

Are Patients Affected By Mitochondrial Disorders At Nutritional Risk?

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Background

Patients suffering from mitochondrial disorders (MD) frequently present gastrointestinal complaints, mainly gastrointestinal dysmotility, that interfere with their food intake¹. Deterioration of their nutritional state may worsen the course of the disease. Our aims were to evaluate a simple screening tool to identify nutritional risk and to perform an extended nutritional assessment to explore the potential presence of deficiencies in this population compared to controls.

Methods

A prospective cohort study comparing outpatients with MD to matched healthy controls was conducted. Nutritional screening and full nutritional assessments were performed, including quantitative and qualitative dietary habits (7-day food recall), body function and composition, resting energy expenditure and quality of life (QoL) measurements. Blood and 24-hours urine analyses were performed in the patient group.

Table 1: Demographics of the study population

Characteristics	Control group	Patient group
Total participants, n	15	11
Caucasians, n (%)	15 (100)	11 (100)
Sex, [male/female], n (%)	8 (53) / 7 (47)	4 (36) / 7 (64)
Age [years], mean ± SD	49±12	60.7±8.0
BMI [kg/m ²], mean ± SD	23.8±2.6	27.8±4.6

Table 2: Nutritional Assessment

Nutritional Assessment	Control group	Patient group
Anthropometrics		
- MAMA [cm ²], mean, SD	49.8 ± 12.6	53.0 ± 15.6
- Hand grip strength [kg], mean, SD	41.7 ± 18.2	30.0 ± 17.9
Gastrointestinal complaints		
- Food intolerances, n (%)	0 (0)	4 (36)
- Bloating, n (%)	3 (20)	8 (72)
- Gut dysmotility, n (%)	1(6)	8 (72)
NRS 2002 score		
- Total score, mean	0	1.09
24-hrs urine analysis		
- Nitrogen Balance [g], mean±SD	N/A	-2.5 ± 2.8
- Creatinine Height Index [%], mean±SD	N/A	63.9 ± 31.4

Literature

¹ El-Hattab AW, Scaglia F. Mitochondrial DNA depletion syndromes: review and updates of genetic basis, manifestations and therapeutic options. *Neurotherapeutics*. 2013;10(2):186-98.

² Cederholm T, Barazzoni R, Austin P, et al. ESPEN Guidelines on definitions and terminology of clinical nutrition. *Clin Nutr*. 2017 Feb;36(1):49-64.

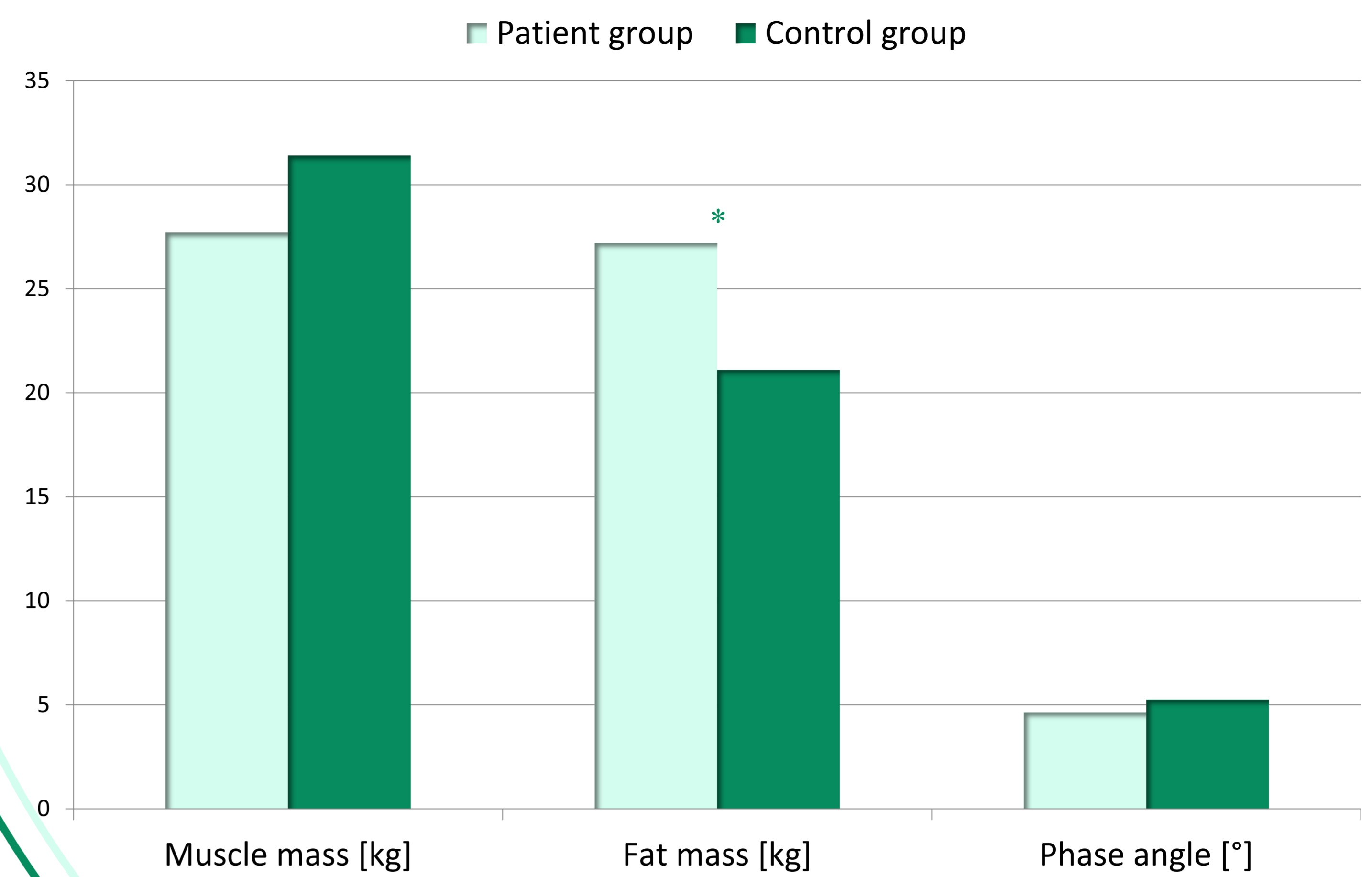


Figure 1: Results of the BIA analysis

* : p=0.005
** : p<0.001

Results

Twenty-six subjects were included: 11 in the patient group and 15 in the control group. None of the patients was screened as malnourished according to the NRS-2002, but compared with controls, they had a lower muscle mass in the BIA analysis (p=0.13), reduced handgrip strength (p=0.07), significant changes in QoL and pathologic creatinine height index, which indicate malnutrition. The patients also had a significantly lower protein intake (p=0.01).

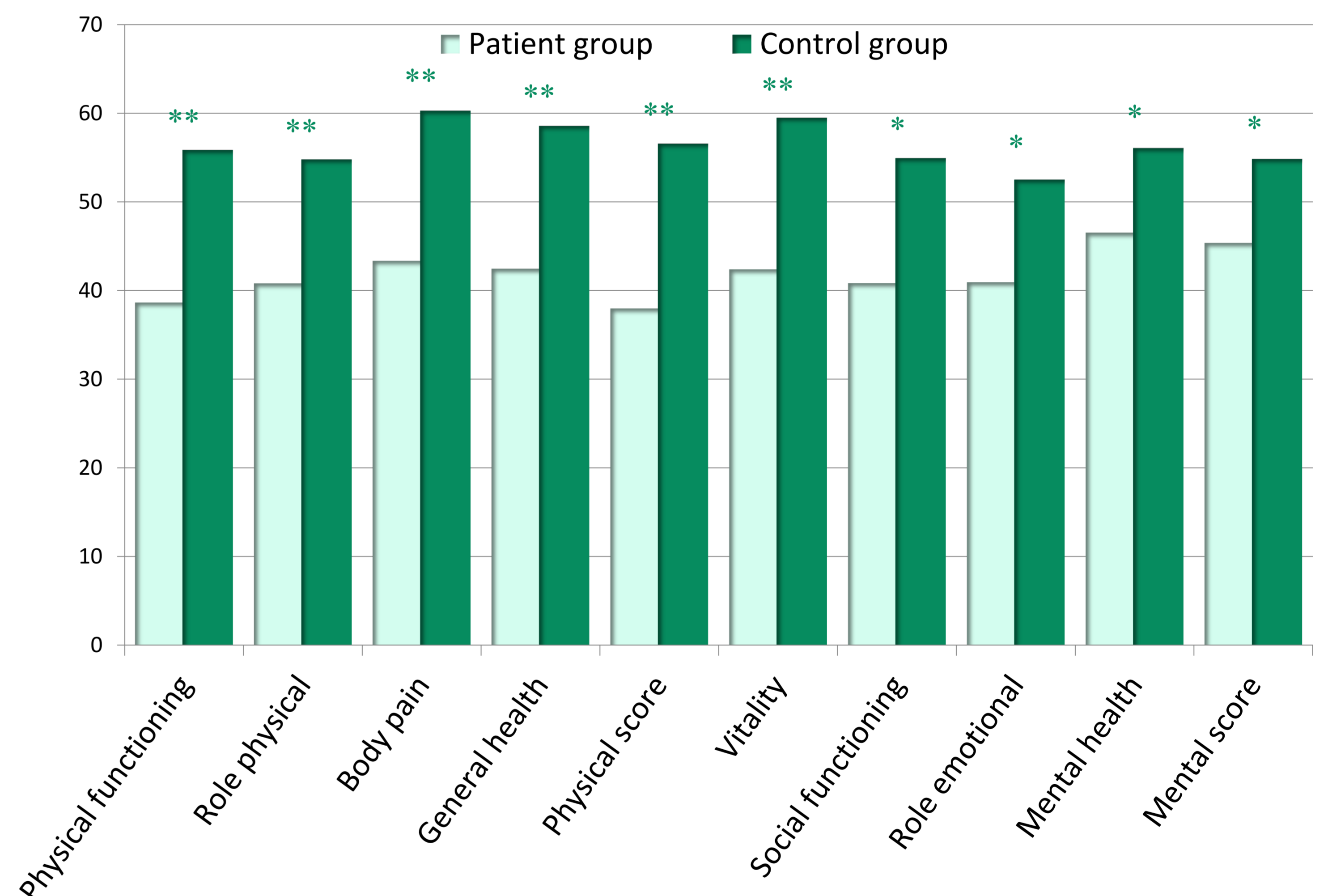


Figure 2: Results of the QoL analysis with SF-36v2™

Conclusion

According to the current definition from the international societies of clinical nutrition and metabolism ESPEN and ASPEN, all patients fulfilled the criteria for manifest malnutrition². Thus, the usual nutritional screening tool is less sensitive for chronically ill outpatients. These results provide a rationale to increase protein intake and adapt patients' energy supplies to improve bodily symptoms and QoL.