Children's haematology

Children and sickle cell disease

Information for patients, relatives and carers

What is sickle cell disease (SCD)?

SCD is a lifelong genetic disorder of the body's red blood cells. Genes are a small part of DNA. This means that people are born with SCD and it is passed on from parent to child.

It is caused by an abnormality in haemoglobin (the part of red blood cells that carries oxygen round the body), which makes the red blood cells become inflexible and sticky. This then makes other blood cells, and the vessels they travel, in sticky. Blood flow becomes interrupted, and inflammation happens.

This process can cause pain and problems in the organs and may affect the immune system. In SCD, this can happen suddenly and is often painful. These are known as sickle cell crises (sickling). The long-term effects of crises and background sickling (that may have no symptoms) are known as chronic complications. Treatment involves trying to stop sickling from happening as much as possible.

If your child has a sickle cell disease, it means that they have inherited two abnormal haemoglobin genes from you and your partner. One of these genes has to be a sickle gene. The other gene can be a sickle gene, a beta thalassaemia gene or another abnormal haemoglobin gene, such as haemoglobin C, haemoglobin D, haemoglobin O Arab.

Carriers of sickle cell have one sickle cell gene only and the other haemoglobin gene is normal. If both parents are carriers, the chance that their child will have SCD is one in four for each pregnancy. If one person has the disease and the other is a carrier, the risk is greater – one in two for each pregnancy.

We will discuss this in more detail with your child when they enter their teenage years. This is to make sure they understand the risk of their children inheriting SCD too.

How common is SCD?

SCD is the most common inherited condition in the world and mostly affects people whose family origins are in the Middle East, Sub-Saharan Africa, parts of India and parts of the Mediterranean.

Between 12,000 and 15,000 people in the UK have SCD and just fewer than 300 babies are born with the condition in this country every year.

Parents of all babies born in the UK are given the opportunity to have their baby tested for SCD in the first week of life (the heel prick test).

What kind of problems can happen in SCD?

- infection
- gallstones
- joint damage or destruction
- loss of spleen function (making your child prone to infections)
- damage to kidneys
- leg ulcers
- visual impairment
- neurological damage which can lead to stroke
- priapism (a painful erection of the penis lasting more than 30 minutes)

The signs and symptoms of crisis may include:

- fatigue (feeling tired or weak)
- pain
- jaundice (yellowing of the white of the eyes)
- paleness of skin, or inside the mouth or eyes
- dizziness
- headaches
- symptoms of infection, such as fever

What to do in an emergency

When you get to A&E, tell the receptionist your child has SCD. Please go to the Emergency Department (A&E).

If you go to an A&E in another hospital and are admitted, we might be able to transfer your child to St. Mary's Hospital, depending on how unwell they are. Though we are always happy to support other teams, we would not be able to manage your care at another hospital.

Note: If your child has an infectious illness, such as cough, cold, diarrhoea, vomiting or chicken pox, please inform the A&E team as soon as you arrive.

If you think your child's condition may be life threatening **call 999**.

If you develop any of the following symptoms, please seek medical attention straight away:

- Temperature: 38°c or above
- Shortness of breath or difficulty in breathing
- Feeling very unwell
- Abdominal pain
- Pain that cannot be controlled with your usual painkillers
- Pain that is more severe than your child's usual sickle pain, is different from their usual sickle pain, or both
- Limb weakness, particularly if felt more on one side of their body than the other, dragging of feet
- You think your child is much more anaemic than usual.
- Priapism – a painful erection of the penis lasting more than 30 minutes

For a non-urgent medical problem or a medical problem unrelated to SCD, for example a rash or a twisted ankle, you should contact your GP or take your child to your local Emergency Department (A&E). If necessary, they can contact us.

What can trigger a sickle cell crisis?

- infection
- stress
- dehydration
- cold or hot weather or sudden changes in temperature
- nothing at all – sometimes crises happen out of the blue

Treatment

A simple painful crisis is when your child has pain but is otherwise well. You may be able to manage it at home by giving your child painkillers, fluids, allowing them to rest and keeping them warm.

However, if the pain becomes worse your child may need stronger painkillers in hospital to help control the pain and treat the cause and any complications relating to their condition.
Sometimes your child may feel unwell from the start of the crisis even if it's not very painful. This is not a simple painful crisis. If your child is unwell even if they have no pain, you should always contact us so that we can see if they need treatment or are developing any problems.

We might also suggest other ways of managing their condition with preventative treatments. We usually recommend this if the frequency of sickle cell crises increases or if an episode was particularly severe. These may include starting hydroxycarbamide or a long-term blood transfusion programme. Some people have also had a bone marrow or stem cell transplant that is a cure for sickle cell disease.

Gene therapy is another treatment option currently used in clinical trials. Gene therapy can treat sickle cell disease by removing your child's own cells, and changing them to add or turn on non-sickle haemoglobin or to help faulty genes work properly. Then they are put back (re-infused) into your child. We hope that this will offer a cure to more patients in the future.

How to manage pain at home

Get to know how sickle cell affects your child. This helps you to recognise the early signs that a sickle crisis is starting. Get them to rest, keep warm, take painkillers, and keep them hydrated. Taking a warm bath or applying a heat pad to the painful area may help. Use distraction, such as listening to music, doing puzzles or watching a film.

Be mindful of the kind of pain your child is experiencing. Is it their usual sickle crisis pain, chronic pain or non-sickle pain?

If the pain becomes worse, they may need stronger painkillers in hospital so it's important for you to contact us using the details on page 7 of this leaflet. We can help to control the pain in hospital and treat the underlying cause (trigger) and any complications relating to their condition.

Medications

Medications your child will need to take at home are:

- Penicillin V or erythromycin prophylaxis antibiotics
- Folic acid for development of the red blood cells
- Painkillers such as paracetamol, ibuprofen for pain when required

In addition, they might need to take:

- Hydroxycarbamide to manage sickle cell
- Tilefrine for management of priapism
- Deferasirox (Exjade®)
- Deferiprone (Ferriprox®) for iron overload
- Desferrioxamine (Desferal) an infusion pump inserted under the surface of skin for iron overload

Other medications should be prescribed by your GP.

Helping your child stay well

There are things you can do to help your child stay well and prevent sickle cell crises.

It's important that:

- They take all their medications and follow their instructions.
- Your child drinks enough fluids and has a good balance of nutrition and activity.
- Infections are treated quickly.
- Their vaccinations are up to date.
- Your child is warm at all times. If they go swimming, make sure you dry them off quickly.
- You help your child to develop strategies to cope with life's ups and downs. We know that stress is an important factor in sickle cell disease.
- You come to all your child's clinic appointments so that we can review their health and monitor them for any complications that may occur.

Hydroxycarbamide, has been offered to people who have problems with sickle cell. A recent study, known as 'baby HUG', demonstrated good effects and tolerability of hydroxycarbamide in babies. Because of this, we now offer parents/carers the option to start hydroxycarbamide as a preventative treatment before any problems start.

Attending your child's clinic appointments is a requirement stated in the National Standards produced by the Sickle Cell Society together with the Department of Health and Social Care. These appointments are every three months until your child is two years old, and then every six months. Please note that if your child is taking hydroxycarbamide, we will see them every two to three months. If there are any problems or complications, we will see your child more often. If your appointment is inconvenient, please let us know, and we will do our best to reschedule.

Travel advice

Check if any travel vaccines are needed for the country you are travelling to. Arrange these through your GP practice or a travel clinic at least six weeks in advance. If you are visiting an area that requires yellow fever vaccination and are currently taking the drug hydroxycarbamide please talk to your hospital specialist, as we might recommend that you don't have it.

For more information, please see our leaflet: Vaccinations, medication and travel, for children with thalassaemia and sickle cell disease. If you don't have this, let us know.

Protect your child against infection

Stay up to date with their routine immunisations including these extras:

- Pneumococcal polysaccharide vaccine (PPV23)- given every 5 years against pneumococcus infections
- Haemophilus influenzae type b (Hib) and meningococcal C
- Meningococcal ACWY conjugate
- Influenza (given every year)

Carry a vaccine record card. You can get one of these from your GP practice.

Transfusion and iron chelation

Due to complications of SCD, some patients need regular blood transfusions or red cell exchanges. Blood contains iron. A gradual build-up (accumulation) of iron in the body is called iron overload.

Iron cannot be removed by the body naturally, resulting in complications affecting the heart, the liver, the endocrine glands (adrenal and pancreas). If allowed to keep building up, it will cause serious complications.

Regular blood tests and scans are done to monitor and detect when the iron levels are raised and require treatment.

We use medication to remove iron from the body to reduce and prevent damage to organs, this is called iron chelation therapy.

There are three types of iron chelators, which can be used alone or combined. These are:

- Deferasirox (Exjade®), taken as tablets by mouth
- Deferiprone (Ferriprox®), taken as tablet by mouth
- Desferrioxamine (Desferal®), given by an infusion pump inserted under the surface of the skin

Transition

As your child gets older, we will support them to take more responsibility for their health. This is to make sure that they can look after themselves when they leave home to live independently.

It's as important for them to learn the skills needed for a day-to-day adult life, such as paying bills or working, as it is to be able to navigate their own health needs. This process is called transitioning and we will help and guide them throughout that time. Transitioning happens gradually over their teenage years so that by the time your child is 18, they will feel confident in:

• Knowledge about their condition

• Attending clinic appointments on their own if so they wish

• Ordering their medicines

• Organising their vaccinations.

As your child gets older, they may want to talk to the team looking after them in private. They may have questions they would like to ask but may feel uncomfortable discussing them with you. This is normal and we will be happy to see them on their own.

Contact details

- **Haemoglobinopathies admin team:** 020 3312 6157

  Haemoglobinopathies clinical nurse specialist team (09.00 to 17.00): 07795 651 153
  imperial.paediatrichaemoglobinopathies@nhs.net

- **Paediatric haematology day unit (PHDU):** 020 3312 5080/5081

- **Paediatric site practitioner:** 07824 625 419

- **Address:** Imperial College Healthcare NHS Trust, St Mary's Hospital, Praed Street, London W2 1NY

The team

- Dr Kirstin Lund, consultant paediatric haematologist,
- Professor Josu de la Fuente, consultant paediatric haematologist
- Dr Adam Gassas, consultant paediatric haematologist
- Dr Toni Petterson, consultant paediatric haematologist
- Dr Leena Karnik, consultant paediatric haematologist
- Miss Catherine Mkandawire, matron (Haemoglobinopathies and apheresis children and young people)
- Miss Kajal Rai, clinical nurse specialist (Haemoglobinopathies children and young people)
- Ms Nancy Huntley, clinical nurse specialist (Apheresis children and young people)
- Miss Ristell Fernandes, clinical nurse specialist (Apheresis children and young people)
- Mr Renzo Tiong, clinical nurse specialist (Apheresis children and young people)

Further information

https://www.imperial.nhs.uk/our-services/adolescent-and-young-adult-healthcare---11to25

https://www.readysteadygo.net/

Sickle Cell Society

020 8861 7795
sicklecellsociety.org

UK Thalassaemia Society

020 8882 0011
office@ukts.org
ukts.org

Sickle Cell and Young Stroke Survivors (SCYSS)

0800 084 2809 or 020 7277 2777
info@scyss.org
scyss.org

Contact

The charity for families with disabled children
https://contact.org.uk/

Family Fund

https://www.familyfund.org.uk/

Grants are available for families raising a child or young person with a long-term disability, disabling condition or life-limiting illness, and living on a low income.

The Hope Project Scotland

07756 266691
via website
thehopeprojectscotland.org.uk

Citizens Advice Bureau

https://www.citizensadvice.org.uk/

The Care Information Exchange (CIE)

CIE gives secure online access to information from the hospital such as test results, appointments and letters. Young people and their parents can also add things like symptoms the young person is experiencing to give a more complete record. To register and access the record QR code below to download the application.

Everyone registering will need to complete a form, show proof of identification, and have an email address. The email address will be the login name, and the address for notifications when something new is added to the record. Go to www.careinformationexchange-nwl.nhs.uk/ or scan this QR code with your phone camera to find out more and register:

How do I make a comment about my visit?

We aim to provide the best possible service and staff will be happy to answer any of the questions you may have. If you have any **suggestions** or **comments** about your visit, please either speak to a member of staff or contact the patient advice and liaison service (**PALS**) on **020 3312 7777** (10.00 – 16.00, Monday to Friday). You can also email PALS at imperial.pals@nhs.net The PALS team will listen to your concerns, suggestions or queries and is often able to help solve problems on your behalf.

Alternatively, you may wish to complain by contacting our complaints department:

Complaints department, fourth floor, Salton House, St Mary's Hospital, Praed Street
London W2 1NY

Email: ICHC-tr.Complaints@nhs.net

Telephone: **020 3312 1337 / 1349**

Alternative formats

This leaflet can be provided on request in large print or easy read, as a sound recording, in Braille or in alternative languages. Please email the communications team: imperial.communications@nhs.net

Wi-fi

Wi-fi is available at our Trust. For more information visit our website: www.imperial.nhs.uk

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