

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 2, NOVEMBER 2020

Arginase deficiency

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).

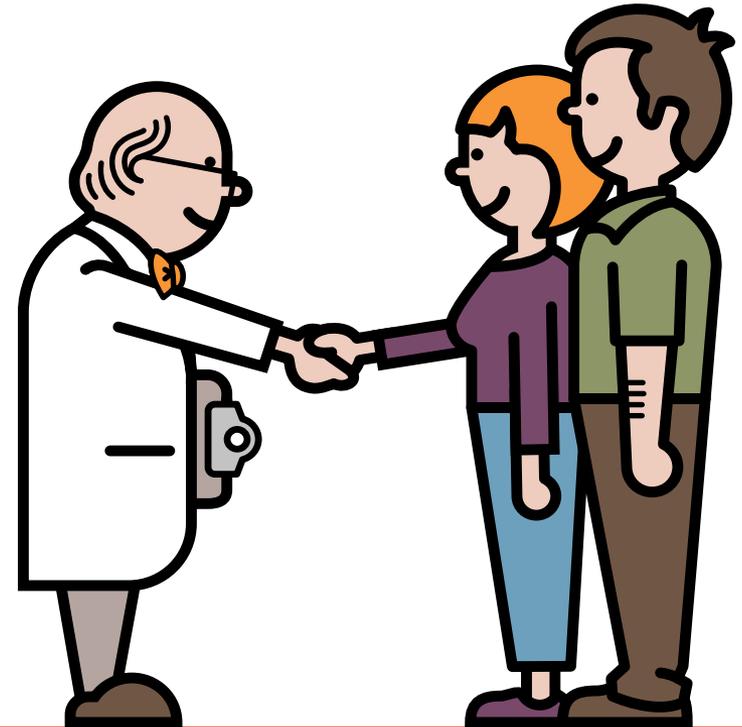
The team are Rachel Skeath, Karen van Wyk, Pat Portnoi and Anita MacDonald. The group is facilitated by Heidi Chan from Nutricia.

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.

Arginase deficiency

Information for families following a new diagnosis



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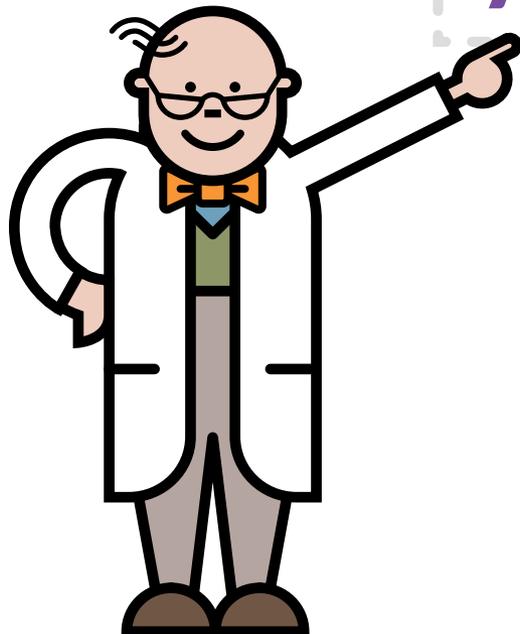


Tools Enabling Metabolic Parents LEarning

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What is Arginase deficiency?

It is an inherited metabolic condition.



Argi nase deficiency

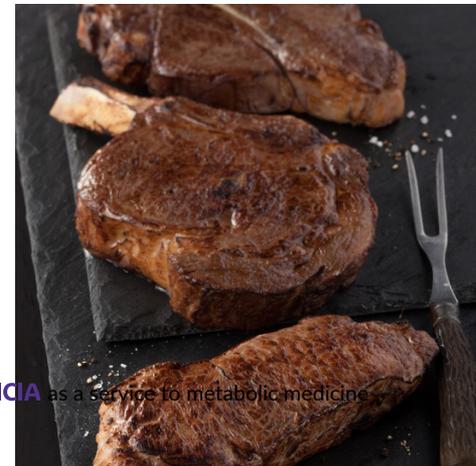
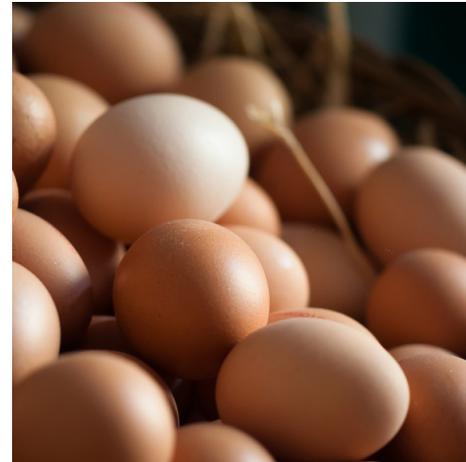
Arginase deficiency

What is protein?

Many foods contain protein.

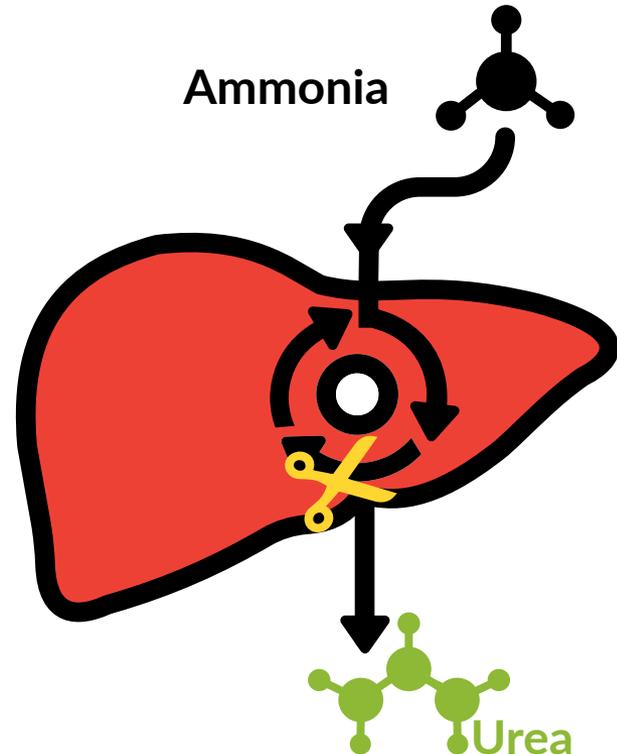
The body needs protein for growth and repair.

Many people eat more protein than the body needs.



How do we remove waste protein from the body?

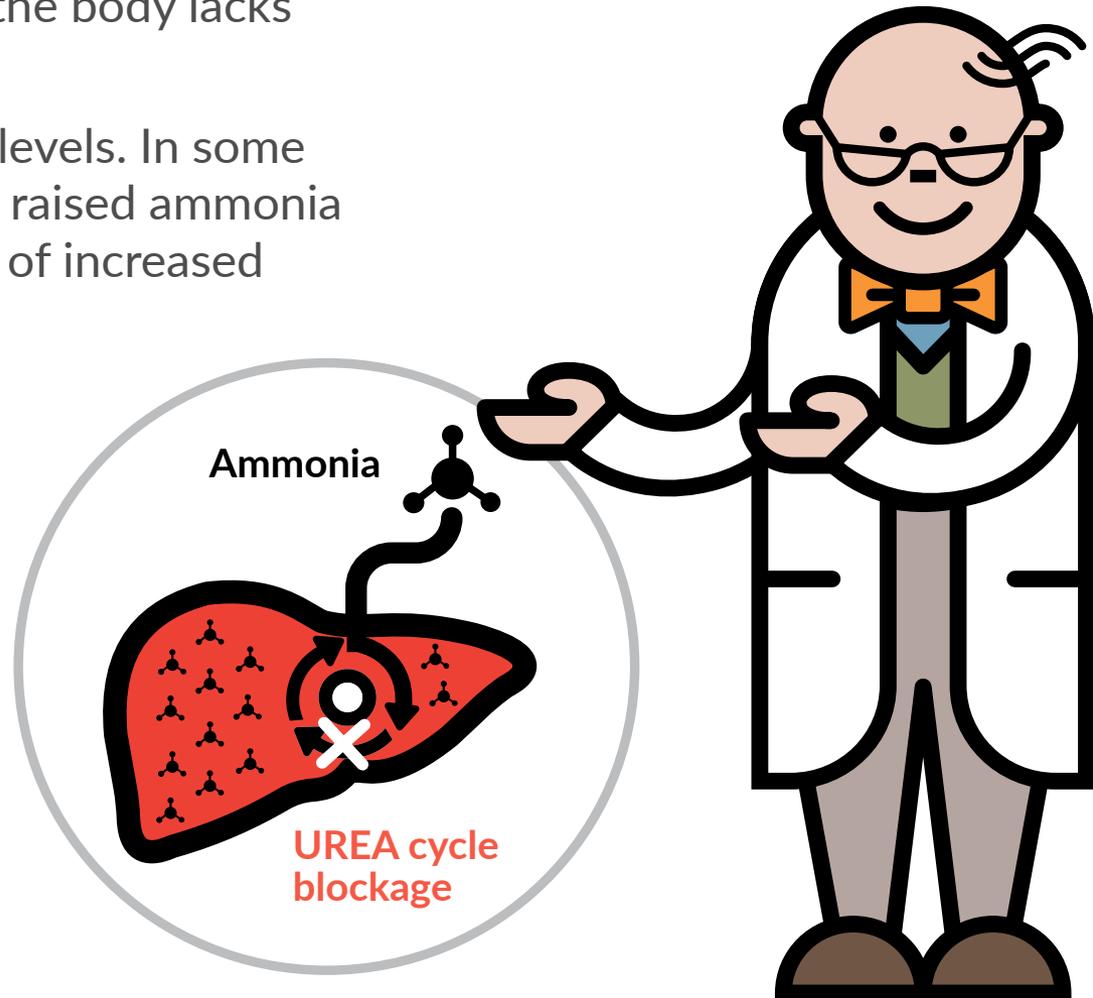
- First, the body converts waste protein to a toxic chemical called **ammonia**
- Ammonia is then converted into a non-toxic chemical (**urea**) in the liver
- This process occurs via the **urea cycle**
- In the urea cycle, several steps have to take place. Each step needs an enzyme (like chemical scissors) for it to work
- Arginine is formed in the urea cycle and then broken down to release urea. The enzyme that **breaks** down arginine is called Arginase 1.



What happens in Arginase deficiency?

In the Arginase deficiency, the body lacks the enzyme **arginase 1**.

This leads to high arginine levels. In some patients, it can also lead to raised ammonia levels, particularly at times of increased protein breakdown.



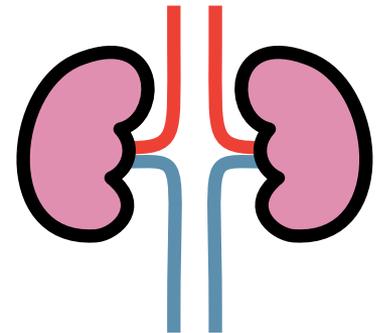
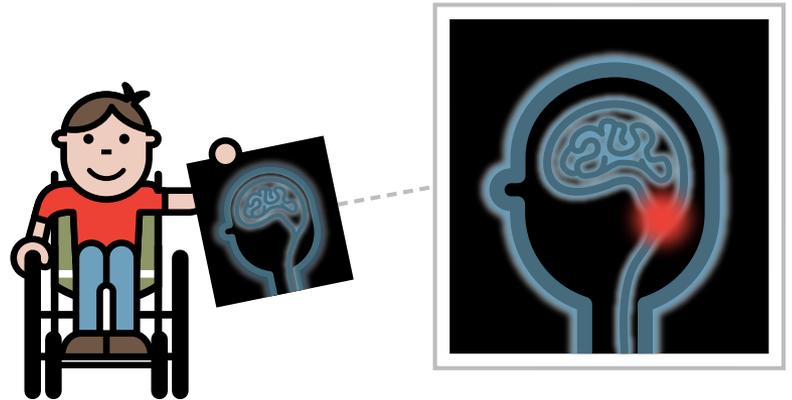
What are the signs of Arginase deficiency?

Arginine is one of the building blocks from which protein is made, so we need some arginine in our blood. It is harmful, however, if the levels are much too high.

Children with arginase deficiency may get:

- Stiff legs (spasticity)
- Learning difficulties
- Seizures
- Poor growth

Arginase deficiency sometimes leads to high ammonia levels, causing the patient to become drowsy or even comatose. High ammonia levels are most likely during illnesses.

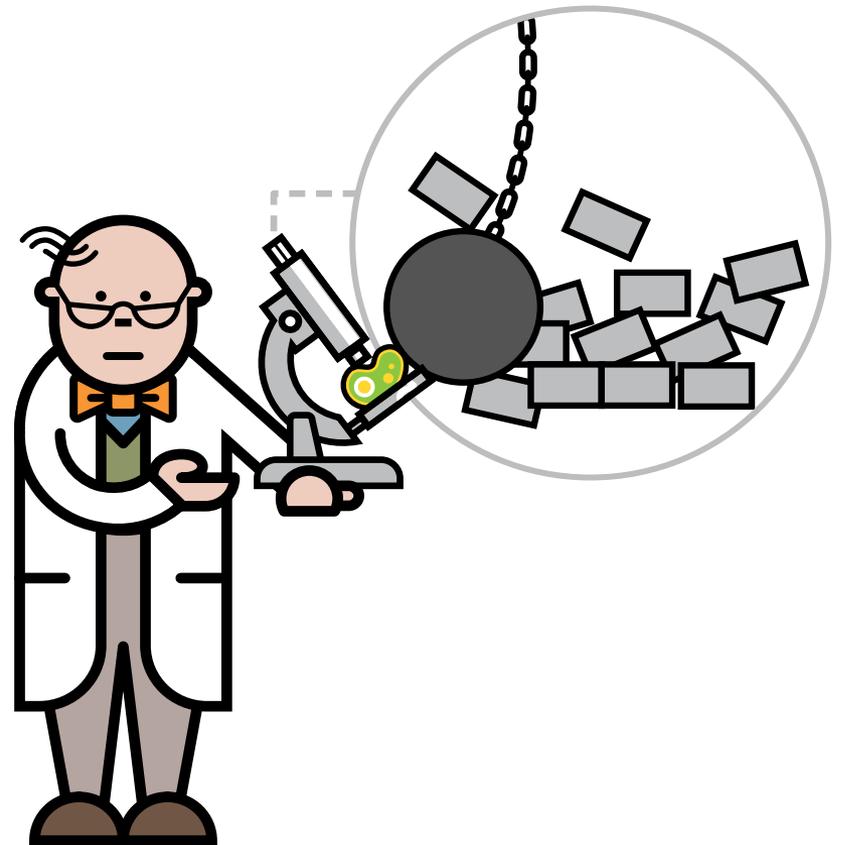


When does Arginase deficiency cause high ammonia levels?

Ammonia levels can rise when there is an increased break down of protein. This may happen if too much protein is eaten.

It commonly results from break down of the body's own protein. This is often triggered by infections, particularly if there is vomiting.

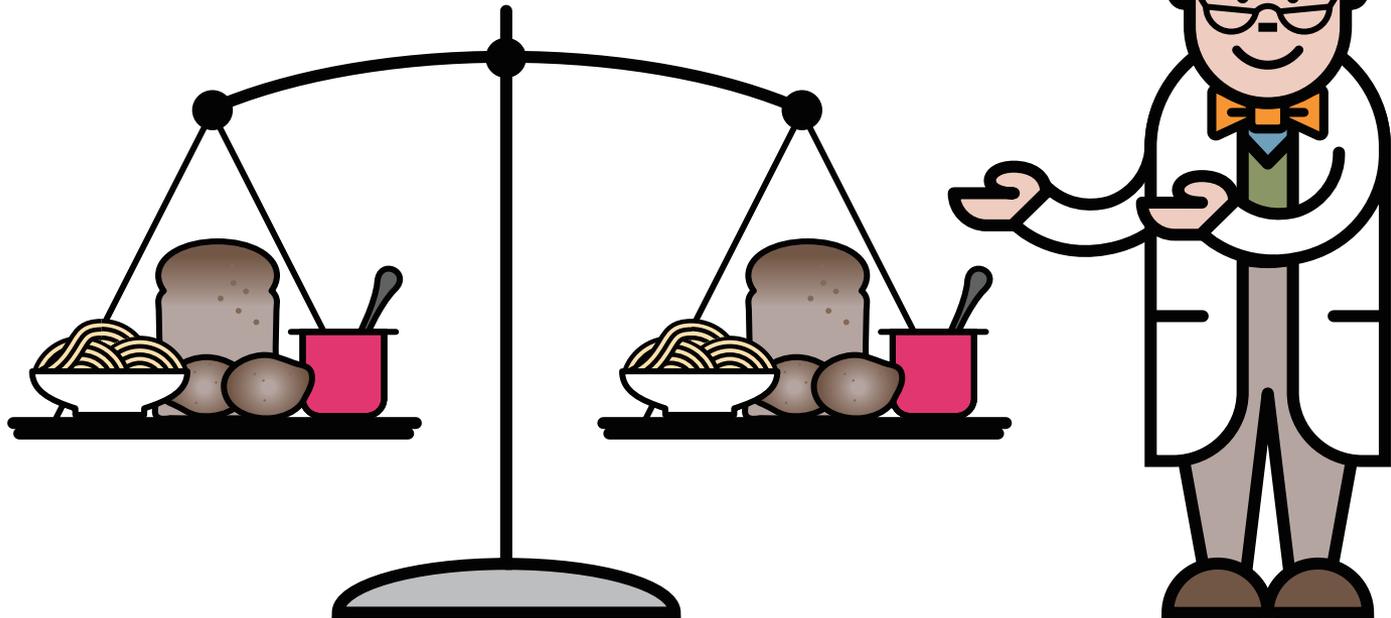
This causes **catabolism** which is a break down of body protein and can lead to a metabolic crisis.



Protein balance is needed in Arginase deficiency

In **Arginase deficiency** it is important that enough protein is given to grow

...but not too much as its breakdown will increase the arginine levels



How is Arginase deficiency diagnosed?

The diagnosis is suspected in a patient with high arginine levels in the blood.



The diagnosis is confirmed by finding the mutation in the ARG1 gene .



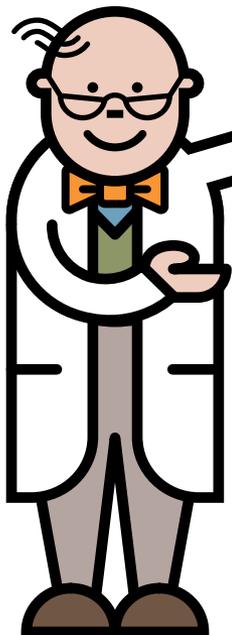
How is Arginase deficiency managed day to day?

Arginase deficiency is managed with the following:

A protein restricted diet

A special amino acid supplement

Sufficient energy supply from food and feeds



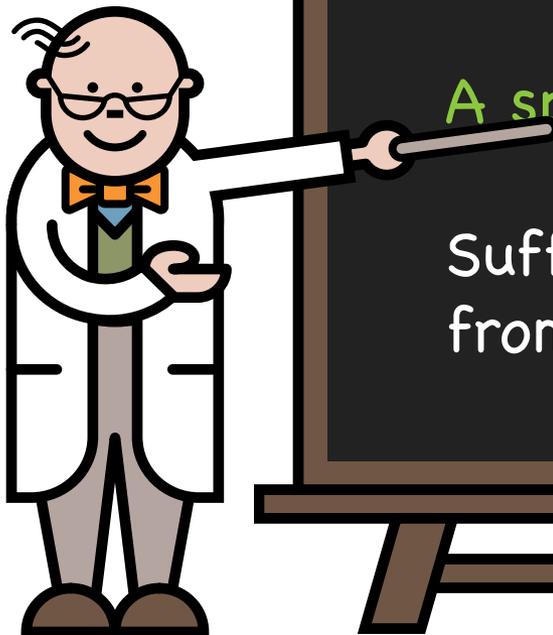
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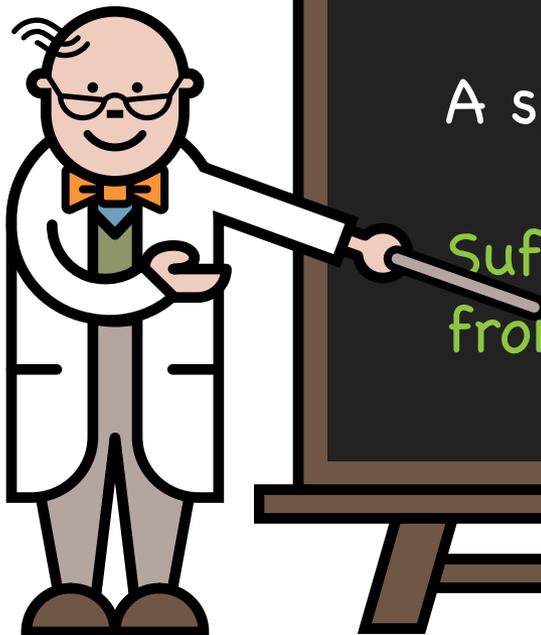
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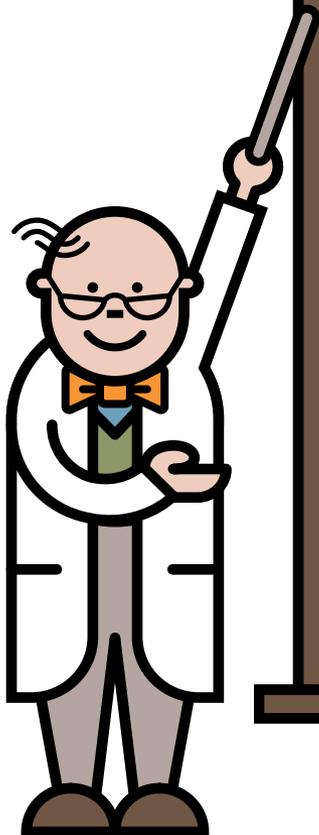
Regular feeding



Vitamin and mineral supplements



Other medications to control the level of ammonia in the blood



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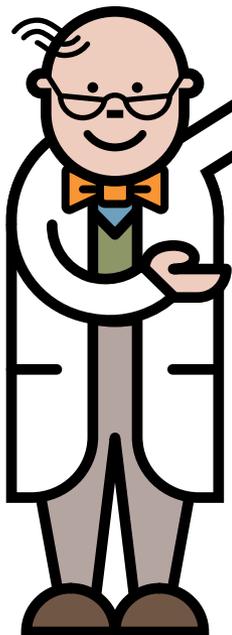
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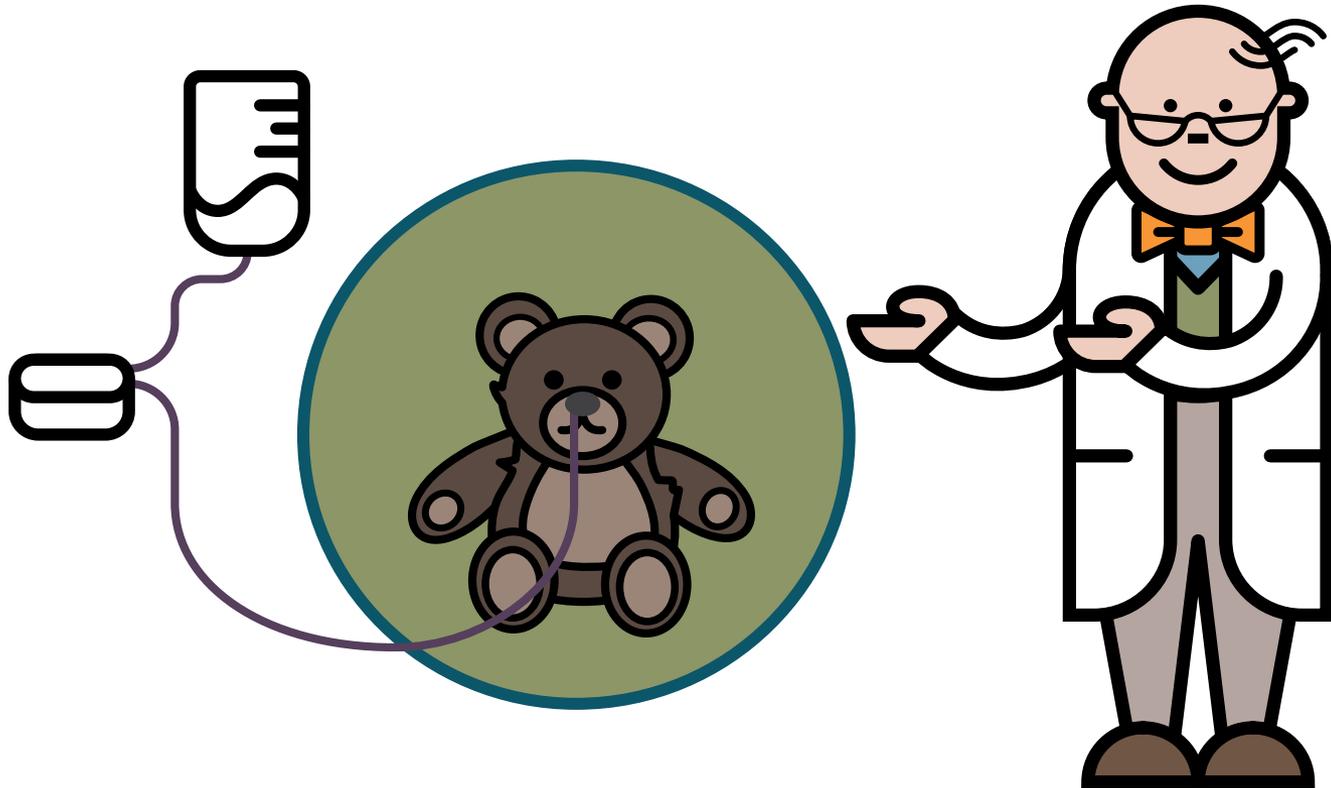


Other medications to control the level of ammonia in the blood



Is tube feeding needed?

Tube feeding may be necessary to give regular feeds. This will ensure energy, nutrient and fluid needs are met.



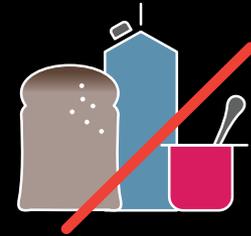
How is Arginase deficiency managed during illness?

- During any childhood illness, an emergency regimen is given
- This is to avoid a lack of energy supply and build-up of ammonia



How is Arginase deficiency managed during illness?

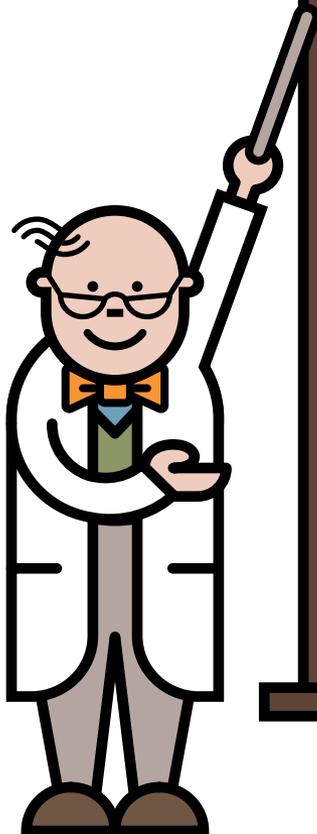
Stop all protein in food & drink



Start the emergency regimen.
This is made up of glucose polymer

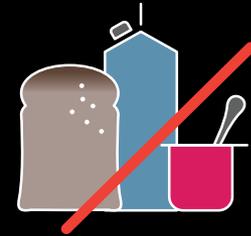


Continue medication as prescribed



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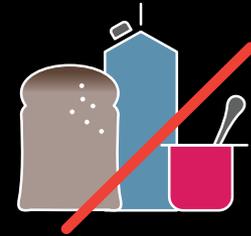


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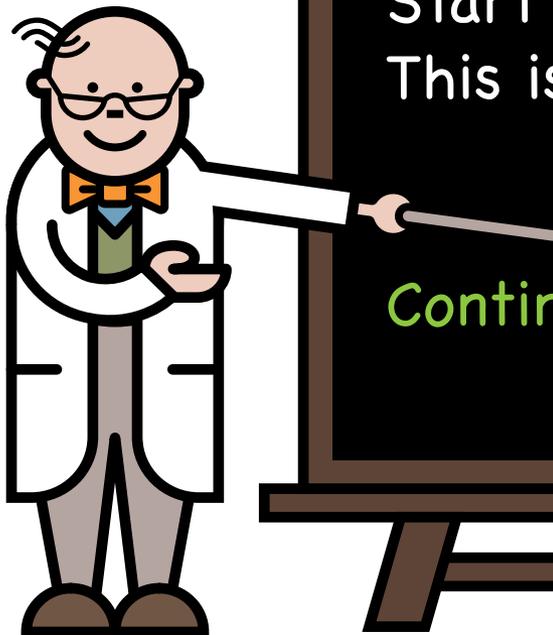
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Continue medication as prescribed



Checklist for illness

Always take full amounts of
emergency feeds as prescribed



If symptoms continue and/or you are
worried, go immediately to the hospital



Regularly update your metabolic team



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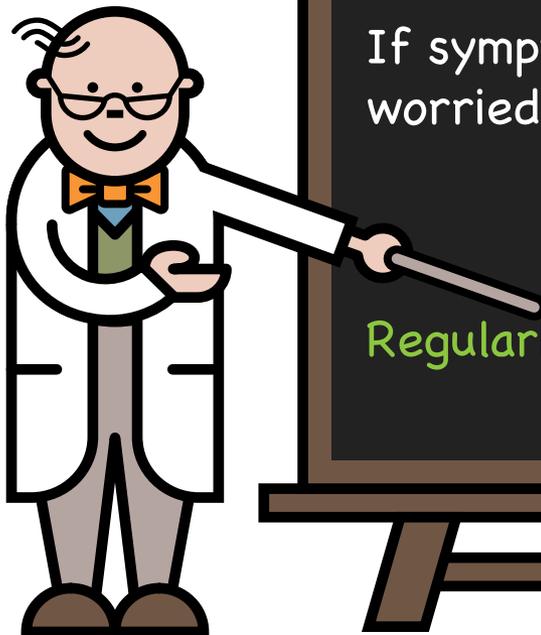
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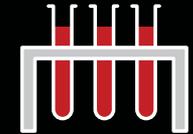
Key message

It is imperative that emergency feeds are started **promptly** and there are **no delays** in management.



How is Argininosuccinic aciduria monitored?

Frequent blood tests to check amino acids, nutrient and chemical levels



Height and weight



Developmental checks

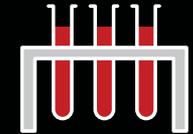


Diet and medications are adjusted according to age, weight and blood chemical levels



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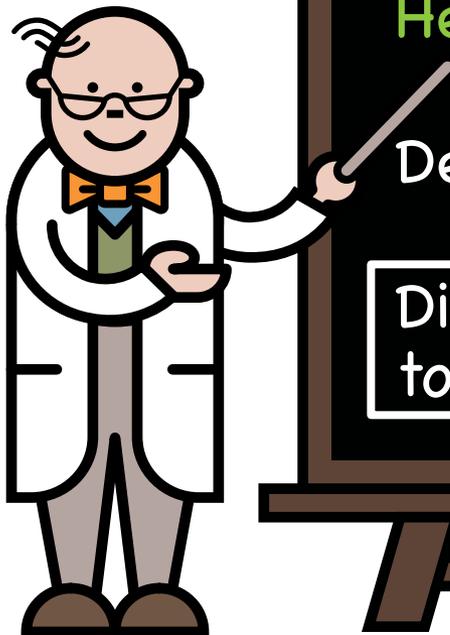
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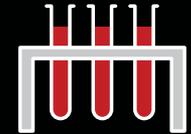


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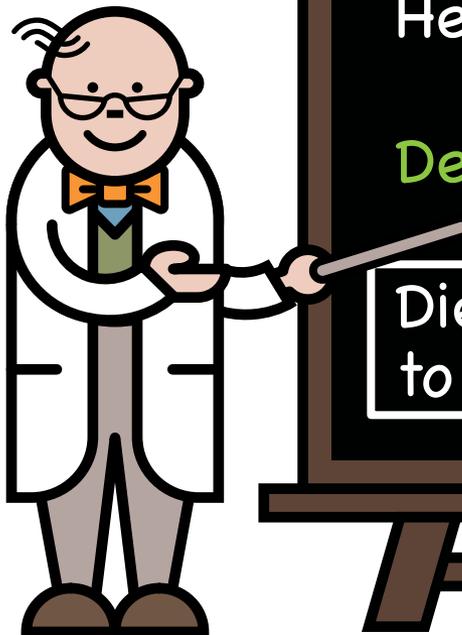
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Chromosomes, genes, mutations



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

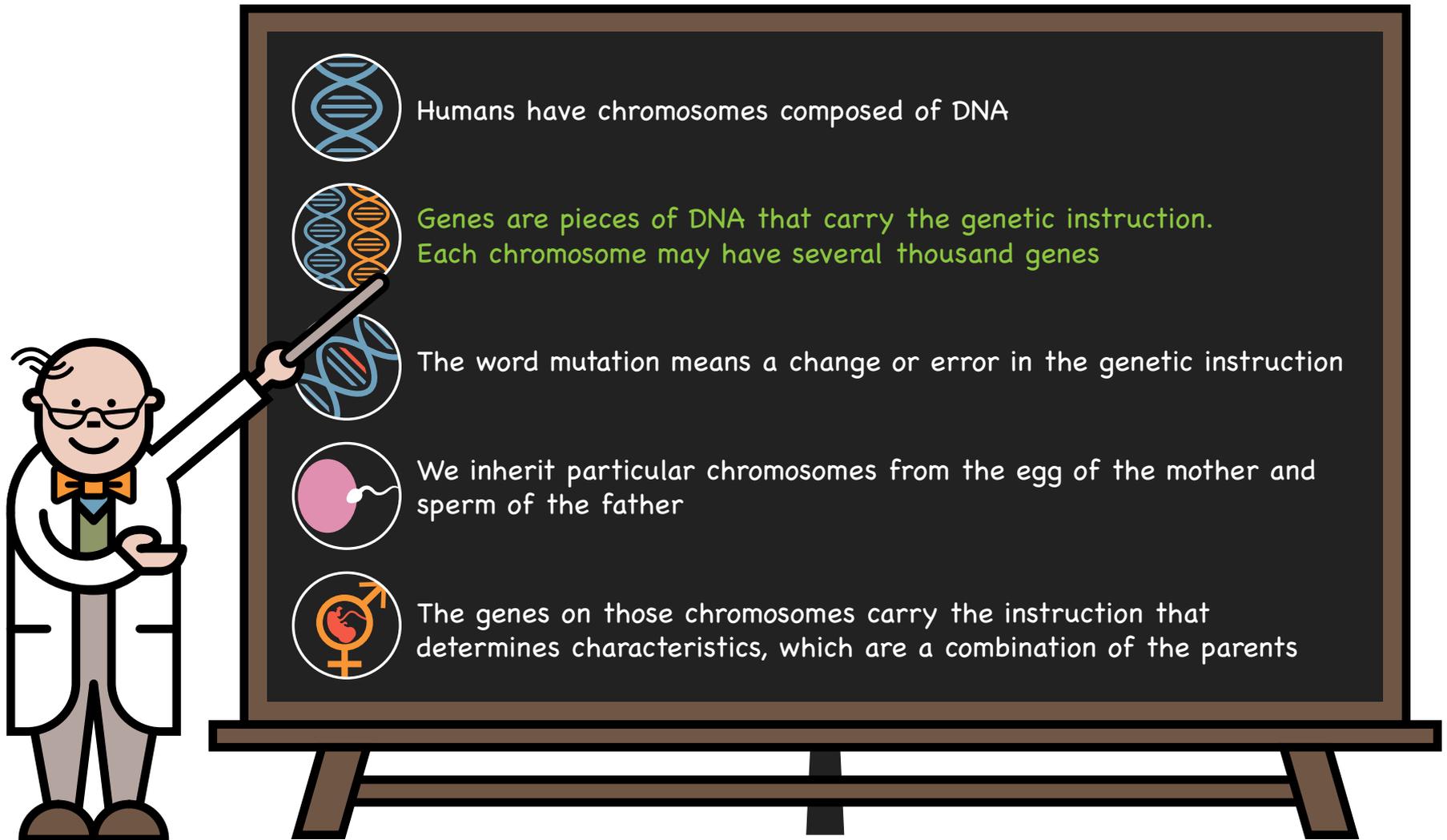


We inherit particular chromosomes from the egg of the mother and sperm of the father



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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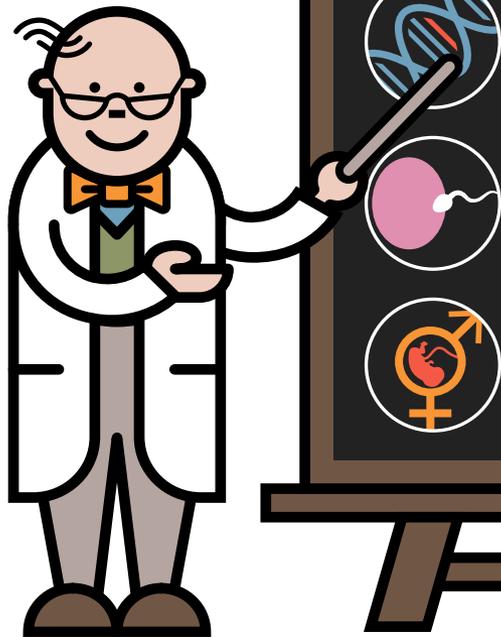
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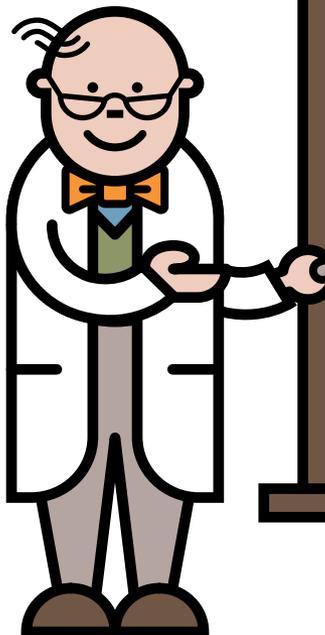
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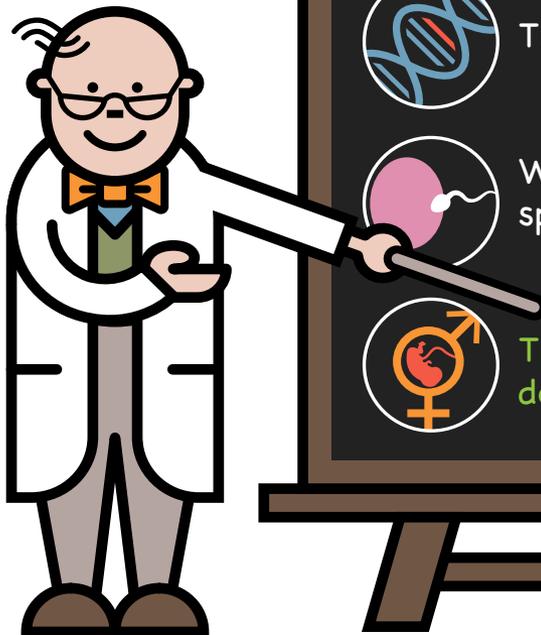
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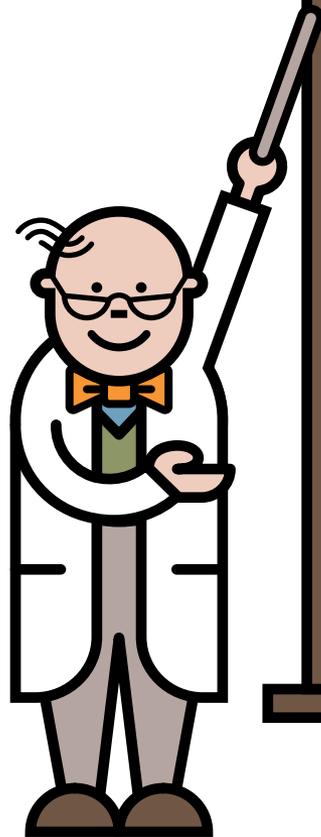
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Inheritance



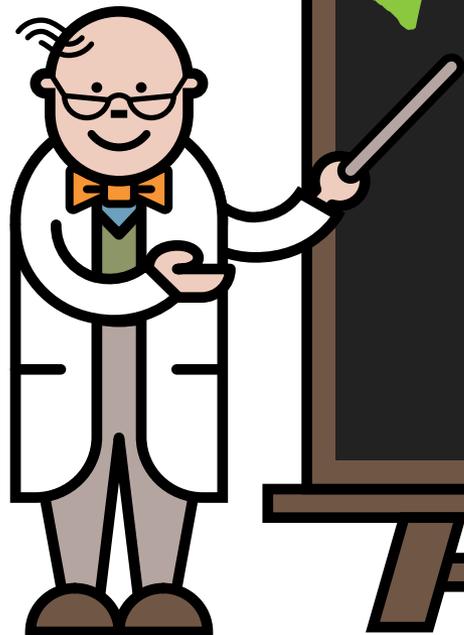
Argininosuccinic aciduria is an inherited condition. There is nothing that could have been done to prevent your baby from having Arginase deficiency

Everyone has a pair of genes that make the arginase 1 enzyme. In children with Arginase deficiency, neither of these genes work correctly. These children inherit one non-working Arginase gene from each parent

Parents of children with arginase deficiency are carriers of the condition

Carriers do not have Arginase 1 because the other gene of this pair is working correctly

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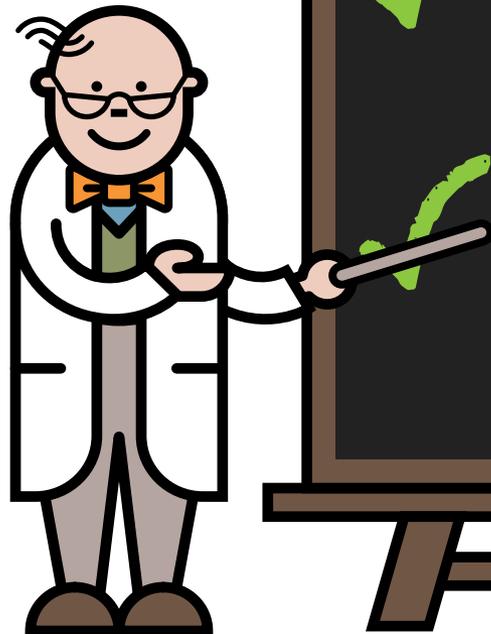


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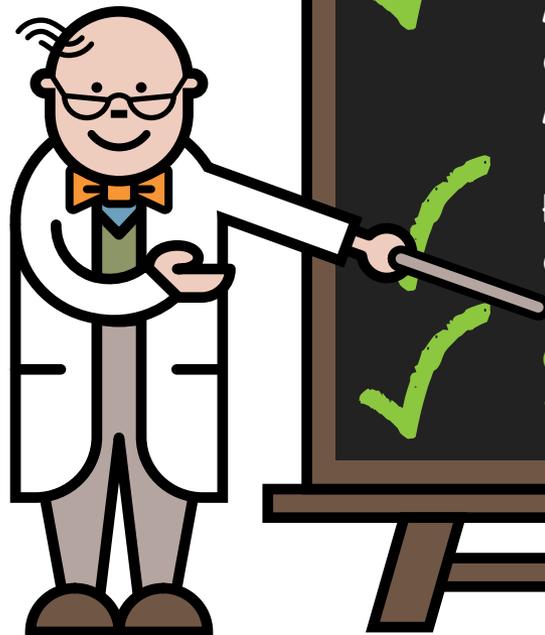
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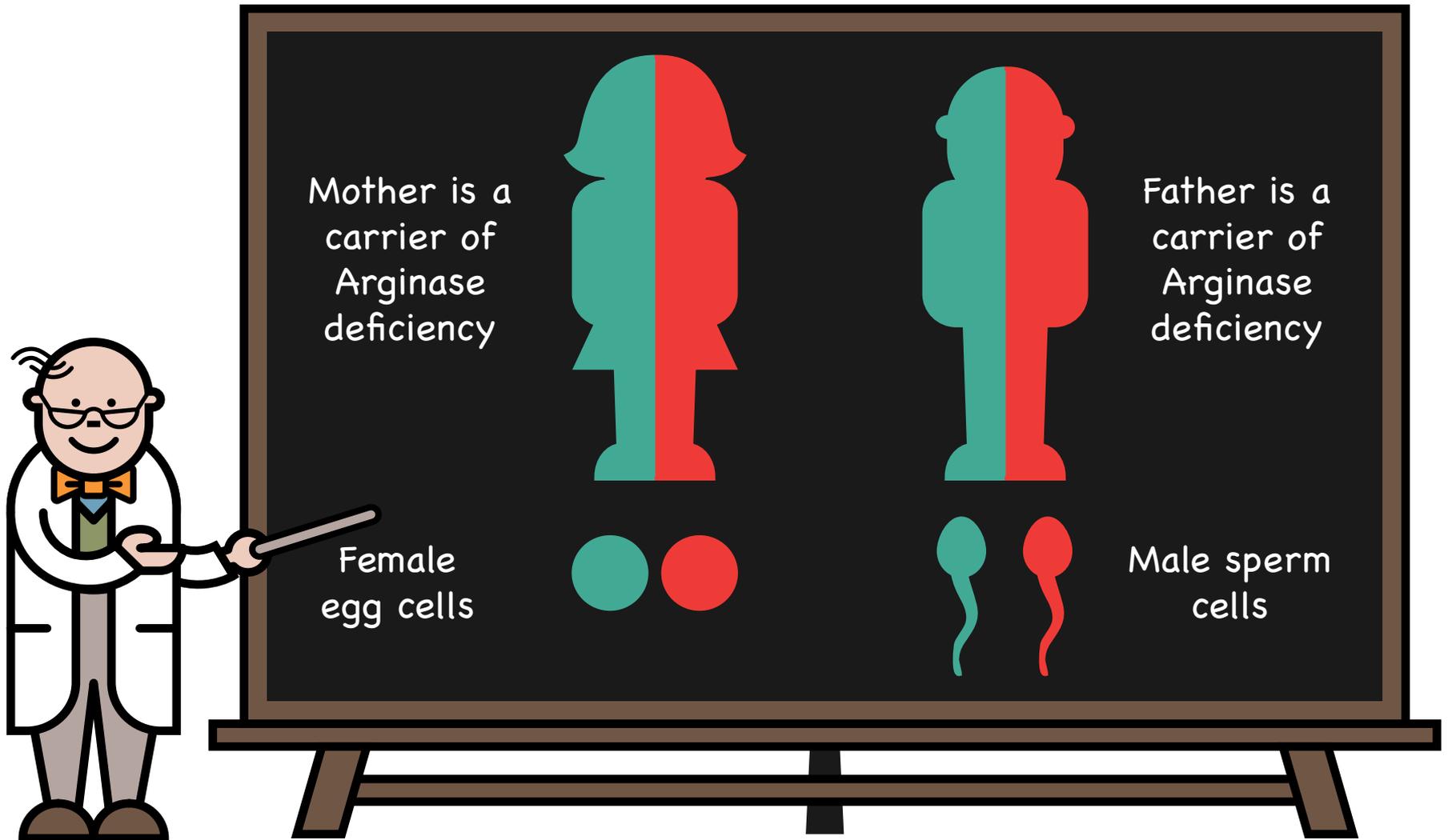
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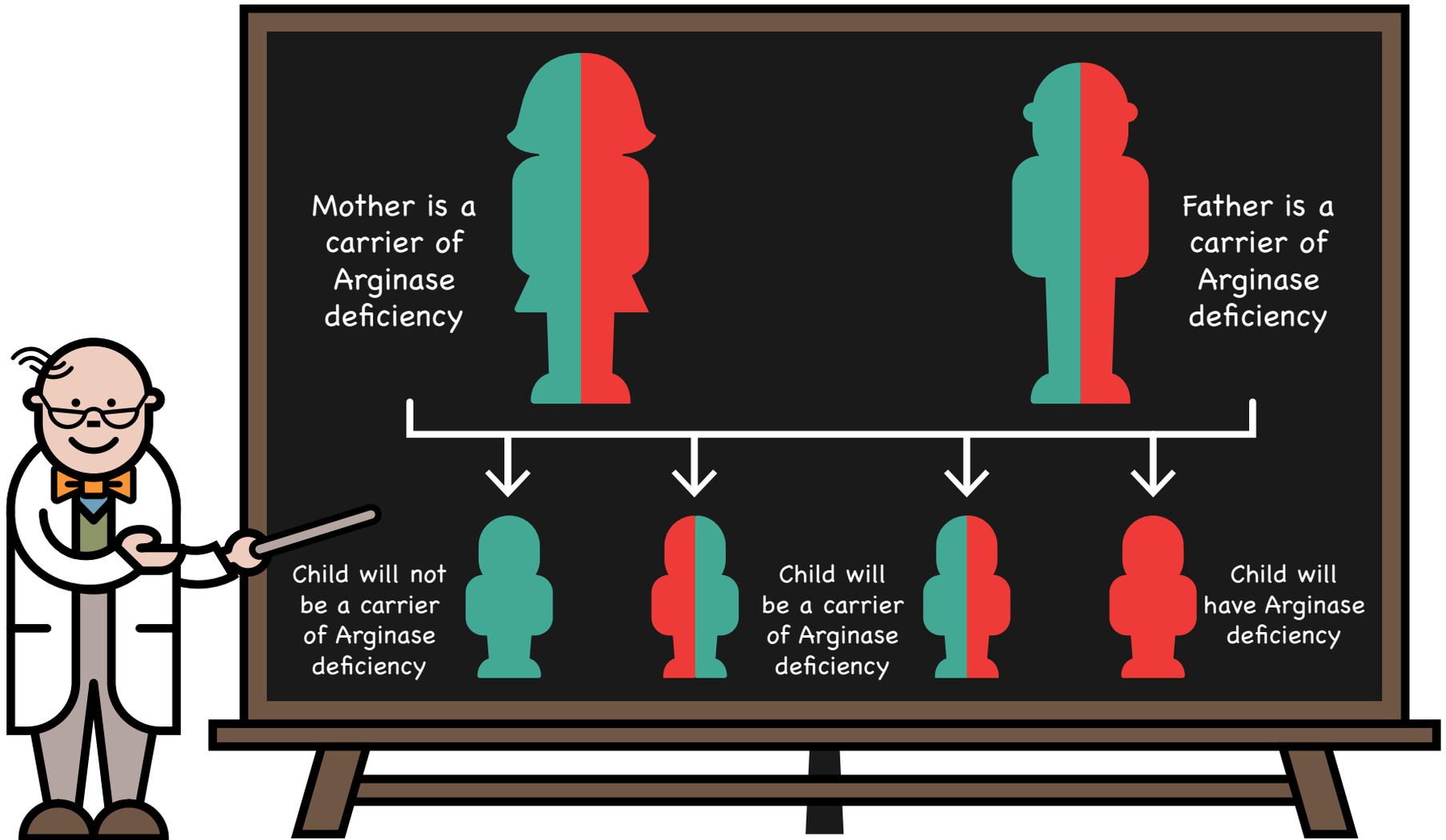
Inheritance

Autosomal recessive (carriers of Arginase deficiency)

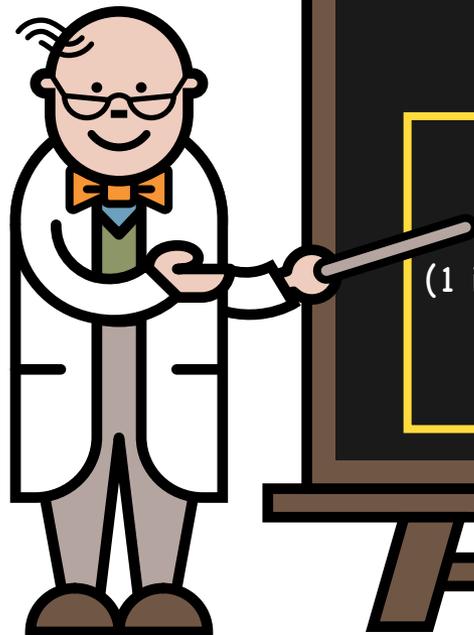


Inheritance

Autosomal recessive – possible combinations



Future pregnancies



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

25% chance (1 in 4) of Arginase deficiency

50% chance (1 in 2) for the baby to be a carrier of Arginase deficiency

25% chance (1 in 4) for the baby to have two working genes and neither have Arginase deficiency or be a carrier

Take home messages



✓ Arginase deficiency is a serious inherited metabolic disorder that can lead to severe problems

The condition is managed with a protein restricted diet, regular feeding and medications

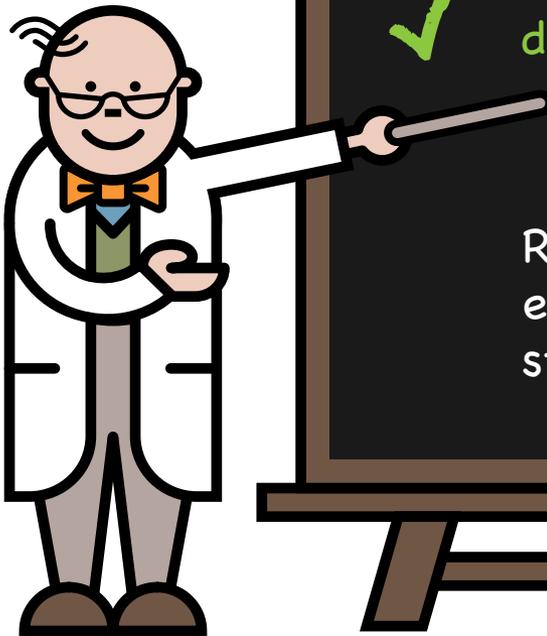
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Take home messages

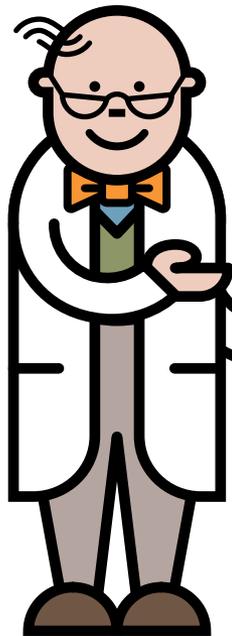
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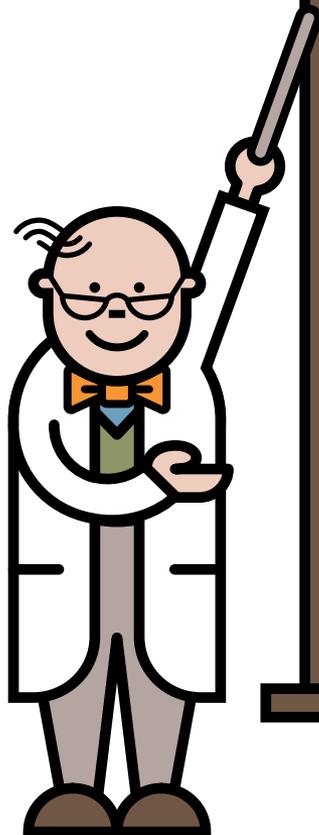


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Helpful hints



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

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

Always ensure you have your emergency feed products and a written emergency plan

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

Helpful hints



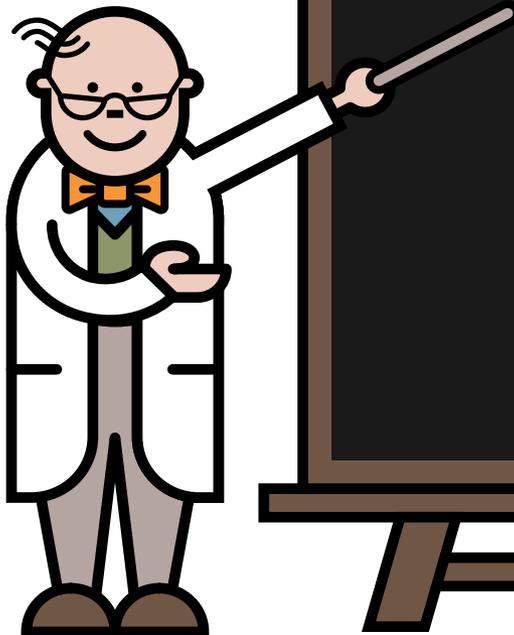
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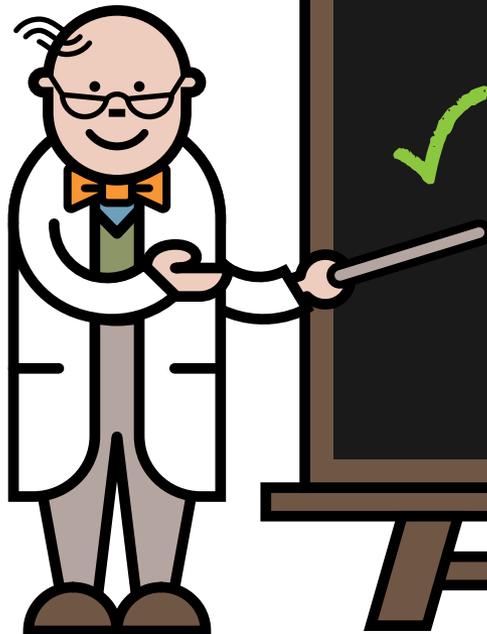
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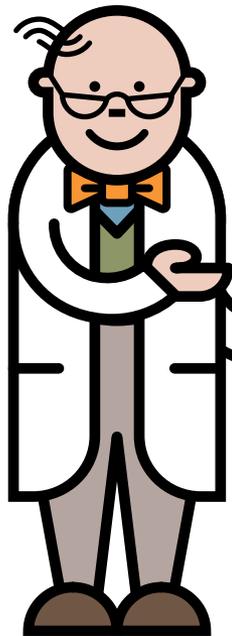
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Who's who

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

Visit www.lowproteinconnect.com 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.



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British Inherited Metabolic Diseases Group



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**METABOLIC
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