

# TEMPLE



Tools **E**nabling **M**etabolic **P**arents **L**Earning

ADAPTED BY THE DIETITIANS GROUP

**BIMDG**

British Inherited Metabolic Diseases Group



BASED ON THE ORIGINAL TEMPLE WRITTEN BY  
BURGARD AND WENDEL

VERSION 4, OCTOBER 2020

HT1

Supported by **NUTRICIA**  
as a service to metabolic medicine

# TEMPLE foreword

TEMPLE (Tools Enabling Metabolic Parents LEarning) are a set of teaching slides and booklets that provide essential information about different inherited metabolic disorders that require special diets as part of their management. These teaching tools are aimed at parents who may have an infant or child that has been recently diagnosed with a disorder. They are also useful when teaching children, extended family members, child minders, nursery workers and a school team.

They have been developed by a team of experienced clinical and research metabolic dietitians from the UK who are members of the British Inherited Metabolic Disease Group (BIMDG).

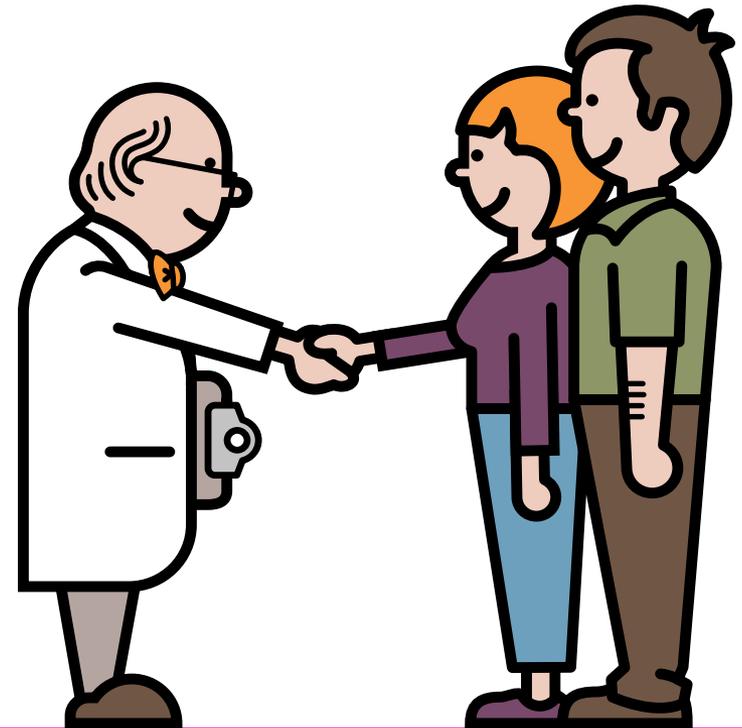
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

Each module produced is reviewed by a consultant clinician who is a member of the BIMDG.

**This teaching tool is not designed to replace dietary information that may be given by a dietitian in clinic.**

# Tyrosinaemia Type 1 (HT1)

Information for families following  
a new diagnosis



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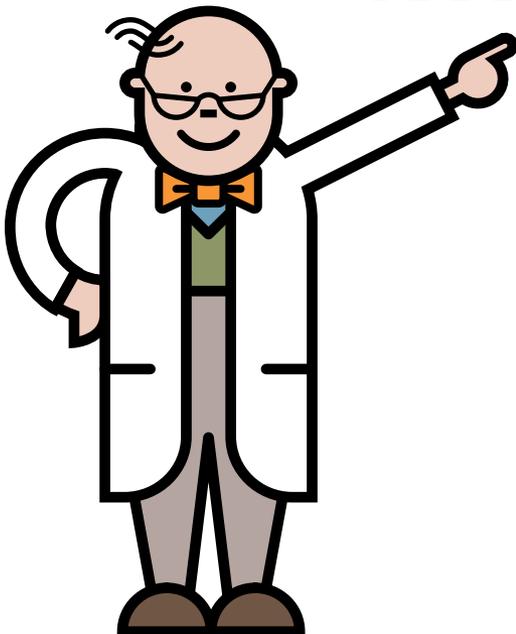
# What is HT1?

HT1 stands for Isovaleric Acidaemia

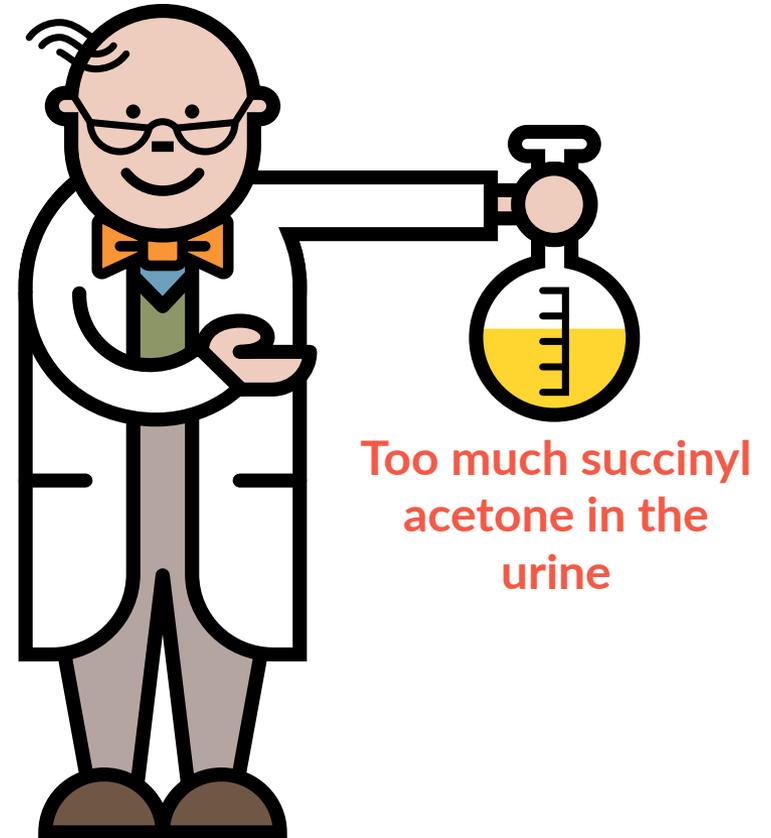
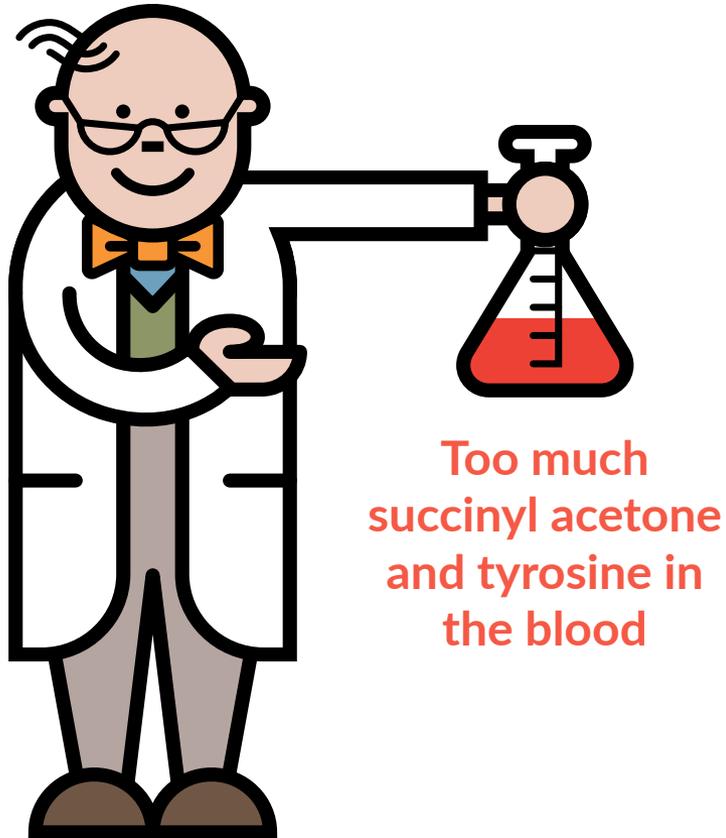
It is an inherited metabolic condition

Hereditary Tyrosinaemia Type 1

HT1



# What is HT1?

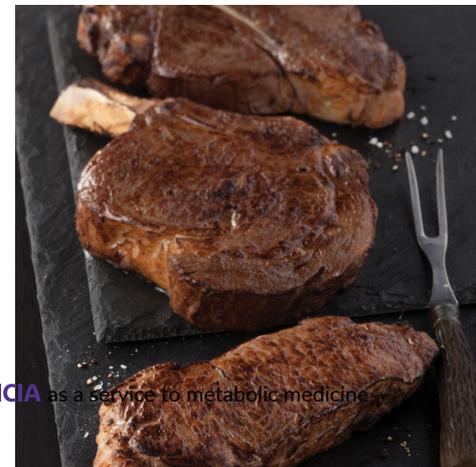
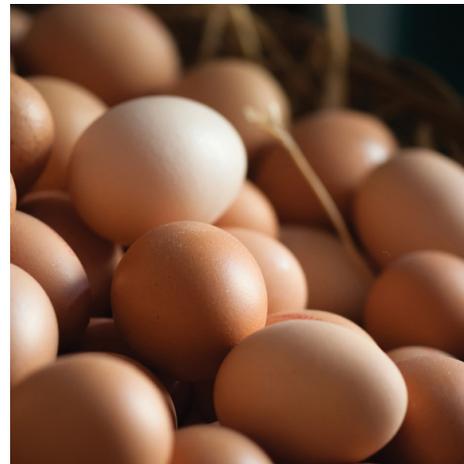


# HT1 and protein

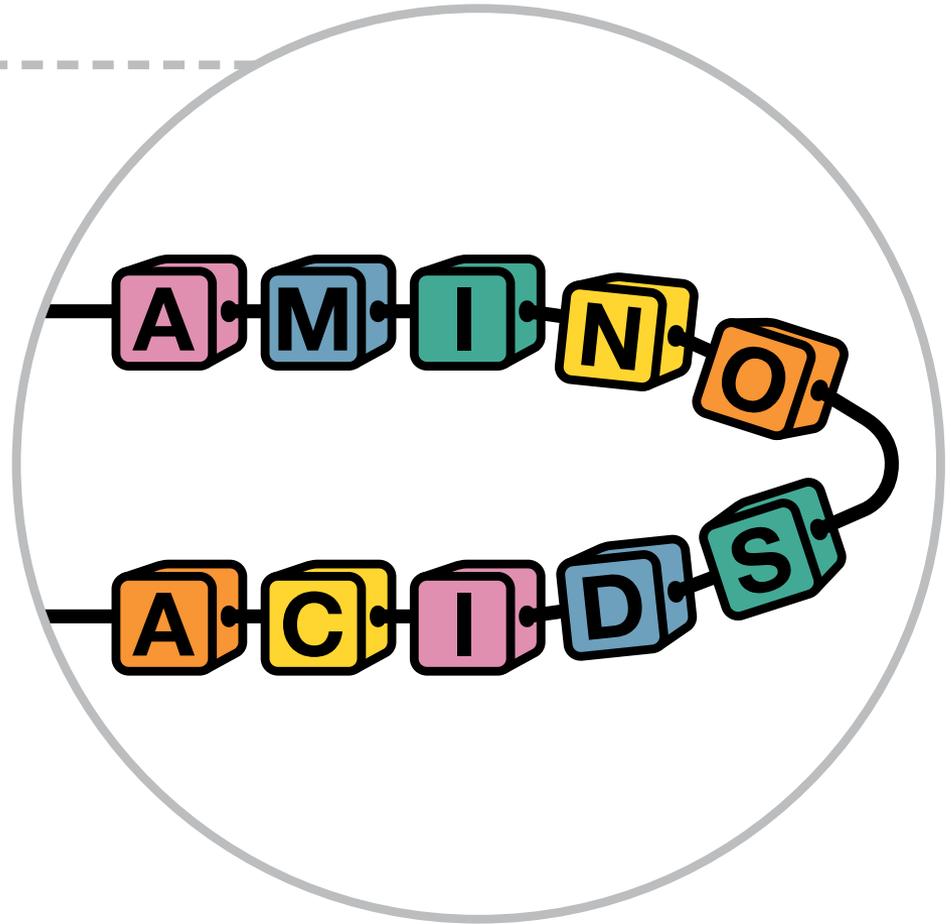
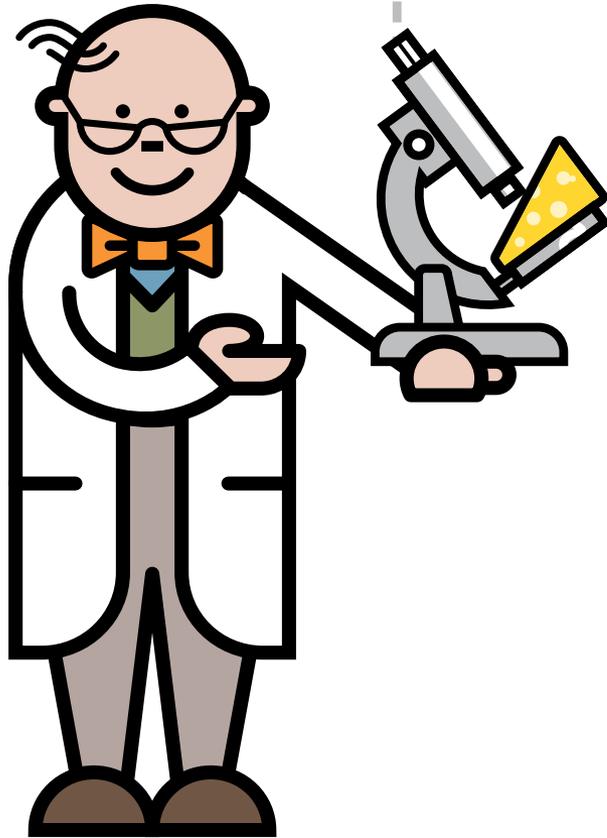
HT1 affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair



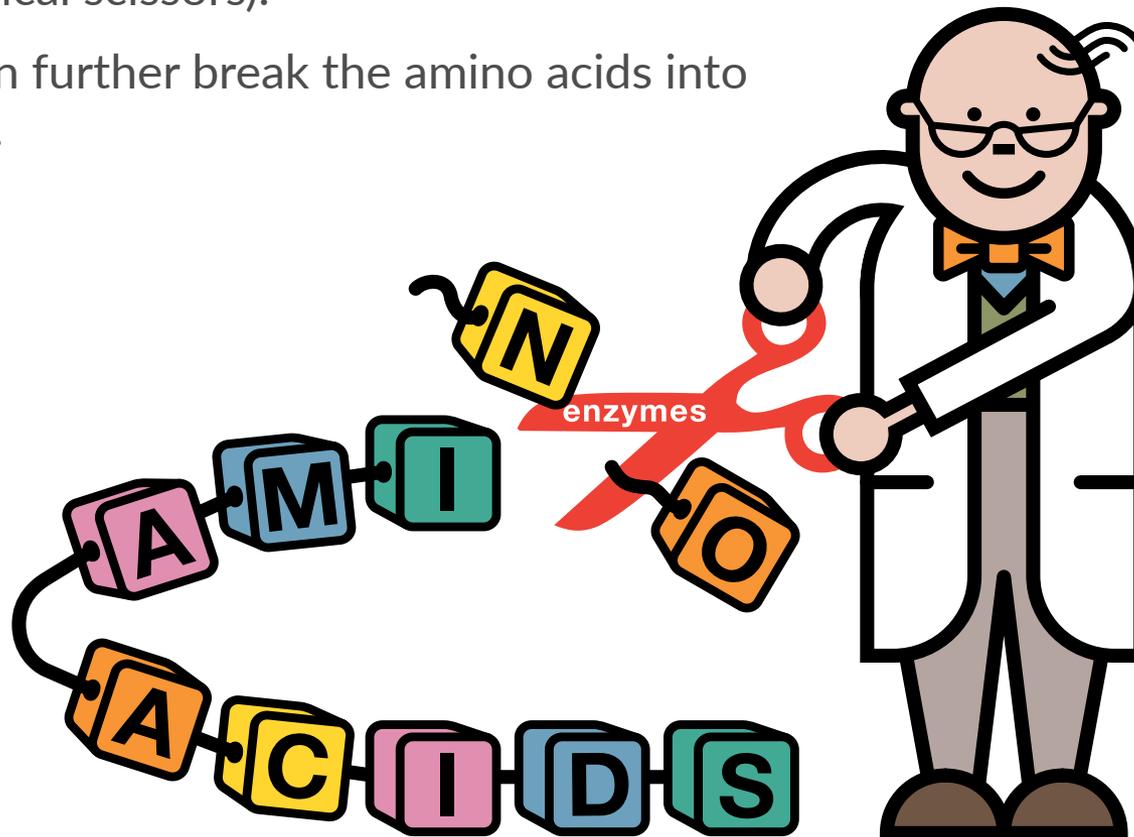
# What is protein?



# Protein and enzymes

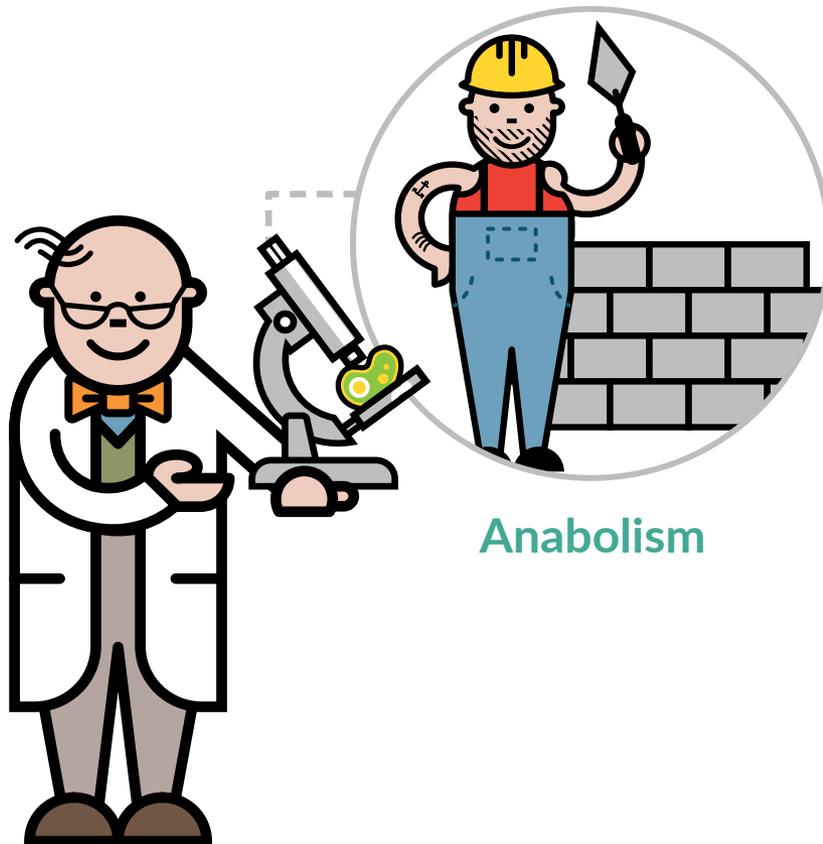
Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.

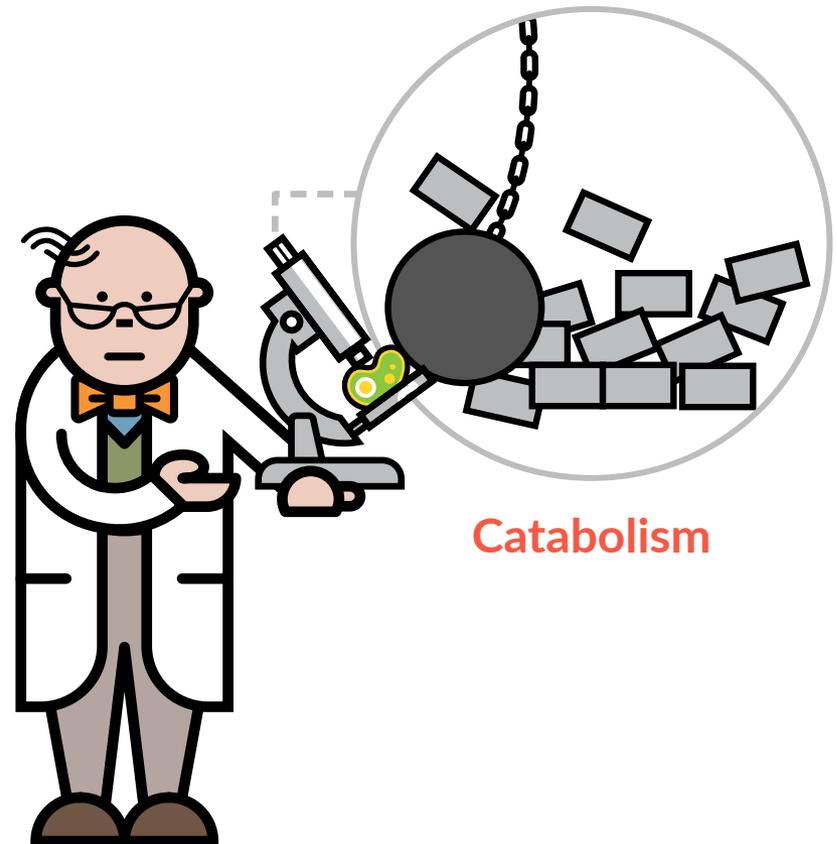


# Protein metabolism

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



Anabolism

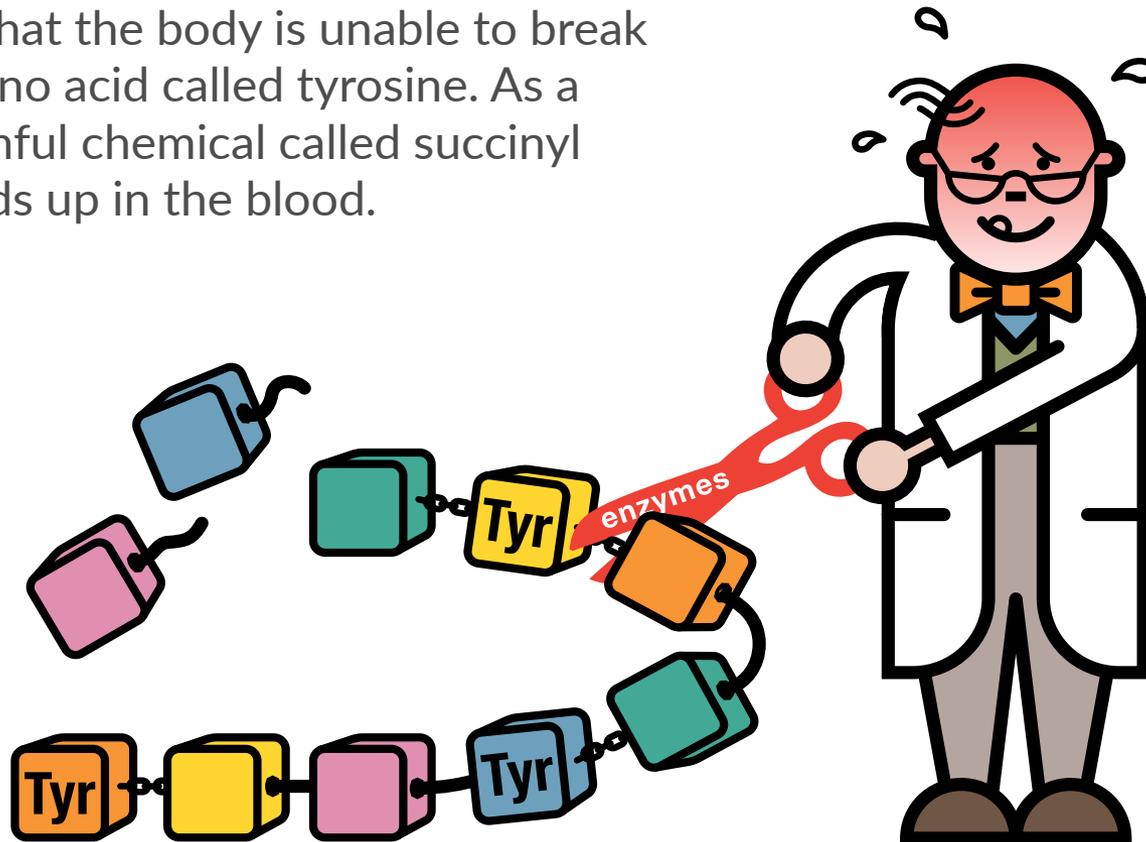


Catabolism

# What happens in HT1?

In HT1, the body lacks an enzyme called **fumarylacetoacetate hydrolyase**.

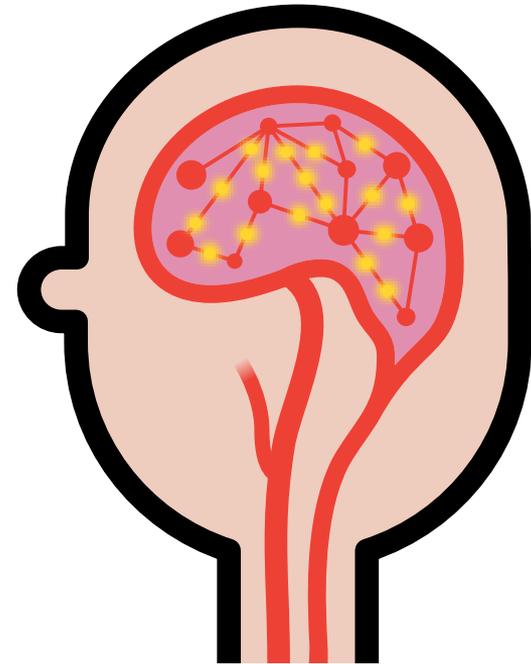
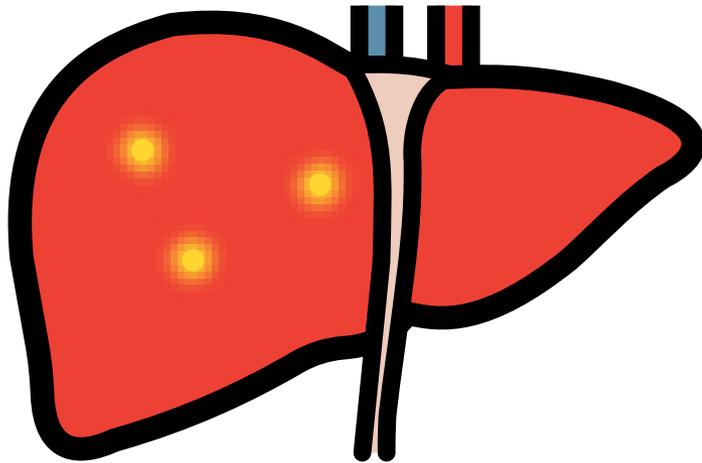
This means that the body is unable to break down an amino acid called tyrosine. As a result, a harmful chemical called succinyl acetone builds up in the blood.



# What can go wrong in HT1?

When unmanaged HT1 can cause liver failure and lead to liver cancer.

Some children may have lower school achievement.



# What are the symptoms in HT1?

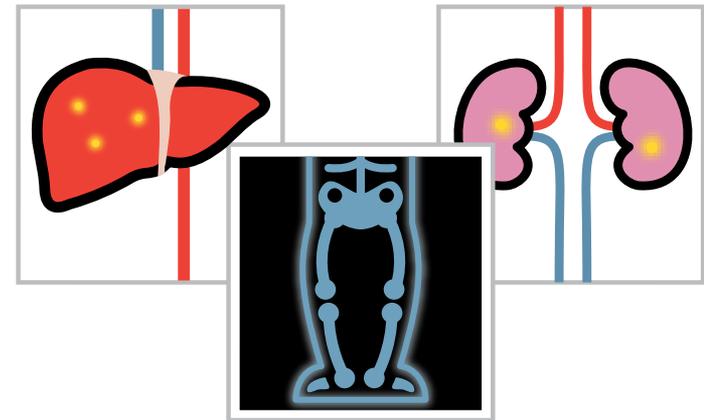
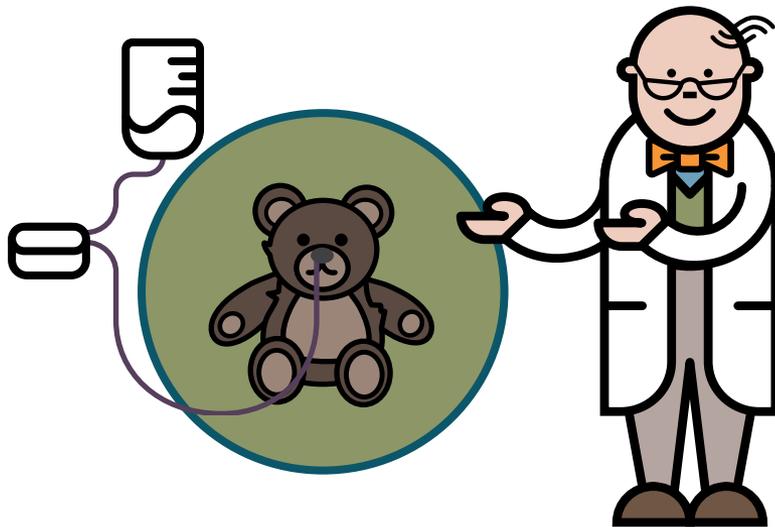
Most babies become unwell in the first few months of life. Symptoms include:

- 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 HT1 diagnosed?

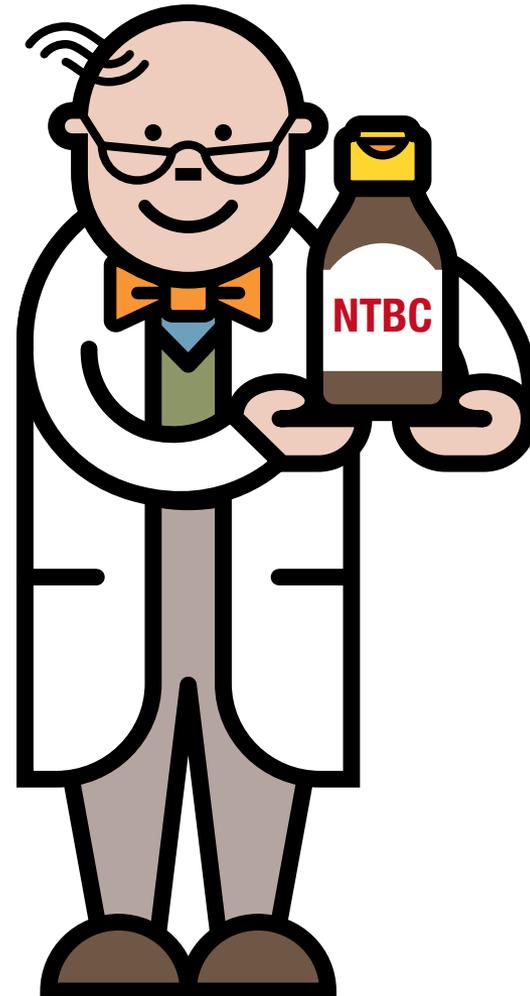
HT1 is diagnosed by high levels of succinyl acetone in the blood and urine.



# Management with nitisinone

This medication is also called NTBC.

- Your child will start taking NTBC/ nitisinone as soon as possible
- NTBC/nitisinone helps prevent liver and kidney damage and lowers the risk of liver cancer



# How is HT1 managed?

HT1 is managed with the following special diet and medication:

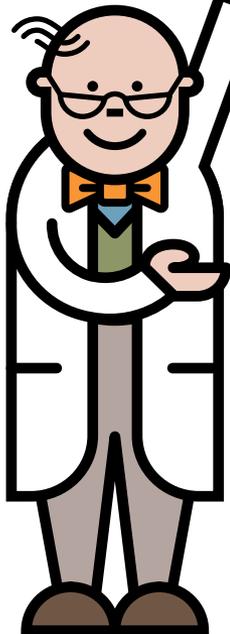
Nitisinone medication

Limited high protein foods

Measured amounts of tyrosine (protein) containing foods

A protein substitute. Sometimes extra phenylalanine is needed

Low protein foods



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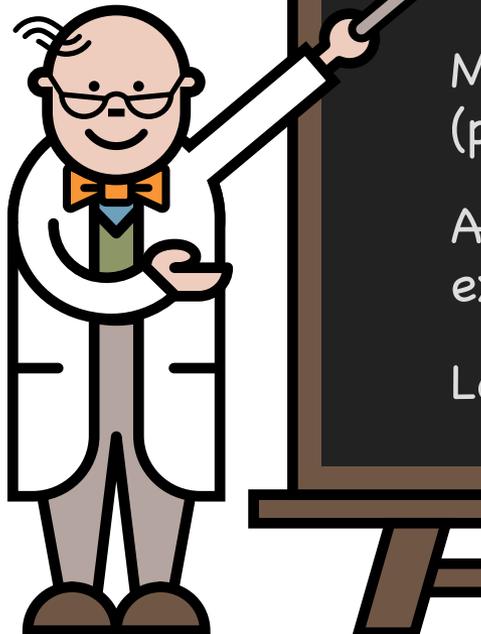
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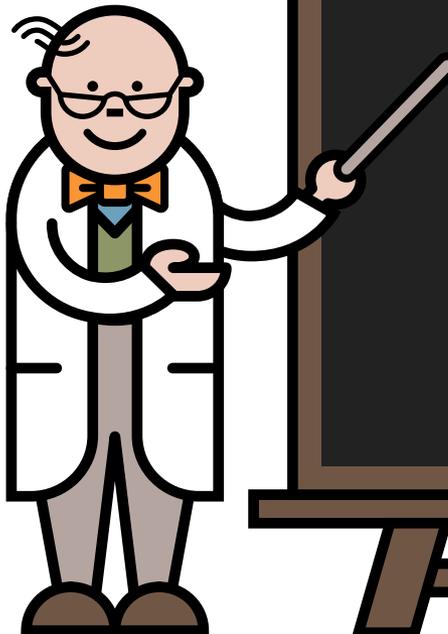
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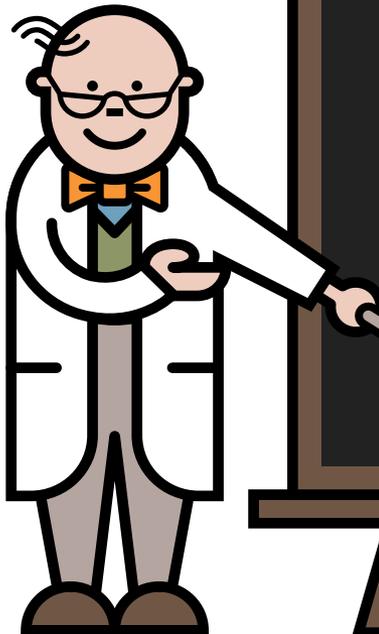
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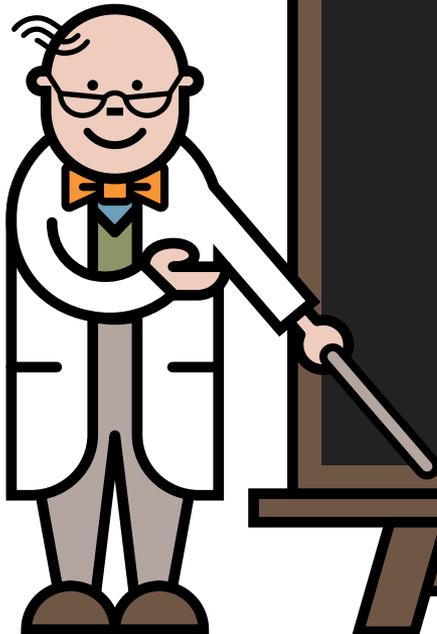
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# High protein foods

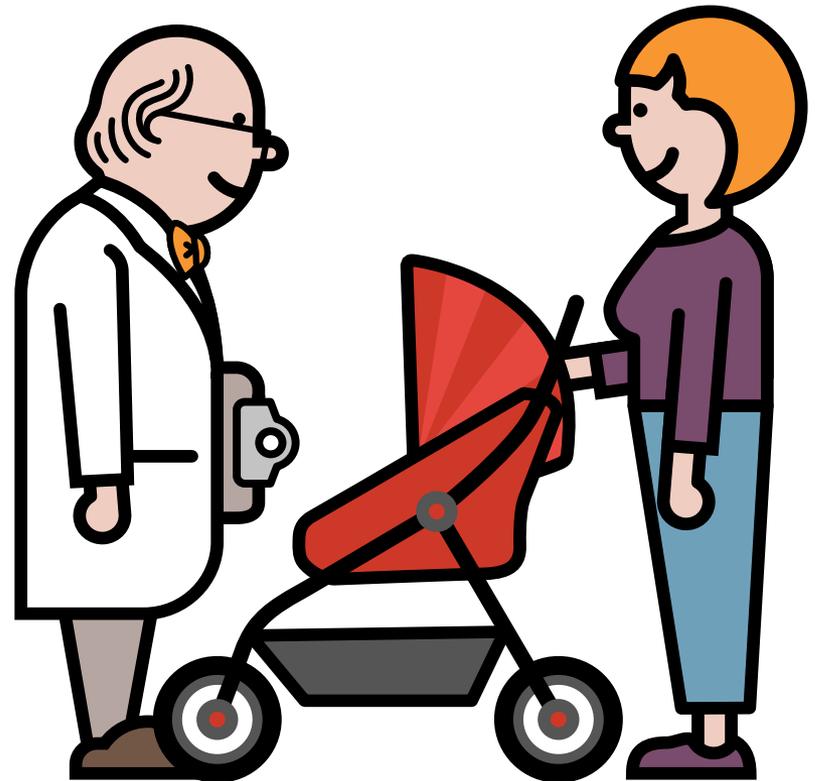
These foods are high in tyrosine (protein) and must be avoided: meat, fish, eggs, cheese, milk, bread, pasta, nuts, seeds, soya, Quorn and tofu.



# Measured phenylalanine intake

In babies, a restricted amount of phenylalanine (protein) is given from breast milk or measured amounts of infant formula.

The amount given will be monitored regularly by your specialist metabolic dietitian.

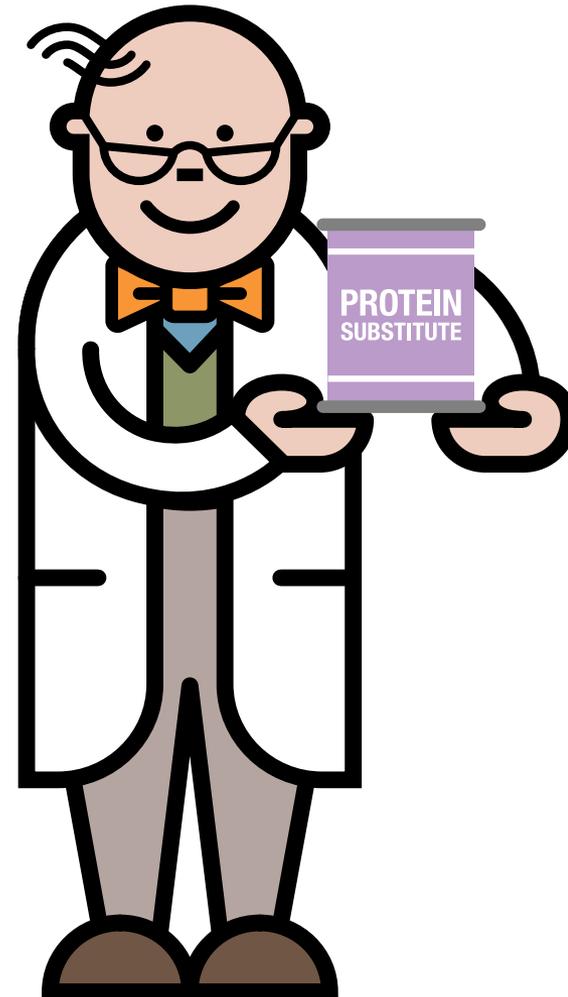


# Protein substitute

Protein substitute is essential for metabolic control.

It will help to meet your baby's protein, energy, vitamin and mineral requirements.

It is available on prescription.

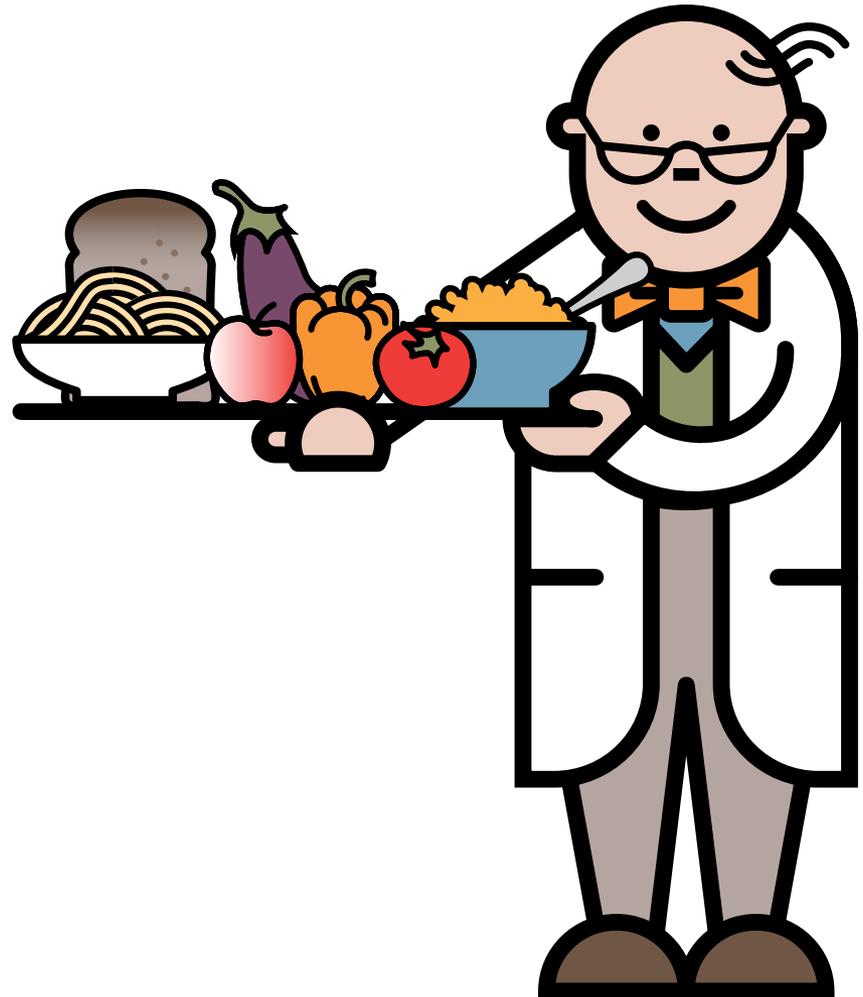


# Low protein foods

There are many low protein foods. These include fruit, many vegetables and prescribable low protein foods such as bread and pasta.

They provide:

- a source of energy
- variety in the diet

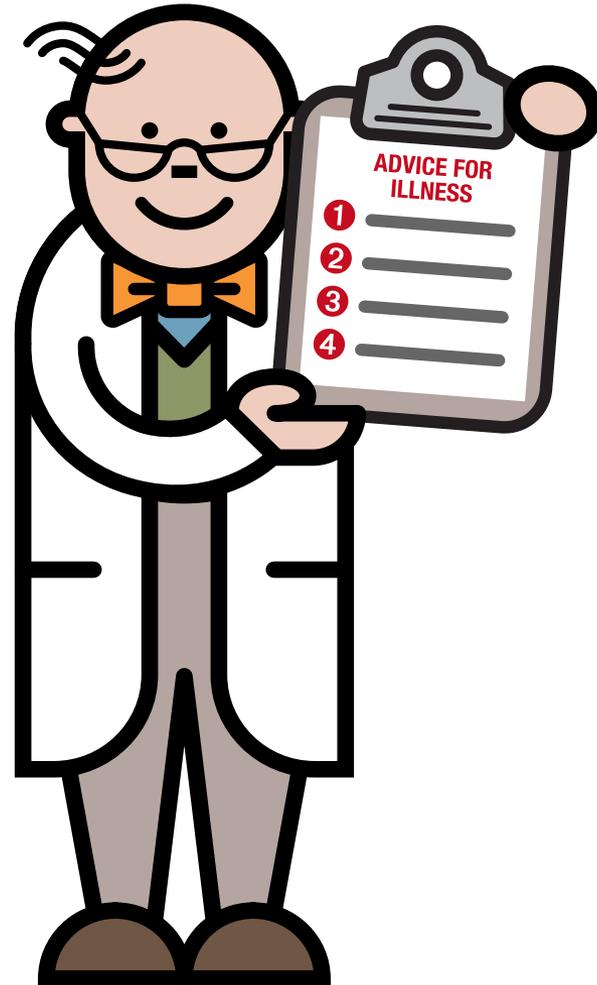


# How is HT1 managed during illness?

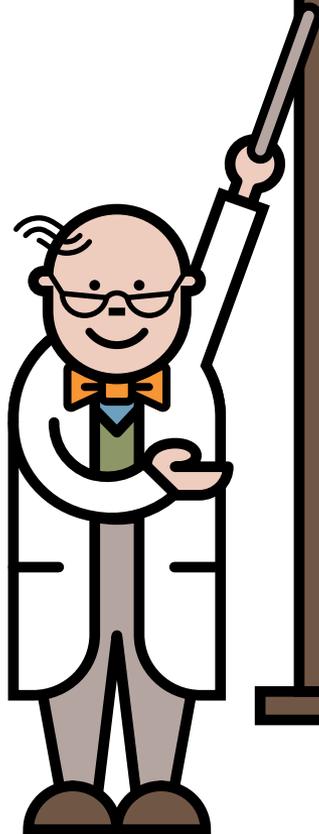
During any childhood illness, catabolism or protein breakdown occurs, causing blood tyrosine levels to increase.

It is important to continue with the usual diet as much as possible.

The drug nitisinone should be given at all times including illness.



# How is HT1 monitored?



Frequent blood tests to check tyrosine, phenylalanine and other chemical levels



Height and weight



Developmental checks



Diet and medication is adjusted according to age, weight and blood tests



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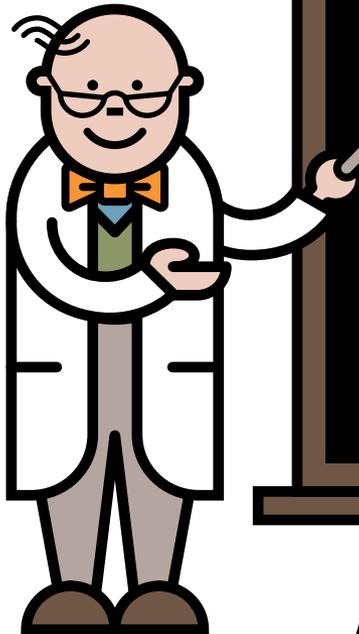
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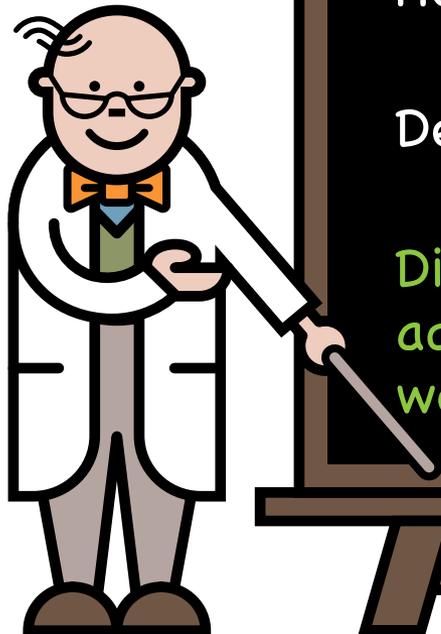
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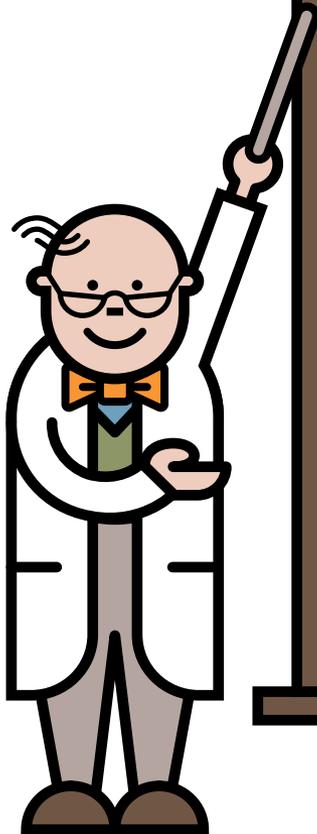
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# Chromosomes, genes, mutations



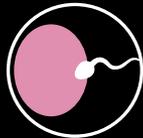
Humans have chromosomes composed of DNA



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

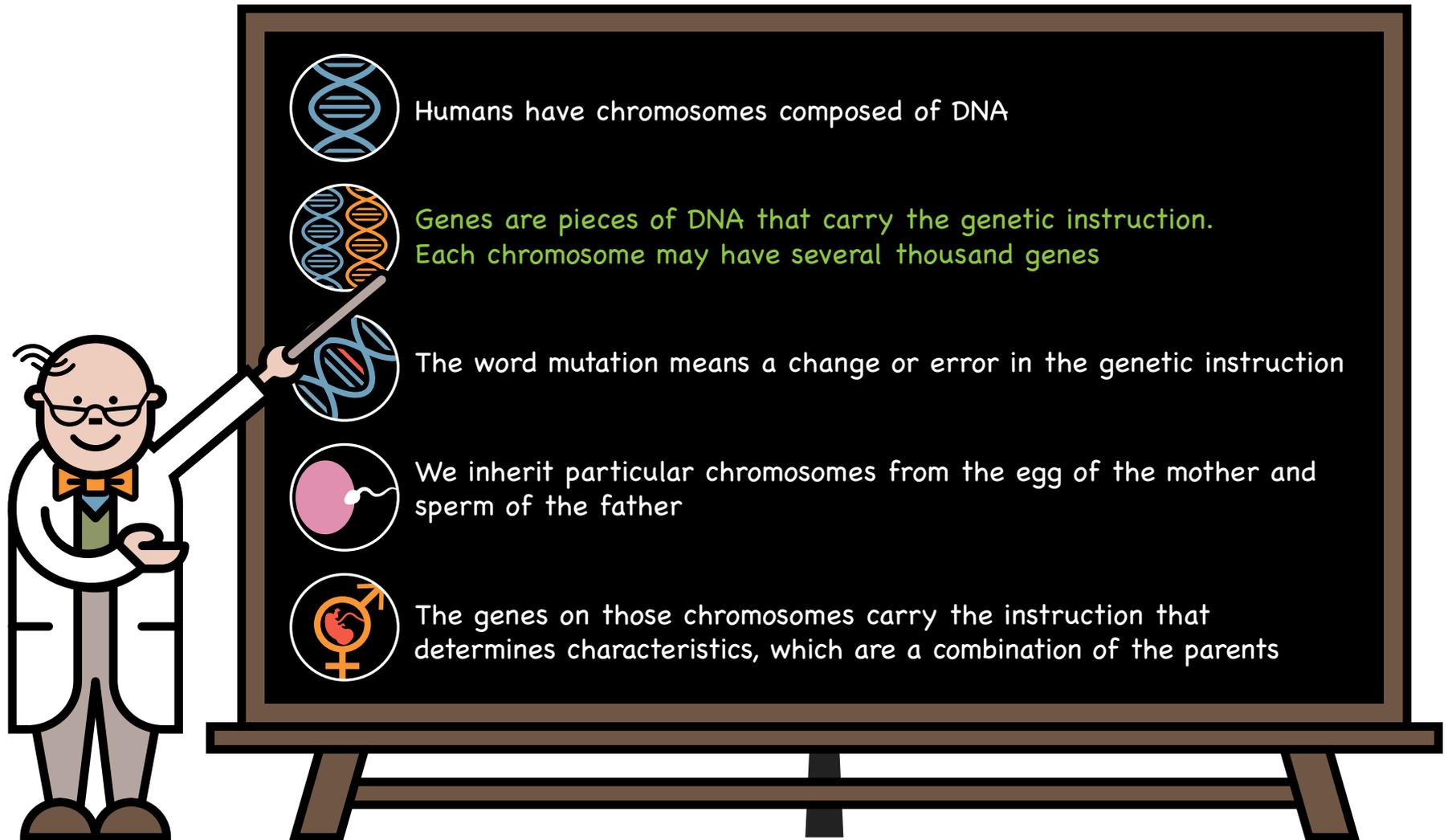


We inherit particular chromosomes from the egg of the mother and sperm of the father



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

# Chromosomes, genes, mutations



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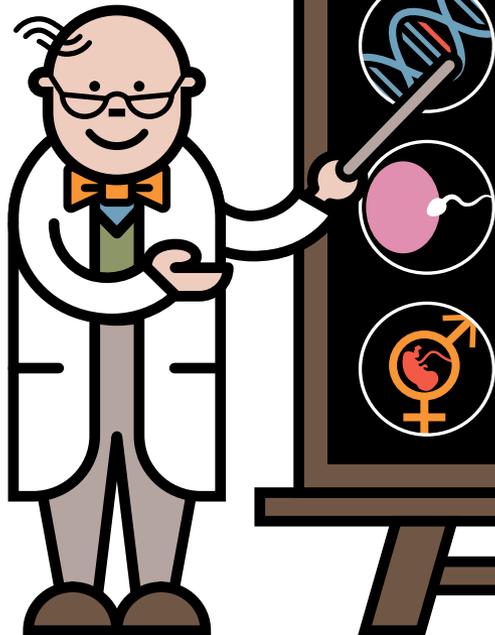
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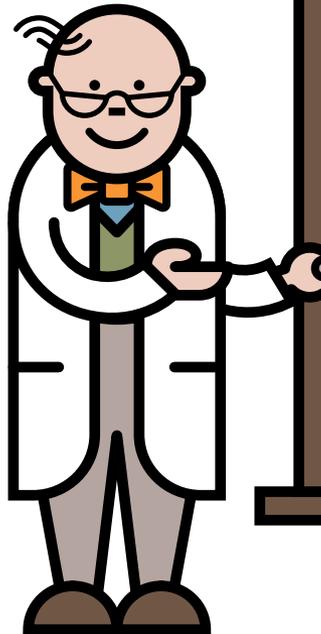


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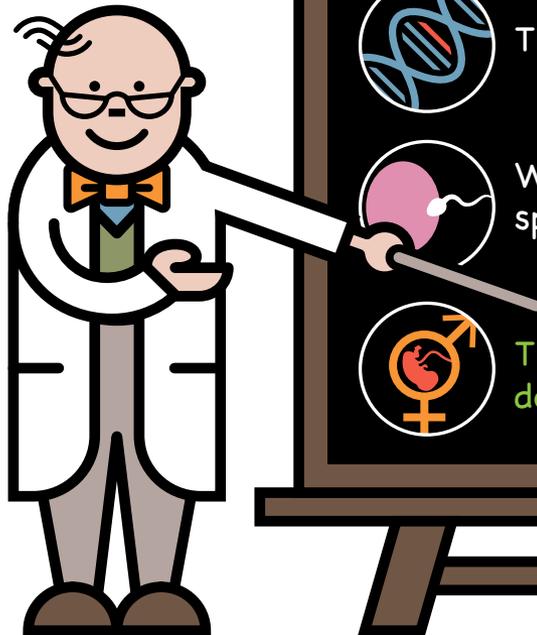


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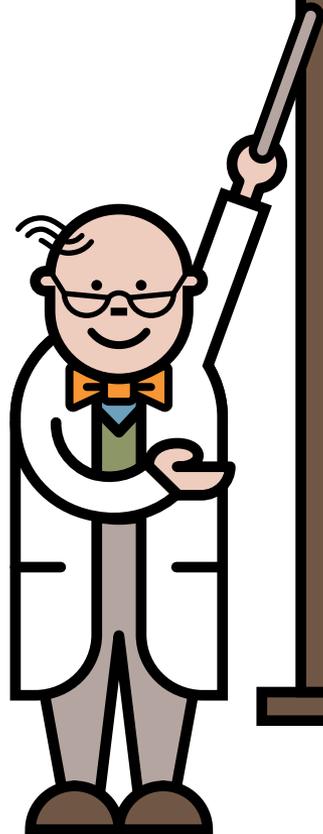


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# Inheritance



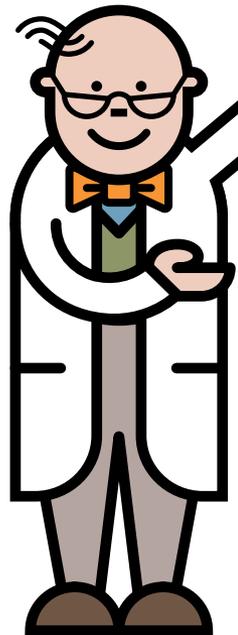
HT1 is an inherited condition. There is nothing that could have been done to prevent your baby from having HT1

Everyone has a pair of genes that make the fumarylacetoacetate hydrolase enzyme. In children with HT1, neither of these genes works correctly. These children inherit one non-working HT1 gene from each parent

Parents of children with HT1 are carriers of the condition

Carriers do not have HT1 because the other gene of this pair is working correctly

# Inheritance



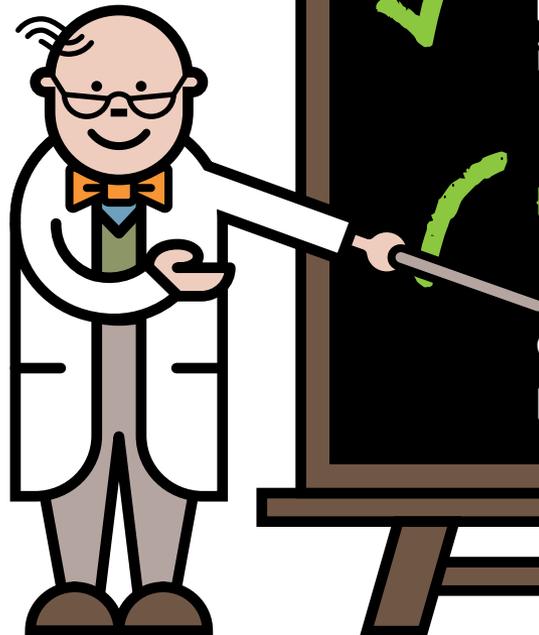
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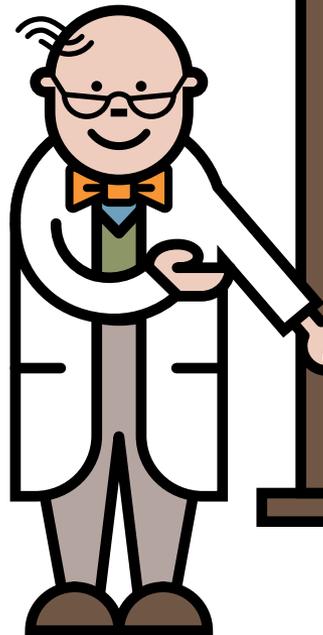
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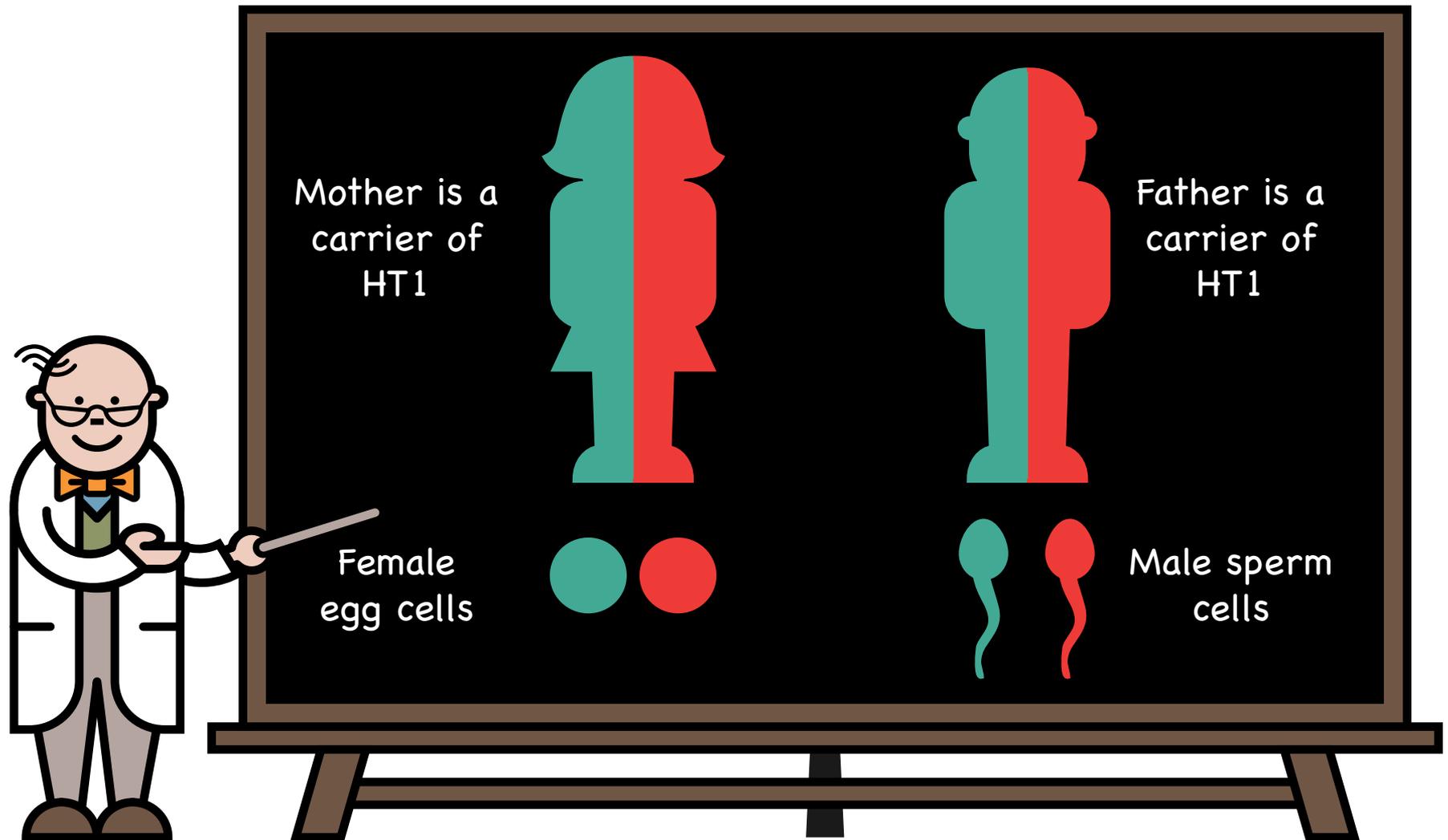
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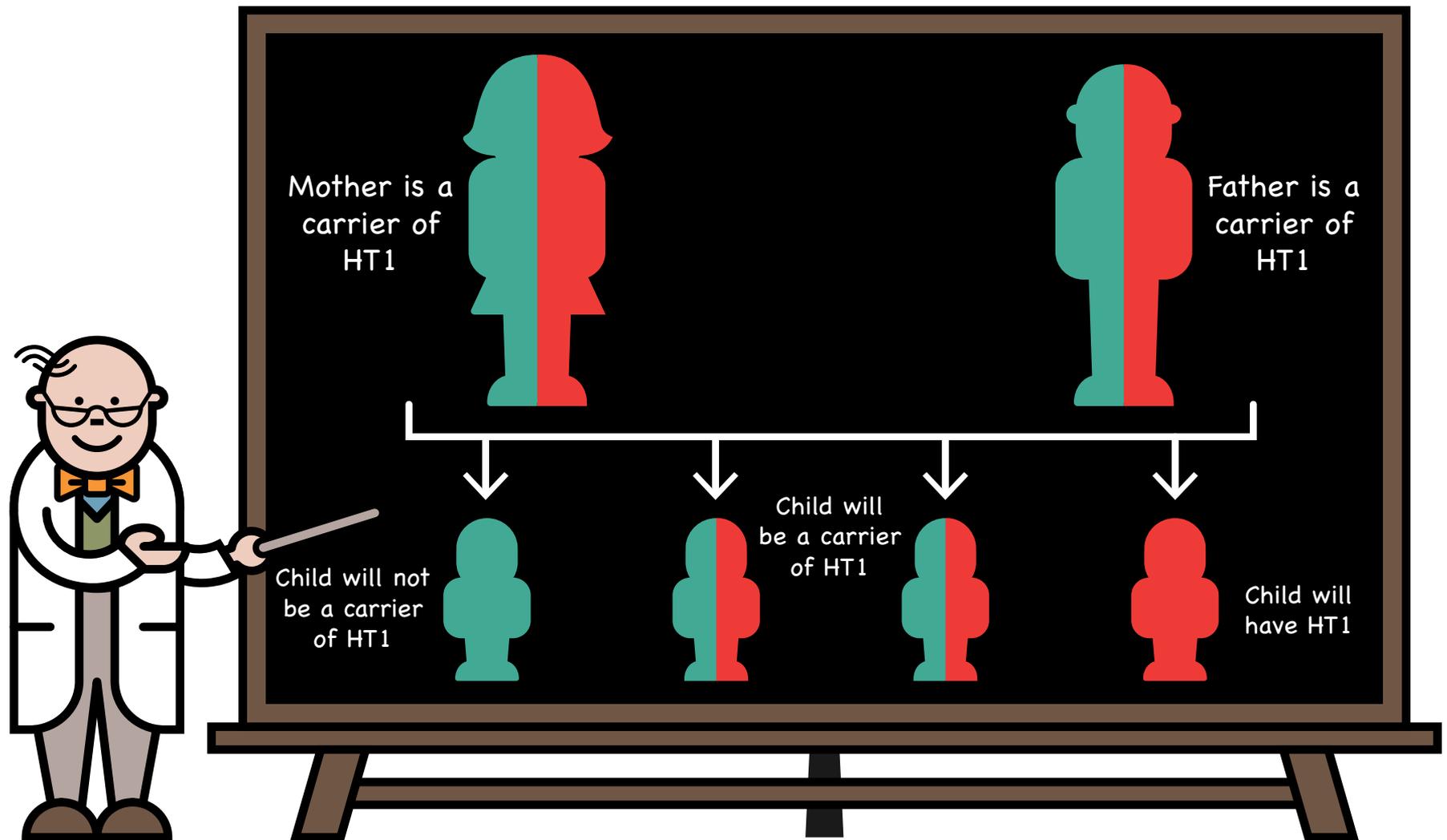
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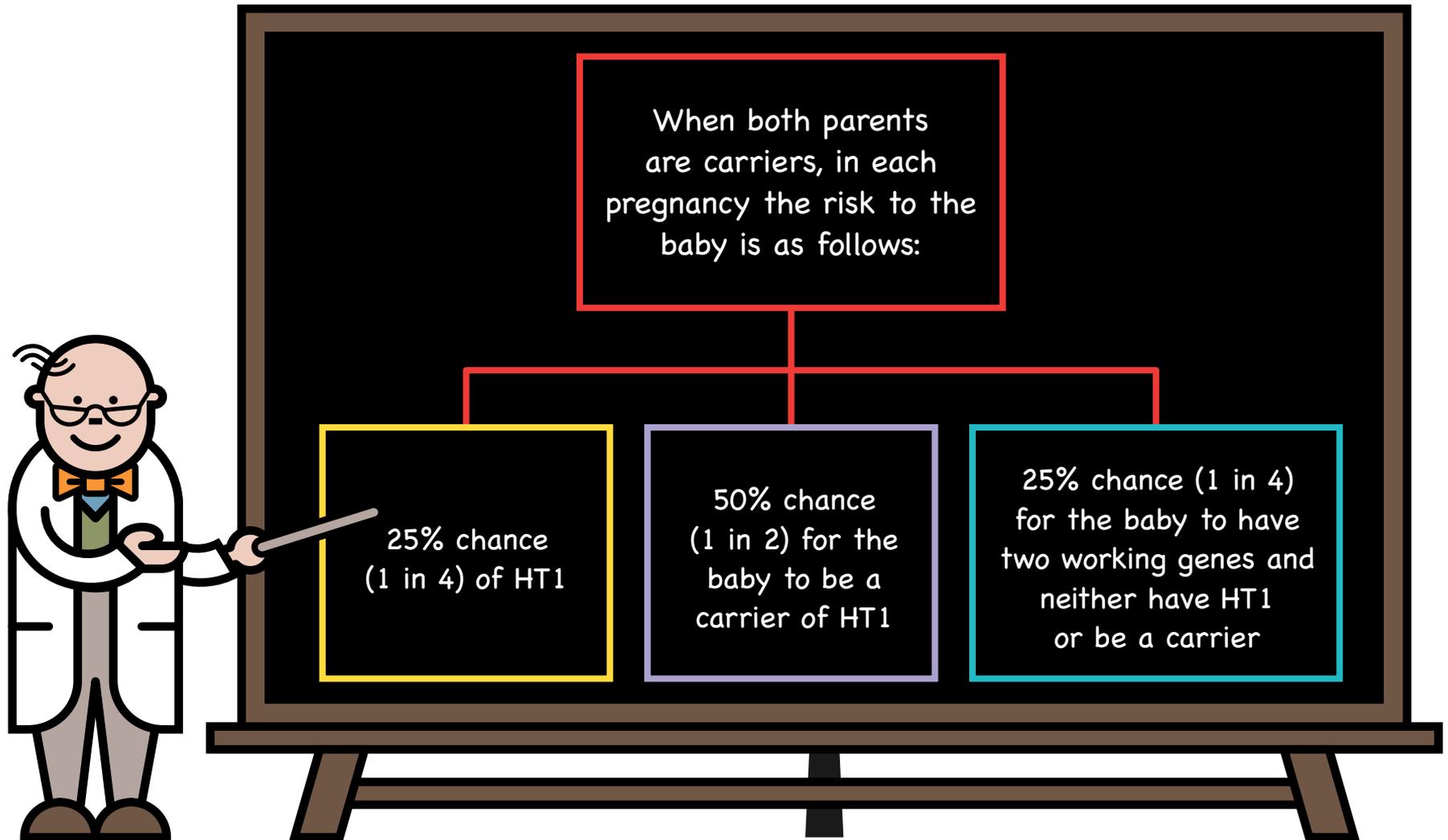
# Inheritance – Autosomal recessive (carriers of HT1)



# Inheritance – Autosomal recessive – possible combinations



# Future pregnancies



# Take home messages



HT1 is a serious inherited metabolic disorder that can lead to severe liver problems

Damage can be prevented with nitisinone, a diet low in tyrosine and a protein substitute

It is important that blood levels of tyrosine and phenylalanine are regularly checked

Remember, during illness, it is imperative that nitisinone is still given

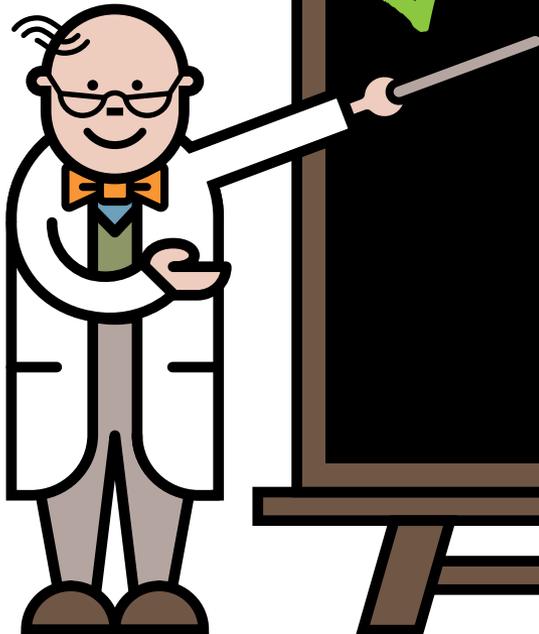
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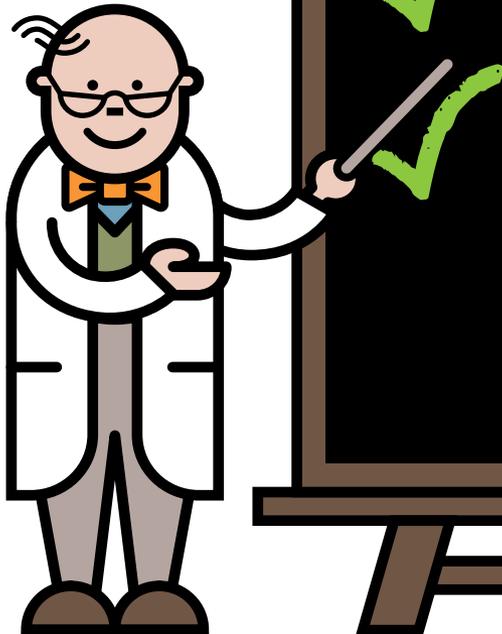
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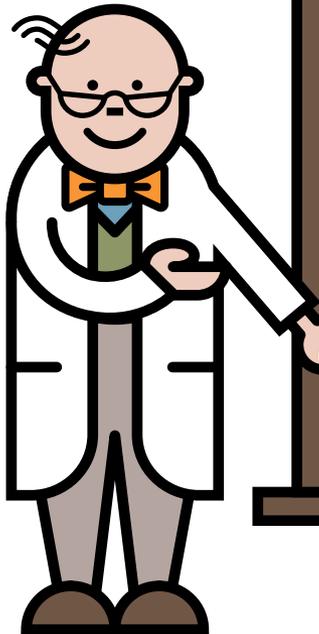
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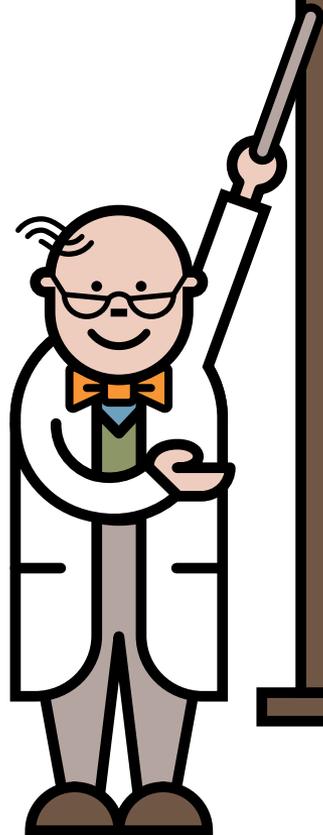


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# Helpful hints



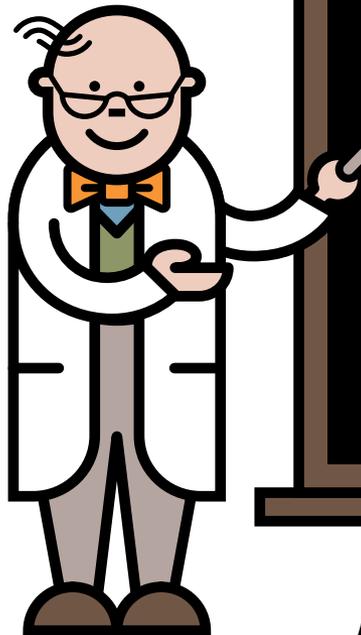
Always ensure you have a good supply of your dietary products and medicines and that they are in date

Your dietary products and medicines are prescribed. These are obtained via a pharmacy or home delivery

Always ensure you have sufficient blood testing equipment and send samples on a regular basis

Medications to control fever should be given as normally recommended – always keep supplies available

# Helpful hints



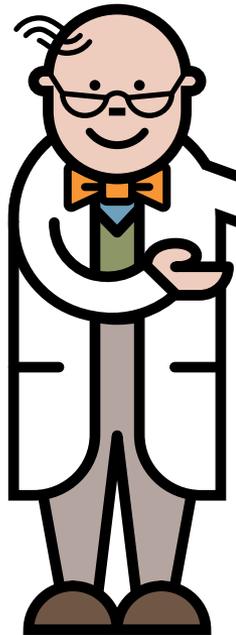
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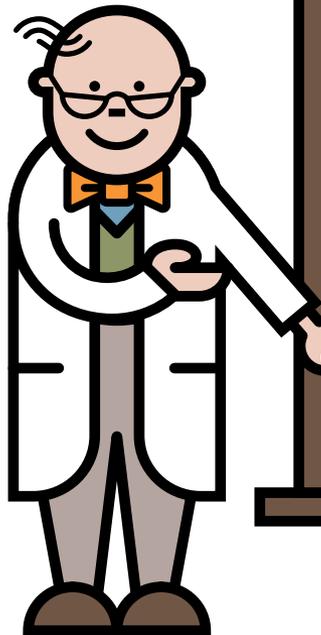
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# Who's who

- My dietitians
- My nurses
- My doctors
  - Contact details, address, photos

Visit [www.lowproteinconnect.com](http://www.lowproteinconnect.com) and register to get access to support and practical advice for those living on a low protein diet.

The site also provides information on upcoming events and personal stories from others on a low protein diet.



[www.bimdg.org.uk](http://www.bimdg.org.uk)

[www.nutricia.co.uk](http://www.nutricia.co.uk)

[www.metabolicsupportuk.org](http://www.metabolicsupportuk.org)