

## Welcome to the Boston Children's Hospital Transition Toolkit!

As a teenager getting ready for adulthood, or as a young adult, you can begin to take control of how you handle your metabolic condition and your health in general. This Toolkit is designed to help you! To start, read through these materials and fill out the forms. You can also begin to look for an adult-focused doctor to replace your pediatrician who only deals with childhood problems. This process of switching to an adult-focused doctor is called *medical care transition* and takes time and planning – but once it's done, it will be great for your future health and success.

### Use the following forms to help achieve your medical care transition



**Health Readiness  
Assessment**



**Metabolic  
Condition Basics**



**Medical Health  
Summary**



**Transition Plan**

What am I supposed to do with these **forms**?

**Measure** your health independence by taking the [Health Readiness Assessment](#) and over time try to answer yes to every question.

**Read** up on your [Metabolic Condition Basics](#) and discuss possible adult-health problems with your doctors or nurses.

**Ask** a doctor or social worker for help filling out the [Medical Health Summary](#) and [Transition Plan](#) at your next appointment.

**Save** these forms on your computer and USB flash drive and bring the Transition Toolkit to every medical appointment.

**Keep** the flash drive in your wallet or purse and save any changes or updates.



Save these forms and bring them to future medical appointments



Ask a doctor to help you fill out your forms

How will The **Transition Toolkit** help me? With the Toolkit you can –

**Decide** when you're ready to act as your own health spokesperson.

**Prepare** for doctor's appointments and fill out health forms.

**Talk** to doctors and nurses about your medical condition and what roles they should play in your medical care.

**Keep track** of all of your important medical and condition information.

To get started, fill out the [Health Readiness](#) form.

# Transition Toolkit

## Health Readiness Assessment

Are You *Ready* to Handle Your Own Health Care as an Independent Adult?



To know if you are ready for a health care transition, answer the following questions:

1. I know my height, weight, birth date, and social security number  Yes  No
2. I know the name of my condition  Yes  No
3. I know my genotype or genetic status  Yes  No
4. I can tell you about my condition and explain my health care needs  Yes  No
5. I know what kind of medical insurance I have  Yes  No
6. I know where to find my medical records  Yes  No
7. I know who to ask about getting my own health insurance  Yes  No
8. I know the basic treatment for my condition  Yes  No
9. I know what my diet should include and not include  Yes  No
10. I know how to prepare my own food  Yes  No
11. I know how to shop for my own food  Yes  No
12. I know the names of my supplements and medicines  Yes  No
13. I know how to order or get my medications filled  Yes  No
14. I know the symptoms and medical problems related to my condition  Yes  No
15. I visit my metabolic specialist regularly  Yes  No
16. I have discussed issues about sex with my doctor  Yes  No
17. I know how to get birth control and how to stay protected against sexually transmitted diseases/infections (STDs)  Yes  No
18. I have discussed the use of tobacco, alcohol and drugs with my doctor  Yes  No
19. I know how to schedule a doctor's appointment  Yes  No
20. I keep a schedule of medical appointments on my calendar  Yes  No
21. I can get myself to medical appointments (or can arrange transportation myself)  Yes  No
22. I know who to call in case of an emergency  Yes  No
23. I prepare questions for my health care provider before appointments  Yes  No
24. I have a support network for my condition  Yes  No

## Transition Toolkit

### Health Readiness Assessment

Are You *Ready* to Handle Your Own Health Care as an Independent Adult?



Now count the number of times you checked YES.

If you checked YES for:

#### **17 to 24 statements – great work!**

You are already taking full responsibility for your health care. The transition forms in this Toolkit will help you become a stronger health advocate.

#### **8 to 16 statements – you are on the right path!**

You are actively taking on many important responsibilities in your health care. The following Transition Toolkit forms will help you answer YES to many more of these questions.

#### **0 to 7 statements – let's get started!**

Transitioning health care to adult providers and becoming a better health advocate for yourself takes time and planning. Now's a perfect time to start! Pick a few of the previous responsibilities from the list and spend the next month doing them. The Toolkit forms will make this much easier.

Now what?

Update and save this assessment every month until you've answered all of the tasks with a YES!

Next, take a look at some [basic information about your metabolic condition](#).



#### Overview of the Condition:

NAGS deficiency is a very rare metabolic condition in which a chemical called N-acetylglutamate synthase (NAGS) does not work correctly in your body. This can cause ammonia to build up in your blood over time. When ammonia builds up too much, it can cause serious health problems. You can manage NAGS deficiency throughout your life with a special low-protein diet, drug therapy, and ongoing health care.

NAGS deficiency is also sometimes called N-acetylglutamate synthase deficiency.

#### Medical Problems for Babies and Children:

- If NAGS deficiency goes undetected, babies and children may have: extreme sleepiness, irritability, vomiting, poor appetite, trouble with breathing or body temperature, unusual body movements, seizures, or coma. These symptoms can be triggered by eating a lot of protein, or after an illness or surgery, or by steroid medication.
- If untreated, developmental problems for babies and children with NAGS deficiency can include delayed growth or learning delays.
- Newborns in some US states are now tested for NAGS deficiency. If the condition is identified early and treatment and diet are followed carefully, health problems can be lessened.

#### Medical Problems for Teens and Young Adults:

- Teens and young adults with NAGS deficiency may have learning disabilities.
- If you have an illness, fever, surgery, or are pregnant, *contact your health-care provider right away.*
- Watch for signs of ammonia build-up (hyperammonemia) including: nausea, vomiting, sleepiness, or unusual problems with your mood or thinking. If you have any of these, *get medical care right away.*
- Some teens and young adults with NAGS deficiency have anxiety or depression. Counseling and medication can help. If you have anxiety or depression, *it is important to get in touch with your health care provider to get treatment.*

#### How to Minimize Medical Problems and Complications:

- Follow a low-protein diet recommended by your dietician.
- Take medicines recommended by your health care providers, which may include Carbaglu® (also known as carglumic acid).
- Avoid steroids in medication or supplements.
- Avoid medicines that contain valproic acid, including Depakote®. Check with your doctor to find out which other medicines contain valproic acid.
- Keep your immunizations up-to-date.
- Stay in regular contact with your health care providers and health specialists.
- Keep an **Acute Illness Protocol** that you can bring with you to the emergency room: <http://newenglandconsortium.org/for-professionals/acute-illness-protocols/urea-cycle-disorders/neonate-infant-child-with-hyperammonemia/>



#### Fertility and Pregnancy:

- NAGS deficiency does not affect your ability to have children.
- Genetic counseling can help you and your partner understand the risks to your children. Your doctor can refer you to a genetic counselor who has a special understanding of your health issues.
- Pregnant women with NAGS deficiency must follow a careful diet with special guidance from a dietician and health care providers.

#### How To Get Support:

- Get education support from teachers and specialists at your school.
- Join a support group with people who have NAGS deficiency or similar metabolic conditions. This group of health conditions is called **urea cycle disorders**.
- A lot of research is being done on NAGS deficiency and other urea cycle disorders. Get up-to-date information about new treatments and discoveries at the **Urea Cycle Foundation** website: <http://nucdf.org/>

#### Resources:

AAP/AAFP/ACP Transition Clinical Report

<http://aappolicy.aappublications.org/cgi/reprint/pediatrics:128/1/182.pdf>

Transition Toolkit (New England Consortium of Metabolic Programs)

<http://newenglandconsortium.org/for-families/transition-toolkit/>

Got Transition

<http://www.gottransition.org/>

National Urea Cycle Disorders Foundation

<http://www.nucdf.org/>

Acute Illness Protocol

<http://newenglandconsortium.org/for-professionals/acute-illness-protocols/urea-cycle-disorders/neonate-infant-child-with-hyperammonemia/>

National Institutes of Health – Genetics Home Reference – NAGS deficiency

<http://ghr.nlm.nih.gov/condition/n-acetylglutamate-synthase-deficiency>

#### Genetics Referrals:

Clinical Services

<http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests>

Genetic Services

<http://www.acmg.net/gis>

## Transition Toolkit

### Metabolic Condition Basics: NAGS deficiency

Medical information you need to know as an adult with NAGS deficiency



#### What Next?

Now you can fill out the [Medical Health Summary](#), print it, and save it. This will help you keep important medical information in one place.

This guide was adapted with permission from the American College of Medical Genetics ACT Sheet <http://www.acmg.net/>

**Transition Toolkit**  
**Medical Health Summary**

Use this form to save important health information



Date Updated: \_\_\_\_\_

Last Name		First Name	
Birthdate	Gender	Weight	Height
Address			
City		State	Zip Code
Insurance Name			
Insurance ID & Group #			
Insurance Address & Phone Number			
Metabolic or Genetic Condition			
Medications/Supplements and Dosages			
Allergies to Food, Drugs and Other Substances			

Emergency Contact Name & Relationship		Emergency Contact Phone Number
Pharmacy Name		Pharmacy Phone Number
Immunizations:	Type	Date



**Transition Toolkit**  
**Medical Health Summary**

Use this form to save important health information



Other medical information:

What next?

A final step toward completing your medical transition is meeting with your health care providers to discuss and fill out your [Transition Plan](#).





## Transition Toolkit

### Transition Plan

Make an ongoing health plan with your health care provider



Fill out this plan with your health care provider, to help you transition to being in charge of your own health

**Patient Name:** \_\_\_\_\_

**Boston Children's Hospital Medical Record #:** \_\_\_\_\_

Primary Care Physician	Phone Number
	Email
Pediatrician	Phone Number
	Email
Metabolic Specialist	Phone Number
	Email
Other Health Care Provider	Phone Number
	Email
Emergency Contact Name & Relationship	Phone Number

#### Medical Findings

Genotype/Laboratory Findings
Treatment Status
Current Medications/Treatments
Allergies/Adverse Reactions
Past Medical Concerns or Hospitalizations
Living Situation/Employment
Mental Health Status/Cognitive Status



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### Transition Plan

Make an ongoing health plan with your health care provider



Other Health Risks

#### Contract:

To enhance collaboration, each person should agree on the following responsibilities:

Metabolic Specialist will:

- Obtain labs and tests: For example...
- Prescribe condition-related medications:
- Work with PCP to develop an emergency protocol:

Patient will:

- Remember to take medications: For example...
- Order and pick up supplies/supplements:
- Show-up to appointments:

Primary Care Physician will:

- Monitor patient's general health:
- Update metabolic specialist with important patient information:
- Contact metabolic specialist with medical concerns: For example...
- Other:

When you're done filling out this Transition Plan, print it and save it in a safe place.

Congratulations! You are well on your way to being in charge of your own health.