

# Identifying Practice Gaps and Opportunities in Dietitian-Led Management of Homocystinuria: Nutrition Lessons from Three Cases

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## Introduction

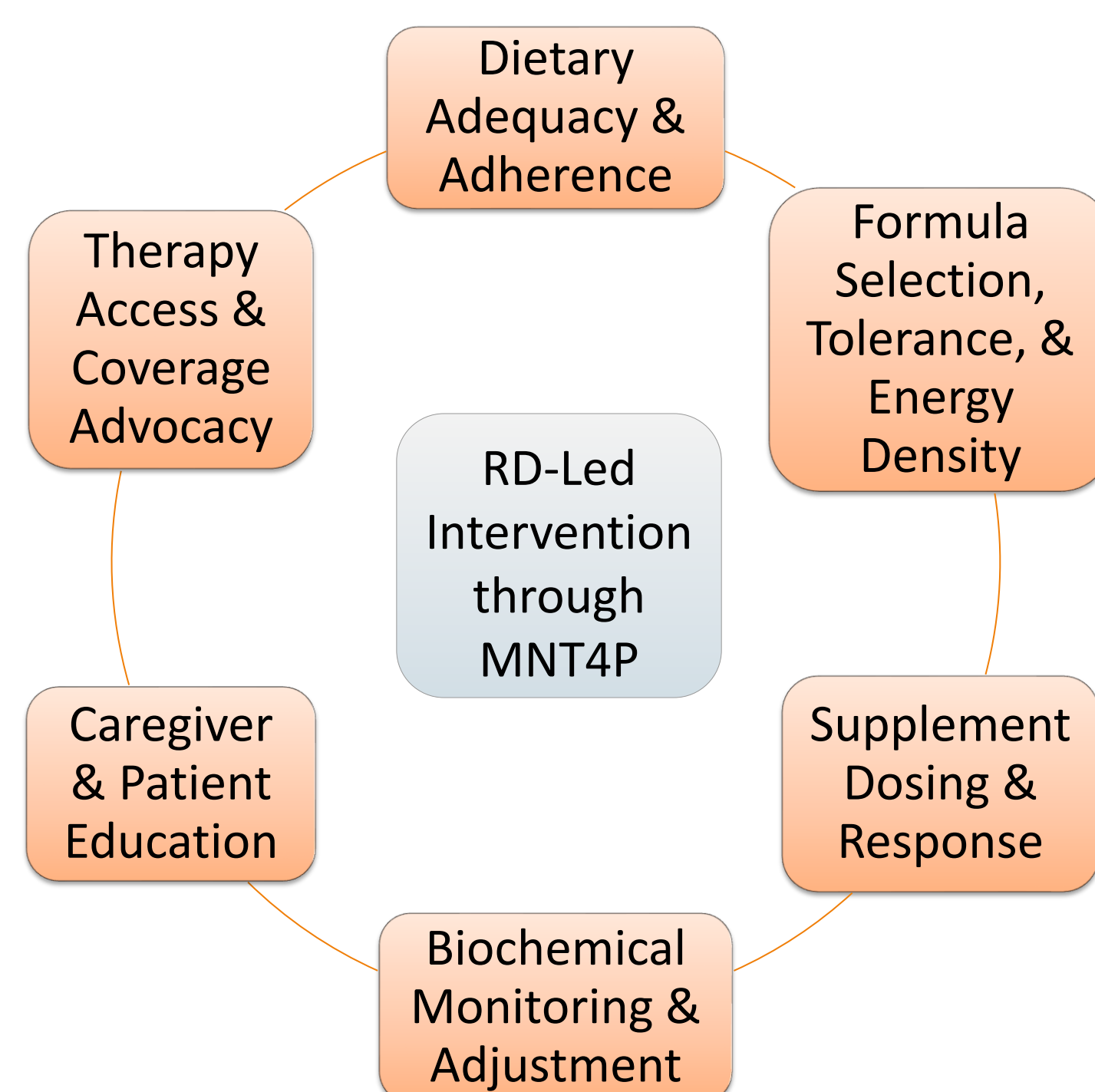
- Homocystinuria (HCU) due to cystathionine-beta synthase deficiency results in elevated total homocysteine (tHcy) from impaired methionine metabolism.
- Clinical severity varies and is influenced by B6 responsiveness.
- Management includes diet, specialized formula, and pharmacologic therapy (i.e., betaine).
- Lack of standardized B6 trial protocols in the literature complicates clinical decision-making.<sup>1</sup>
- Dietitians play a central role in long-term care.

## Objectives

- Compare clinical and nutritional management across 3 HCU cases.
- Identify practice gaps in dietitian-led care.

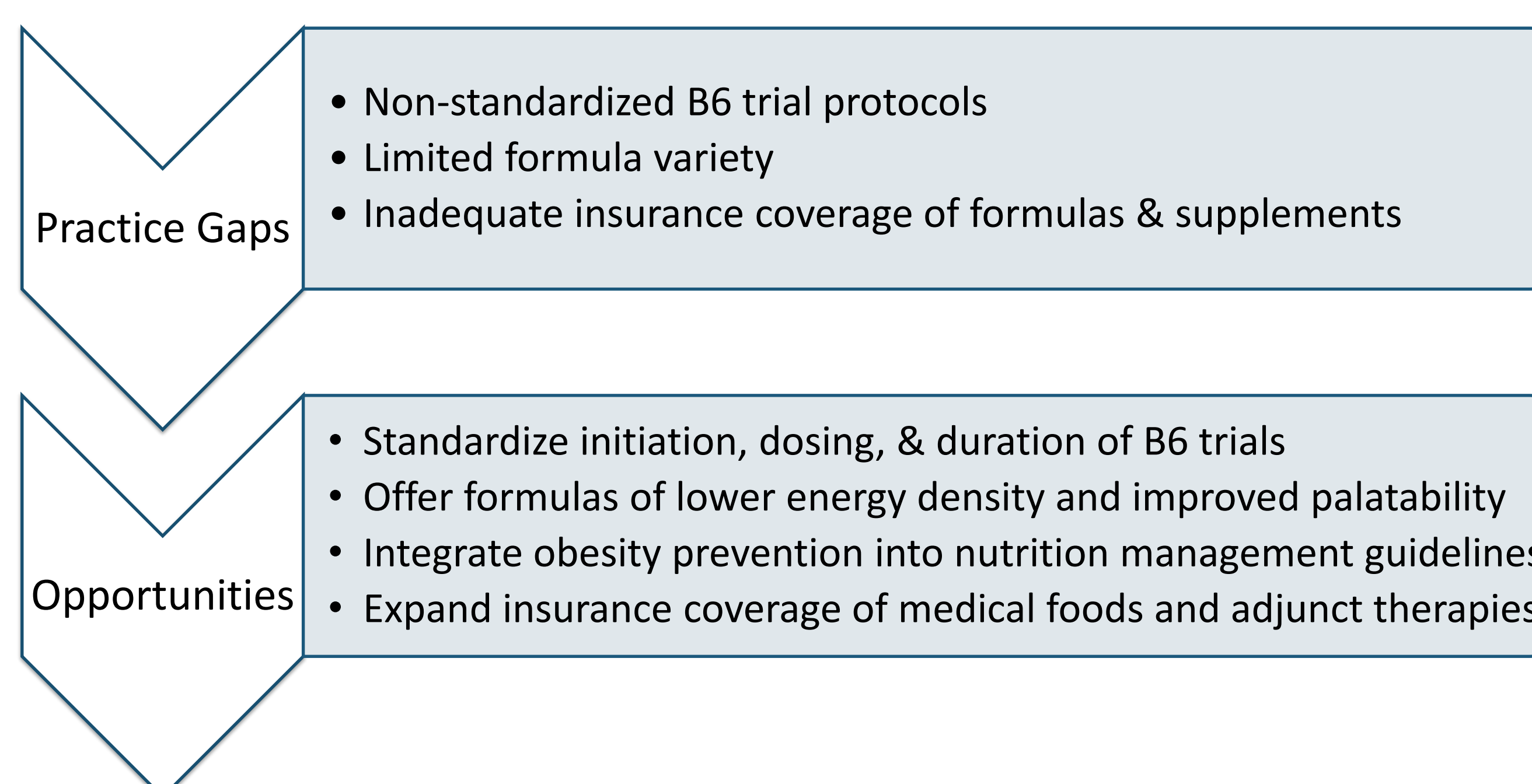
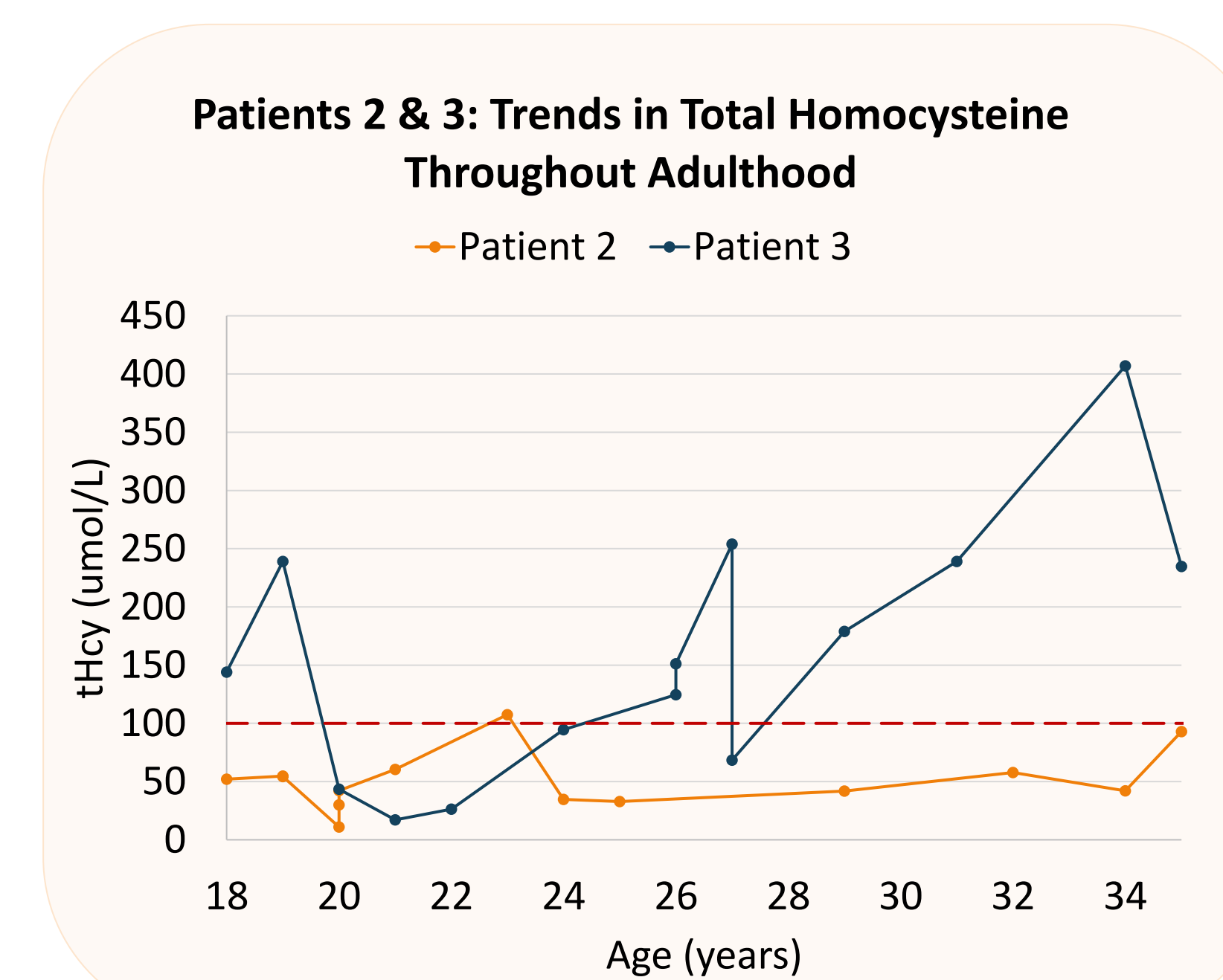
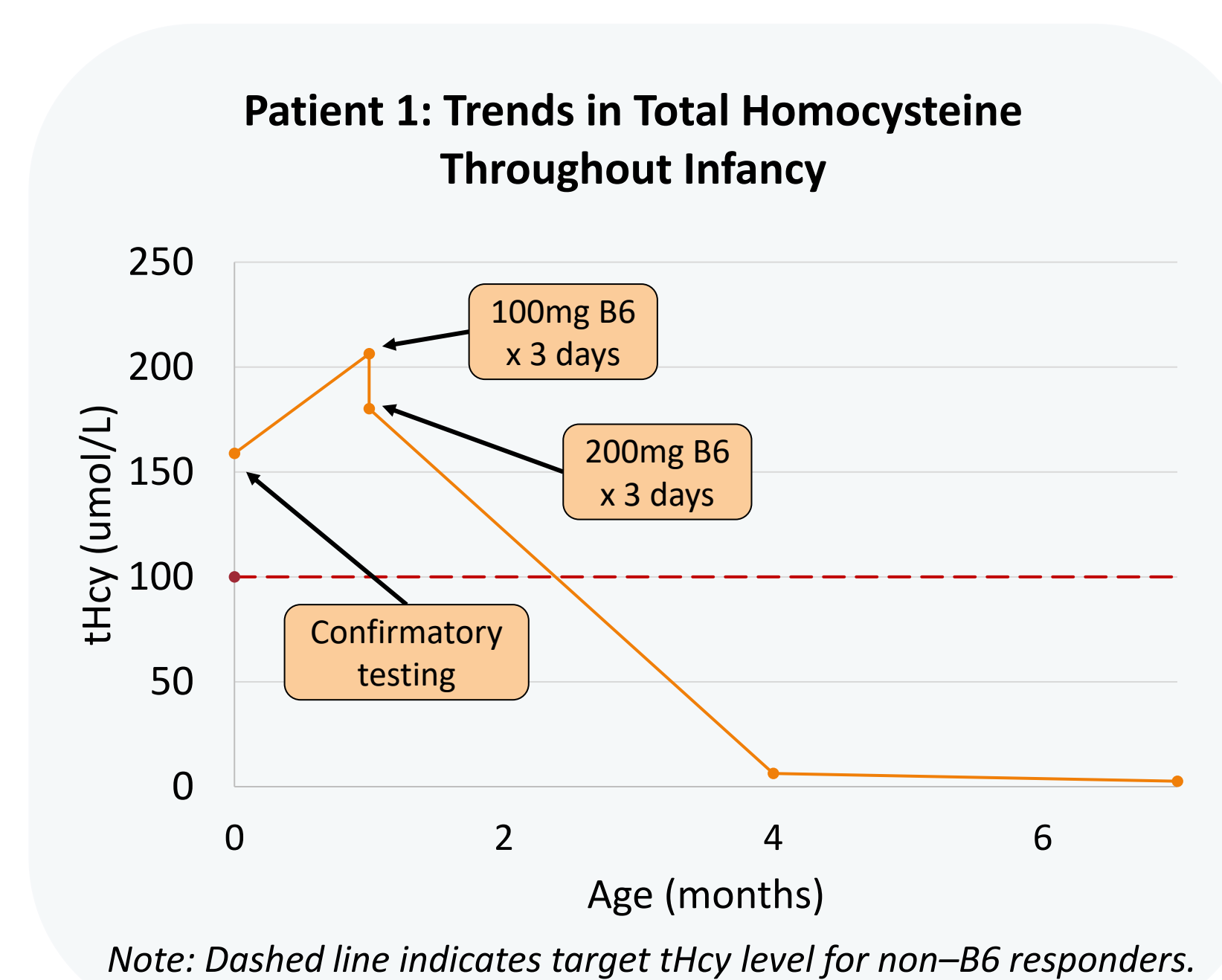
## Methods

- Performed retrospective chart review of 3 patients with molecularly confirmed HCU.
- Extracted data from medical records:
  - NBS results and biochemical trends (tHcy and related markers)
  - Clinical and genetic characteristics (genotype, B6 responsiveness, trial approaches)
  - Nutrition prescriptions, adherence, and barriers of care
- Evaluated dietitian-led interventions and patterns.



	Patient 1: 10 m.o. M	Patient 2: 35 y.o. M	Patient 3: 35 y.o. F
<b>Diagnosis</b>	NBS	NBS	Detected with ectopia lentis at age 2
<b>Molecular</b>	p.G85R (P) & p.D376dup (VUS)	p.I278T (P) & p.D376N (P/LP)	Homozygous p.V320A (P)
<b>B6 Response</b>	Non-responsive	Partially-responsive	Non-responsive
<b>Anthropometrics</b>	<b>Weight:</b> 12 kg <b>Weight-for-length:</b> 96%tile	<b>Weight:</b> 108 kg <b>BMI:</b> 33 kg/m <sup>2</sup>	<b>Weight:</b> 92 kg <b>BMI:</b> 34 kg/m <sup>2</sup>
<b>Nutrition Prescription</b>	<b>Intact protein:</b> 5g <b>Formula:</b> 110g Hominex-1 + 20g Enfamil Gentlease <b>Therapies:</b> Betaine, folic acid	<b>Intact protein:</b> 40g <b>Formula:</b> 2 HCU Lophlex LQ + 44 HCU Easy Tablets <b>Therapies:</b> Betaine, B6, B12, folic acid	<b>Intact protein:</b> 12g <b>Formula:</b> 160g Hominex-2 <b>Therapies:</b> Betaine, B6, B12, folic acid
<b>Barriers</b>	<ul style="list-style-type: none"> <li>• <b>Energy</b> estimation in setting of accelerated growth</li> <li>• <b>Access:</b> <ol style="list-style-type: none"> <li>1. Supplements</li> <li>2. Medication</li> <li>3. Culturally-tailored educational materials</li> </ol> </li> </ul>	<ul style="list-style-type: none"> <li>• <b>Poor tolerance</b> to energy-dense formulas</li> <li>• Lack of <b>access</b> to low-energy formulas</li> <li>• Difficulty maintaining protein restriction while pursuing <b>weight loss</b></li> </ul>	<ul style="list-style-type: none"> <li>• Treatment <b>non-adherence</b> related to intellectual delay, frequent guardianship changes, and lost to follow-up</li> <li>• <b>Access</b> to formula due to insurance denial</li> </ul>

## Metabolic Control Differs Across Cases and Is Associated with Early Detection and RD Support



## Results

- Patients 1 and 2 demonstrated better metabolic control than Patient 3, associated with early NBS detection, family support, and ongoing dietitian involvement.
- Protein restriction adherence remained challenging for Patients 2 and 3, with concerns of excess energy intake from formulas.
- B6 trial protocols varied across cases, reflecting lack of standardization.
- MNT4P supported care through education, supplement access, WIC coordination, and translated materials.

## Conclusion

- Dietitian-led care is critical to effective HCU management.
- Early detection, consistent RD involvement, and structured support programs promote improved metabolic outcomes.
- Inconsistent B6 trial practices highlight a need for standardization.
- Key challenges include adherence, risk of obesity, and access to medical nutrition therapies.
- Expanded RD-led follow-up models and reimbursement are needed to support frequent monitoring and improve long-term outcomes.

## References

1. Morris AA, Kožich V, Santra S, et al. Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency. *J Inherit Metab Dis.* 2017;40(1):49-74. doi:10.1007/s10545-016-9979-0

## Acknowledgments

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