For your doctor

You, Me, and OTC

What you need to know about OTC when making treatment decisions

Ornithine transcarbamylase (OTC) deficiency

OTC deficiency is a rare metabolic disorder of the urea cycle that results in elevated ammonia, putting patients at risk of neurological complications and hyperammonemic crises.[1]

OTC deficiency is the most common subtype of urea cycle disorder (UCD). It is an X-linked disorder; however, carriers of OTC deficiency **can and do experience elevated ammonia and associated symptoms.**[1-3]

Heterozygous OTC deficiency carriers can present outside of childhood with nonspecific symptoms such as lethargy, headache, and natural protein aversion. Elevated ammonia and the associated symptoms can range from asymptomatic to severe due to the pattern of X-chromosome inactivation in the carrier's liver.[1,3]

OTC deficiency can become life-threatening at any age. **Triggers such as dieting, protein consumption, exercise or overexertion, puberty, menstruation, pregnancy/postpartum, menopause, and stress can result in elevated ammonia and a hyperammonemic crisis.**[3-7]

Immediate consequences of elevated ammonia can include lethargy, nausea, mood or behavioral changes, slurring words, headaches or migraines, forgetfulness, vomiting, hyperactivity, coma, and death.[8-11]

Implications of elevated ammonia over time can include **neuropsychological complications** like learning disabilities, intellectual disabilities, and executive function deficits.[2,12]

Carriers who experience symptoms may need to follow a low-protein diet, take dietary supplements, or take a nitrogen-scavenger medication.

Considerations for someone with OTC deficiency

- **Nutrition and calories:** People on a low-protein diet can have difficulty getting enough calories and nutrients and/or maintaining a healthy diet.
- **Exercise and activity:** Overexertion can lead to catabolism, resulting in elevated ammonia.
- **Stress:** Stress can trigger a rise in ammonia levels, so its management is important.
- **Menstruation:** OTC carriers are at risk for metabolic decompensation and elevated ammonia during menstruation. Women may require increased caloric intake or medication adjustment during that time.[7]
- **Postpartum:** OTC carriers should be closely monitored for any signs of postpartum psychosis, which could be an indicator of life-threatening hyperammonemia.[5]
- **Surgery/medications:** Consider OTC deficiency and consult the patient's metabolic specialists when planning for procedures or prescribing medications. Surgery, trauma, or certain medications can trigger elevated ammonia and associated symptoms.[3]

My OTC Information

This sheet will help you learn more about how I am managing my OTC deficiency so you can consider this information when making health-related decisions.

My name: _____

My date of birth: _____

My OTC healthcare team

Name	Type of Doctor	Phone	Email

My OTC management plan

My daily protein allowance: _____ grams

My dietary supplements and medications		
Supplement/Medication	Dose	Time of Day

Other things I do to manage my OTC deficiency

- _____
- _____
- _____
- _____
- _____

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