What do you need to know if your child is diagnosed with CDA-1?

What is CDA-1?

CDA-1 is an inherited condition that affects the development of red blood cells. Due to an 'error' (also known as a mutation in scientific terms) in one of the genes involved in the production of red blood cells, the cells do not mature normally. They die in the bone marrow before they get into the circulation.

The condition is ultra-rare and it's estimated that one in a million people are affected. For a child to develop CDA-1, both parents must pass on this mutation in their genes. It is possible for someone to be a carrier of CDA-1, which happens if they have only inherited one copy of this mutated gene. A carrier does not present with the condition themselves, but they can pass it on to their offspring.

What are the symptoms of CDA-1?

The main symptom of CDA-1 is anaemia, which means there is not enough red blood cells circulating in the blood.

Therefore, people with CDA-1 can feel tired and weak, lacking energy. This may mean that your child might require more sleep in comparison to other children their age. It could also mean they get more tired during physical activities, for example in PE lessons at school, so it could be a good idea to make the teachers aware of your child's condition.

People with CDA-1 may look pale and can have jaundice – their skin and the whites of their eyes become a yellow colour. How serious this is varies from person to person.

10-20% of children with CDA-1 may have differences in the shapes of their fingers and toes. As they mature they may have poor appetite, low mood and not grow very tall. It's not clear if this is directly due to CDA-1 or due to treatment with blood transfusions.

How is CDA -1 diagnosed?

Blood tests can detect whether a person has anaemia. In the past, a bone marrow sample would be required to test if someone had CDA-1, however this can now be done via genetic tests (using blood samples). These tests are used to identify whether a patient has this inherited condition and which genes are involved. The diagnostic process is carried out in specialist centres, where it can take up to 3 months to get a result.

In some cases, CDA-1 can be detected before birth, but it is usually diagnosed in childhood or adolescence.

We are aware that a diagnosis of CDA-1 can come as a shock to patients and their families, remember to reach out for support if you need it.

What are the complications of CDA-1?

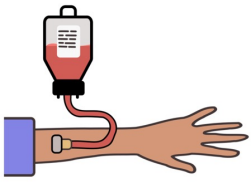
- Child development: Children with severe anaemia may not grow very tall and have difficulty concentrating. If the anaemia is causing problems, your child will be offered treatment which can take the form of transfusions or injections to boost blood cell production.

- Gallstones: These are small stones that form in the gallbladder as a result from the breakdown of red blood cells. This doesn't always need treatment, but the gallbladder may need to be removed in a minor operation if the stones are causing problems.

- Making bone marrow outside the bones: The bone marrow is where blood cells are made. With very severe anaemia the body tries to compensate by making extra bone marrow in other areas of the body, which can cause pain by pinching nerves or putting pressure on organs. This is usually treated by transfusions, or sometimes surgery, or sometimes low dose radiotherapy.

What is the treatment for CDA-1?

Blood transfusions can be used to treat the anaemia experienced in CDA-1. Depending on how severely people are affected, they may require transfusions:



If severely affected, children with CDA-1 may need transfusions from birth or even in the womb. They will need transfusions every 3-4 weeks throughout their lives. Since there is lots of iron in red blood cells, receiving blood transfusions can lead to excess iron in the body. This needs to be removed, to stop it building up in the organs. The removal of the excess iron is done by a process called iron chelation. This involves using medicines which bind to the iron, so more is excreted in the urine and faeces.

If moderately affected, people with CDA-1 don't usually need transfusions, except if they become unwell for another reason. This group of people are more likely to develop an enlarged spleen (an organ in the abdomen), because the spleen tries to compensate for the lack of red blood cells which are being produced in the bone marrow. This can be uncomfortable, and can cause low levels of immune cells (white blood cells), but it is not dangerous.

Some moderately affected people may get treated with a protein called interferon, which is given as a weekly injection under the skin. Interferon helps to increase the production of haemoglobin. Haemoglobin is what makes red blood cells red, and it's the molecule that helps to carry oxygen around in the blood. These patients may also need iron chelation to remove excess iron, as their body tends to absorb too much iron from their diet.

If mildly affected, people with CDA-1 don't need transfusions. They may get treated with interferon. They are unlikely to get severe iron overload and need chelation, or if they do it will only be for a short time.

People with CDA-1 may be affected differently at different times in their lives. For example, your child may not need regular transfusions until they reach puberty, when a growth spurt puts extra demands on the body.

Its also beneficial for CDA-1 patients to receive the hepatitis B vaccine and vitamin supplements for folic acid – this could be something to discuss with your haematologist.

It's important to manage CDA-1 patients with a holistic approach, which considers all aspects of life. Often people with more complex cases will be managed by a variety of health care professionals, for example haematologists, psychologists, dieticians etc, in the hopes to achieve the most effective management outcome.

What does the future hold for my child with CDA-1?

Getting diagnosed is only the first hurdle. Because it's so rare, often GPs and hospital consultants won't know anything about it. It's also rare to meet anyone else who has the condition. Finding specialist care and support can make all the difference. Often patients with more complex cases will be managed by a variety of health care professionals, for example haematologists, psychologists, dieticians etc, in the hopes to achieve the most effective management approach.

Everyone is affected so differently so it can be difficult to get accurate information about what is likely to happen to any particular person. But if the condition is well-managed with transfusions and iron chelation, having CDA-1 does not mean giving up on living a full and active life.

Where to find support - *CAN (congenital anaemia network)*

CAN is a charity for people with inherited anaemias and their families. It was started by a group of parents and doctors, working together to tackle some of the problems of having an inherited anaemia. It aims to give people with CDA-1 accurate information about their condition and advice about where to find specialist care.

CAN runs family days for people to meet each other, as well as providing support and services for patients who need that extra bit of help.

If you want to know more visit www.togetherwecan.uk or email us on info@togetherwecan.uk

Yes we can

CONGENITAL ANAEMIA NETWORK