Iron overload without blood transfusions:information for patients and parents

Can I really have too much iron if I have never had a transfusion?

Yes. This surprises almost everyone, because it sounds like a contradiction.

You can be anaemic and have too much iron at the same time. The two are not opposites. In several inherited anaemias, the anaemia is the reason for the iron overload.

Why does iron build up?

In many inherited anaemias, the bone marrow is working extremely hard but not succeeding. It makes red blood cells that are faulty and that die before they ever reach the bloodstream, or soon after. Doctors call this ineffective erythropoiesis – erythropoiesis means making red cells, and here it is happening but not working.

The body notices that there are not enough red blood cells in circulation. It cannot tell that the problem is faulty manufacturing rather than a shortage of raw materials. So it does the sensible thing for the wrong problem: it turns up iron absorption from food.

The result is that iron keeps arriving, keeps not being used properly, and keeps accumulating – for years, without a single transfusion.

Who does this affect?

This kind of iron overload is seen in thalassaemia intermedia, the congenital dyserythropoietic anaemias, inherited sideroblastic anaemia including ALAS2 mutations, pyruvate kinase deficiency, and other rare red cell enzyme and membrane conditions.

It builds up more slowly than transfusional iron overload, but over a lifetime it can reach levels that cause real harm.

Why does it matter if I feel fine?

Iron overload causes no symptoms for many years. By the time it does, damage has often already occurred. This is why your team will check for it before there is any sign of a problem, and why they may want to treat it while you still feel perfectly well.

Over time, untreated iron overload can cause liver damage and scarring, heart problems, diabetes and other hormone problems, joint pain, and delayed growth or puberty in children.

How is it monitored?

Serum ferritin is a blood test that estimates stored iron. It is easy to repeat, so it is checked regularly, but it is less reliable in non-transfusional iron overload than it is in people who are transfused – it can read lower than the true amount of iron in the liver. For that reason a ferritin in the normal range does not always mean all is well.

Transferrin saturation measures how much iron is circulating in the blood.

MRI scans measure iron in the liver and heart directly. These are much more accurate than ferritin, and importantly, liver iron and heart iron do not always go up and down together – you can have a reassuring liver scan and still be accumulating iron in the heart. This is why heart MRI is done as well, usually every one to two years, and more often if the level is high. The scan is painless and does not use radiation (however, for some people with severe claustrophobia it can be difficult to sit through. Ferriscans measure only iron in the liver and T2* MRIs measure iron in the liver and the heart.

Liver function tests and hormone tests are also checked periodically as well as looking for diabetes (eg fasting glucose test). This is because There is no scan that can measure iron in the pancreas or the glands, so the only way to monitor is see if these are working normally is measuring their function.

How is it treated?

The aim is to bring iron down and keep it from causing organ damage.

Iron chelation therapy uses medicines that bind iron and remove it from the body through urine or stool. The medicines used are deferasirox (a once-daily tablet), deferiprone (a tablet or liquid taken three times a day), and desferrioxamine (an infusion under the skin). Our leaflet on iron overload from blood transfusions describes these in more detail.

An important difference from transfusional iron overload is that chelation here may not be lifelong. Some people need it for a period of months, or in courses, rather than continuously, and it can sometimes be stopped once iron levels have come down and then restarted later if they rise again. Your team will base this on your scans and blood tests.

Venesection removing a unit of blood (in the same way as donating) is a simple way to remove iron in some conditions. It is only possible if the anaemia is mild enough to tolerate it, so for many people with inherited anaemias it is not an option. It is worth asking whether it is one for you.

Treating the underlying anaemia can reduce iron absorption at its source, because it turns down the signal that is driving the body to absorb more. Where a treatment exists for the underlying condition, this is part of the picture.

Never take iron supplements or multivitamins containing iron unless your haematologist has specifically told you to. This applies even though you are anaemic. Iron tablets will not correct this kind of anaemia, and they add to a load your body cannot clear.

If another doctor prescribes you iron – which happens easily, because anaemia usually does mean iron deficiency – it is worth checking back with your haematology team before starting.

What can I do to help myself or my child?

Attend the monitoring, including scans, even while you feel well.

Ask what your ferritin and scan results are, and whether the plan is watchful monitoring or active treatment. In non-transfusional iron overload the plan often changes over time, so it is a fair question to ask again periodically.

Be careful with alcohol, which adds to liver strain.

Vitamin C supplements can increase the iron released into the blood, so ask your team before taking them. Fruit and vegetables in a normal diet are fine.

Read supplement labels. Iron is added to many over-the-counter products that are not sold as iron tablets.

Iron overload is manageable. People who chelate consistently can expect their iron levels to come down and their organs to be protected.

When to seek medical advice

Contact your team if you notice:

- Increasing tiredness that is different from your usual
- Abdominal pain, especially on the upper right side
- Joint pain
- Palpitations or breathlessness
- Increased thirst or passing a lot of urine
- In children or teenagers: delayed growth or delayed puberty
- That another doctor has prescribed you iron

Can we help?

CAN (Congenital Anaemia Network) is a charity specifically for patients with inherited anaemias. It was started by a group of patients and doctors, working together to tackle some of the problems of having an inherited anaemia – the lack of information and the loneliness of not knowing anyone else with your condition. We run events for patients to meet each other, as well as provide some additional information, 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: info@togetherwecan.uk

Do you know your care should involve a specialist centre for inherited anaemias? These conditions are rare enough that few doctors see many of them and all patients with rare inherited anaemias should be under the care of a haemoglobinopathy specialist. Please read our 'How red cell services are organised in England' information sheet. If you don't know who your specialist service should be, please get in touch with us so we can help you find out.