Classical Homocystineuria
Nutritional advances and challenges

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Overview

- Classical Homocystinuria (HCU) is an autosomal recessive disorder of methionine metabolism.
- It is caused due to a deficiency of cystathionine $\beta$ synthatase enzyme (CBS) which leads to an accumulation of homocysteine and Methionine.
- The prevalence of HCU in Qatar is 1:1511.
- Dietary management is an integral part in the management of Homocystinuria along with the medication and supplements.
- The dietary treatment is for lifelong.
Homocystinuria is classified as:

- Vitamin B6 Responsive
- Non-Vitamin B6 Responsive (Classical)

- In Qatar, the majority of children with Homocystinuria due to CBS deficiency are not responsive to pyridoxine therapy. (P.R336C mutation)
Goals of the Dietary Management

- Maintain Total homocysteine to the target treatment ranges.
  - HMC experience (HCU Study): Some patients have achieved normalization especially NBS

<table>
<thead>
<tr>
<th>Groups</th>
<th>No</th>
<th>Mean</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>NBS</td>
<td>44</td>
<td>61.19</td>
<td>19</td>
</tr>
<tr>
<td>LDS</td>
<td>67</td>
<td>119.76</td>
<td>42</td>
</tr>
</tbody>
</table>

Al-Dewik et al., “Natural History with clinical, biochemical, and molecular characterization, of classical Homocystinuria in the Qatari population”. Journal of Inherited Metabolic Diseases (n press)
Goals of the Dietary Management

- Maintain Methionine to the target treatment ranges.
  - HMC experience (HCU Study):

<table>
<thead>
<tr>
<th>Groups</th>
<th>No</th>
<th>Mean</th>
<th>Range</th>
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<tbody>
<tr>
<td>NBS</td>
<td>44</td>
<td>388.17</td>
<td>20</td>
</tr>
<tr>
<td>LDS</td>
<td>67</td>
<td>815.93</td>
<td>231</td>
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</tbody>
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- Maintain Cysteine to the normal age related reference range
- Ensure normal growth, development and nutritional status (including growth, signs of protein deficiency and lab results)
- Prevent catabolism

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Challenges & Advances

• Illiteracy

  • Make it simple & easy
  • Use illustrations
  • Food packaging samples
Cont.

- **Language barrier**
  - No English/ No Arabic!
- Dietitians available from different backgrounds
- Bilingual staff
• Socio-economic issues
  • Government funds (all products are free of charge)
Cultural aspects

- Non-disclosure between family members
  (Separate appointments for siblings)

- Hiding diagnosis from extended family
  (noncompliance with diet)
Cultural aspects; Cont.

- Family gatherings
  - Weekends
  - Ramadan
  - Travel
  - Weddings

- Consequence: impact on compliance, thus poor prognosis
To overcome challenges

- **Experimental Kitchen**
  - Preparing low protein meals
Education Materials & Booklets

Booklets: available and distributed among patients

- 1 gm protein
- Low protein recipes
- Amino Acid booklet
Cont.

• Products testing panel
  • Patients testing ---- Feedback received ---- actions taken!
• The Annual Homocystinuria Family gathering day
  • Cooking competitions (families own recipes)
  • Lectures and workshops
  • Activities & gifts
Conclusion

- Dietary management is lifelong for individuals with classical homocystineuria.
- It plays an integral part in the treatment of Homocystinuria.
- It is important to correct the metabolic imbalance and promote growth and development by providing adequate nutrition thus preventing complications.
- It helps to lead a normal life.
- It’s important to actively involve patients and their relatives to overcome any challenges.
References


• Homocystinuria, Online Mendelian Inheritance in Man (OMIM)


• Mandava P et al; Homocystinuria/ Homocystinemia, Medscape, Mar 2011


• Genetic Disease Foundation. Available at: http://www.knowyourgenes.org/genetic-testing-full.shtml


Thank you!