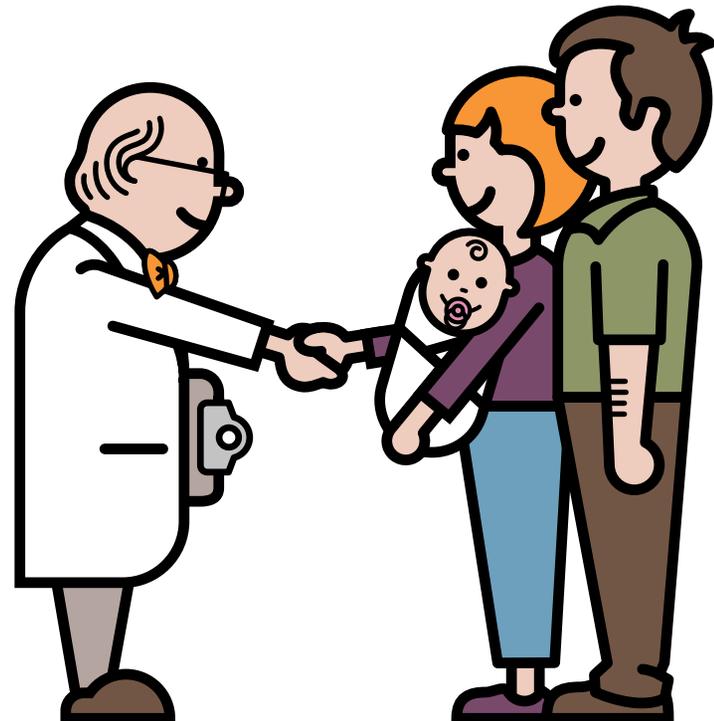


# Homocystinuria

# HCU

Information for families following  
a positive newborn screening



Adapted by the Dietitians Group BIMDG

**BIMDG**

British Inherited Metabolic Diseases Group



BASED ON THE ORIGINAL TEMPLE WRITTEN BY  
BURGARD AND WENDEL

Reviewed & revised for North America by: A. Huber

This version of the TEMPLE tool, originally adapted by the Dietitians group of the BIMDG for use within the UK and Ireland, has been further adapted by Nutricia North America for use within United States and Canada. This version no longer represents clinical or dietetic practice in the UK or Ireland.

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Tools Enabling Metabolic Parents LEarning

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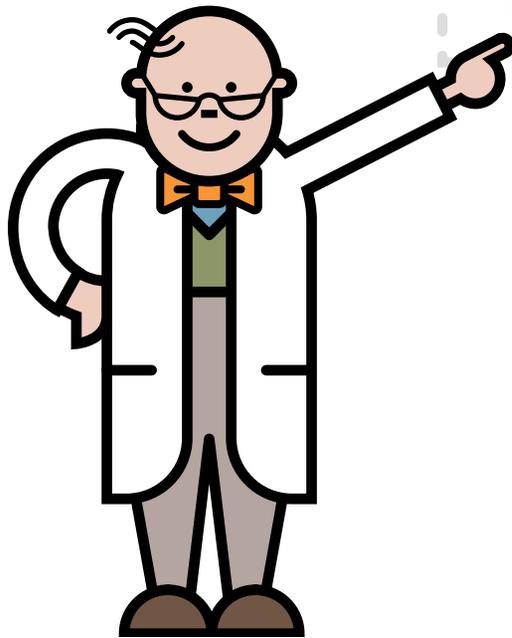
**NUTRICIA**

# What is HCU?

HCU stands for homocystinuria.

It is pronounced ho-mo-sis-tin-ur-ee-a.

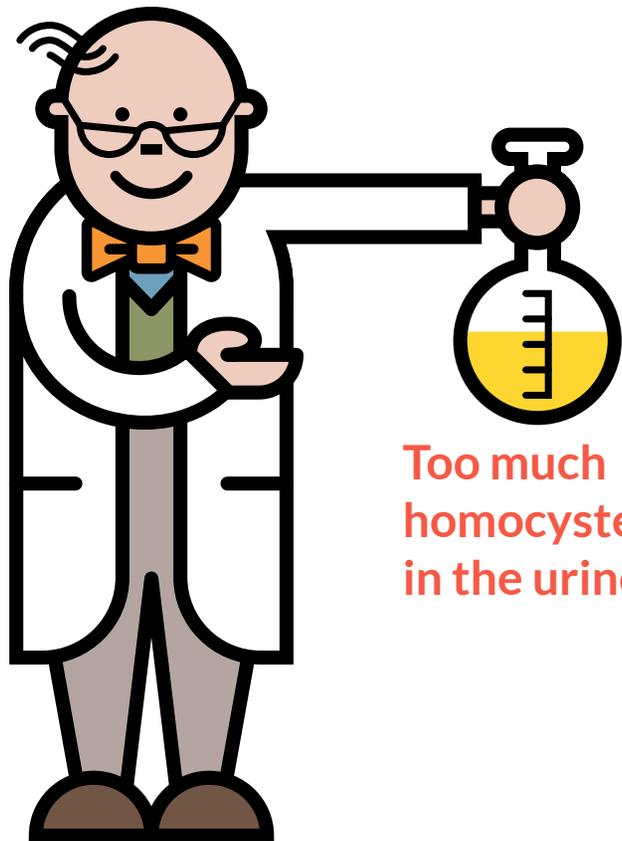
**It is an inherited metabolic condition.**



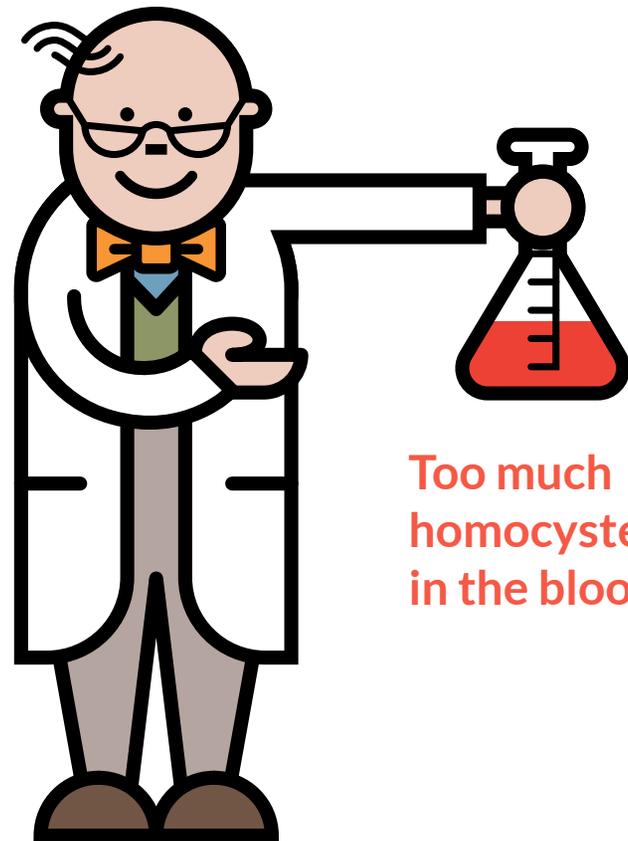
Homo cystin uria

HCU

# What is HCU?



Too much  
homocysteine  
in the urine



Too much  
homocysteine  
in the blood

# How does HCU affect the body?

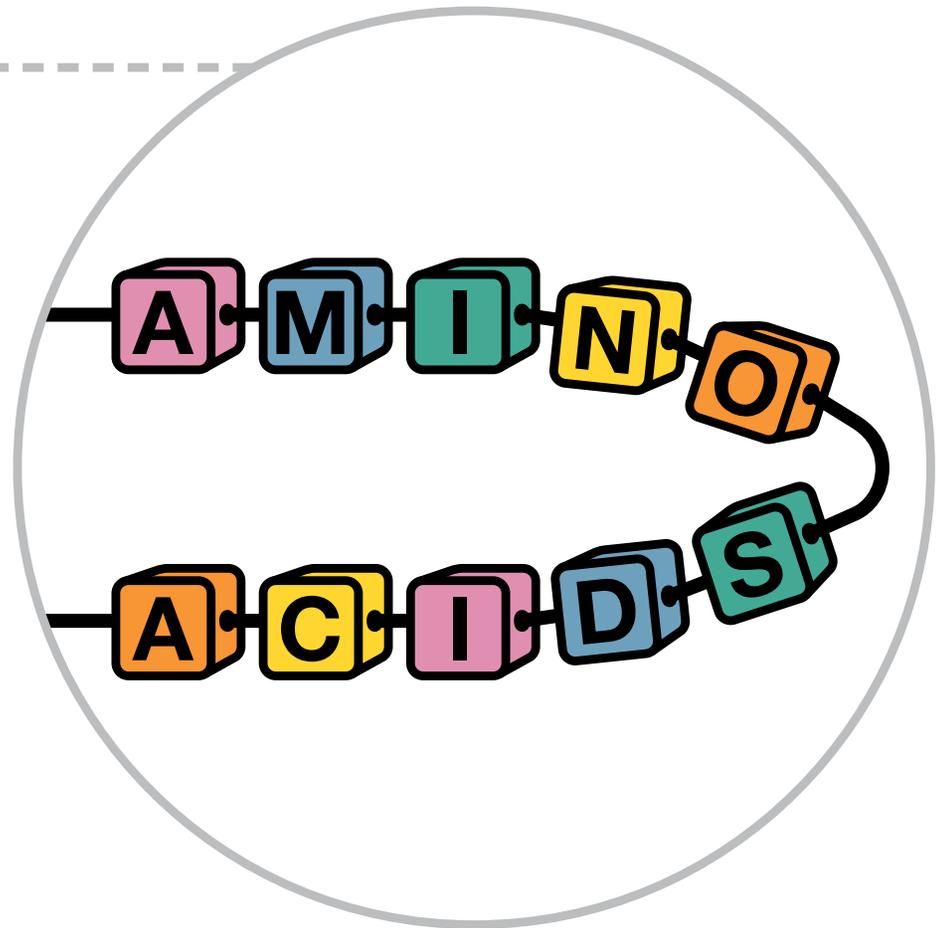
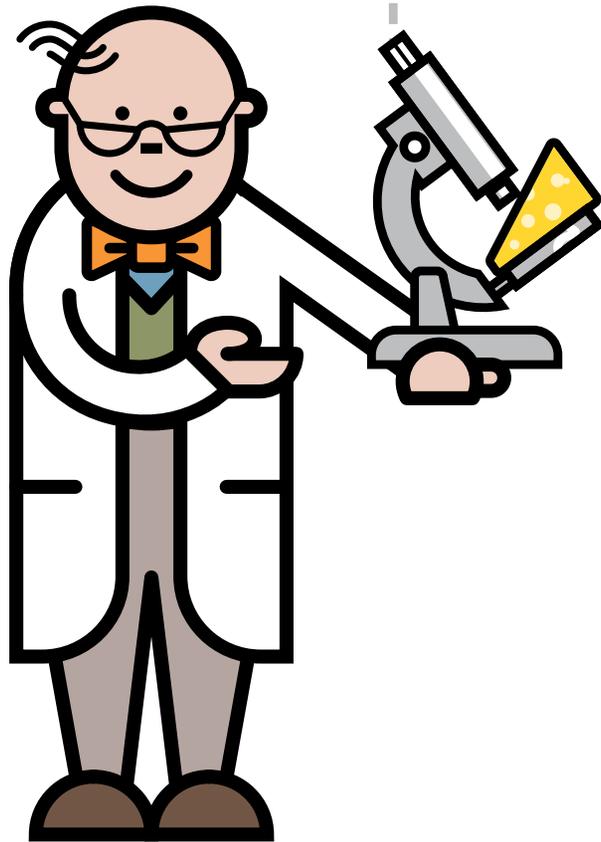
HCU affects the way the body breaks down protein.

Protein is found in our bodies and in many foods. The body needs protein for growth and repair.



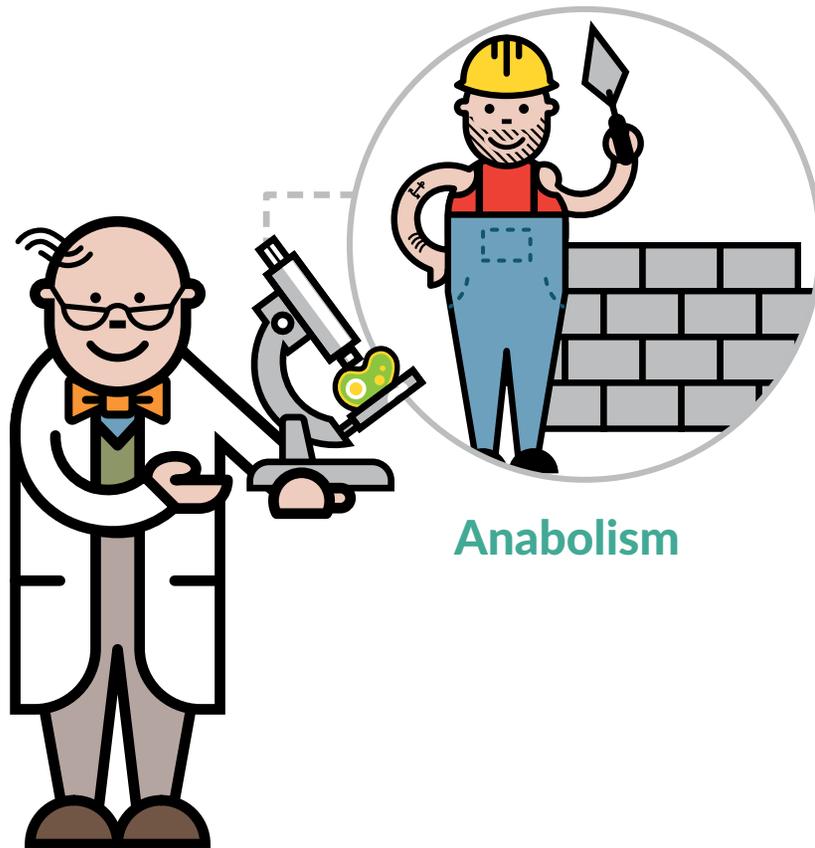
# What is protein?

Protein consists of chains of many smaller units called amino acids.

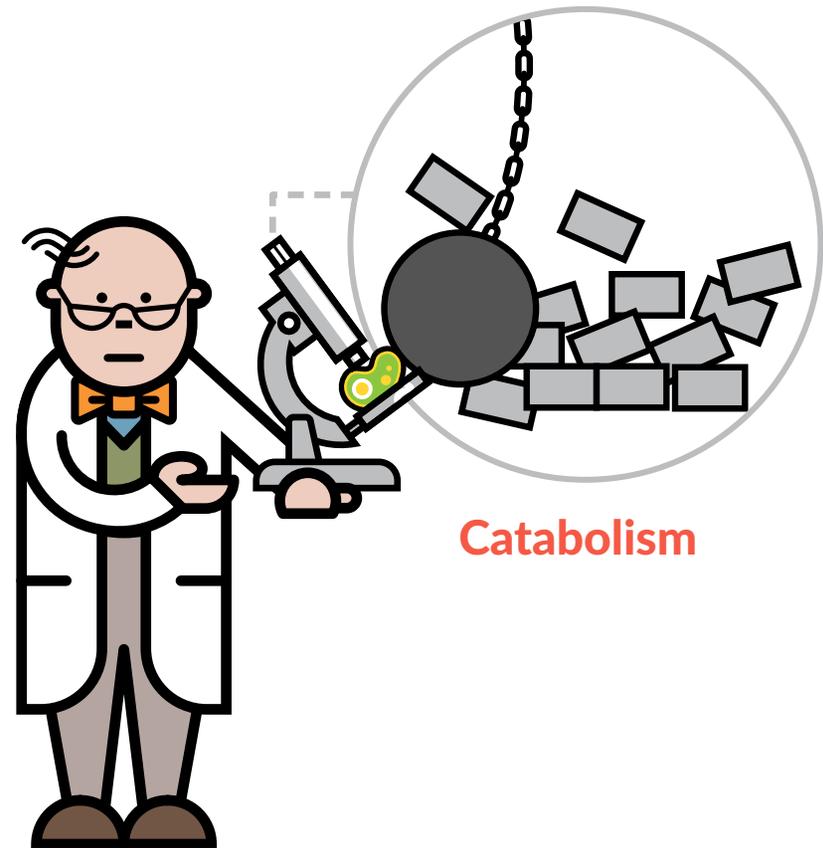


# Protein metabolism

**Metabolism** refers to the processes that occur inside the cells of the body.



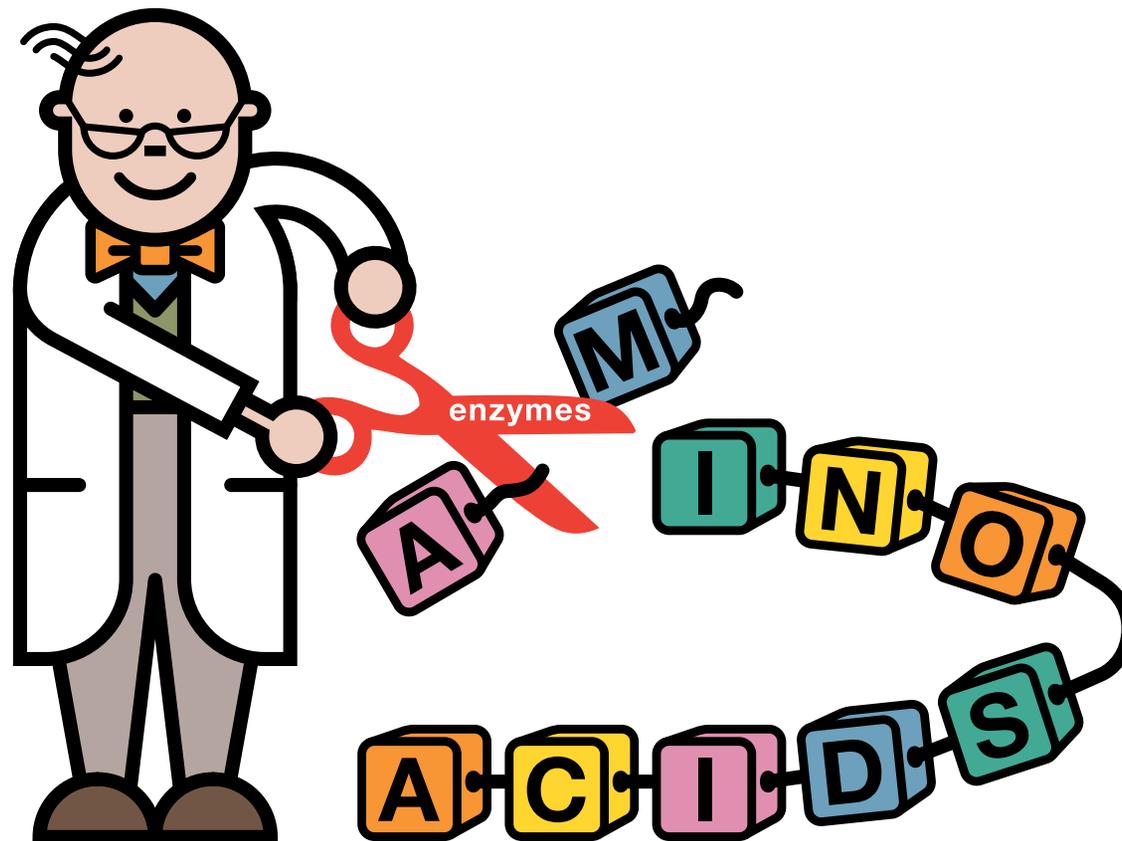
Anabolism



Catabolism

# What do enzymes do?

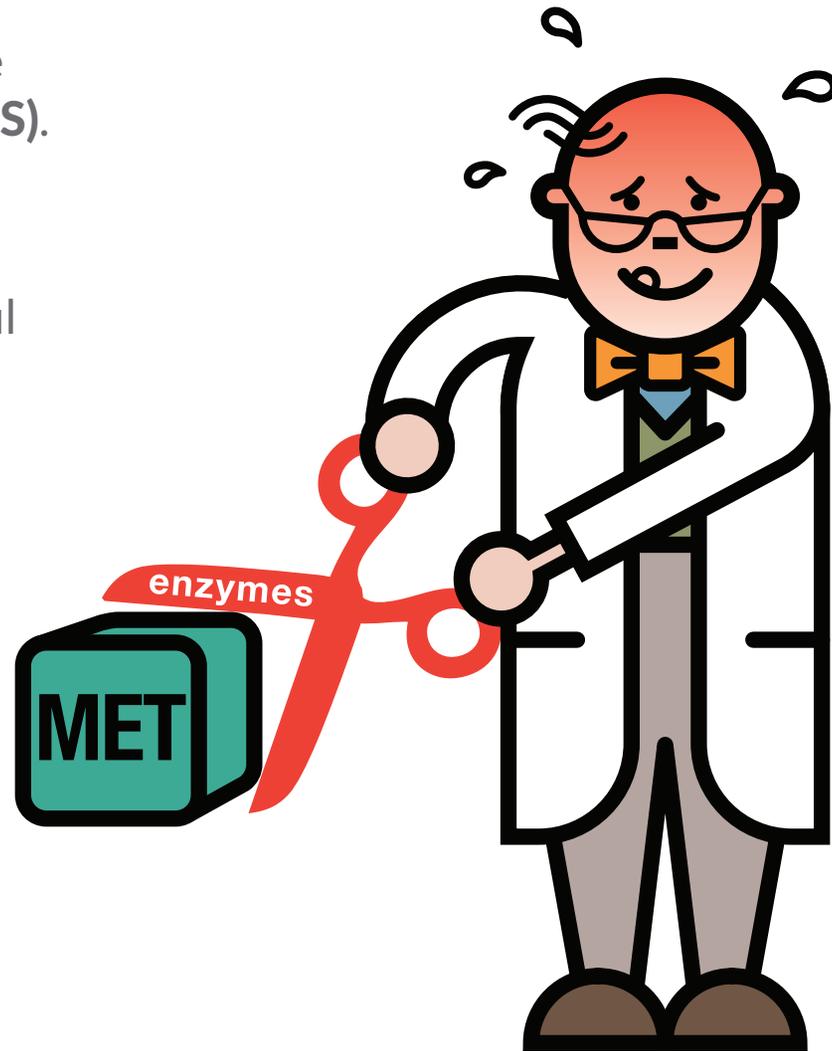
Enzymes help with metabolism by functioning like scissors. They break down proteins into smaller parts, including amino acids.



# What happens in HCU?

HCU is due to a deficiency of an enzyme called **cystathionine beta-synthase (CBS)**.

This results in the body being unable to break down an amino acid called methionine (MET). This causes a harmful buildup of methionine and another substance called homocysteine (HCY) in the blood.



# What can go wrong in HCU?

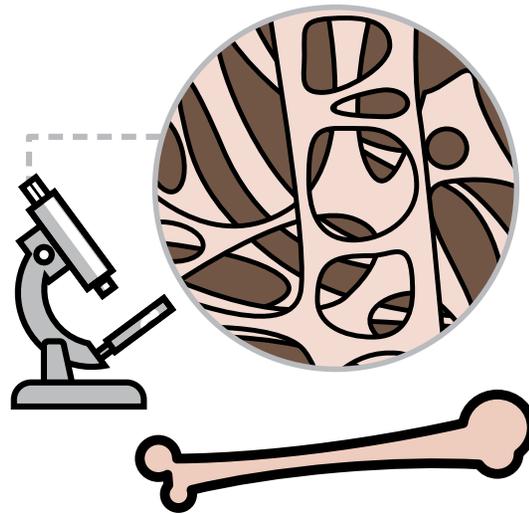
Nearsightedness  
and dislocated  
eye lenses



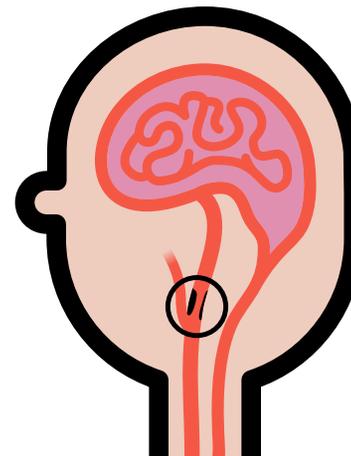
Difficulties  
with learning  
and behavior



Long and  
thin bones



Blood clots and  
strokes



# How is HCU diagnosed?

As part of newborn screening, a few drops of blood are collected.

The blood sample is then analyzed.

High levels of methionine on a newborn screen result could mean your child has HCU, which will prompt your clinician to do further testing to confirm the diagnosis.

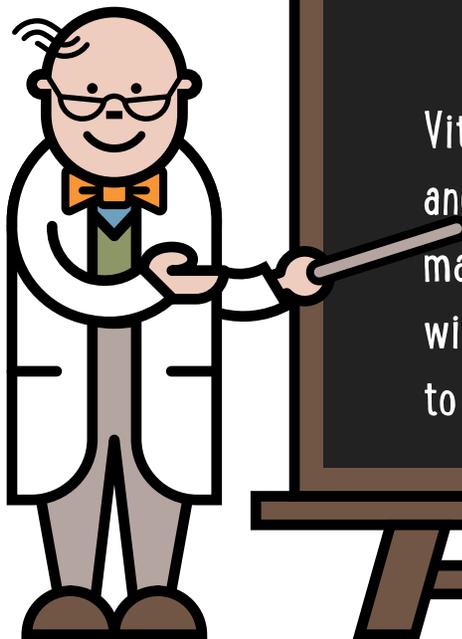


# How is HCU managed day-to-day?

For some individuals with HCU, the enzyme requires the help of a vitamin called Vitamin B<sub>6</sub>, or pyridoxine, together with folic acid to work properly.



Vitamin B<sub>6</sub> helps the enzyme work better, and, if successful, supplementation is the only management needed. Roughly 50% of individuals with HCU are responsive or partially responsive to supplementation.



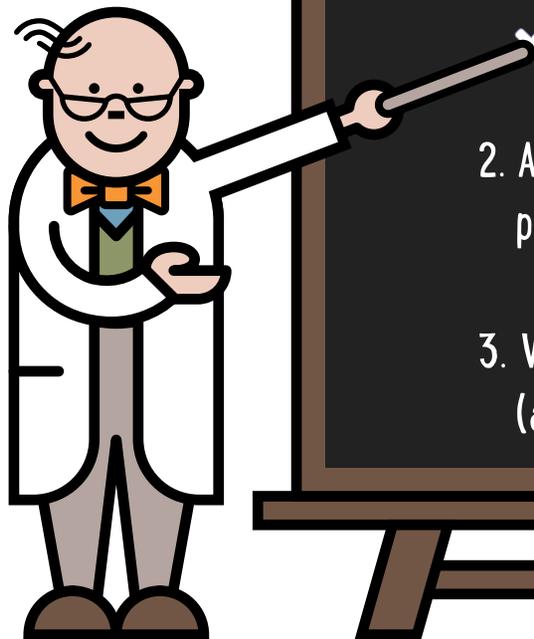
# How is HCU managed day-to-day?

If Vitamin B<sub>6</sub> supplementation is not sufficient, HCU is managed with the following:

1. A methionine-restricted diet
  - ✓ Avoid high protein foods
  - ✓ Include foods low in protein

2. A metabolic formula,  
prescribed by your clinic

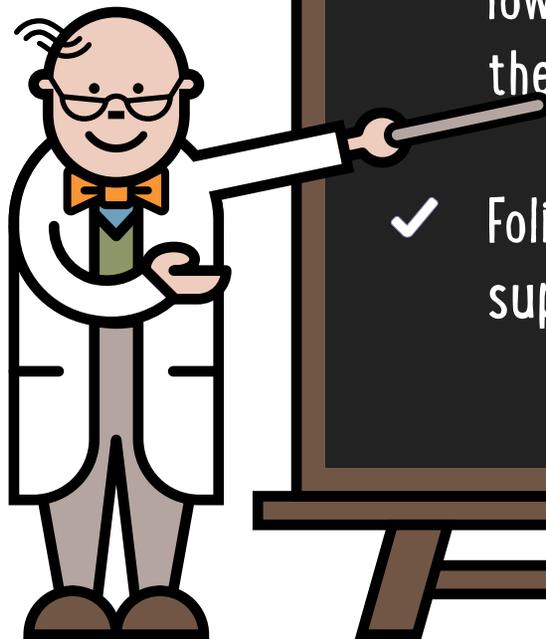
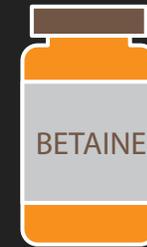
3. When recommended, sometimes cystine  
(an amino acid) supplementation is needed



# How is HCU managed day-to-day?

Other forms of management that may be given are:

- ✓ A drug called betaine. Betaine can help lower the amount of homocysteine in the blood
- ✓ Folic acid, vitamin C, and vitamin B<sub>12</sub> supplements



# Avoid high protein foods

Foods rich in protein are also rich in methionine, and therefore, should be avoided. This includes **meat, fish, eggs, cheese, milk, bread, pasta, nuts, soy and tofu.**



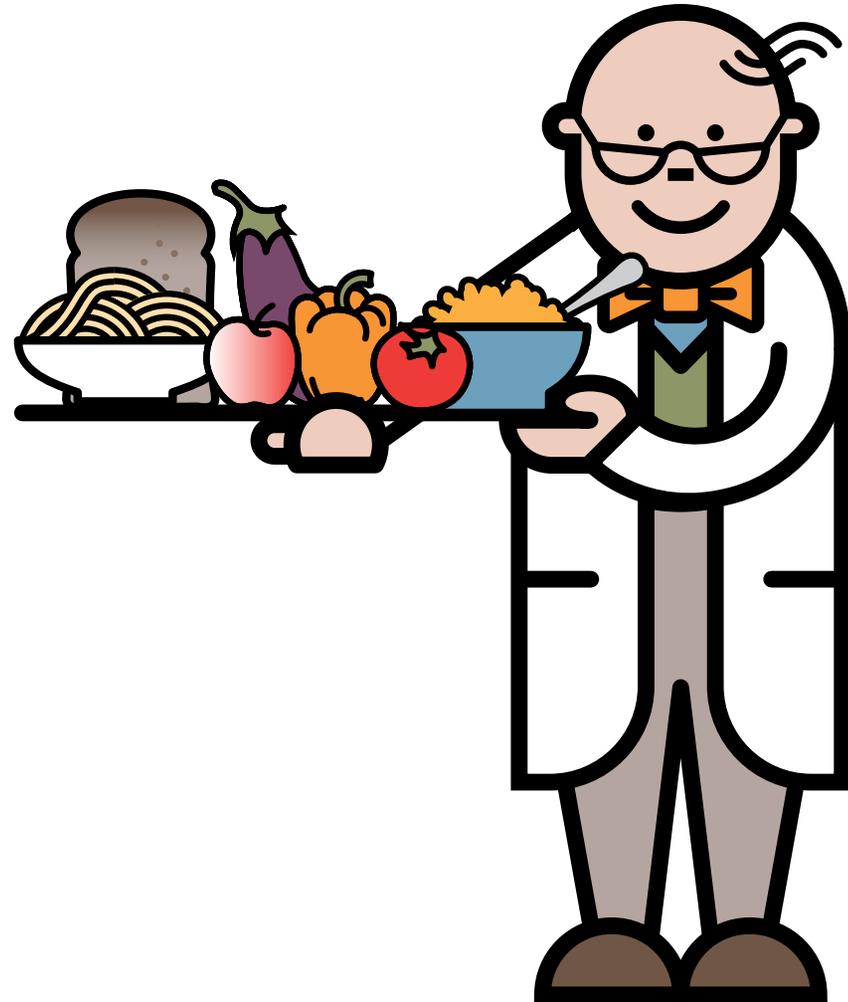
# Include foods low in protein

These are foods that contain small amounts of methionine which can be used in typical quantities.

They include many fruits and vegetables, and specially formulated low protein foods.

They provide:

- An important source of energy
- Variety in the diet



# Low protein cooking

Cooking low-protein meals for your child can still be appealing to the eye and taste good.

There are many low-protein cookbooks to choose from. Your dietitian may be able to recommend a few favorites.



# Feeding your baby with metabolic formula

Methionine is essential for normal development and therefore a limited and controlled amount must be taken daily.

Breast milk or standard infant formula will provide the methionine required by your baby prior to the introduction of solids, generally around 4-6 months of age.

Your baby will also need a special metabolic formula to provide protein without methionine.

Your dietitian will determine how much breast milk or standard infant formula and metabolic formula to offer.

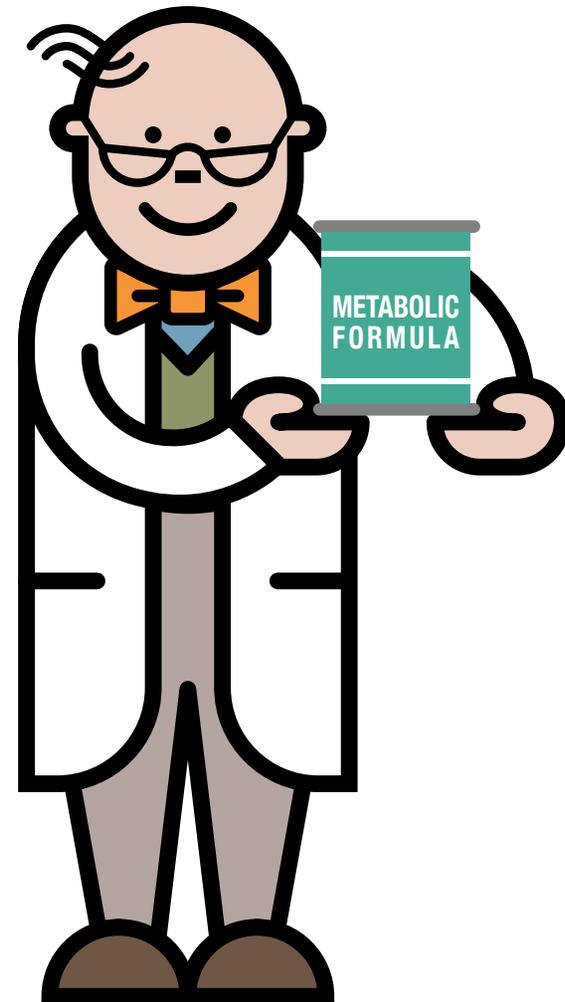


# Methionine-free metabolic formula

Methionine-free metabolic formula is an essential part of meeting your baby's nutritional requirements.

Like breast milk or standard infant formula, metabolic formula has carbohydrate, fat, vitamins, minerals and protein in the form of amino acids without methionine.

Metabolic formula, plus the prescribed amount of methionine, allows your baby to get all the nutrients he or she needs to grow.



# Tracking methionine

As your baby starts to eat solids your clinic will work with you to track methionine.

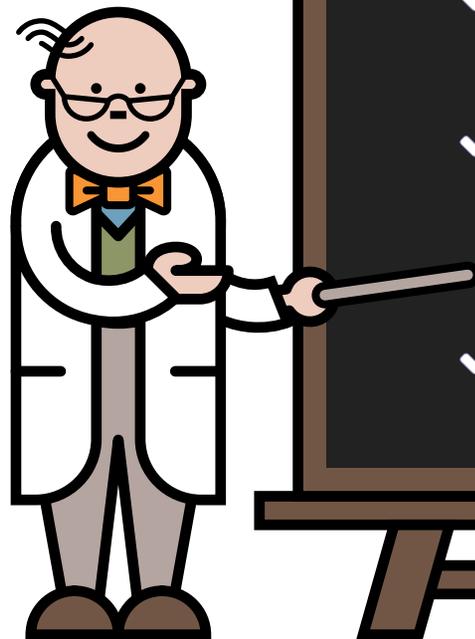
Foods must be weighed or measured using household measures (1 cup, 1 tablespoon, etc.) to determine methionine content.

Your clinic can help you find the best tools to help determine the methionine content of foods.

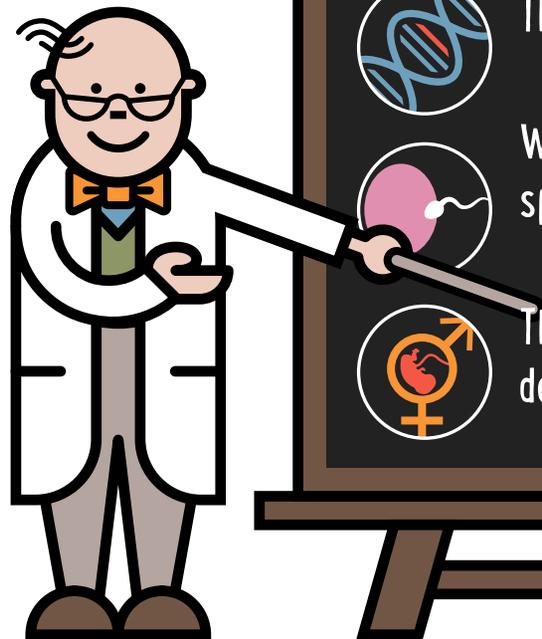


# What happens during a clinic check-up?

- ✓ Blood tests for methionine, homocysteine and cystine levels
- ✓ Height and weight are measured
- ✓ Diet is adjusted according to growth and blood tests
- ✓ Developmental check



# What happens in human genetics?



Humans have chromosomes composed of DNA.



Genes are pieces of DNA that carry the genetic instruction.  
Each chromosome may have several thousand genes.



The word mutation means a change or error in the genetic instruction.

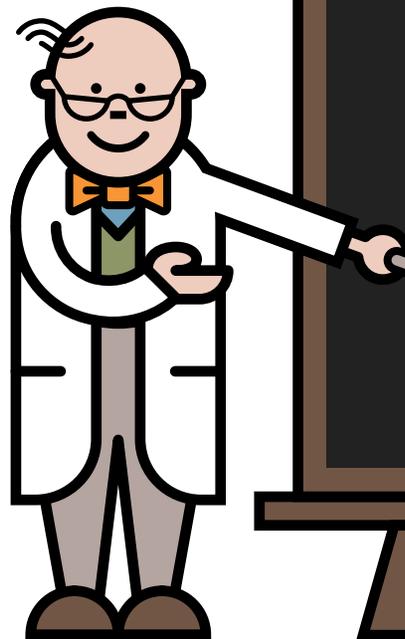


We inherit particular chromosomes from the egg of the mother and sperm of the father.



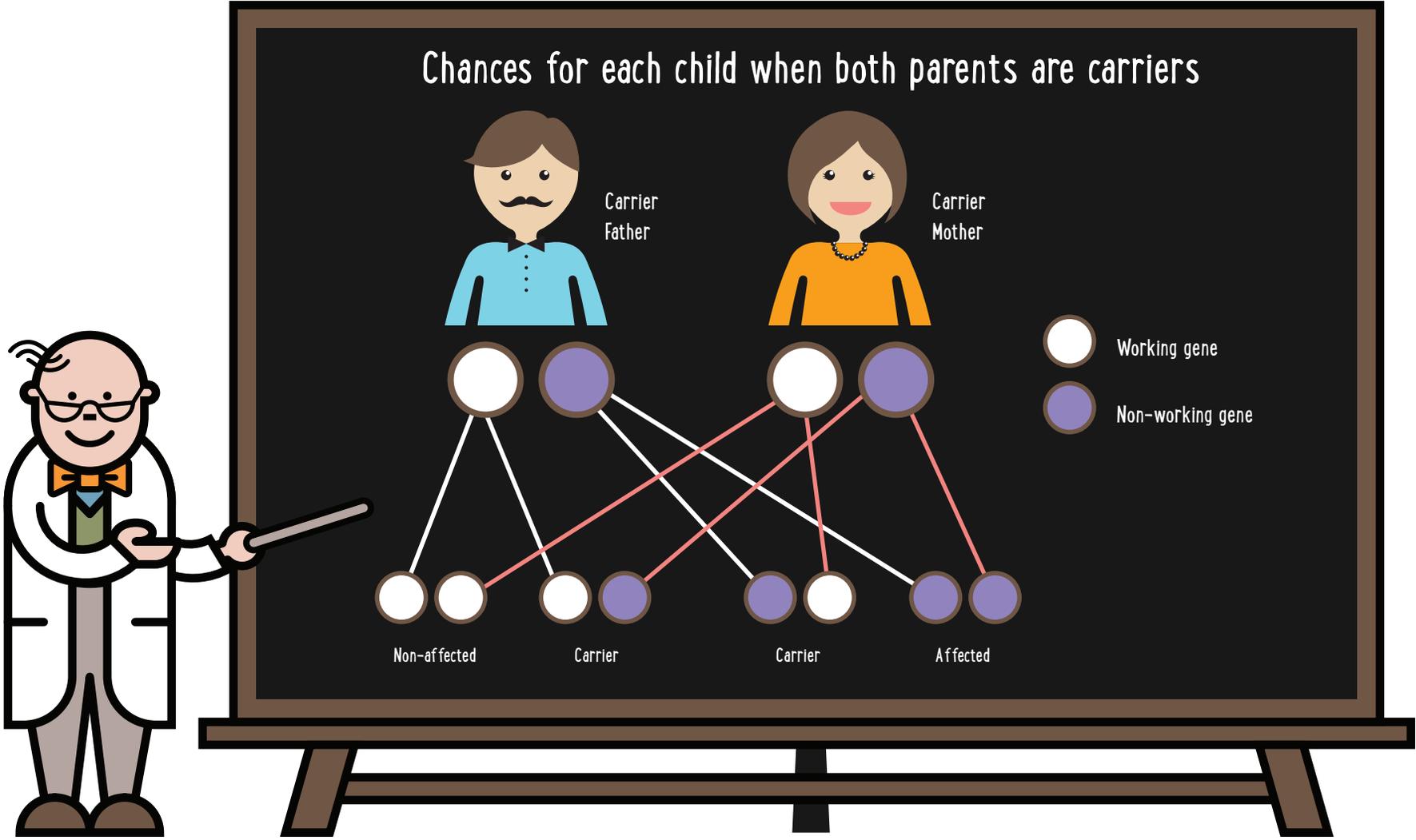
The genes on those chromosomes carry the instruction that determines characteristics, which are a combination of the parents.

# How does one inherit HCU?



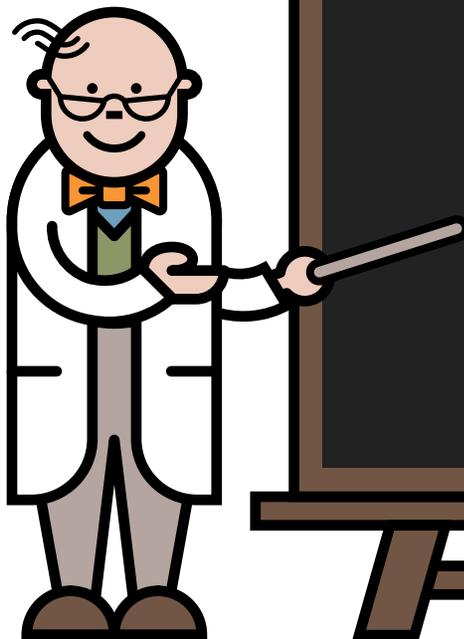
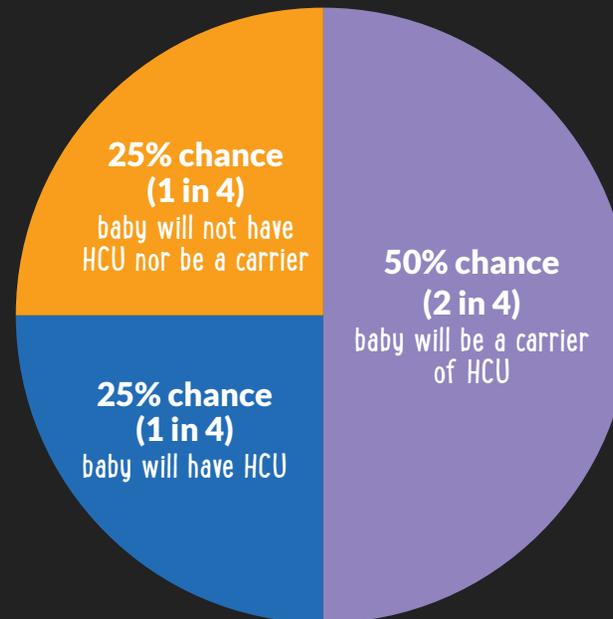
- ✓ HCU is an inherited condition. There is nothing that could have been done to prevent the child from having HCU.
- ✓ Everyone has a pair of genes that make the cystathionine beta-synthase enzyme. In children with HCU, neither of these genes works correctly. These children inherit one non-working HCU gene from each parent.
- ✓ Parents of children with HCU are carriers of the condition.
- ✓ Carriers do not have HCU because the other gene of this pair is working correctly.

# Inheritance – Autosomal recessive – possible combinations

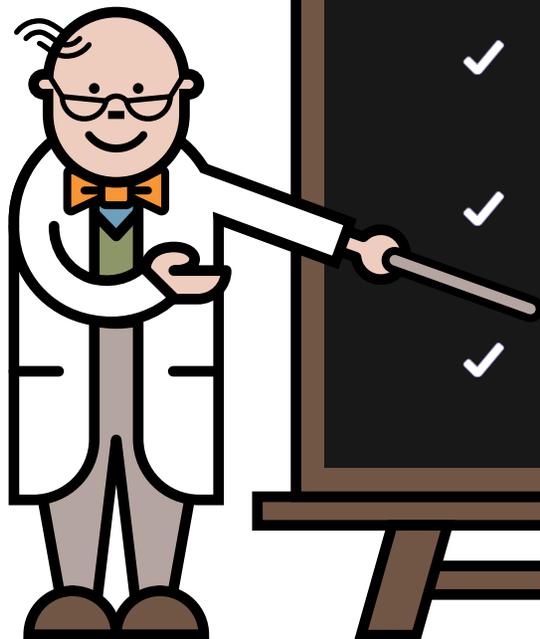


# Future pregnancies

When both parents are carriers, *in each pregnancy*  
the risk to the baby is as follows:

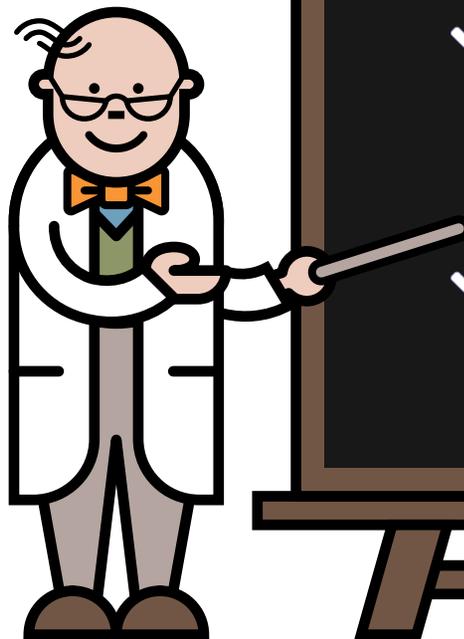


# Take home messages



- ✓ HCU is a serious inherited, metabolic disorder that can lead to a number of problems if unmanaged.
- ✓ Damage can be prevented with either:
  - Vitamin B<sub>6</sub> (roughly 50% of children)
  - Protein restricted diet, with a special metabolic formula
- ✓ Follow your metabolic team's recommendation to offer your child the best opportunity for normal growth and development.
- ✓ Management should also prevent long term complications such as osteoporosis (thin bones), blood clots and strokes.
- ✓ Management started in late childhood cannot reverse problems that already exist but can avoid life-threatening blood clots and strokes.

# Helpful hints



- ✓ Always ensure you have a good supply of your special dietary products and medicines and that they are not expired.
- ✓ Your special dietary products and methionine-free metabolic formula are prescribed by your metabolic clinic.
- ✓ It is important to keep hydrated, especially when ill, and to avoid a sedentary lifestyle.

# Who's who (contact details)

## My dietitian

Name: .....

Phone #: .....

Email: .....

## My nurse

Name: .....

Phone #: .....

Email: .....

## My doctor

Name: .....

Phone #: .....

Email: .....

# Notes

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