrating improvements in their functional assessments. We now have very good clinical experience in managing the condition. One aspect which we have noticed for some time is that patients frequently complain of chronic pain and fatigue accompanied with poor concentration and forgetfulness. Because of this we sought collaboration with Dr Edelstyn, a Neuropsychologist based at Keele University, and have completed a pilot study of cognitive testing in McArdle disease. The results have shown significant differences between people with McArdle disease and other people who have different muscle disorders. We are not sure why this should be the case, however, it does suggest that our anecdotal observation represents a real effect. This pilot study involved only a small number of patients and it will need to be confirmed with a larger study.

Kathryn Wright is making excellent progress with her research to identify a model system for McArdle disease (see separate report). She was successful in winning a prize for her research presentation at the hospital's research day earlier this year. Kathryn is also collaborating with Professor John Howell, who has been undertaking a therapeutic study in McArdle sheep. She will be presenting her research at The World Muscle Society meeting in Sicily this year.

A business case for National Specialist Commissioning (NCG) to fund a diagnostic and clinical service McArdle disease and other rare glycolytic disorders affecting anaerobic energy metabolism (GSDV11 and GSD IXD) has been prepared. The business case reflects a consortium bid and includes the Oswestry McArdle disease clinic and diagnostic services available through The Children's Hospital Birmingham, The Queen Elizabeth University Hospital and Sheffield Children's Hospital. We have been informed that our chances of success are 50%.