Malnutrition and nutrition related problems in adults with mitochondrial diseases

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Mitochondrial diseases

prevalence 1:10,000

treatment supportive

Nijmegen Center Mitochondrial Disorders

Cross section mitochondrion
source: www.ncmd.nl
Malnutrition:

State of nutrition: deficiency
- energy
- protein
- other nutrients

Adverse effects on:
- body composition and function
- clinical outcome

Mitochondrial disorder = malnutrition on cellular level

Stratton R.J. USA 2003
Malnutrition in Mitochondrial Disorders

- Secondary mitochondrial dysfunction
- ↑ existing complaints → ↓ Quality of Life

Dysphagia in Mitochondrial Disorders

Cause:
- muscle weakness
- fatigue
- ptosis
- slow
- accelerated aging
- centrally controlled: strokes and epilepsy
- coordination problem

Result:
\[ \downarrow \text{nutritional state} \]
Aspiration $\rightarrow$ pneumonia $\rightarrow \uparrow$ mortality
Research aim

Nutrition related complaints

Nutritional status

Evidence-based nutrition intervention

Quality of Life↑
Method:

Pilot:

• 23 adults with mitochondrial disease
• June 2010-June 2011 evaluation dietician
• food diary:

<table>
<thead>
<tr>
<th>Kcal</th>
<th>Protein</th>
<th>Carbohydrates</th>
<th>Fat</th>
<th>Water</th>
<th>Fiber</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>use special diet products</td>
<td>use vitamin or mineral supplements</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>food</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

• anthropological data
• diet-related complaints
• nutritional assessment (n=10)
Method

Nutritional Assessment (NA): IC with Delta Track (REE (kcal) and RQ) BIA (FFM and % fat) Anthropometry : height / weight
Method

Status research:
anthropological data of 77 MELAS, CPEO and MIDDS patients

23 CPEO patients:
Swallow Speed Test
(Hughes and Wiles)

B. Smits, J Neurology (2011)
Nutrition related complaints

n = 23 (6 ♂ 17 ♀)
mean(range): 38 (18-58) years

- Gastrointestinal Problems
- Cardiomyopathy
- Diabetes
- Dysphagia
- Overweight BMI > 25
- Underweight BMI < 20
Food diary
n = 20 (6 ♂ 14 ♀)
mean (range): 38 (18-58) years

• No adequate intake
• Special diet products (n=5)
• Low protein intake (n=4)
• High sugar intake (n=12)
Food diary compared to the Recommended Dietary Allowance (RDA)

Milk products:
- Mitochondrial patients: mean 34% RDA, range 0-81%
- Dutch healthy adults: mean 56% RDA, range 0-230%
Body composition

n=12 3 ♂ 9 ♀
mean( range): 40(20-58) years

<table>
<thead>
<tr>
<th>Sex</th>
<th>Fat Free Mass (%) healthy</th>
<th>Fat Free Mass (%) patients</th>
<th>Fat Mass (%) Healthy</th>
<th>Fat Mass (%) Patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>M</td>
<td>80.8</td>
<td>76.1 (SD 11.6)</td>
<td>19.2</td>
<td>23.9 (SD 11.6)</td>
</tr>
<tr>
<td>V</td>
<td>71.9</td>
<td>63.1 (SD 10.4)</td>
<td>28.1</td>
<td>36.9 (SD 10.5)</td>
</tr>
</tbody>
</table>

Status research

<table>
<thead>
<tr>
<th>Sex</th>
<th>N</th>
<th>Min.</th>
<th>Max.</th>
<th>Mean (SD)</th>
<th>Dutch healthy (CBS)</th>
</tr>
</thead>
<tbody>
<tr>
<td>M</td>
<td>32</td>
<td>1,62</td>
<td>1,96</td>
<td>1,78 (0,8)</td>
<td>1,81</td>
</tr>
<tr>
<td>V</td>
<td>45</td>
<td>1,45</td>
<td>1,82</td>
<td>1,65 (0,7)</td>
<td>1,68</td>
</tr>
</tbody>
</table>
Energy requirements and energy intake

n=10  1 ♂ 9 ♀
mean( range): 38(18-58) years

REE Harris and Benedict
mean 158 kCal = 11% overestimation of
REE Indirecte calorimetric ( -25 - +386 kCal  SD 143 kCal)

- Intake - REE Indirecte calorimetric + activity allowance (kCal)
- Intake – Harris en Benedict + activity allowance (kCal)
- Intake – Dutch energy reccomendations (kCal)
Underweight / overweight

n= 77  32 ♂ 45 ♀
mean( range):46(19-69) years .
51 patients with m3243AG mutation:  mean age 44 years
26 CPEO patients:  mean age 50 Years

<table>
<thead>
<tr>
<th>Status</th>
<th>CPEO (%)</th>
<th>m3243AG mutation (%)</th>
<th>Total (%)</th>
<th>Healthy (CBS) (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Underweight</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>BMI&lt;20</td>
<td>27</td>
<td>26</td>
<td>26</td>
<td>1.5</td>
</tr>
<tr>
<td>Normal weight</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>BMI 20-25</td>
<td>31</td>
<td>53</td>
<td>46</td>
<td>51</td>
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<tr>
<td>BMI &gt; 25</td>
<td>42</td>
<td>22</td>
<td>29</td>
<td>47.5</td>
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<td>BMI&gt;30</td>
<td>12</td>
<td>4</td>
<td>6.5</td>
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Dysphagia and BMI

n= 39  17 ♂  22 ♀
16 patients with m3243AG mutation and 23 CPEO patients
mean( range): 47(28-69) years

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<td>dysphagia BMI</td>
<td>16 (41%)</td>
<td>16.8</td>
<td>27.8</td>
<td>20.4</td>
<td>3.4</td>
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<tr>
<td>no dysphagia BMI</td>
<td>23 (59%)</td>
<td>16.9</td>
<td>34.7</td>
<td>24.3</td>
<td>4.7</td>
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Conclusion

Adults with mitochondrial disease:
• low energy requirement
• need for high nutrient density
• adequate intake not achieved

Cause:
• dysphagia,
• bowel problems
• fatigue

Increased risk of malnutrition.
Discrepancy between need and intake → nutritional intervention useful
Future

More research
• Actometre
• Validated Fatigue questionnaire
• Gastrointestinal questionnaire

Before and after diet intervention
- body composition,
- muscle strength
- Quality of Live
Questions?

VOILA!

your average favorite dish
Dysphagia and BMI

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Mann-Whitney test  p = 0.07
Supplementation vitamins and minerals

• 12 of 23 patients takes multivitamins
• 8 of 23 patients takes Q10
Underweight / overweight

n = 77  32 men 45 women 19-69 mean 46 years
mean age m3243AG mutation: 44 Years
mean age CPEO: 50 Years

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<td>%</td>
<td>M (%)</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>BMI 20-25</td>
<td>2 (15)</td>
<td>6 (46)</td>
<td>31</td>
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