



# Children's Hospital Boston Transition Toolkit

Welcome to the **Children's Hospital Boston Transition Toolkit**! As a teenager ready for adulthood, you should begin to take control of how you handle your metabolic condition and your health in general. To start, read through these materials. You should begin to look for an adult-focused doctor to replace your pediatrician who only deals with childhood problems. This process of finding an adult-focused doctor is called a medical care transition and takes time and planning — but once it occurs, it is a great 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](#) and 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?

*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.

## Personal Health 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 |

## Personal Health Assessment

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



Now count the number of times you circled YES. If you circled YES to:

17 to 24 statements, great work! You are already taking full responsibility for your health care. The transition documents 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 documents 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 transition documents 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](#).

## Metabolic Condition Basics: Biotinidase Deficiency

Medical information you need to know as an independent adult with a biotinidase deficiency



### Overview of the Condition:

This is a genetic problem that causes the body to not be able to breakdown and process a vitamin called biotin.

### Medical Problems for Babies and Children:

- Symptoms of this condition in babies and children include seizures, loss of hair, learning problems and developmental problems, hearing loss, and skin rashes
- If the condition is *not* found when a baby is born and not treated, kids may experience any or all of these problems

### Medical Problems for Teens and Young Adults:

- To avoid developing these problems, teens and young adults must take biotin as prescribed by a doctor
- Some females reported losing their hair but this was fixed by increased doses of biotin as prescribed by their metabolic doctors
- Adults are at risk for developing the symptoms if they do not maintain good levels of biotin in their blood
- Some adults may have less-serious forms of this condition and they may experience weak muscles or skin rashes during times of stress

### How to Avoid Medical Problems and Complications:

- Take supplements of biotin as prescribed by a doctor, usually 5 to 10mg per day
- Avoid raw eggs
- Schedule regular eye-sight and hearing check ups

### Pregnancy and Other Medical Events

Most medical experts believe that fertility is not affected in men and women who take their medications and stick to special diets.

### Resources:

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition=biotinidasedeficiency>

Biotinidase Deficiency Family Support Group: <http://biotinidasedeficiency.20m.com/>

### 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.

## Metabolic Condition Basics: Galactosemia

Medical information you need to know as an independent adult with galactosemia



### Overview of the Condition:

Galactosemia is a rare genetic condition in which the body cannot break down an important substance called galactose. Galactose is a sugar most commonly found in milk and dairy products such as cheese, yogurt, and ice cream. Because people with galactosemia can't break down galactose, it builds up in the body and can cause serious health problems.

### Medical Problems for Babies and Children:

- If galactosemia goes unnoticed, babies are at risk for serious medical conditions like uncontrolled shaking or convulsions, yellow skin and whites of the eyes or jaundice, and vomiting
- Untreated newborns with galactosemia are at increased risk for *E. coli septicemia*, a life-threatening blood infection
- All newborns are now tested for galactosemia and if this condition is caught early and dairy products are avoided, the most serious health problems can be avoided.

### Medical Problems for Teens and Young Adults:

- Almost all people with galactosemia have some difficulties in speech and language such as speaking clearly and coming up with the right word quickly
- Other adults have difficulties quickly understanding and processing what they hear or read
- Other symptoms of galactosemia in adults include tremors and anxiety – some young adults may be shy and feel lonely

### How to Avoid Medical Problems and Complications:

- Avoid milk and other dairy products
- Check labels for foods that include nonfat dry milk solids, casein, sodium caseinate, whey, whey solids, curds, lactose, or galactose on the list of ingredients
- Replace dairy with soy or rice-based products

### Pregnancy and Other Medical Events

- Almost all women experience premature ovarian insufficiency (POI)
- POI occurs when something has happened to the supply of eggs in a young woman's ovaries – women may stop having their periods and may experience hot flashes – infertility or not being able to get pregnant is also caused by POI
- POI can be treated and some women with galactosemia have been able to have children
- Fertility, or the ability to father children, is not affected in men

### Resources:

Parents of Galactosemic Children, Inc. <http://www.galactosemia.org/>  
Learn Genetics Galactosemia: <http://learn.genetics.utah.edu/content/disorders/whataregd/galactosemia/>  
Moving Forward: Your Guide to Galactosemia and Primary Ovarian Insufficiency (POI):  
<http://newenglandconsortium.org/for-families/galactosemia/galactosemia-and-primary-ovarian-insufficiency-poi/>

## Metabolic Condition Basics: Galactosemia

Medical information you need to know as an independent adult with galactosemia



### What Next?

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## Metabolic Condition Basics: Homocystinuria

Medical information you need to know as an independent adult with homocystinuria



### Overview of the Condition:

Homocystinuria is a rare condition where the body can no longer break down an important amino acid called homocysteine. Because people can't break down this chemical, it builds up in the body and can cause serious health problems.

### Medical Problems for Babies and Children:

- Young kids with this condition are at risk for developmental problems including learning problems, seizures and eye problems
  - Eye problems include partial dislocation of the lens, severe nearsightedness, and quivering of the iris
- Babies who do not have good treatment during development tend to have worse disorder-related problems later in life

### Medical Problems for Teens and Young Adults:

- Teens and young adults who do not monitor their blood homocysteine levels are at risk for blood clots
  - Blood clots can lead to very serious problems like blockages of blood to the heart and lungs
- As patients with homocystinuria grow older, they become at risk for developing serious mental health problems including depression, anxiety and even psychosis in some cases, and seizure disorders
- Some young people may develop scoliosis where their spinal columns begin to curve
- Osteoporosis or low bone density often occurs by age 20 and occurs in half of patients

### How to Avoid Medical Problems and Complications:

- In most people, oral doses of vitamin B6 lower blood homocysteine levels
- Special formulas (low-methionine) are used to prevent buildup of homocysteine
- If there is poor or double vision, an ophthalmologist should perform a dilated eye exam to look for eye problems mentioned above.

### Pregnancy and Other Medical Events

- Pregnancies should be handled normally but the mother should be monitored by a doctor very closely
- The child will not inherit the condition unless the father carries the gene that is responsible for homocystinuria

### Resources:

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition=homocystinuria#treatment>

Google Health: <https://health.google.com/health/ref/Homocystinuria>

## Metabolic Condition Basics: Homocystinuria

Medical information you need to know as an independent adult with homocystinuria



### 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.

# Metabolic Condition Basics: Ornithine Transcarbamylase (OTC) Deficiency

Medical information you need to know as an independent adult with an OTC deficiency



## Overview of the Condition:

OTC deficiency is one of a number of urea cycle disorders that happens when there is a problem with the body's natural way of removing toxic chemicals like ammonia from the bloodstream. OTC deficiency is a genetic condition that is inherited from a baby's mother.

## Medical Problems for Babies and Children:

- Babies must get diagnosed and treated right after birth because their ammonia levels will begin to rise right away which can lead to brain damage and death
- If babies survive the newborn period undiagnosed, children with milder forms of this disorder have a variety of hard-to-identify symptoms like hyperactivity, refusal to eat meat or other high-protein foods, and uncontrollable crying
- Often children get misdiagnosed for having Reye's syndrome

## Medical Problems for Teens and Young Adults:

- Sometimes kids are not diagnosed until the teen or adult years – they may experience symptoms such as severe migraines, instances of high levels of ammonia in the blood, or diagnosis of a family member will lead to doctors diagnosing the condition in them
- If treatment stops or does not occur, ammonia levels rise and young adults are at risk for stroke, extreme weakness, liver and kidney problems and severe confusion
  - These symptoms can then lead to more serious ones like brain damage, coma, and death
- In milder cases of OTC deficiency, side-effects include irritability, migraines, sleep problems and unexplained tiredness

## How to Avoid Medical Problems and Complications:

- Eat a very specific diet with limited protein if prescribed by a doctor
- Take supplements to maintain chemical and amino acid levels in the body as prescribed by a doctor
- Take the medications prescribed by your doctor
- The only cure for OTC deficiency is a liver transplant

## Pregnancy and Other Medical Events

- Men and women with less severe forms of OTC deficiency are able to have children
- Mothers are at risk for developing hyperammonia (or too high levels of ammonia) during childbirth and should be monitored closely by doctors

## Resources:

Genetics Home Reference: <http://ghr.nlm.nih.gov/>

National Urea Cycle Disorders Foundation: <http://www.nucdf.org/index.htm>

## 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.

## Metabolic Condition Basics: Phenylketonuria (PKU)

Medical information you need to know as an independent adult with PKU



### Overview of the Condition:

Phenylketonuria (PKU) is a rare condition in which the body cannot break down an important amino acid called phenylalanine (Phe), which is found in all protein foods. Because people can't break down this chemical, it builds up in the body and can cause serious health problems..

### Possible Medical Problems for Babies and Children:

- All newborn babies are tested for PKU
- Babies with PKU who do not have good treatment while growing up tend to have worse PKU-related problems later in life
- If a phenylalanine-restricted diet is followed, babies will develop normally – if not, babies are at risk for mental retardation, delayed social skills, hyperactivity, seizures, skin rashes, and other medical problems

### Possible Medical Problems & Symptoms for Teens and Young Adults:

- Depression
- Anxiety and Panic Attacks
- Memory loss
- Attention difficulties
- Aggressive or angry outbursts
- Migraines
- Eczema
- Sleep problems
- Inability to finish reading a book
- Troubles in social relationships
- Planning, organization and other executive functioning skills
- Breath and skin may smell “musty”

### How to Avoid Medical Problems and Complications:

- In severe forms of PKU, follow a strict low-phenylalanine (low-protein) diet
- Foods to avoid include: all dairy products (products with milk in them), meat, fish, eggs, nuts, regular pastas, regular rice, and artificial sweeteners like NutraSweet (aspartame) commonly found in diet soda.
- Your diet must also include special formulas and specially prepared low-protein foods, which supply protein building blocks and other nutrients needed to keep you healthy.
- Your diet must be monitored by a dietician who is familiar with PKU.

## Metabolic Condition Basics: Phenylketonuria (PKU)

Medical information you need to know as an independent adult with PKU



### New treatments for PKU:

- **BH4 therapy:** This is a medication that boosts the activity of the enzyme that is not working in PKU. People with milder forms of PKU can lower their blood phenylalanine (Phe) levels and can sometimes eat more protein. Some people with classic PKU also benefit from this medicine.
- **Large Neutral Amino Acid therapy:** This is also a medication that competes with Phe transport across the blood-brain barrier. This means that less Phe enters the brain. Note: This medication cannot replace diet during pregnancy.
- **Formulas:** There are many new formulas (medical foods), some of which taste much better than the old formulas.
- **Enzyme replacement therapy:** This therapy is in clinical trials. This means that it is not yet approved for general use. Its safety and effectiveness are being checked

Ask your metabolic doctor or dietician for more information about these advances in therapies for PKU.

### Pregnancy and Other Medical Events

- Mothers-to-be must follow strict low-phenylalanine (low-protein) diets both before becoming pregnant and throughout the pregnancy to avoid developmental problems and mental retardation in their babies
- Pregnancies should be handled normally but the mother should be monitored by a doctor very closely
- PKU is passed on to babies only when both parents carry a copy of the gene

### 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.

### Resources:

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition=phenylketonuria>

PKU Toolkit: <http://newenglandconsortium.org/toolkit/>

PKU.com: <http://www.pku.com/>

National PKU News: <http://www.pkunews.org/index.htm>

# Medical Health Summary



Use this form to save important health information

Date Updated:

Last Name		First Name	
Birthday	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	

## 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 Plan

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



**Patient Name:**

**Children's Hospital Boston 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 Mediations/Treatments
Allergies/Adverse Reactions
Past Medical Concerns or Hospitalizations
Living Situation/Employment
Mental Health Status/Cognitive Status

## Transition Plan

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



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.