

PROLONGED EXERCISE TESTING IN CHILDREN WITH MULTIPLE ACYL CO-A-DEHYDROGENASE DEFICIENCY

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Objective

Multiple Acyl-CoA-Dehydrogenase deficiency (MADD) is an inherited metabolic disorder characterized by impaired oxidation of fatty acids and some amino acids. We were interested whether children with MADD could tolerate a prolonged low-intensity exercise test and if this test could have any additional diagnostic value.

Methods

We performed a maximal exercise test and a 90 minutes prolonged exercise test in 2 patients with MADD and in 5 control subjects. During prolonged exercise test the subjects exercised on a cycle ergometer at a constant workload of 30% of W_{max} for 90 minutes. During the prolonged exercise test heart rate, oxygen uptake, fuel utilization and changes in relevant blood and urinary parameters were monitored.

Results

The tests were tolerated well. During the prolonged exercise test the fatty acid oxidation (FAO) was quite low (20-30 % of total energy expenditure) in the 2 patients with MADD compared to the control subjects (50-60% of total energy expenditure; see Figure 1), while increased presence of ethylmalonic acid in urine combined with the abnormal mid-chain carnitine ester profile in plasma appeared in plasma and urine (characteristic metabolites of MADD; See Table 1A and 1B). In the urine and plasma of the control subjects, no abnormal metabolites were found in organic acid analyses and acylcarnitines.

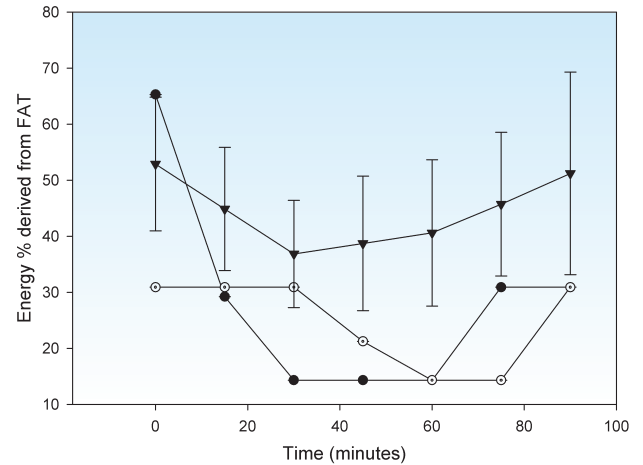


Figure 1. Percentage energy derived from fatty acid oxidation during the prolonged exercise test for patient 1 (filled circle), patient 2 (open circle) and 5 control subjects (black triangles). Values are mean \pm SD.

Table 1A. Results prolonged exercise test subject 1

	T=0	T=30	T=60	T= 75	T=90	T=120
Glucose (mmol·l ⁻¹)	7.1	5.6	4.9	4.8	5.5	4.8
Lactate (mmol·l ⁻¹)	1.1	1.2	1.0	1.3	1.1	1.0
CK (u·l ⁻¹)	103		98	105	119	117
3-keto-B (mmol·l ⁻¹)	0.07	<0.05	<0.05	0.05	0.1	0.15
3-OH-B (mmol·l ⁻¹)	0.09	<0.02	<0.02	0.05	0.12	0.26
FFA (umol·l ⁻¹)	0.76	0.28	0.49	0.88	1.03	0.93
Urine						
Ethylmalonic acid in Mmol·mol ⁻¹ creatine	T= -60-0: 16		T= 120-300: 74			

Table 1B. Results prolonged exercise test subject 2.

	T=0	T=30	T=60	T= 75	T=90	T=120
Glucose (mmol·l ⁻¹)	5.2	4.6	4.3	4.4		4.5
Lactate (mmol·l ⁻¹)	2.0	0.9	0.9	1.0		1.2
CK (u·l ⁻¹)	58		80	78		78
3-keto-B (mmol·l ⁻¹)	<0.05	<0.05	0.07			0.14
3-OH-B (mmol·l ⁻¹)	<0.02	<0.02	0.04			0.24
FFA (umol·l ⁻¹)	0.15	0.31	0.62	0.91		1.34
Urine						
Ethylmalonic acid in mmol·mol ⁻¹ creatinine	T= -60-0: 29		T=120-300: 59			



Conclusion

We suggest that the prolonged exercise test could be of diagnostic importance and might replace the fasting test as a diagnostic procedure in some cases, particularly in patients with anamnestic signs of intolerance for prolonged exercise.