

# Anaemia



- The symptoms of anaemia are tiredness, shortness of breath and being pale.
- The anaemia in CDA is very variable.
  - In some patients, it is very **mild** and does not cause them significant symptoms.
  - In other patients, it is **moderate** and patients may require blood transfusions if they are unwell with an infection.
  - In the remainder of patients, it is **severe** and the patients are dependent on transfusions regularly for their whole life.

# Glossary

Here is an explanation of some key terms:

**Inherited:** this means a condition that is passed on from parents to children. It is coded for by genes. Babies are born with it, but may not appear ill until later in life.

**Gene:** a piece of DNA (the building blocks of our genetic material) that specifically codes for a single protein.

**Chromosome:** String of DNA on which genes are organised next to each other. Everyone has 2 copies of each chromosome- one from their father and one from their mother. Therefore there are 2 copies of every gene (except sex chromosomes)

**Dominant:** A characteristic in a gene that gets expressed regardless of what is in the other gene. For example someone carrying one gene coding for brown eyes and one gene coding for blue eyes will have brown eyes because brown is dominant.

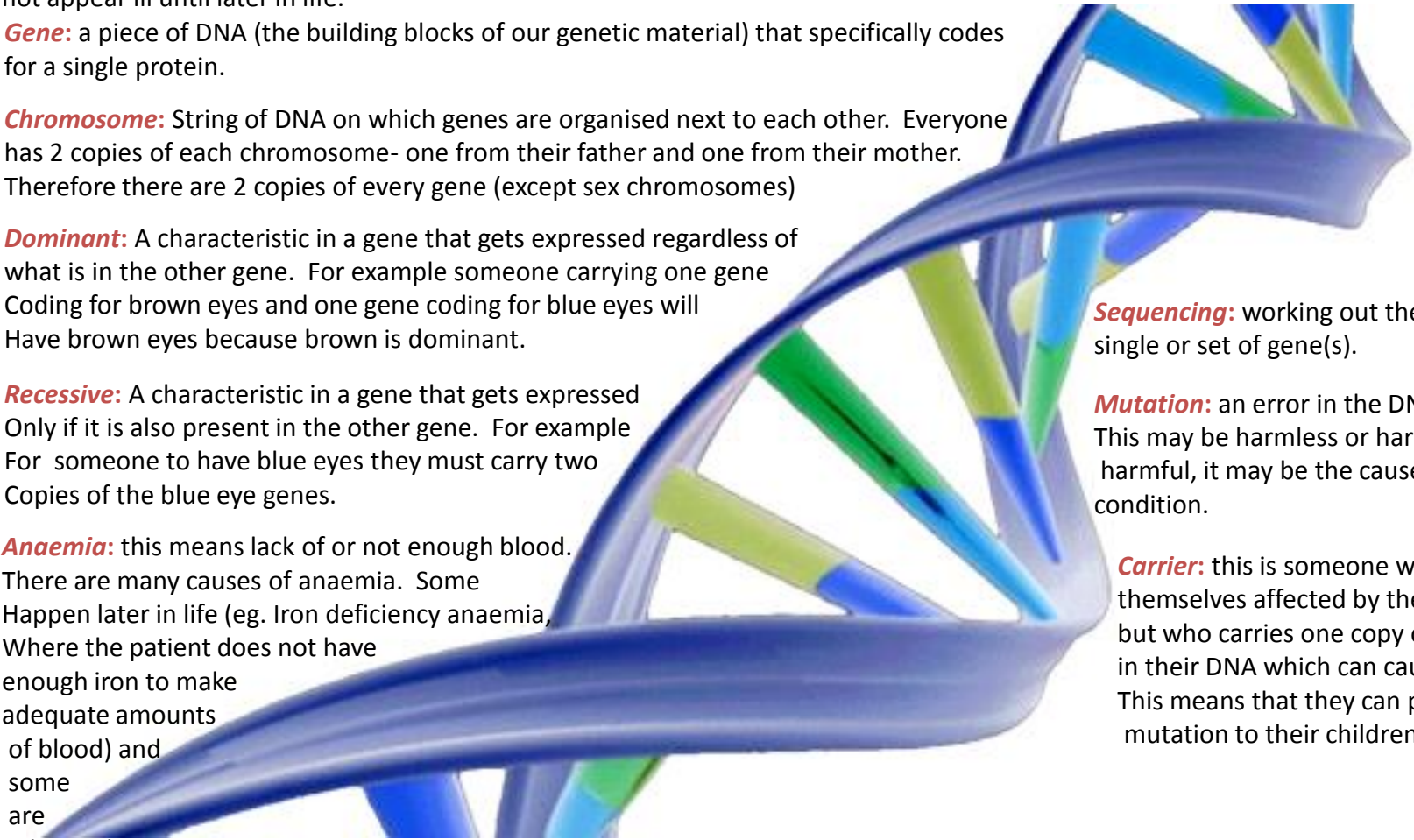
**Recessive:** A characteristic in a gene that gets expressed only if it is also present in the other gene. For example for someone to have blue eyes they must carry two copies of the blue eye genes.

**Anaemia:** this means lack of or not enough blood. There are many causes of anaemia. Some happen later in life (eg. Iron deficiency anaemia, where the patient does not have enough iron to make adequate amounts of blood) and some are inherited.

**Sequencing:** working out the exact code of a single or set of gene(s).

**Mutation:** an error in the DNA code. This may be harmless or harmful. If harmful, it may be the cause of an inherited condition.

**Carrier:** this is someone who is not themselves affected by the condition but who carries one copy of the mutation in their DNA which can cause the condition. This means that they can pass on this mutation to their children.



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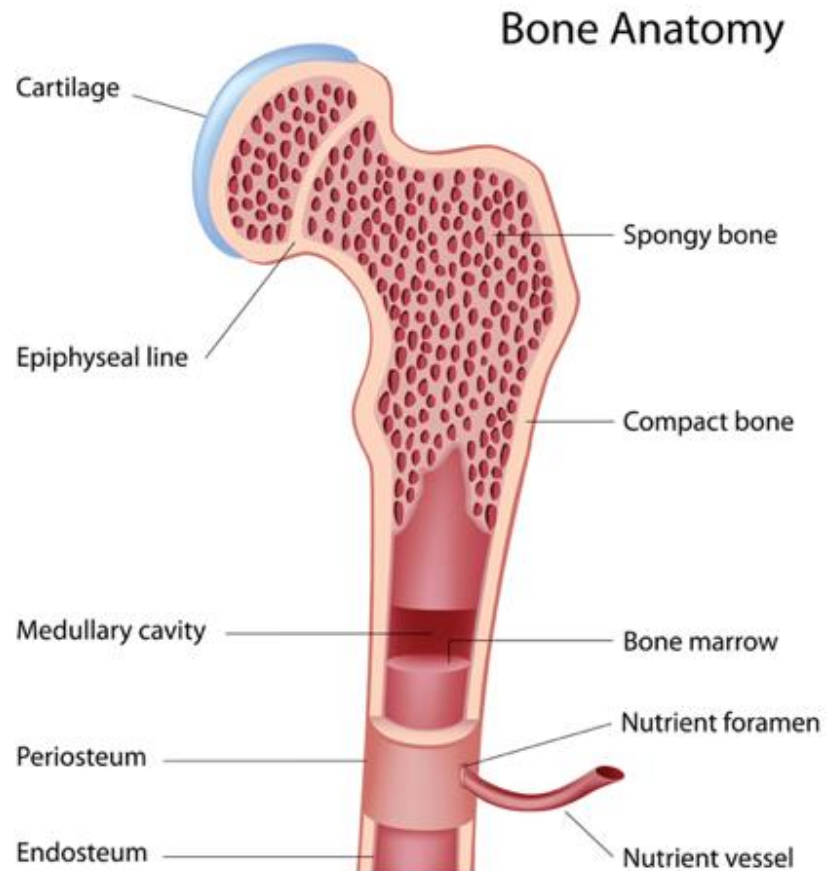
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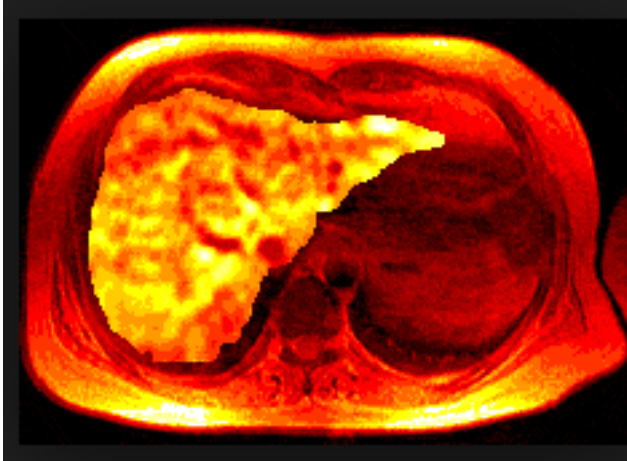
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# The bone marrow

The bone marrow is inside the bones of the body and is the site where blood cells are made. There are 3 main types of blood cells- red blood cells, whose function is to carry oxygen around the body, white blood cells, which are specialised at fighting infections, and platelets, which stop bleeding. Red blood cells are made by a process called erythropoiesis. This means that they start off as immature cells and over the process of a few days become more and more mature and specialised until they are ready, at which point they leave the bone marrow and enter the bloodstream. Once in the bloodstream, they carry out their function for about 3 months at which point they die a natural death.



# Iron overload



A buildup of iron is one of the complications of Inherited anaemias.

This can result from blood transfusions, or can simply be a feature of anaemia without transfusions.

In those cases, patients absorb more iron than they should from their diet. Too much iron can be very harmful to the body as humans don't have any way of getting rid of excess iron. This means iron will accumulate in the liver, the heart, the pancreas and other organs. To prevent the serious complications that would follow, patients with iron overload are treated with medications called iron chelators. These bind the excess iron and get rid of it.

# Congenital dyserythropoietic anaemia (CDA)

- There are 3 main types of CDA. CDA-1 and CDA-2 are recessive and CDA-3 is dominant
- Some people have very mild anaemia, while others may be transfusion-dependent their whole life.
- The diagnosis is made from examination of the bone marrow cells.
- Some people with CDA-1 respond to a treatment called interferon. There is no specific treatment for the other types

# Other features

- Some patients with CDA have other features such as:
  - **Splenomegaly**: this means that the spleen is enlarged and is usually due to the body trying to compensate for the anaemia.
  - **Abnormalities of the hands and feet**: this has been reported in some CDA patients.

# Diamond Blackfan Anaemia

- This type of anaemia usually presents very early in life and when the bone marrow is examined, it is completely empty of red blood cell precursors (precursors are the cells which will turn into red blood cells)
- Some patients may respond to steroids, while others are on life-long transfusions
- Some patients are treated by bone marrow transplantation
- Often, patients with DBA have other inherited health problems (eg cleft palate) which need to be treated in their own right



# Sideroblastic anaemia

- This type of anaemia is named after the appearance of iron granules seen in red blood cell precursors in the bone marrow- they form a ring inside these cells
- Patients with sideroblastic anaemia may or may not be dependent on transfusions, but even if they are not, they almost always accumulate iron in the body
- This condition can be recessive or X-linked (this is where women carry the gene without being affected while men who inherit the gene from their mothers are always affected)

# Red cell enzyme and membrane disorders

- In these types of disorders, the bone marrow is making plenty of red blood cells, but they get destroyed prematurely in the circulation
- This type of anaemia is called haemolytic anaemia
- As a result, instead of surviving about 3 months in the circulation, red blood cells survive only a few weeks, so the bone marrow has to work extra hard to keep up
- Often the spleen is enlarged and sometimes, removing it surgically can help patients get fewer symptoms