Topic: Advances in the treatment of neuromuscular disorders

Effect of a multi-disciplinary approach to diagnosis and management for non-lysosomal skeletal muscle glycogen storage disorders

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The highly specialised national service for rare non-lysosomal skeletal muscle glycogen storage diseases(GSDs) is the only service world-wide offering multi-disciplinary(MD) care for affected patients. The team includes: physician, physiotherapist, clinical nurse specialist, clinical psychologist, dietician, sports and exercise physiologist, patient advocate and research fellow. Provided services include muscle enzyme analysis, genetic studies, a clinical diagnostic and management service. In addition to an MD clinic, telephone clinics, information days for newly diagnosed patients and face-to-face therapy reviews are also offered. Prospective data (audit of outcome measures used for the NHS commissioning for Quality and innovation–CQUIN) from 236 patients will be presented. The majority of patient had McArdle disease(n=164). Other diagnoses include: GSDVII(n=6), GSDIX(n=2), GSDXIII(n=2) and GSDXV(n=1). The incidence of episodes of severe rhabdomyolysis requiring hospital visits was 17% in new referrals and 2.8% in follow-up patients. There was a high frequency of overweight/obesity (52%females, 78%males). Patients who completed the 12 minute walk test(12MWT) and were divided into 4 groups (< 500m(n=12); 500-800m(n=31); 800-1000m(n=24) and >1000m(n=8)). A Patient Liaison Panel oversaw the production of a reference booklet for GPs and assisted with recruitment to the EUROMAC Registry and the Sodium Valproate clinical trial.

This highly specialised MD national service demonstrates reduced frequency of rhabdomyolysis and other complications related to rare GSDs following clinical assessment. Patients could be divided into 4 clear cohorts on the basis of severity as assessed by the 12MWT which showed a positive correlation with quality of life. Improvements in walking distance and quality of life were seen in subsequent visits to the clinic, demonstrating functional improvement over time. Data from anthropometric assessments highlights the need for ongoing dietetic support.