A Pilot Observational Study Investigating The Impact Of Glycogen Storage Disease III On Aerobic Capacity

Philip J. Hennis , David Tomlinson, Gurinder Rayat, Elaine Murphy.

Glycogen storage disease 3 (GSDIII) is a rare inherited metabolic disorder caused by glycogen debranching enzyme (GDE) deficiency which primarily affects the liver, skeletal muscle and heart and results in muscle weakness and profound exercise limitation. Despite exercise intolerance being a major complication associated with the disease, the influence of GSDIII on aerobic capacity is largely unstudied.

PURPOSE: To preliminary describe the impact of GSDIII on aerobic capacity and investigate potential mechanisms responsible for any impairment.

METHODS: In this descriptive study 5 patients (3 female) (39 ± 11 years) with GSDIIIa underwent an incremental cycle exercise test to volitional exhaustion. During exercise breath-bybreath gas analysis took place to determine oxygen utilisation (VO2), carbon dioxide production (VCO2), and minute ventilation (VE) and heart rate (HR) was measured continuously. The study received institutional and NHS ethics approval.

RESULTS: Peak VO2 was lower in the GSDIII patients than predicted based on their demographic data (16.9±8.4 ml/kg/min, 52±23% of predicted), as was peak work rate (WR) (86±59 watts, 52±30% predicted), peak HR (139±26 bpm, 77±11% predicted), and VE peak (30±19 L/min, 36±13%) predicted). Peak RER was low for a test completed to maximal exertion (0.90±0.07).

CONCLUSION: VO2peak is lower in patients with GSDIII than would be expected for their age, height, mass and sex. The mechanisms responsible for this impairment are yet to be fully determined, but the small data set presented here indicate a reserve in respiratory and central cardiovascular function. Previous literature has identified energy deficiency as a primary cause of exercise intolerance in GSDIII due to impaired glycogen breakdown, and these results are supported here by the low RER values at peak exercise.