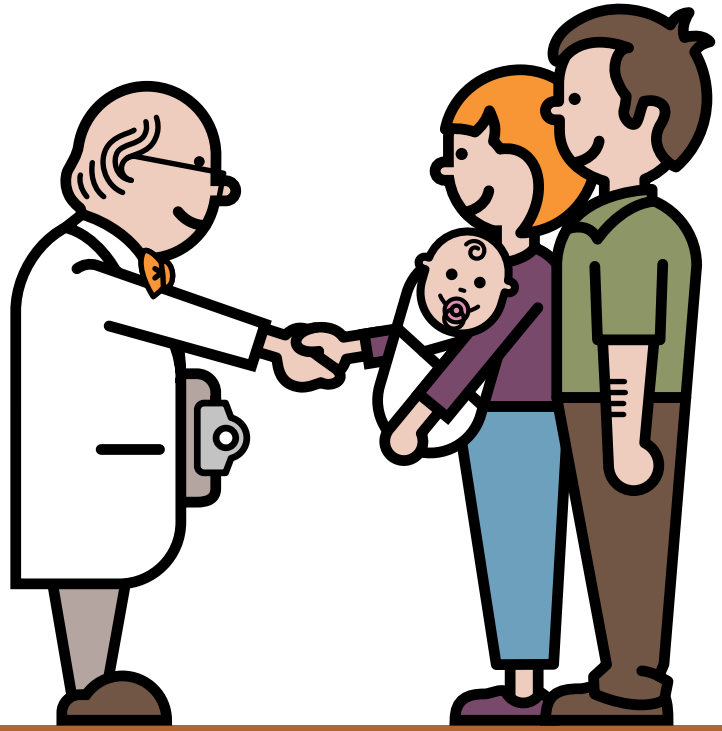
Homocystinuria

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 This version no longer represents clinical or dietetic practice in the UK or Ireland.

TEMPLE
Tools Enabling Metabolic Parents LEarning

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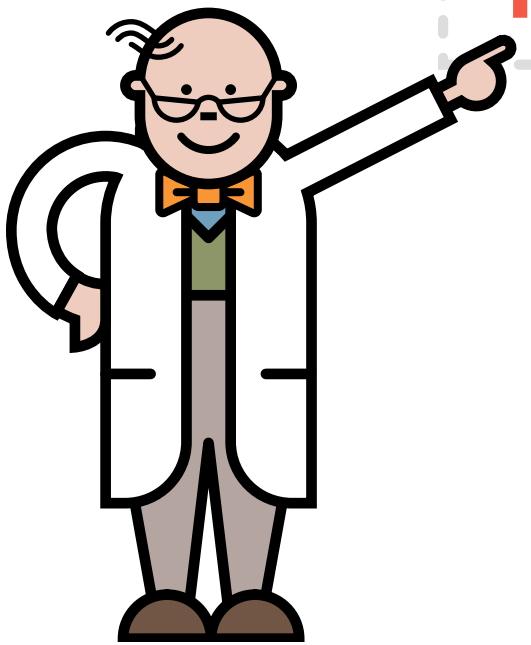
What is HCU?

HCU stands for homocystinuria.

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

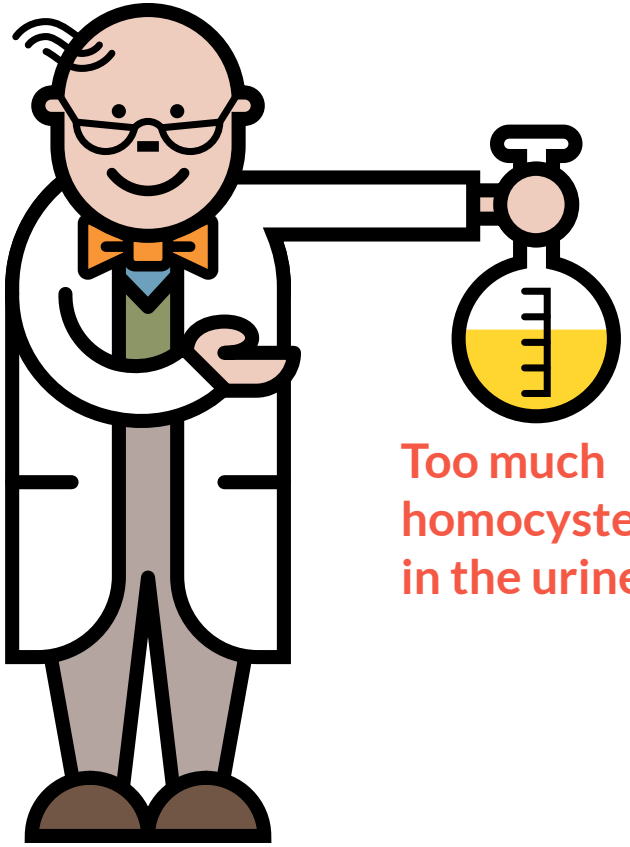
It is an inherited metabolic condition.



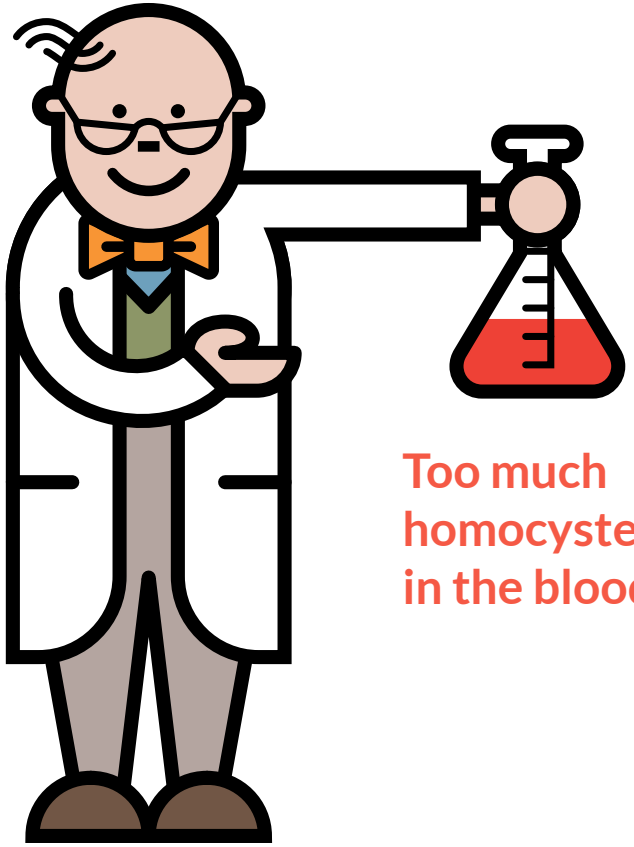
Homo cystin uria

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.

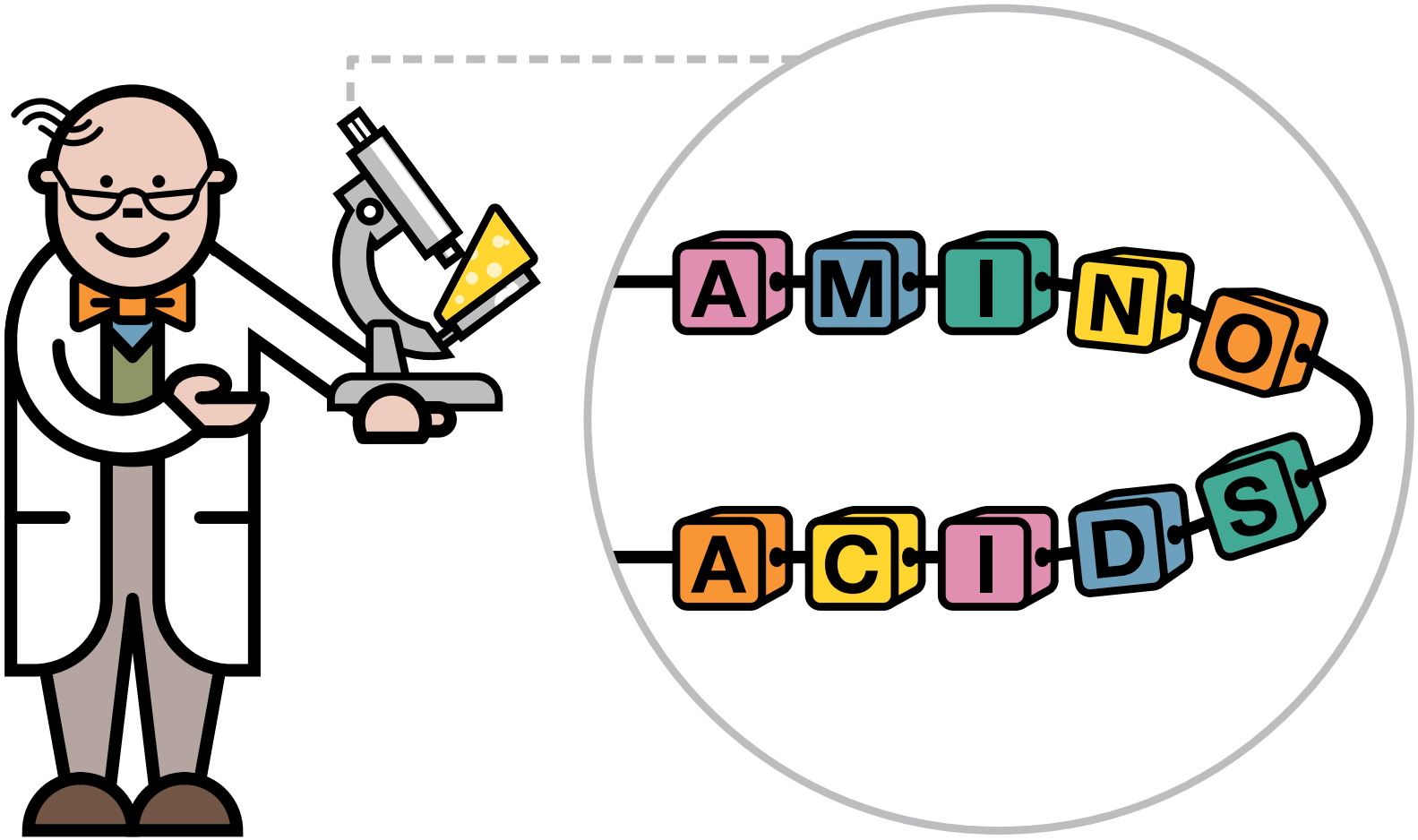
Protein is found in our bodies and in many foods. The body needs protein for growth and repair.





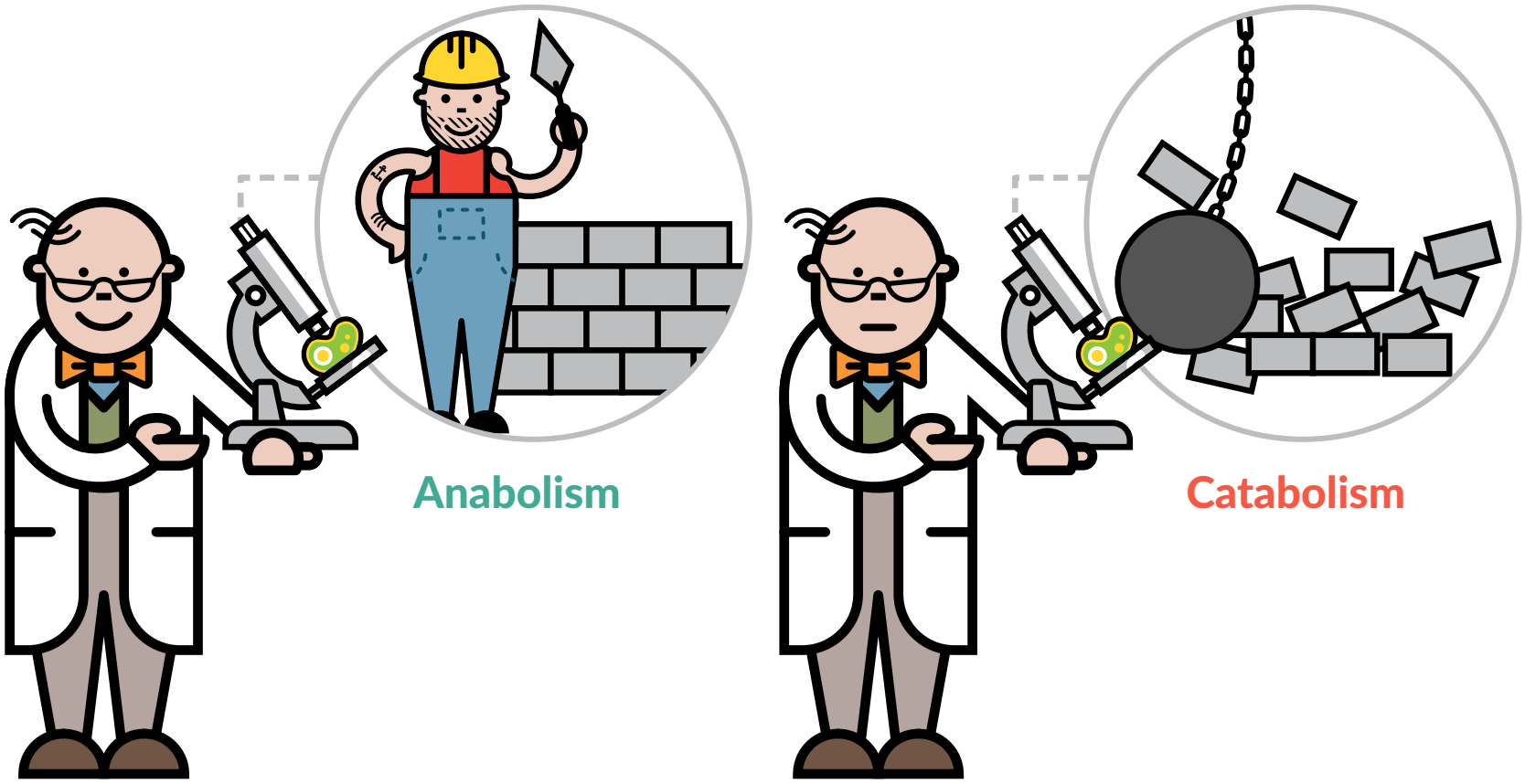

What is protein?

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



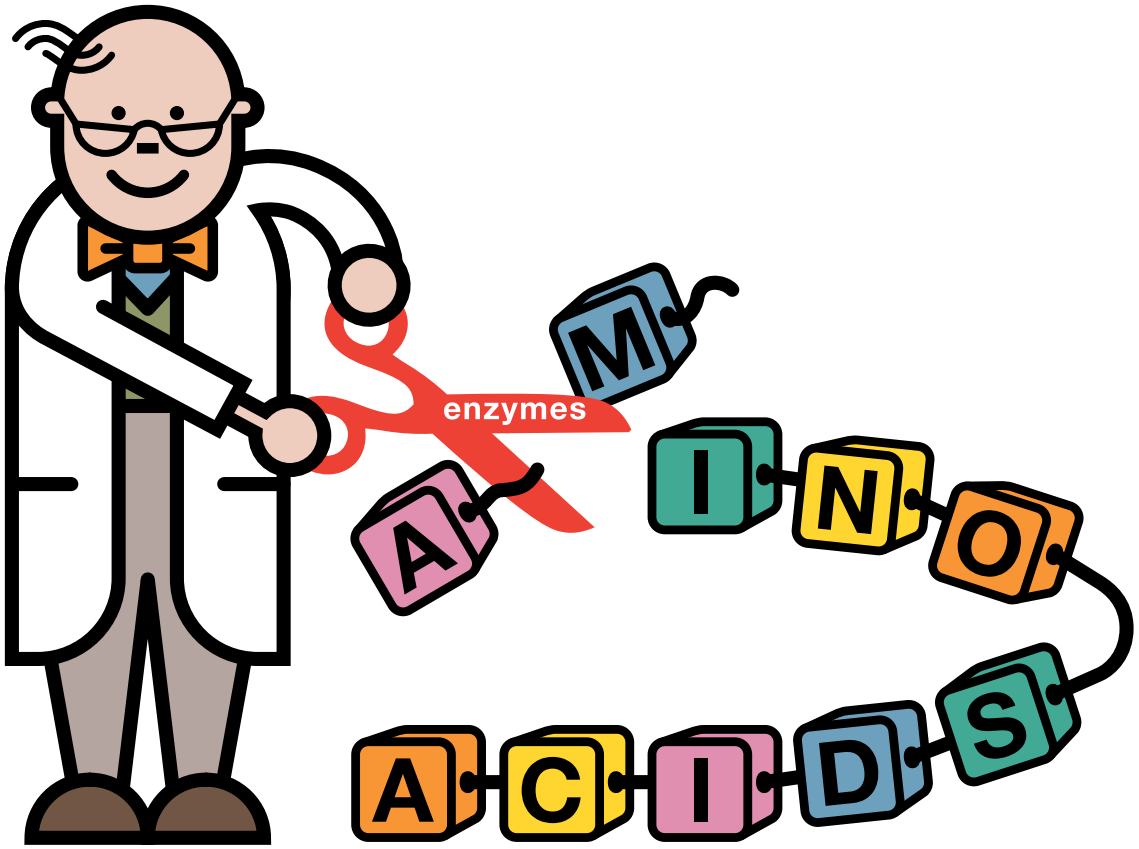
Protein metabolism

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



What do enzymes do?

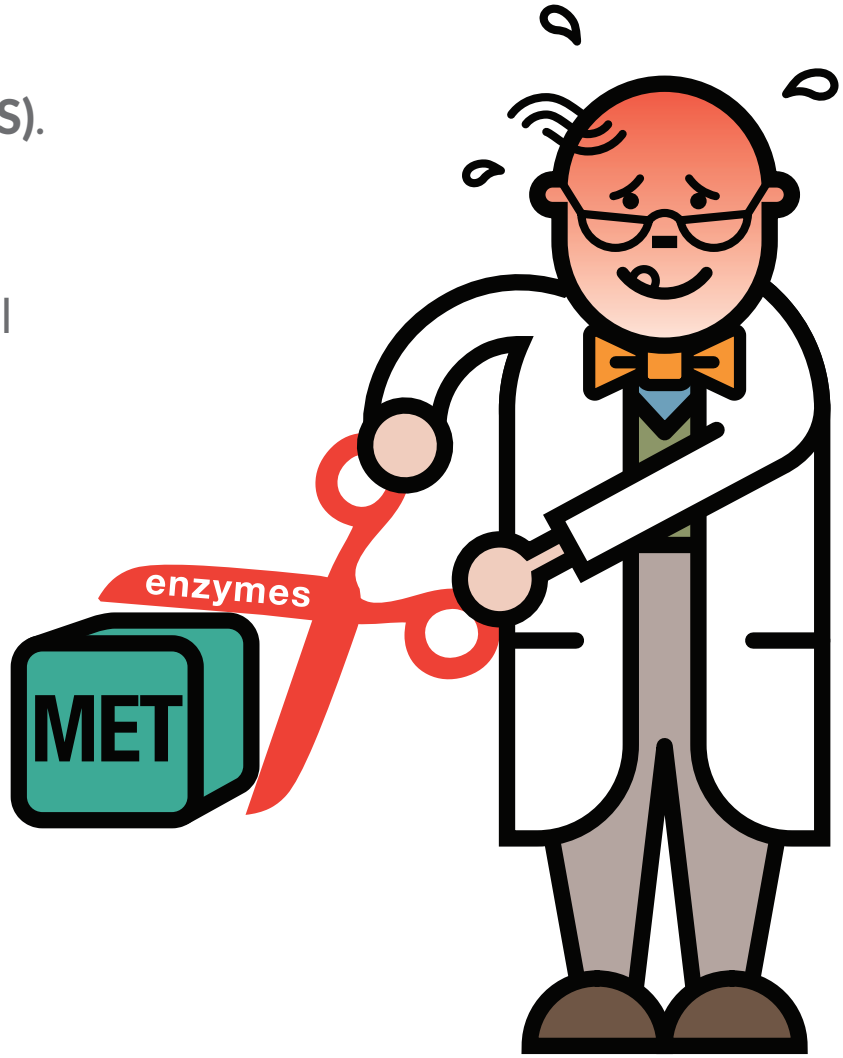
Enzymes help with metabolism by functioning like scissors.
They break down proteins into smaller parts, including amino acids.



What happens in HCU?

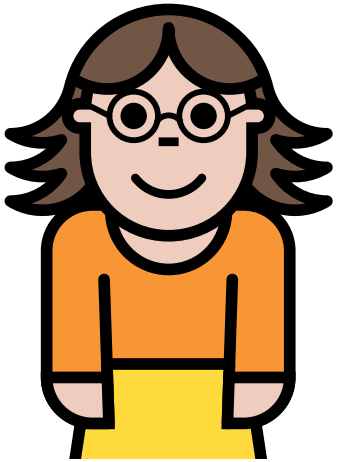
HCU is due to a deficiency of an enzyme called **cystathionine beta-synthase (CBS)**.

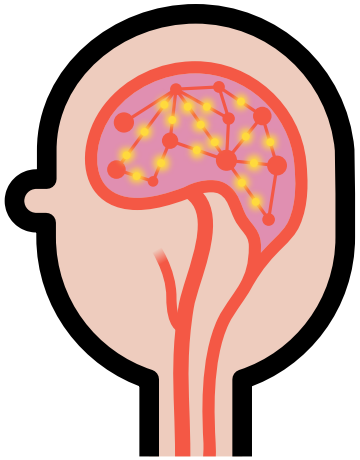
This results in the body being unable to break down an amino acid called methionine (MET). This causes a harmful buildup of methionine and another substance called homocysteine (HCY) in the blood.



What can go wrong in HCU?

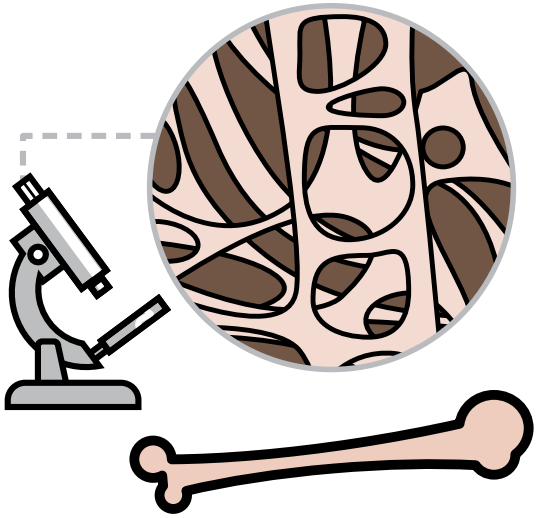
Nearsightedness and dislocated eye lenses

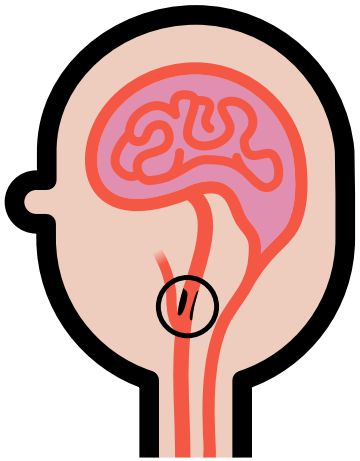




Difficulties with learning and behavior

Long and thin bones





Blood clots and strokes

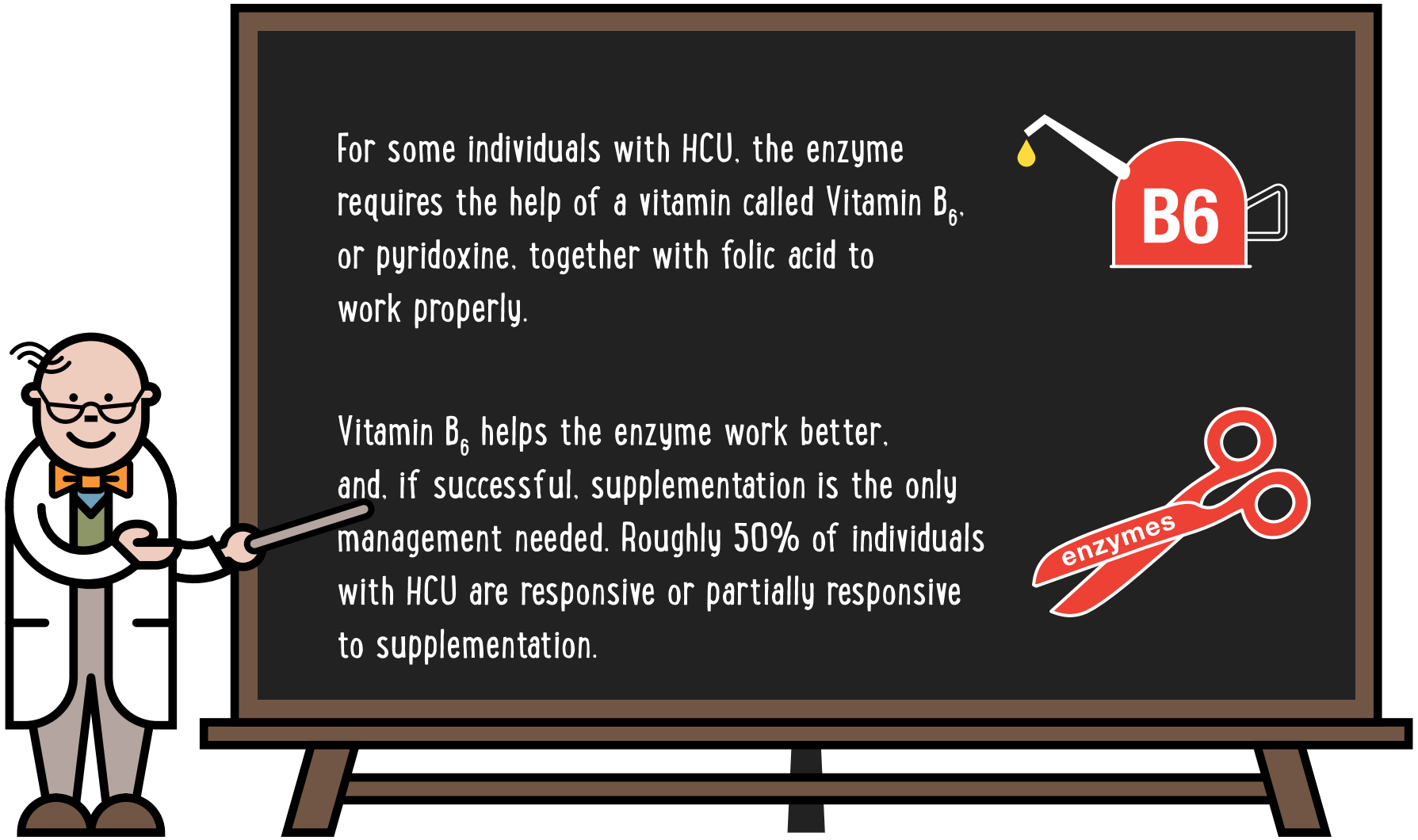
How is HCU diagnosed?

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?



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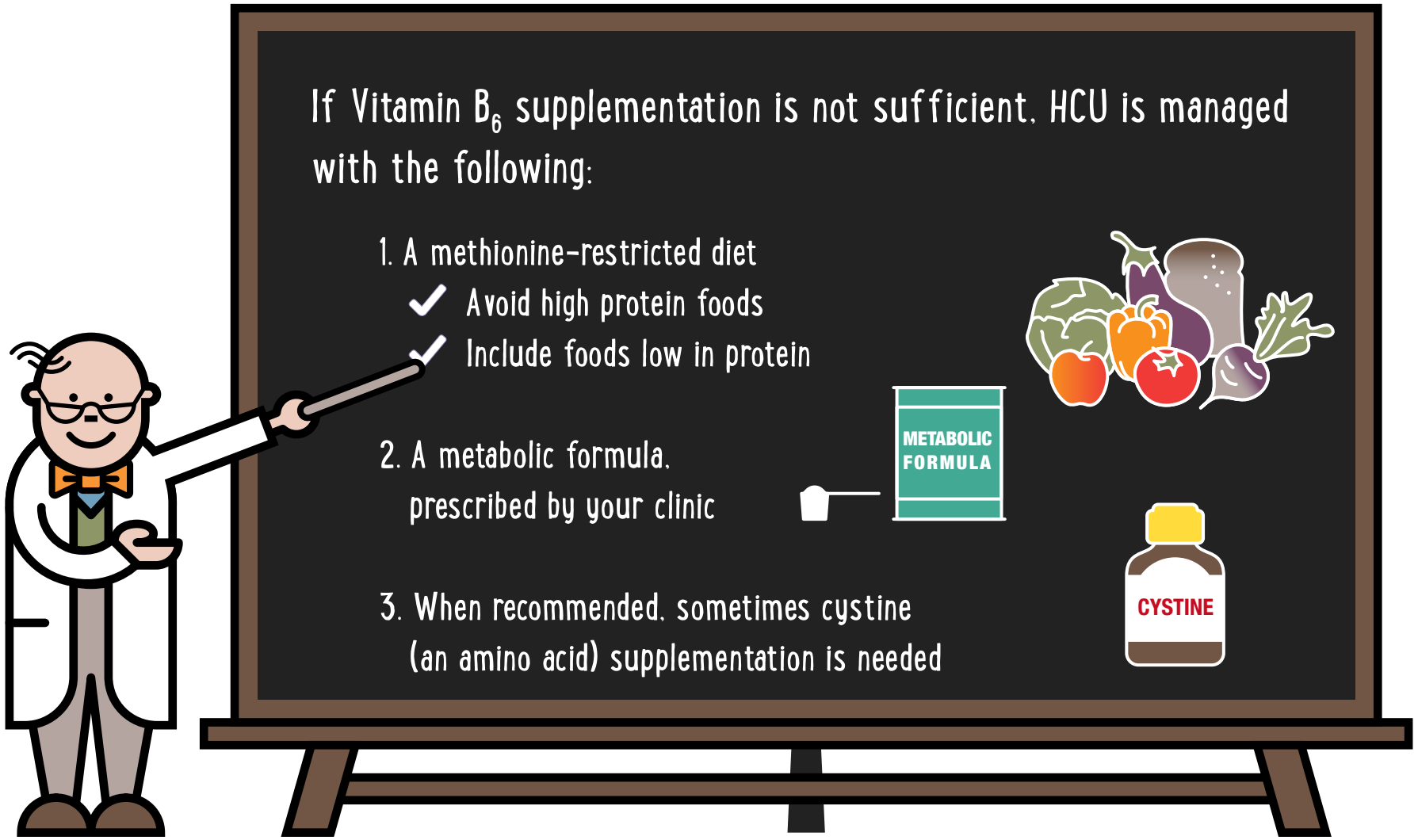

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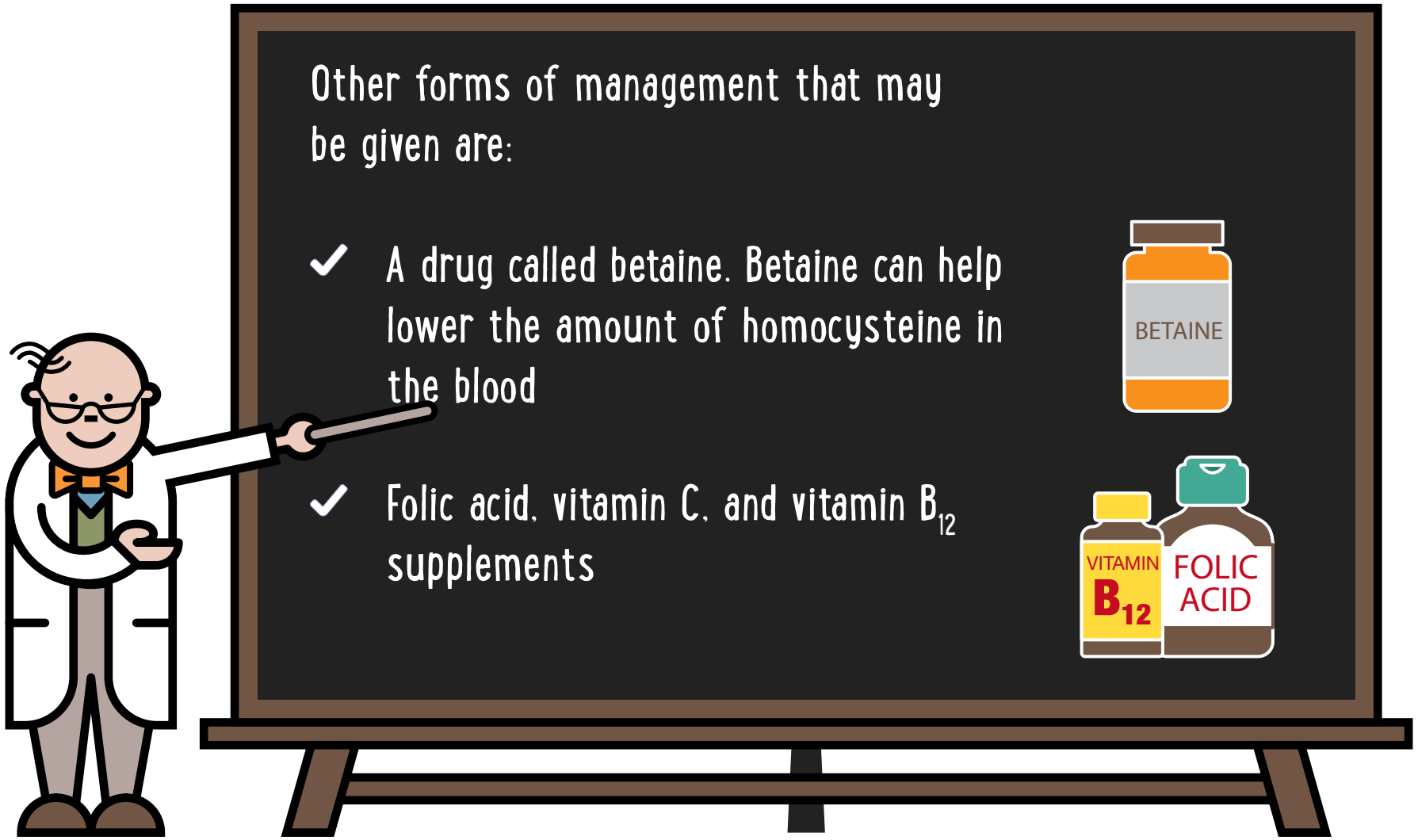


Avoid high protein foods

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**.

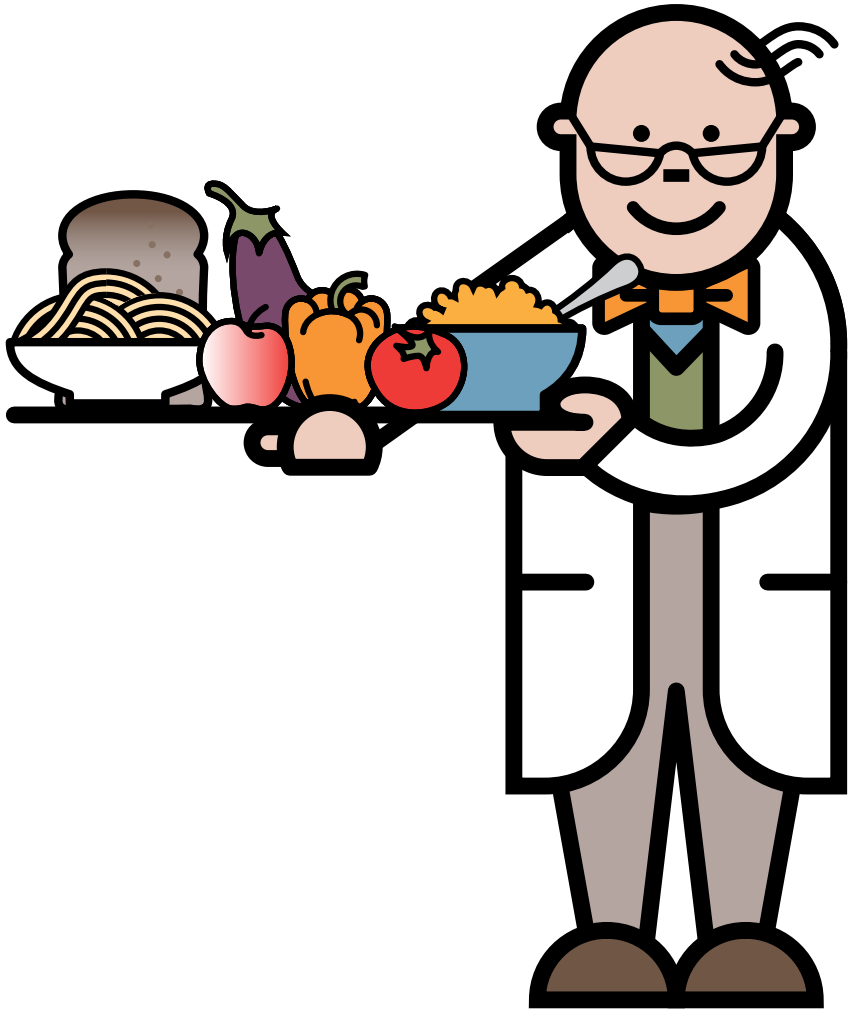
Include foods low in protein

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.

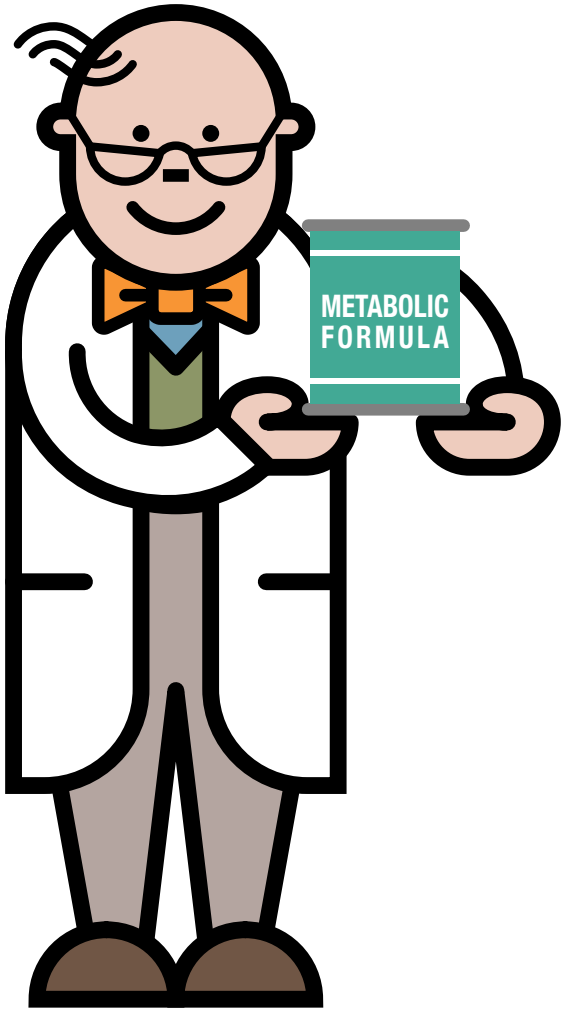
Supported by NUTRICIA as a service to metabolic medicine
Dietary management of the condition should only be done under medical supervision.

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.



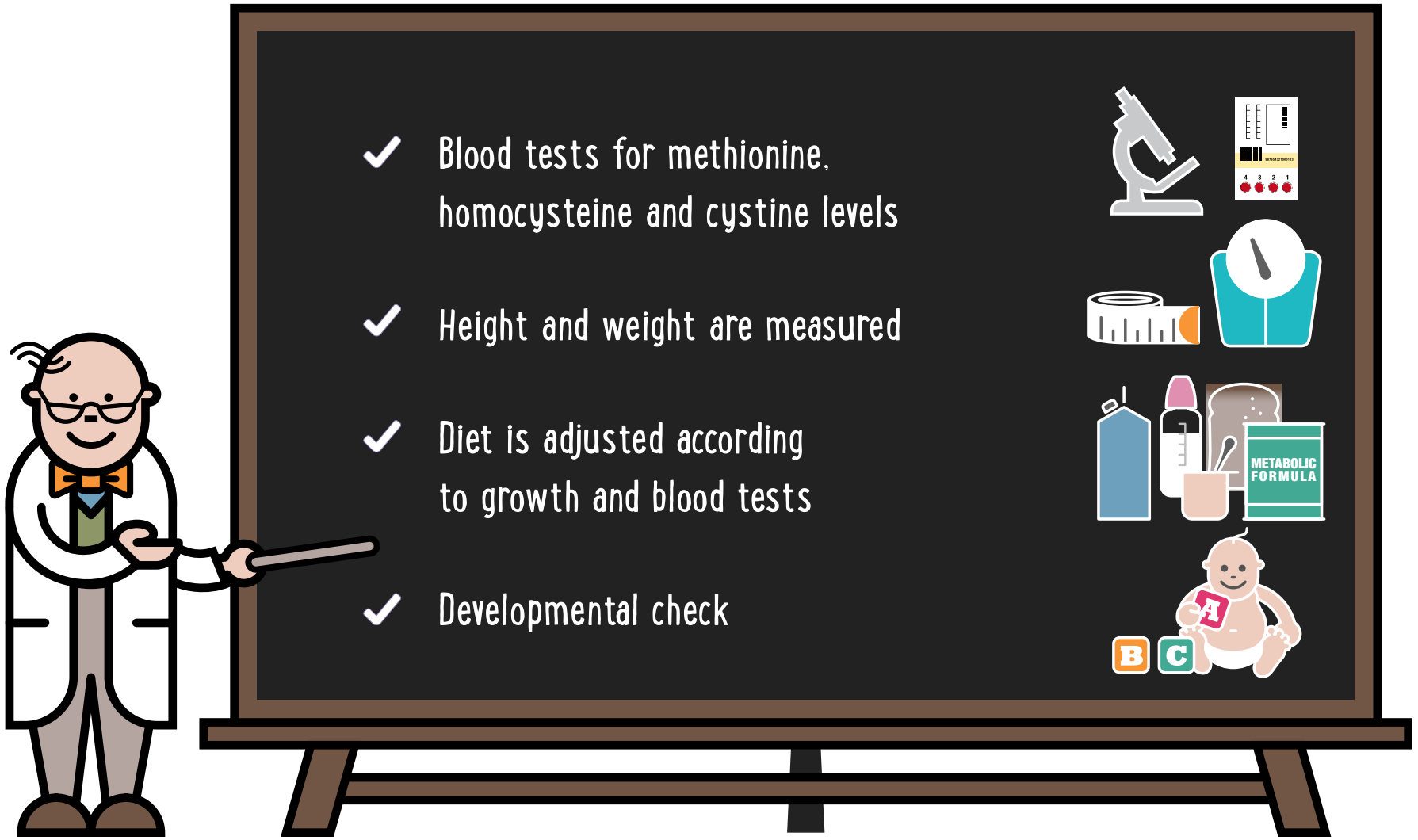
Tracking methionine

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

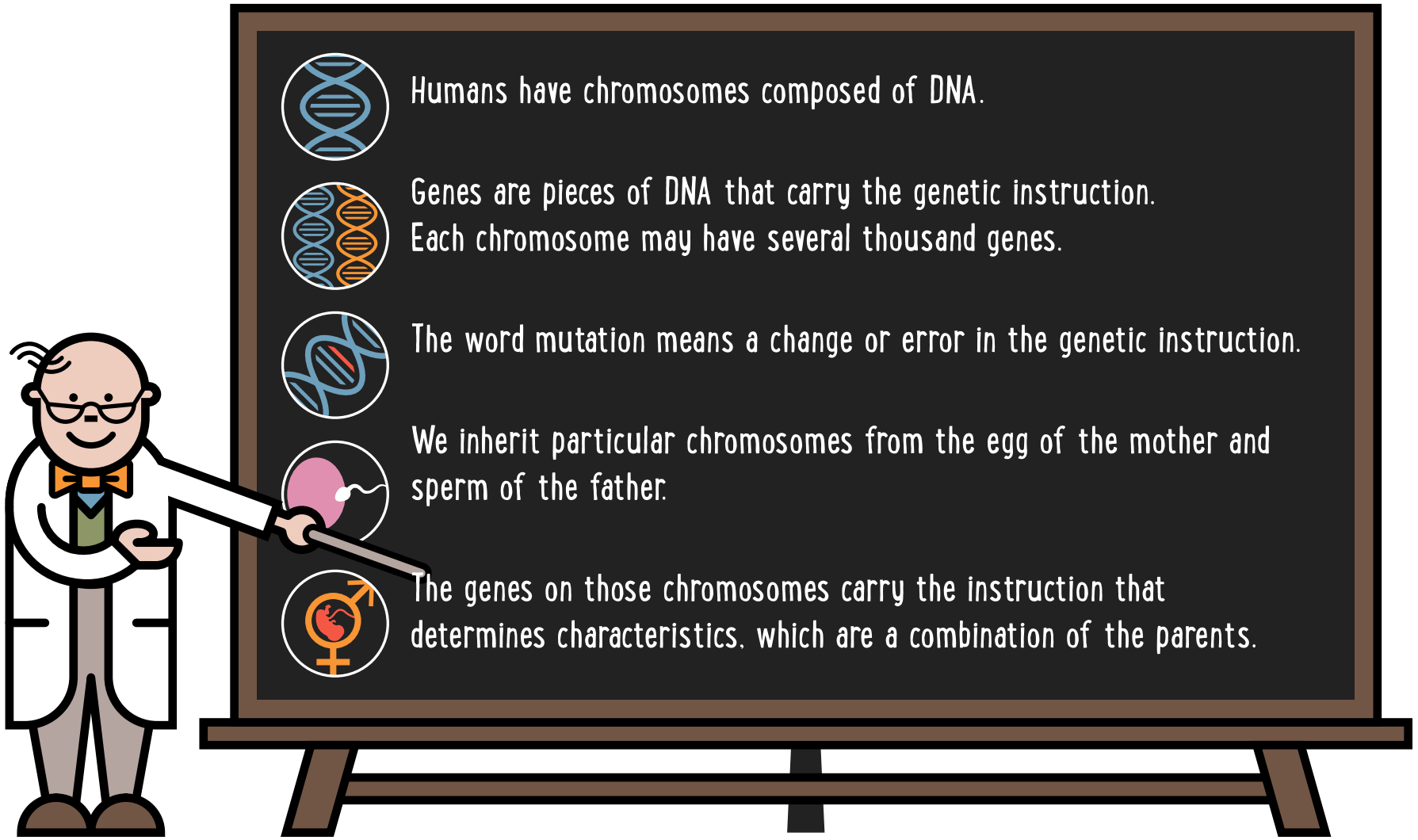
Foods must be weighed or measured using household measures (1 cup, 1 tablespoon, etc.) to determine methionine content.

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

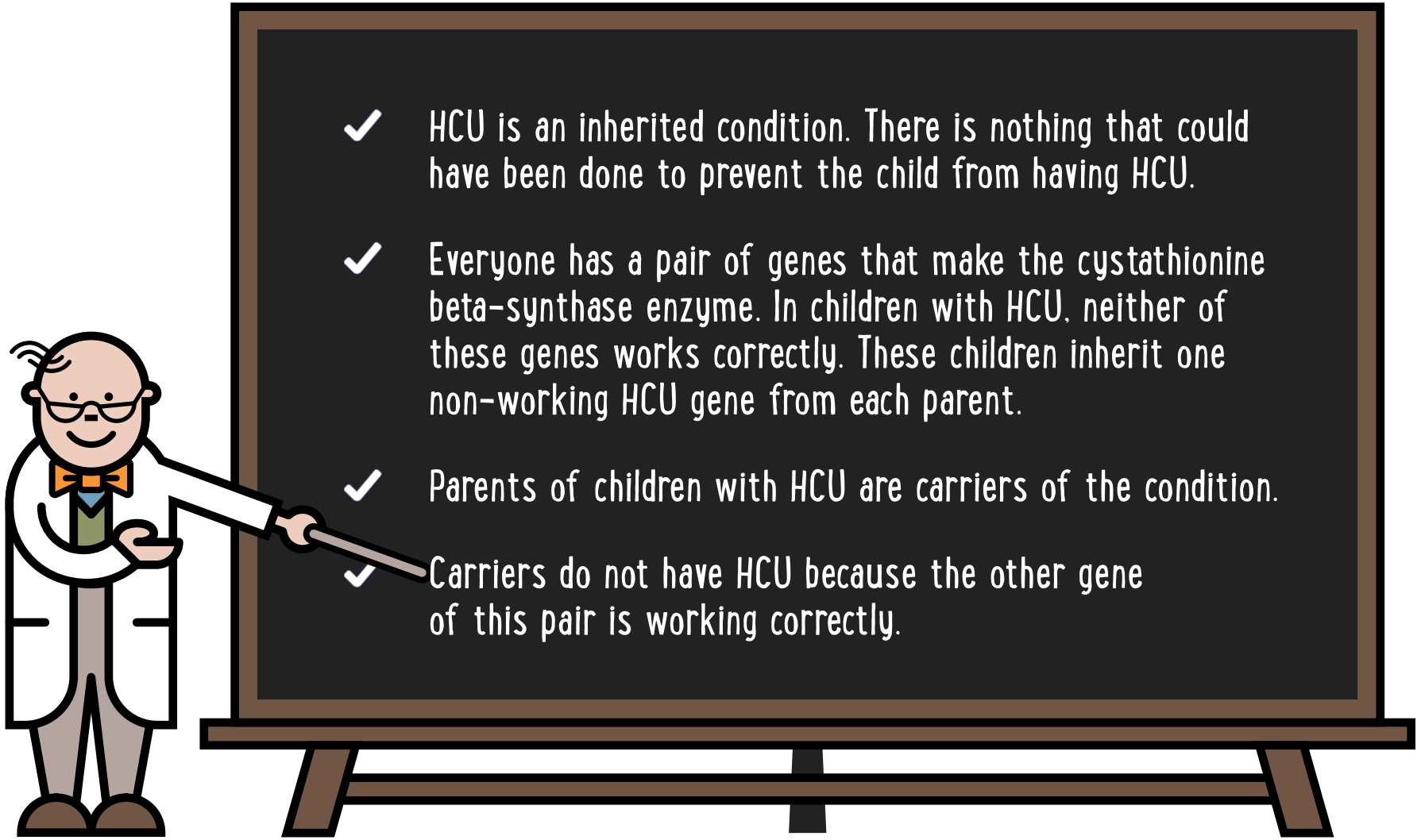
What happens during a clinic check-up?



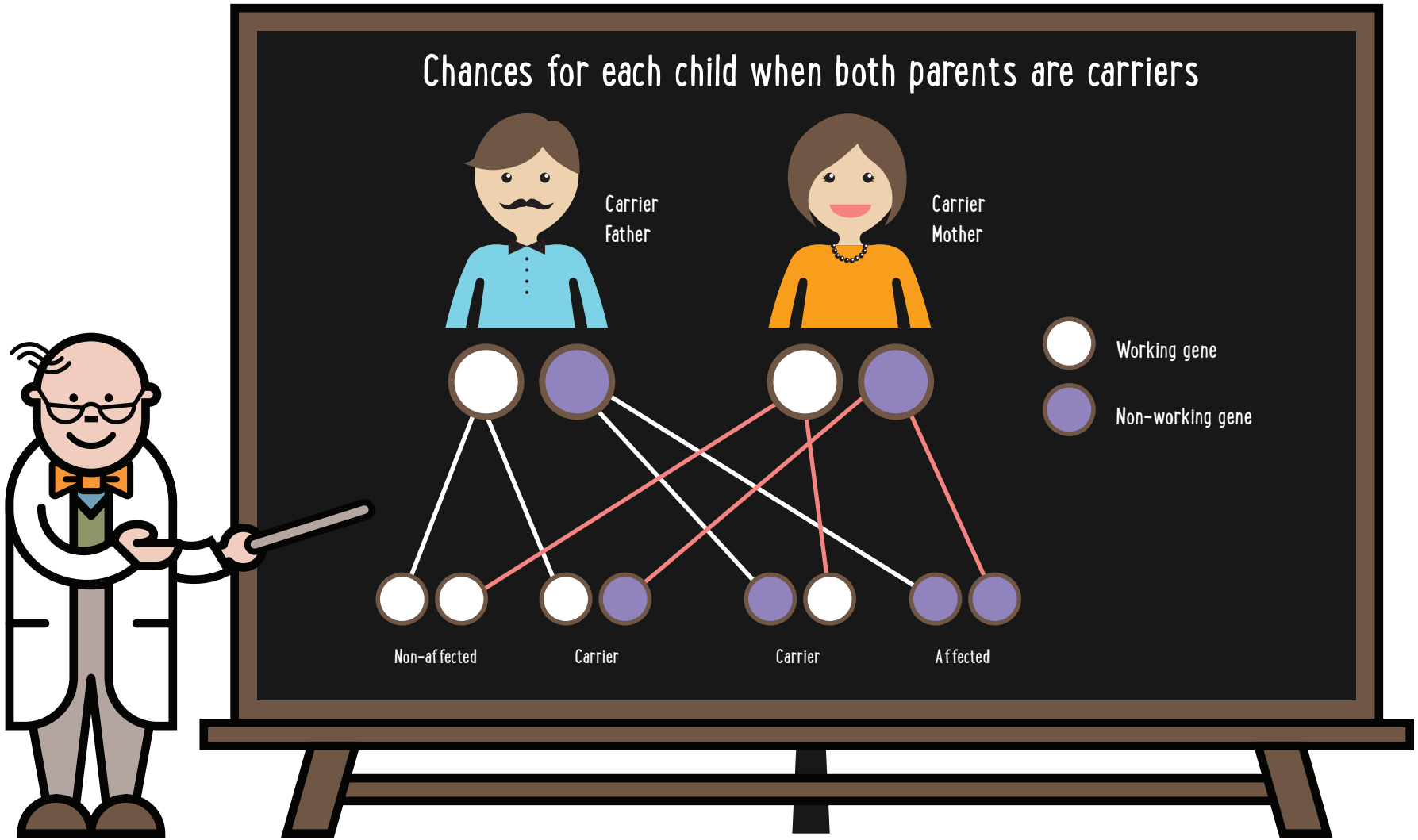
What happens in human genetics?



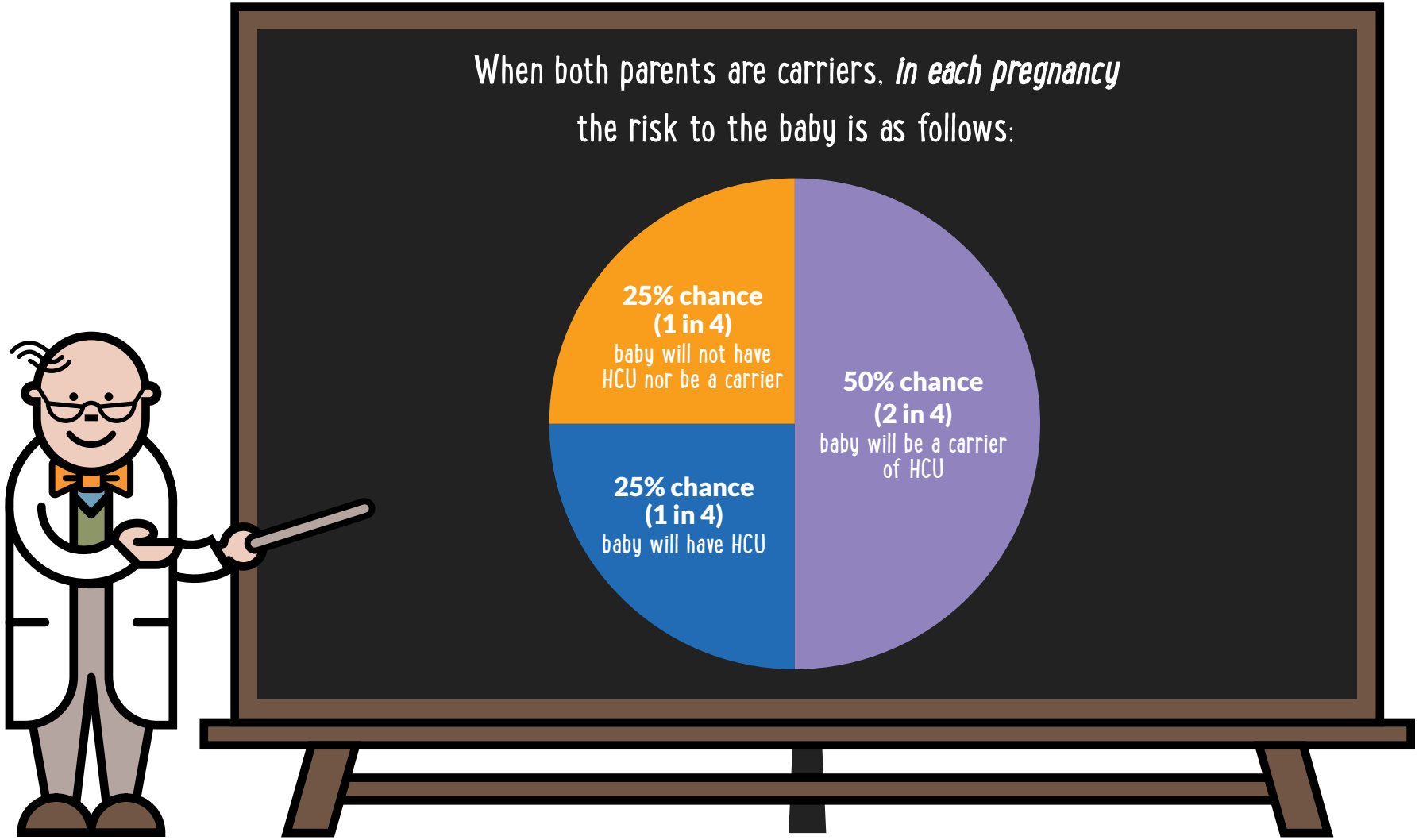
How does one inherit HCU?



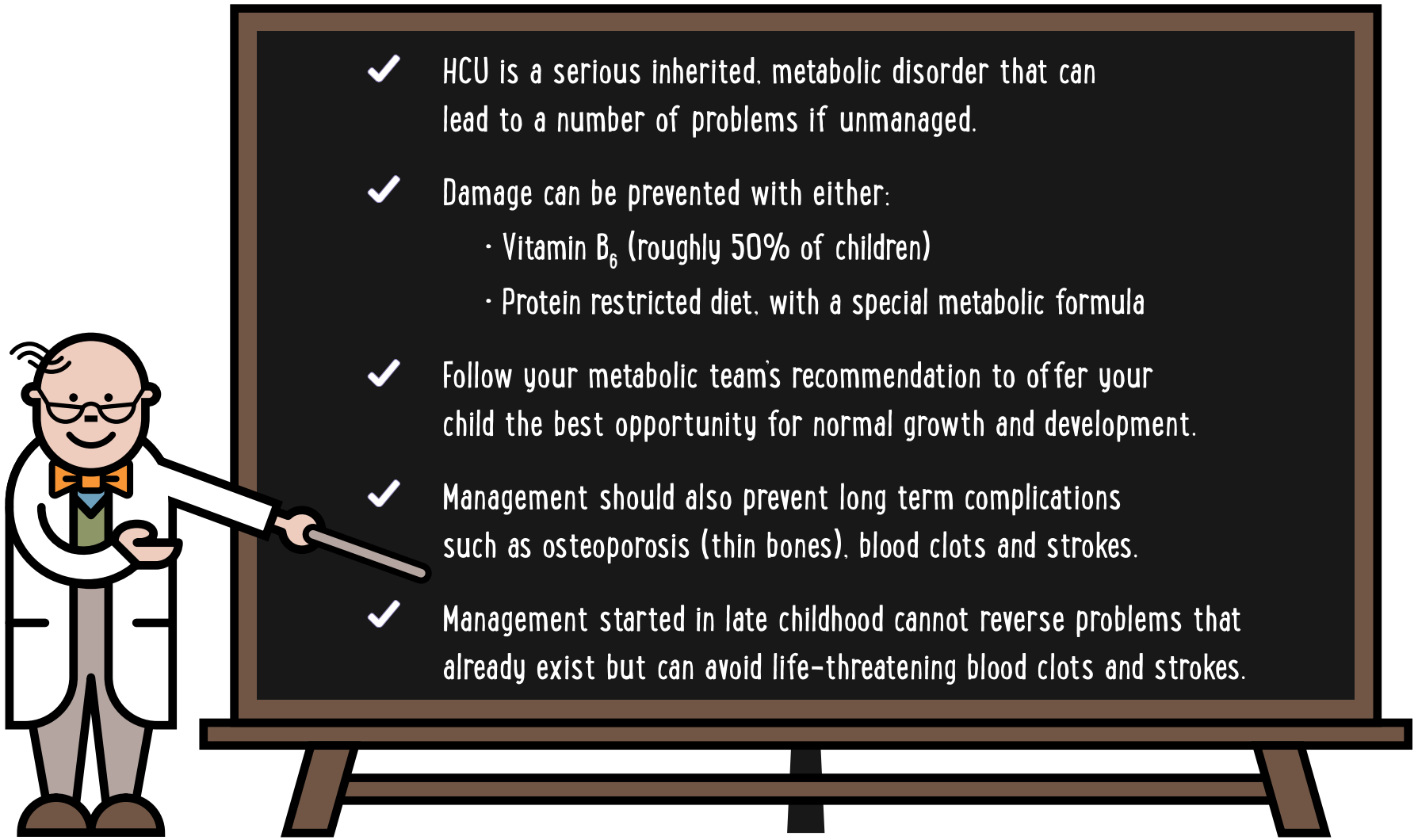
Inheritance – Autosomal recessive – possible combinations



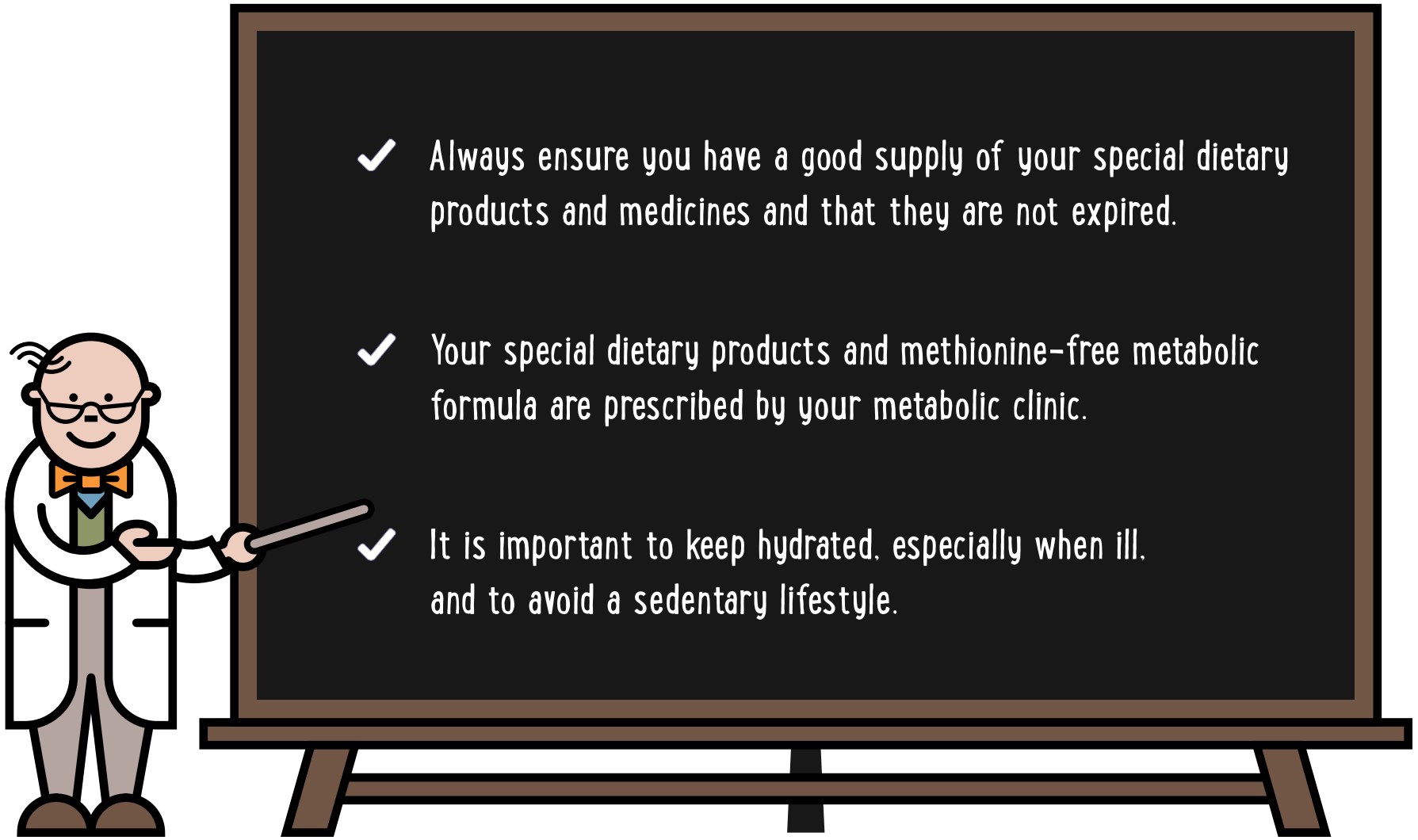
Future pregnancies



Take home messages



Helpful hints



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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