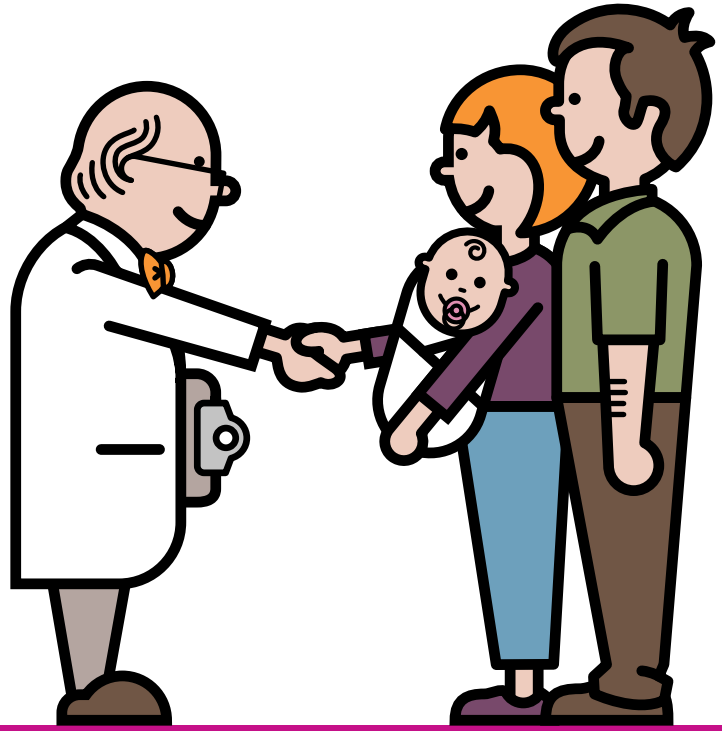
Tyrosinemia Type 1

HT-1

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

TEMPLE
Tools Enabling Metabolic Parents LEarning

Supported by NUTRICIA as a service to metabolic medicine

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 This version no longer represents clinical or dietetic practice in the UK or Ireland.

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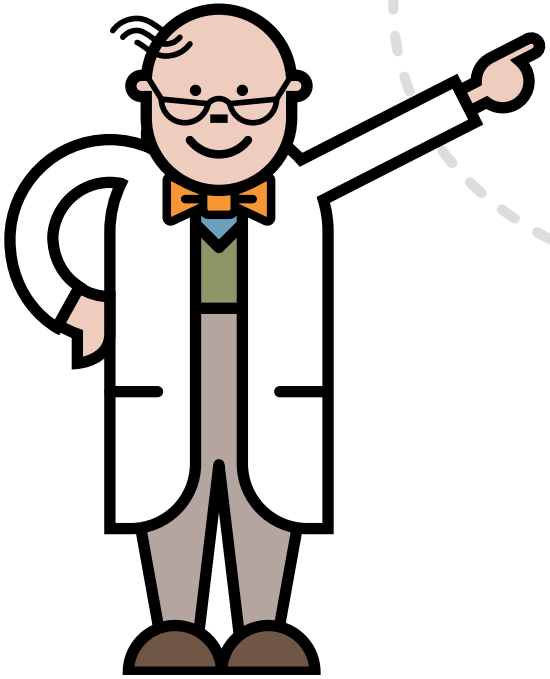
What is HT-1?

HT-1 stands for hereditary tyrosinemia type 1.

It is pronounced ty-ro-sin-emia.

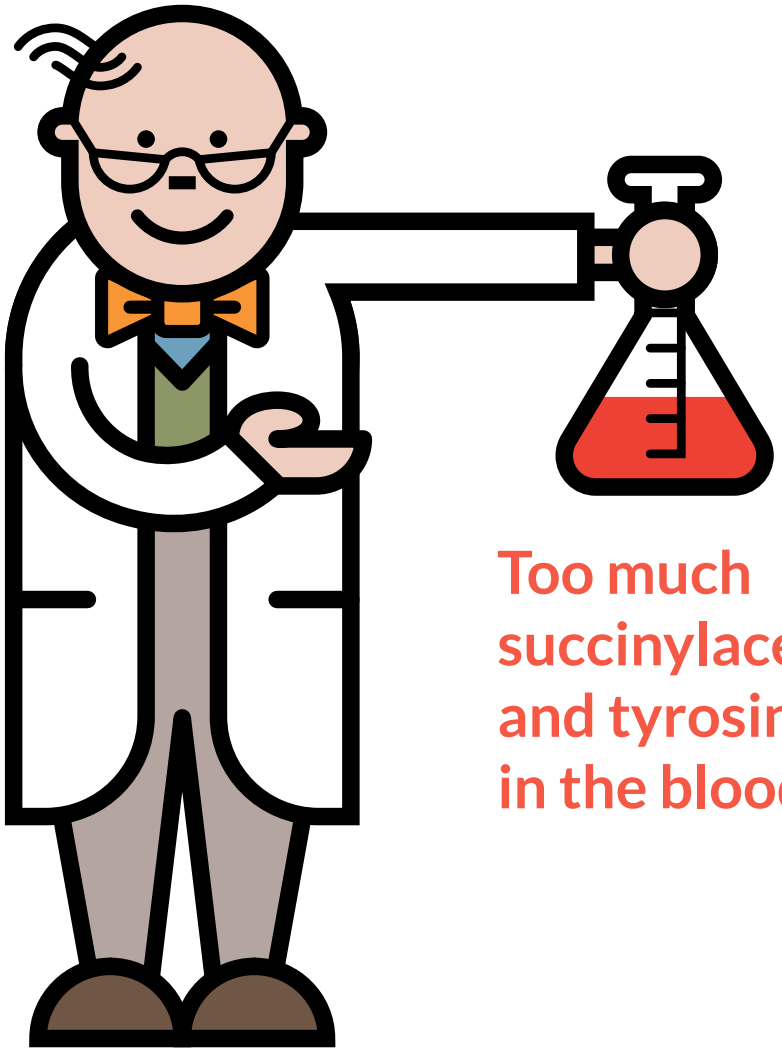
It is an inherited metabolic condition.

Hereditary Tyrosinemia Type 1



HT-1

What is HT-1?



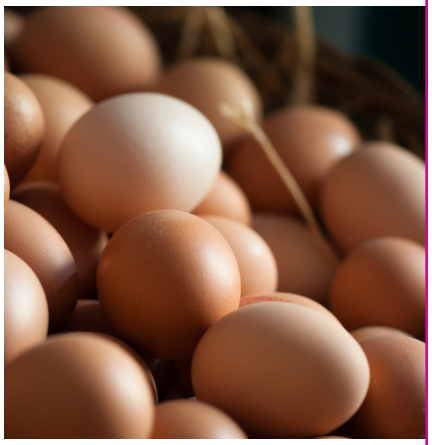
Too much succinylacetone and tyrosine in the blood

How does HT-1 affect the body?

HT-1 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.

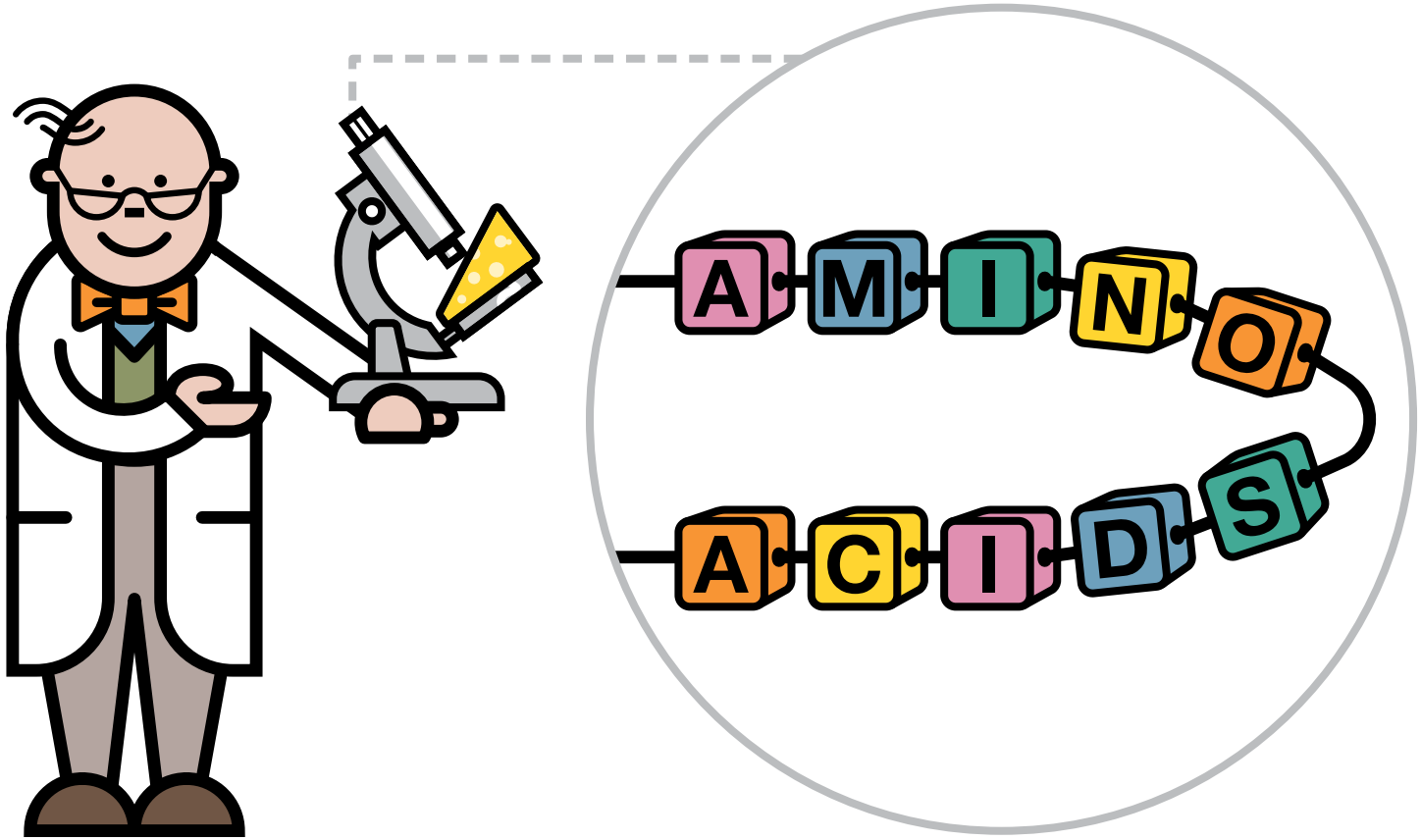


What is protein?

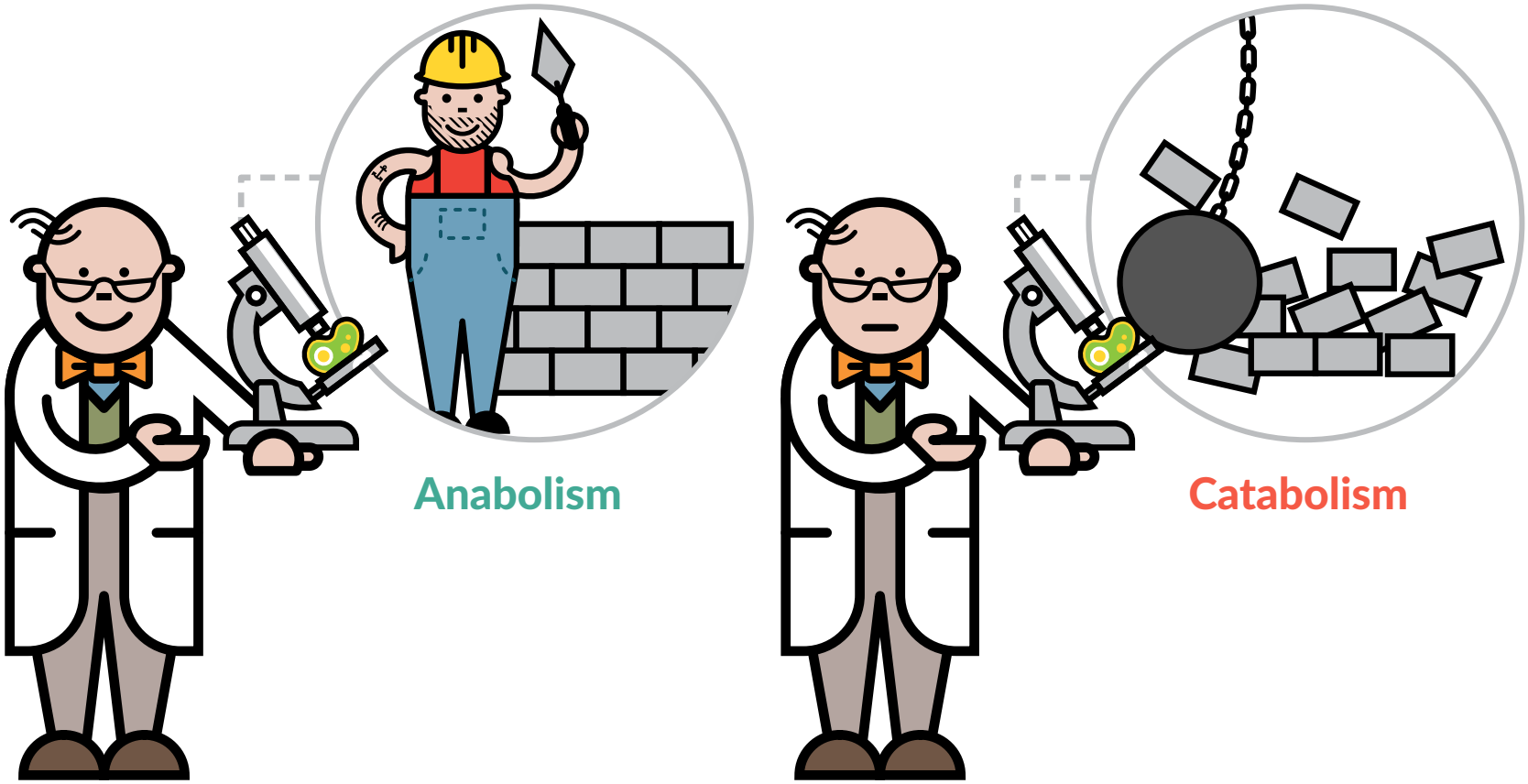
Protein consists of chains of many smaller units called amino acids.

Tyrosine (TYR) and phenylalanine (PHE) are two amino acids.



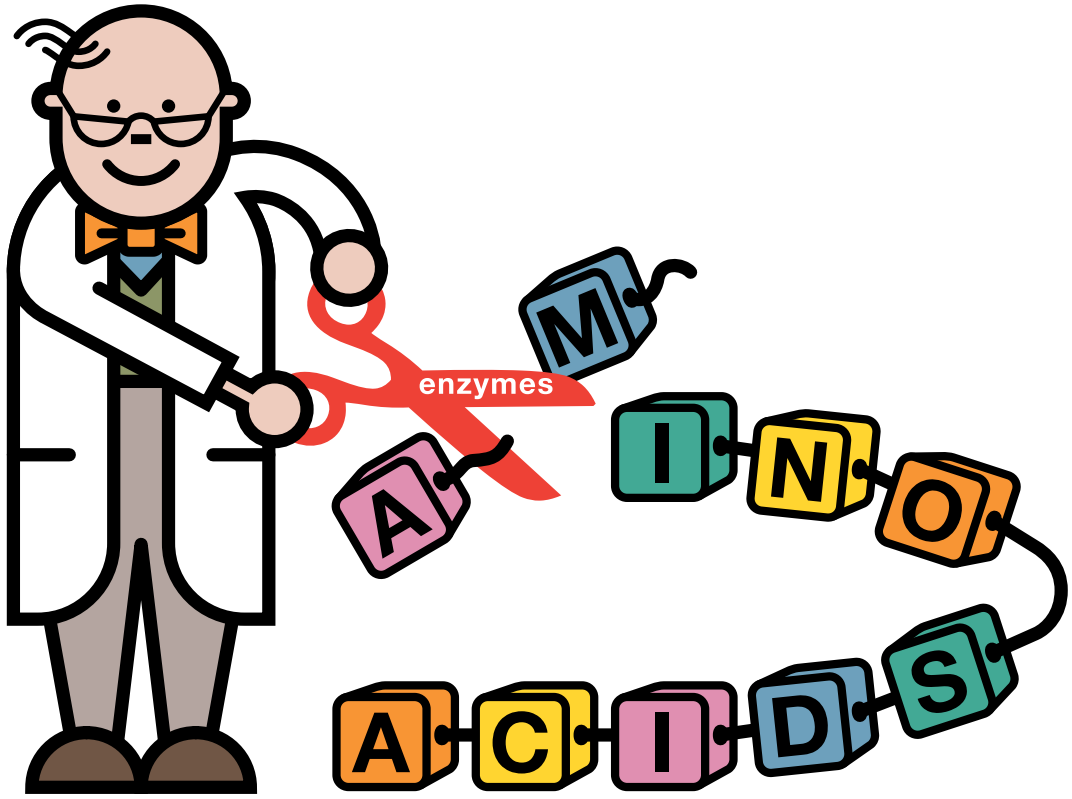
Protein metabolism

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



What do enzymes do?

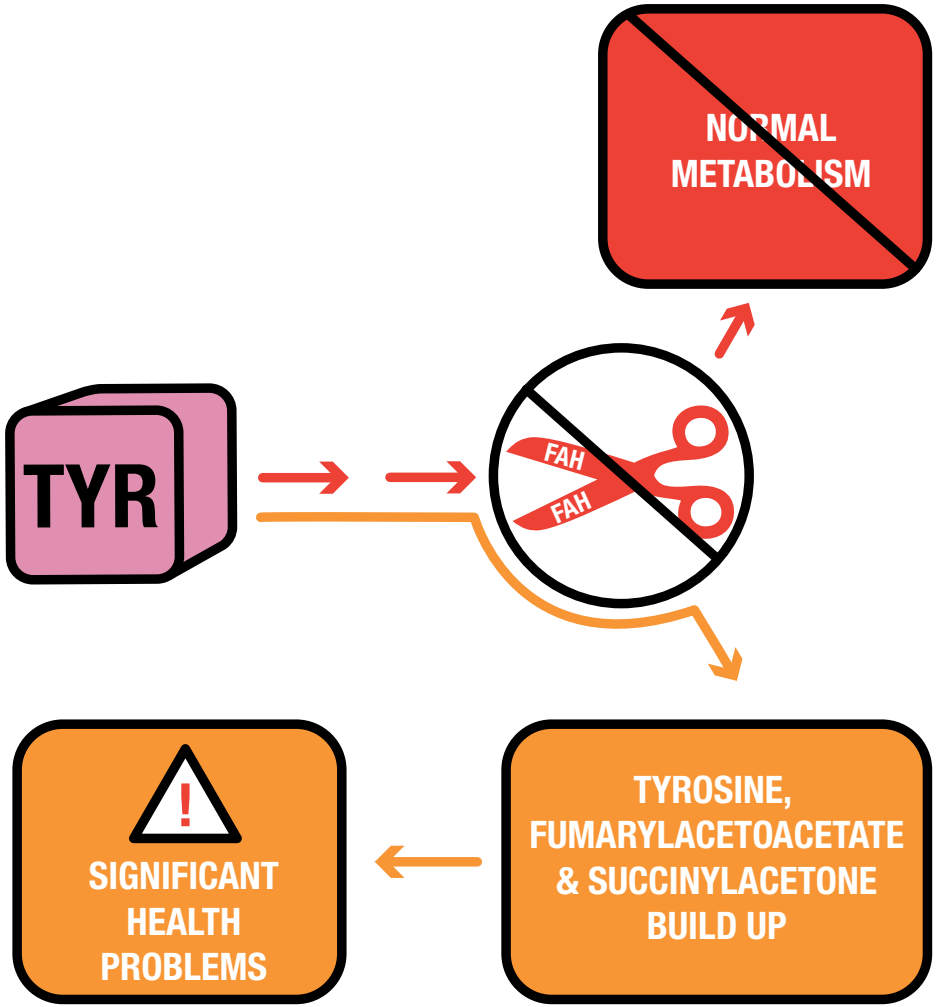
Enzymes help with metabolism by functioning like scissors.
They break down proteins into smaller parts, including amino acids.



What happens in HT-1?

HT-1 is caused by a deficiency of an enzyme called **fumarylacetoacetate hydrolase**, or **FAH**.

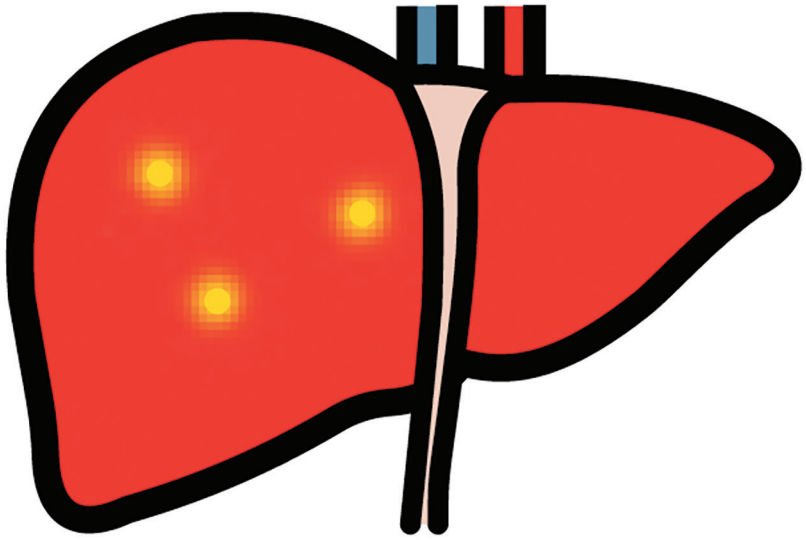
The amino acid tyrosine cannot be broken down as usual and instead toxic substances are created. These toxic substances are called fumarylacetoacetate and succinylacetone.

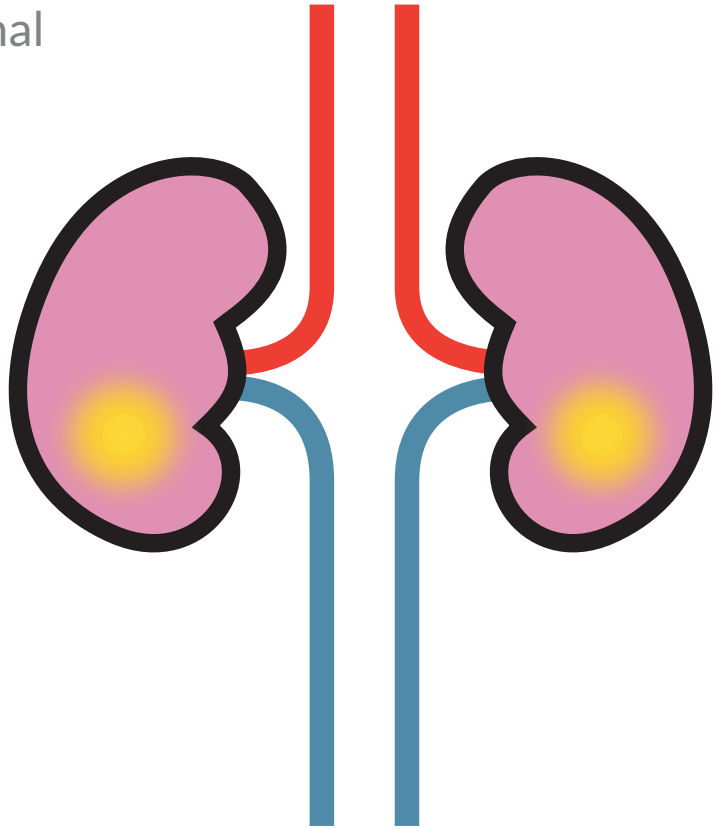


What can go wrong in untreated HT-1?

The buildup of these toxins over time can cause liver failure and can lead to liver cancer and kidney problems.

With proper medical and nutrition management the buildup of the toxins is controlled to support normal growth and development of your child.

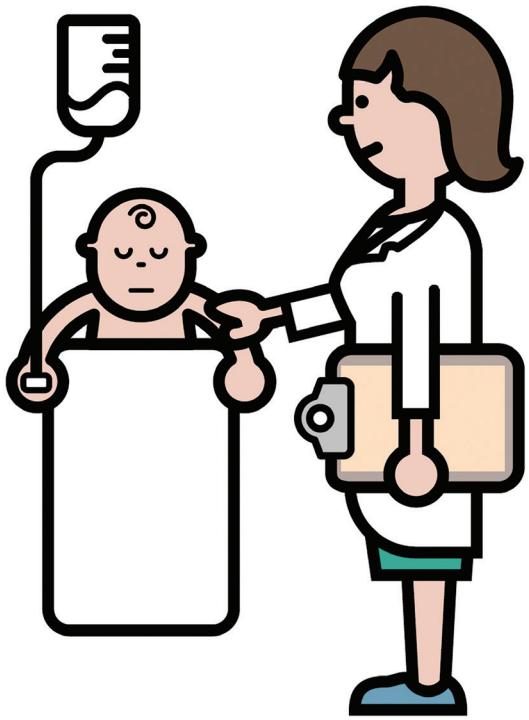




What are the symptoms in HT-1?

Most babies become unwell in the first few months of life. Symptoms include:

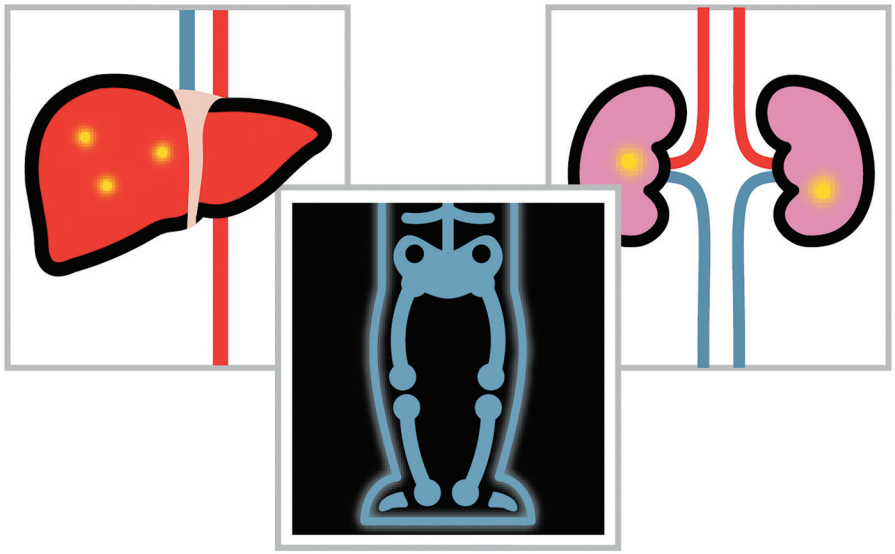
- Poor weight gain
- Liver failure



Other children have a gradual onset of symptoms such as:

- Large liver
- Rickets
- Kidney problems

Some children may develop liver cancer.



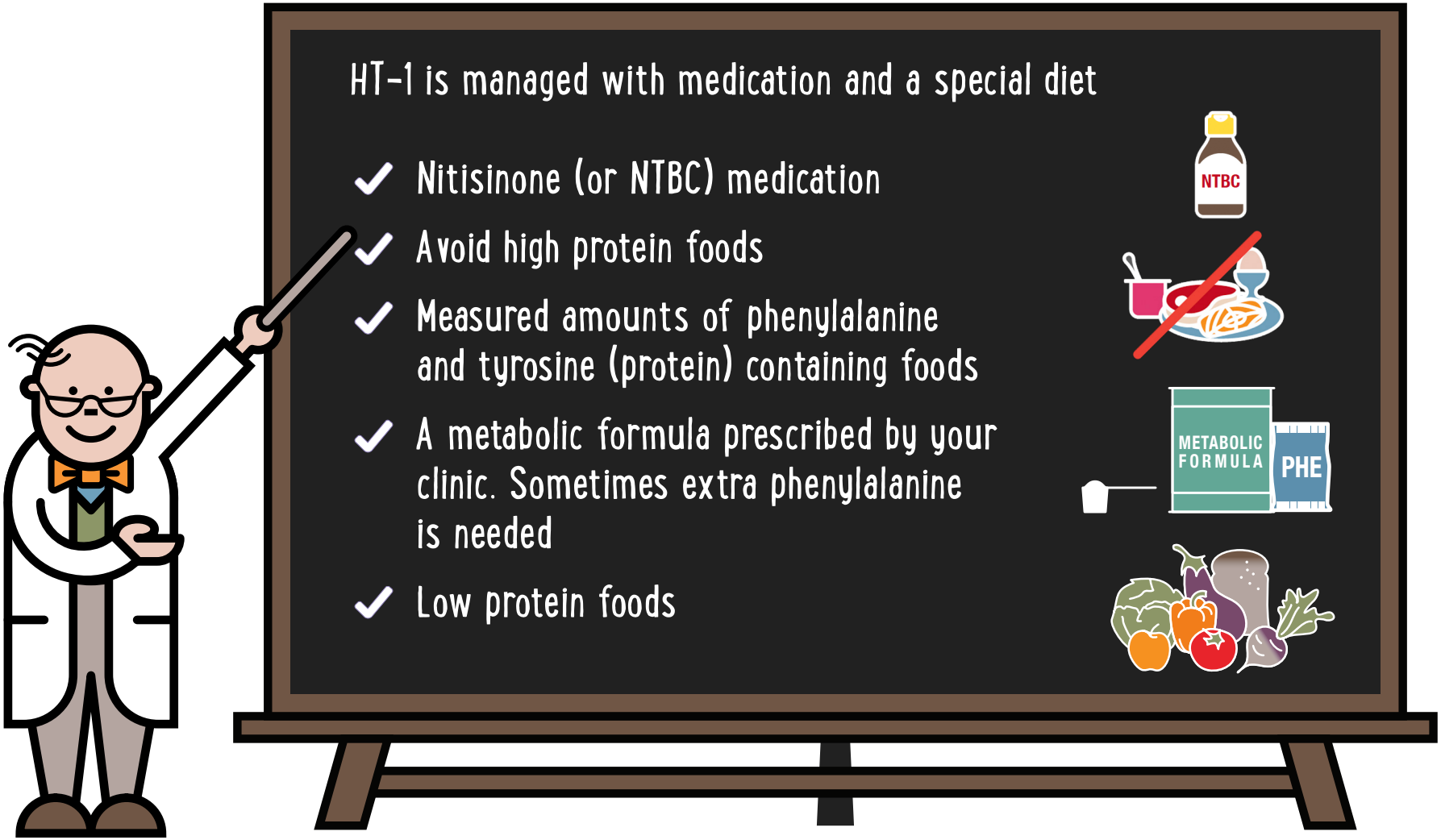
How is HT-1 diagnosed?

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

The blood sample is then analyzed for succinylacetone and/or tyrosine.

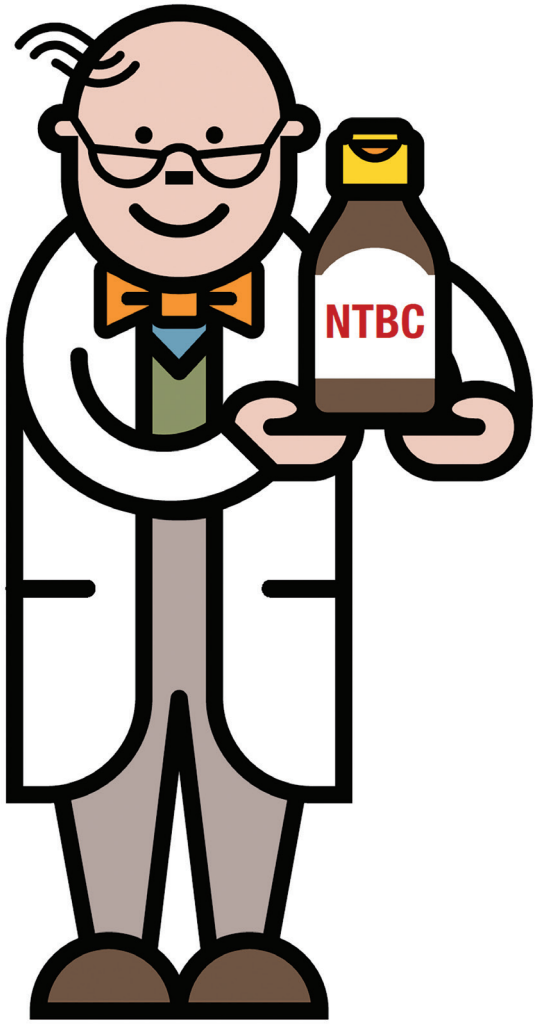
Abnormal results could mean your child has HT-1 which will prompt your clinician to do further testing to confirm the diagnosis.

How is HT-1 managed day-to-day?



Why is it important to take NTBC?

NTBC helps prevent the buildup of the toxins. It also helps prevent liver and kidney damage and helps to lower the risk of liver cancer. Your child will start taking NTBC as soon as possible.



Avoid high protein foods

Taking NTBC medication is important, but it has metabolic effects such as the buildup of tyrosine.

Therefore, the other important part of HT-1 management is a special diet which is restricted in whole protein to limit tyrosine and phenylalanine intake. Foods high in protein include **meat, fish, eggs, cheese, milk, bread, pasta, nuts, soy and tofu.**

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

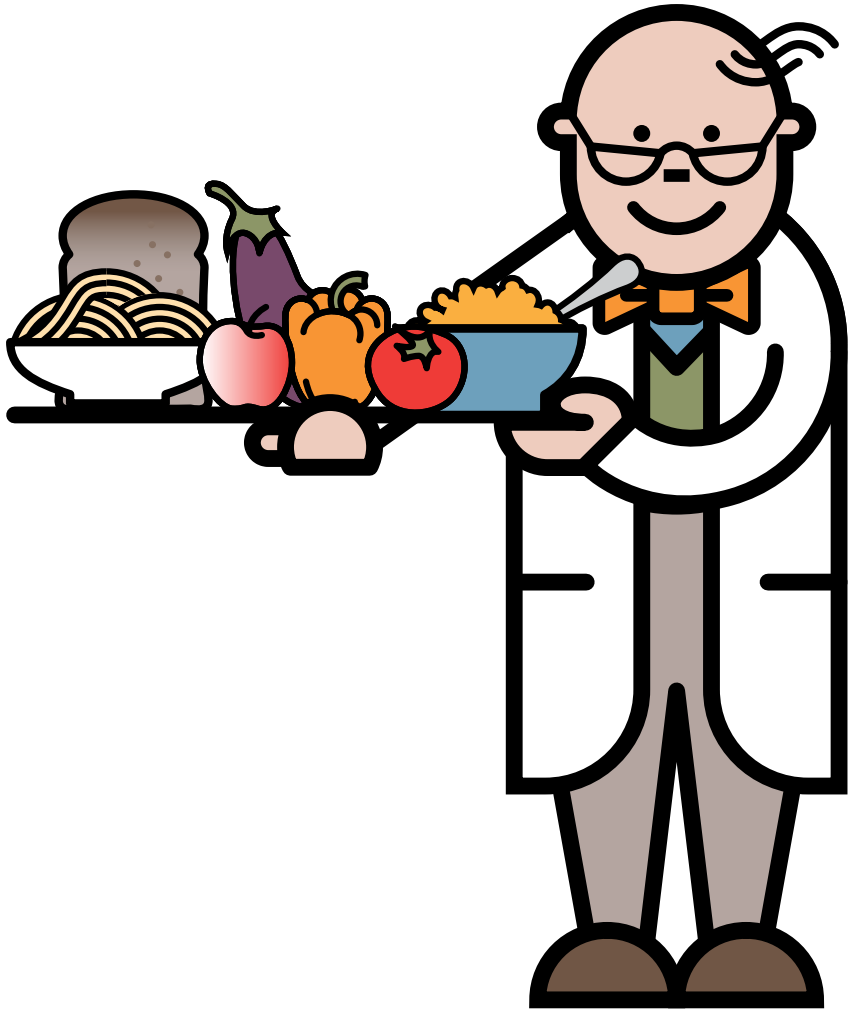
Include foods low in protein

These are foods that contain small amounts of tyrosine and phenylalanine 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

Tyrosine and phenylalanine are essential for normal development and therefore a limited and controlled amount must be taken daily.

Breast milk or standard infant formula will provide the tyrosine and phenylalanine 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 tyrosine and phenylalanine.

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

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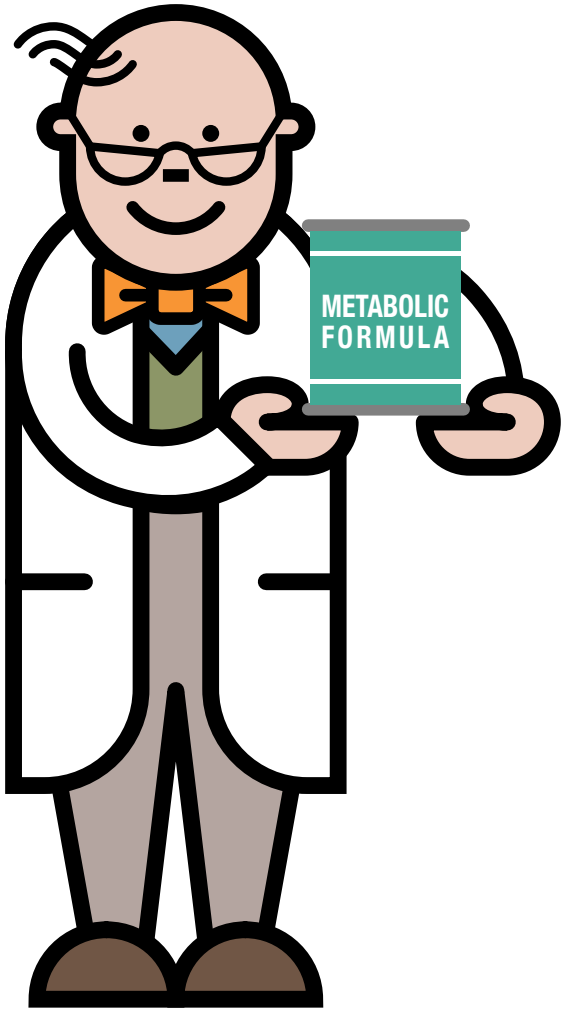
Tyrosine- and phenylalanine-free metabolic formula

Tyrosine- and phenylalanine-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 and minerals; while the protein comes in the form of amino acids without tyrosine and phenylalanine.

Metabolic formula, plus the prescribed amounts of tyrosine and phenylalanine, allows your baby to get the nutrients he or she needs to grow.



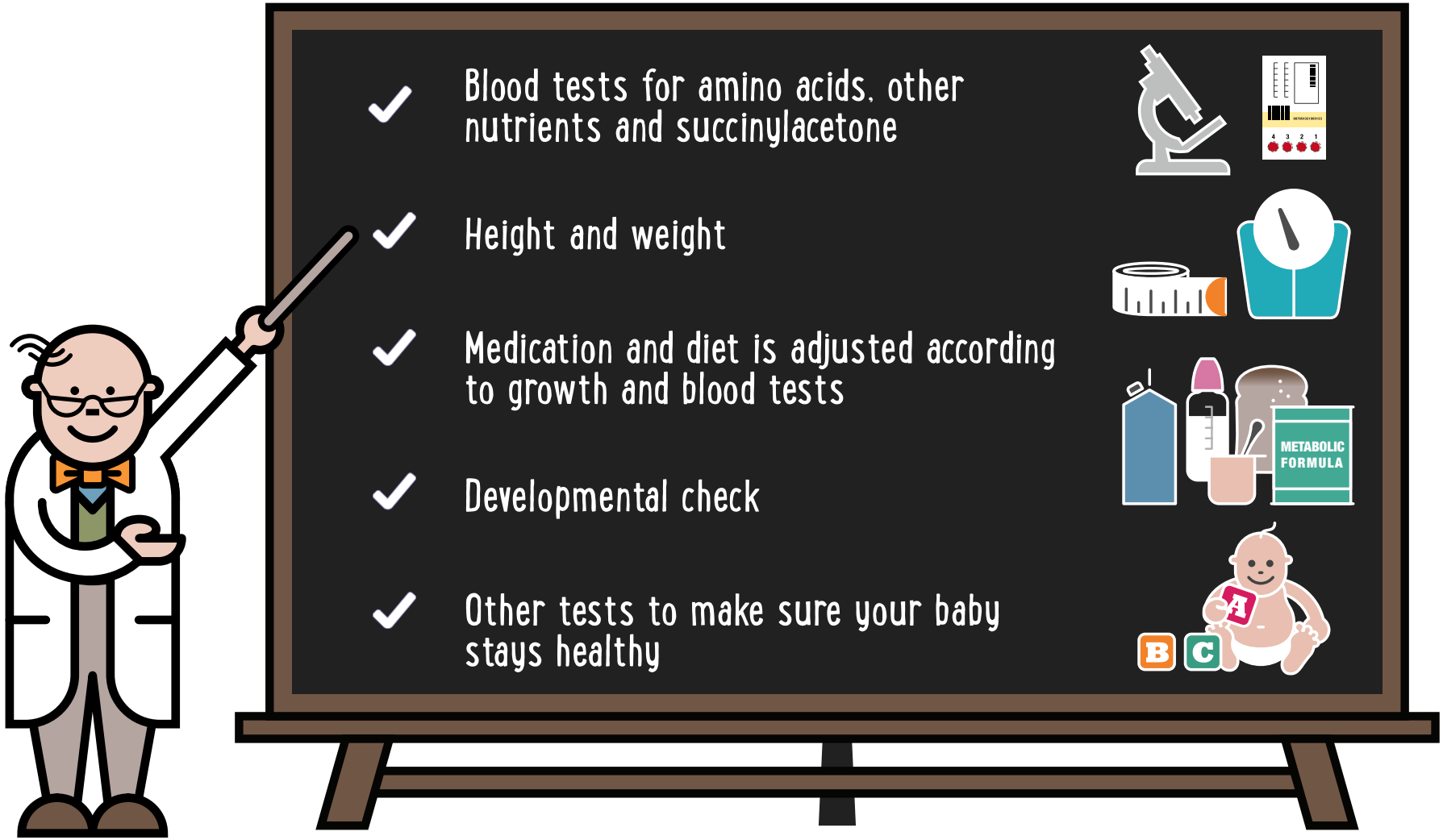
Tracking tyrosine and phenylalanine

As your baby starts to eat solids your clinic will work with you to track tyrosine and phenylalanine.

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

Your clinic can help you find the best tools to help determine the tyrosine and phenylalanine content of foods.

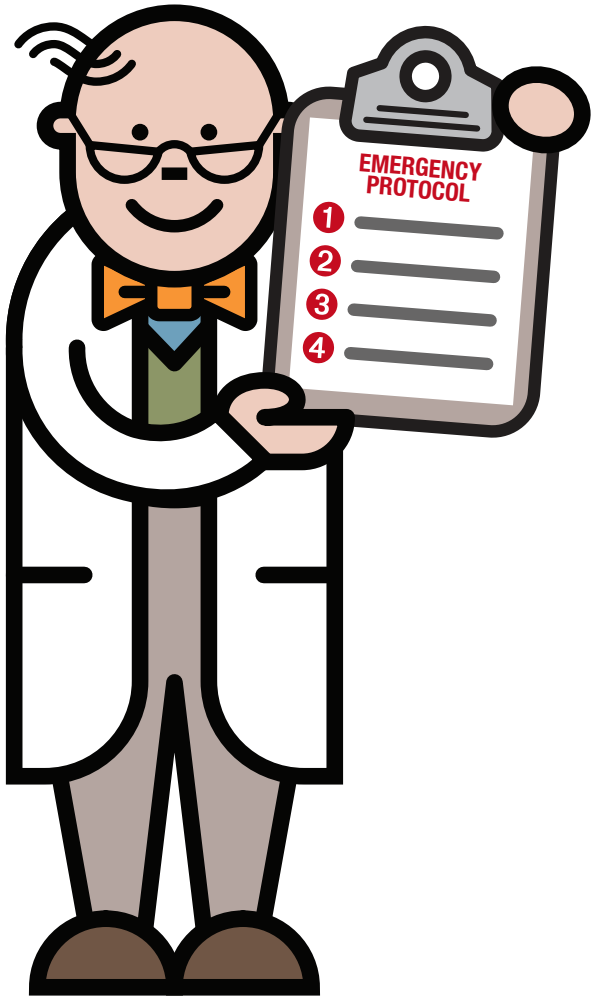
What is monitored in HT-1?



How is HT-1 managed during illness?

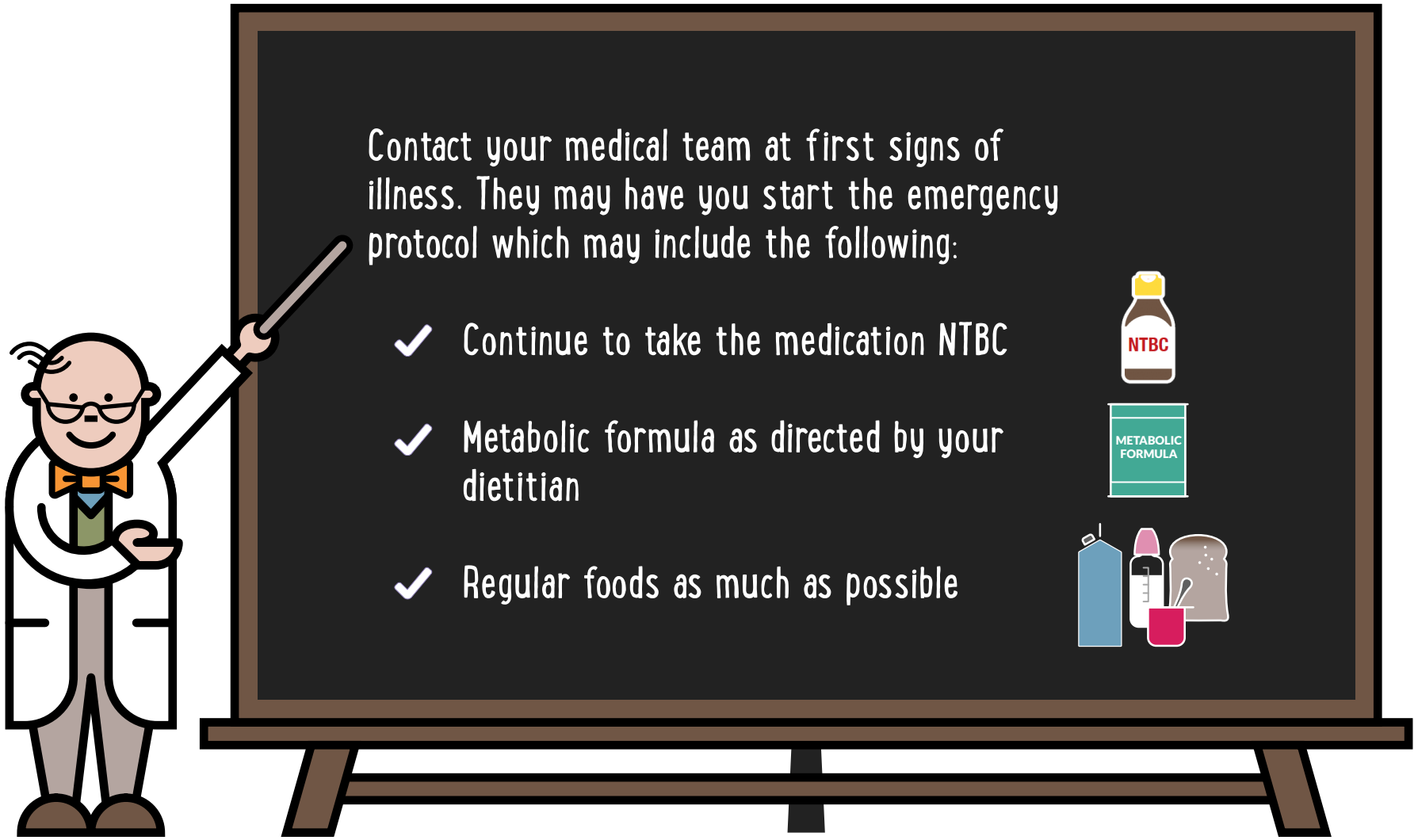
During any illness, our bodies need extra energy. The body will start breaking down cell protein, which will lead to a buildup of tyrosine and succinlyacetone and other toxic substances.

It is important to start the sick day protocol your metabolic team has developed for you and contact them.

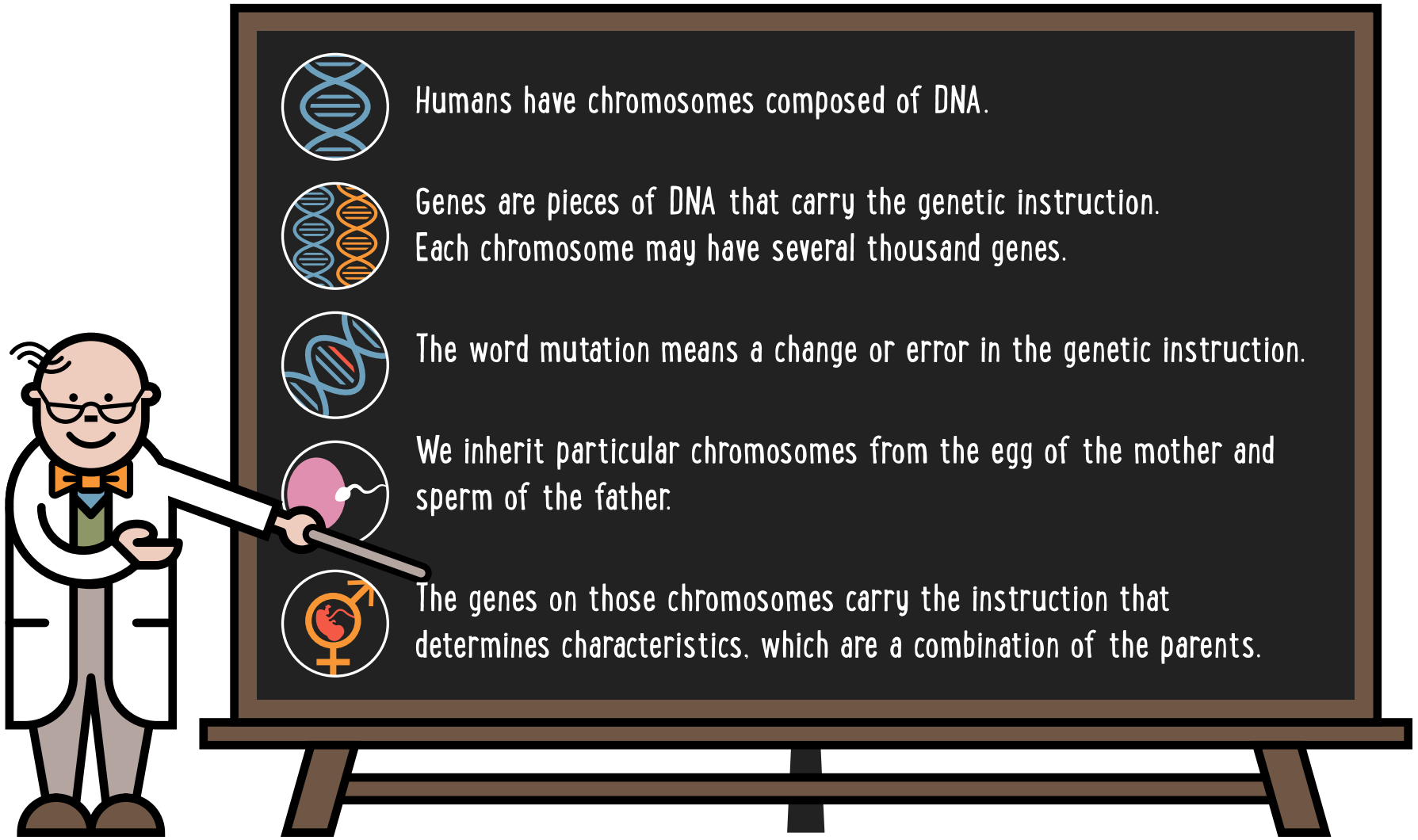


How is HT-1 managed during illness?

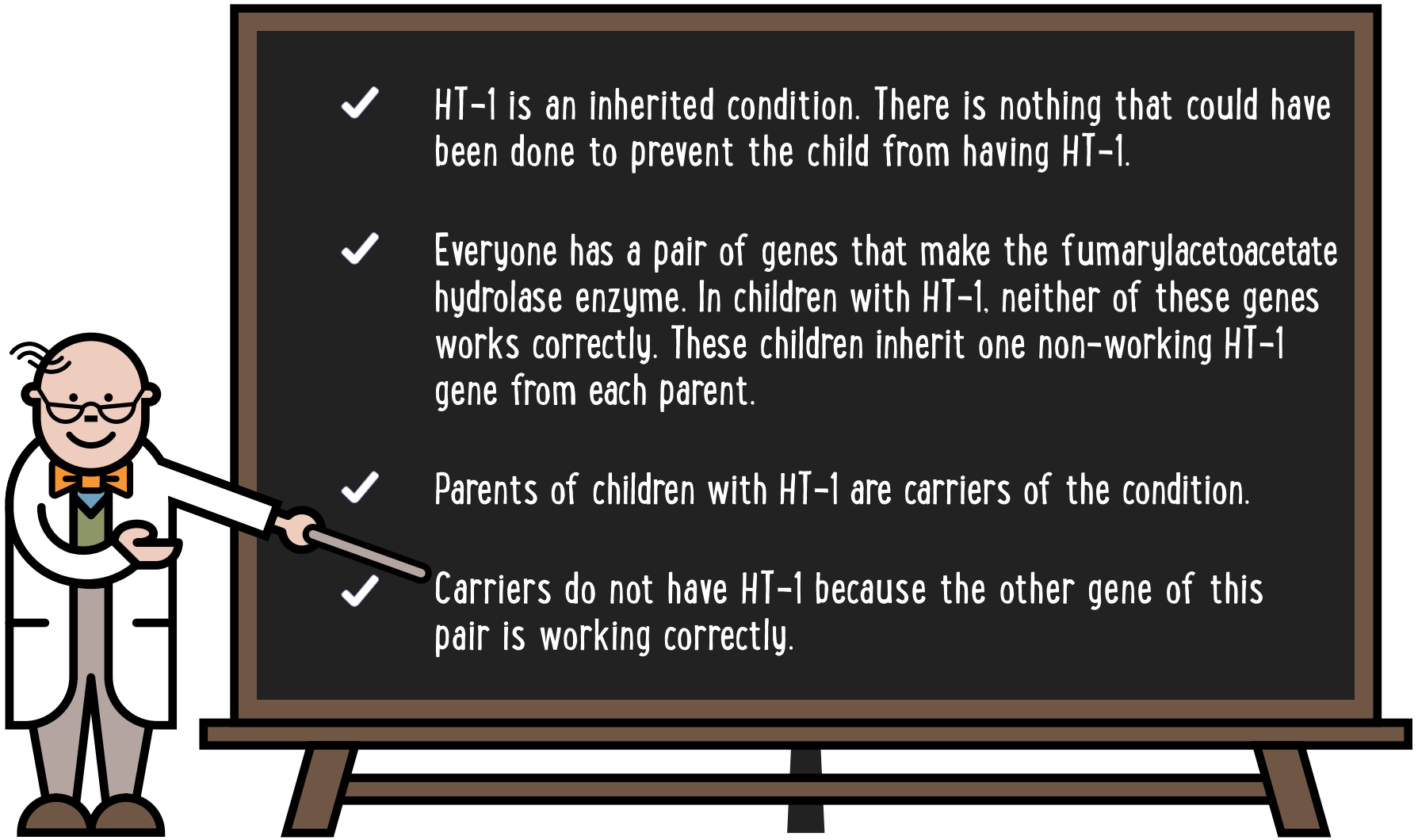
Always follow your medical team's guidance.



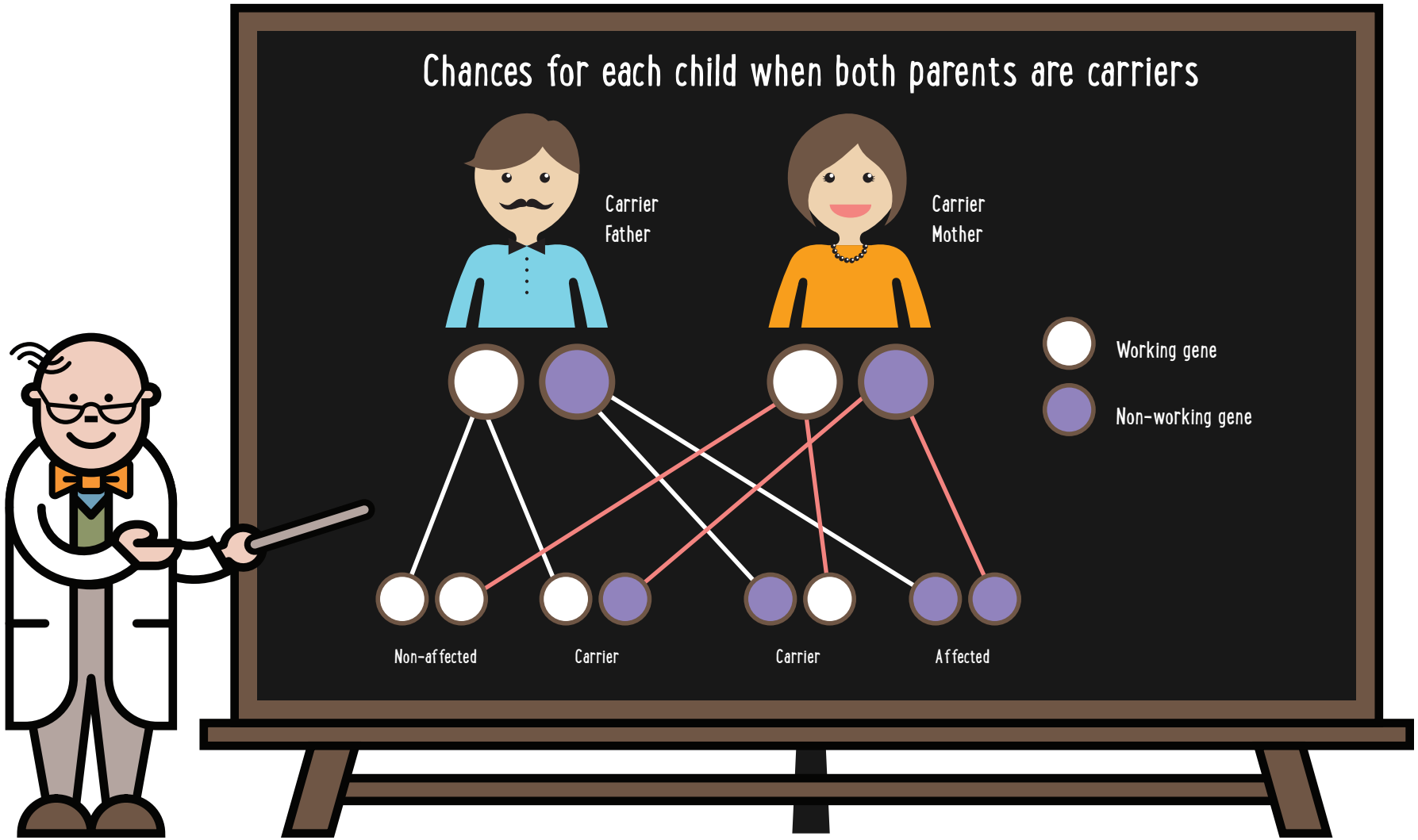
What happens in human genetics?



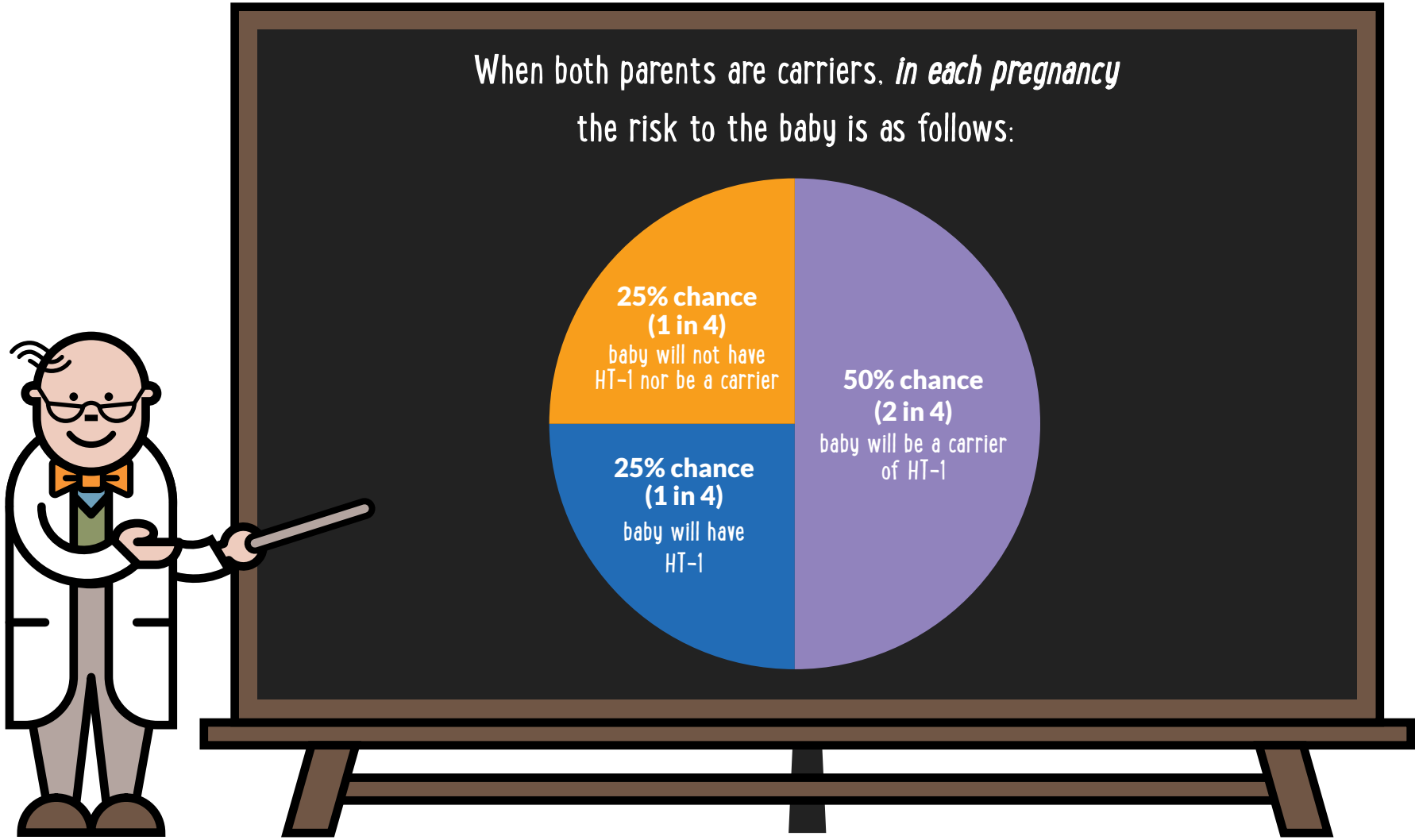
How does one inherit HT-1?



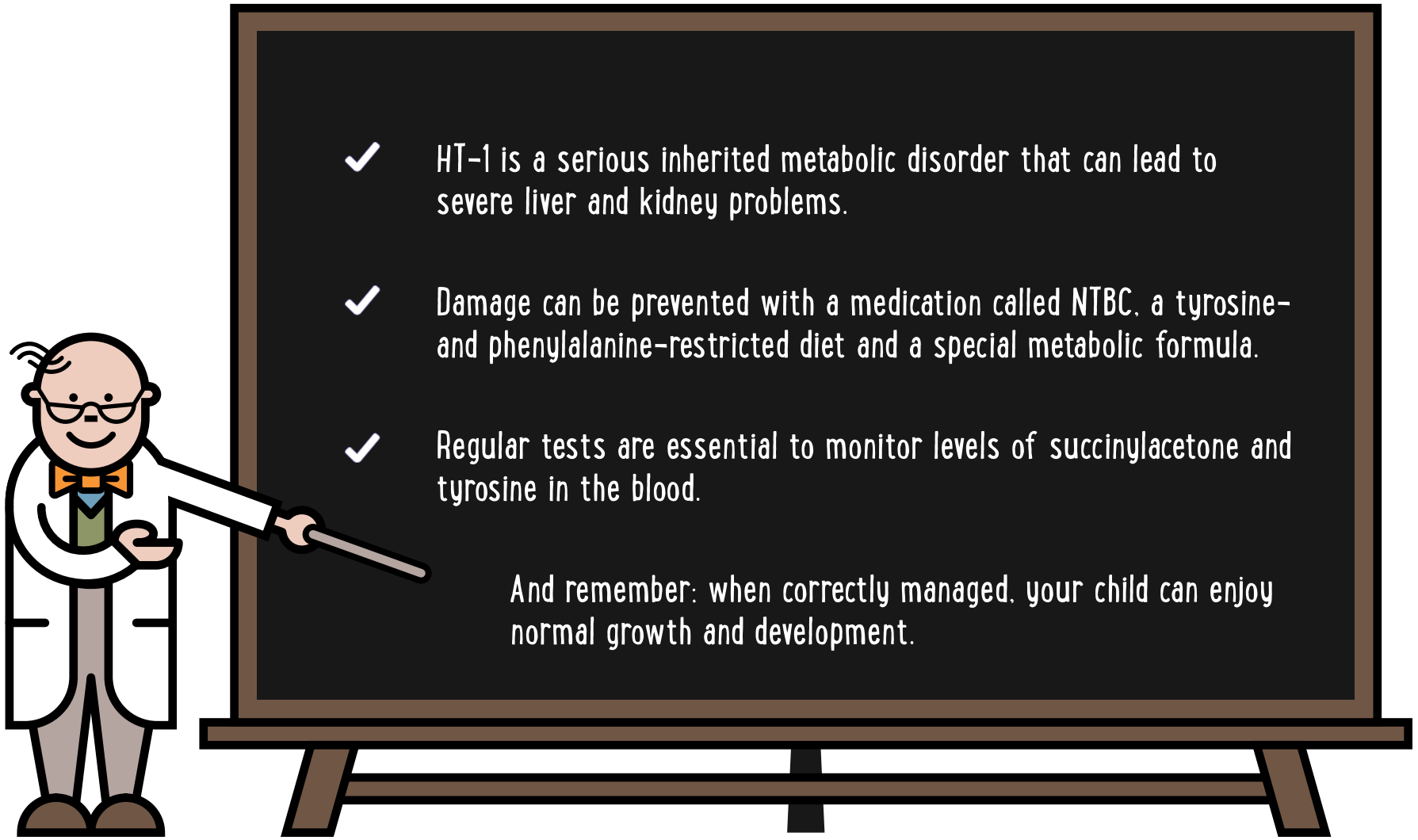
Inheritance – Autosomal-recessive - possible combinations



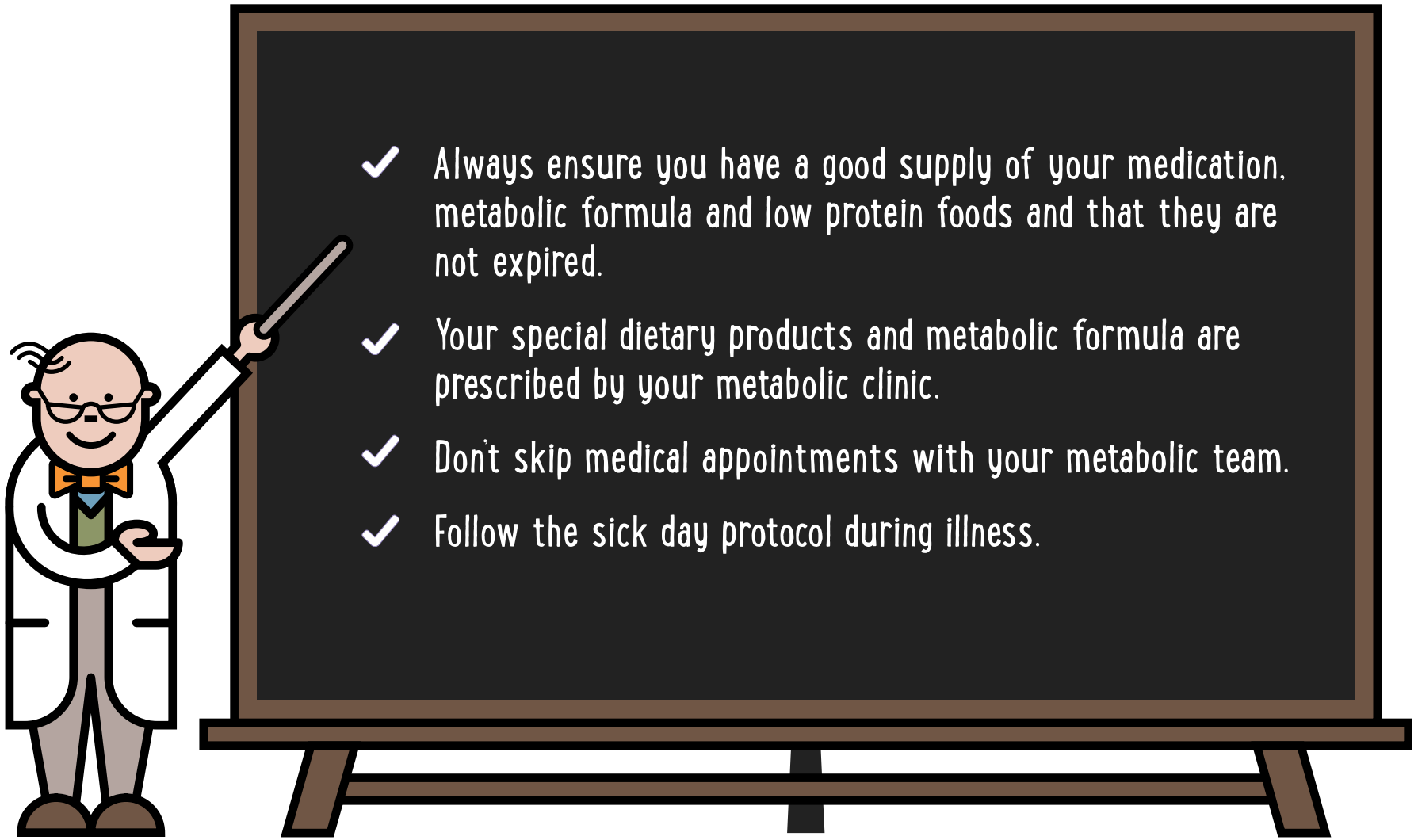
Future pregnancies



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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Tools **E**nabling **M**etabolic **P**arents **LE**arning

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