



The Royal College of Pathologists

Pathology: the science behind the cure

Split Your Genes Online Workshop

Video Transcripts

[Dr Shubha Allard – Consultant Haematologist](#)

I'm Shubha Allard. I'm a consultant haematologist working with NHS Blood and Transplant which is the National Blood Service in England.

NHS Blood and Transplant collects and provides around 2 million units of red cells and components for the management of patients with many different disorders in hospitals across the whole of England. And this includes of course, sickle cell anaemia patients.

Not all patients with sickle need transfusion. They have a so called chronic or ongoing anaemia which they in fact tolerate very well. But some patients with sickle will become severely anaemic and then they need a top-up transfusion whereby blood needs to be given to them through a drip in their arm.

Other patients with sickle may need exchange transfusion where sickle blood is replaced by non-sickle blood. Now this could be in an emergency, for example in an acute sickle chest crisis, or this could be in an ongoing basis where patients may need to go on to an exchange program. Say for example, for the treatment or prevention of stroke in relation to sickle.

Now patients with sickle cell anaemia are much more likely to form red cell antibodies than other patients who need transfusion. So, this means that we have to select and match the blood for patients with sickle even more carefully.

NHS Blood and Transplant is working very hard to get enough blood donors so we can provide safe, adequate and the right blood to meet the needs of all patients. And here we need more blood from donors from a black and/or ethnic minority background so we make sure that we had enough blood to manage patients with sickle.

NHS Blood and Transplant also has clinical and laboratory teams that work closely with hospitals to provide expert help in the management of patients with sickle across the whole country.



Dr Cath Booth – Consultant Haematologist

My name is Dr Cath Booth. I'm a consultant haematologist working with NHS Blood and Transplant and Barts Health NHS Trust in East London.

NHS Blood and Transplant is the organisation in England involved with collecting, processing and then distributing blood products for transfusion and organs for transplant. They also do a lot of research into new treatments and processes and that includes getting the best blood products for patients like those that sickle cell disease.

My work involves working with hospitals to make sure they use blood products wisely and choosing the best blood products for people with special requirements or people who've had reactions to transfusions. I also do a lot of work around education of patients, the public and other clinicians about blood transfusion.

Sickle cell disease is a genetic condition where there's a single base pair change in the DNA and that causes a change in the haemoglobin molecule in the red blood cells so that when the oxygen levels fall it forms polymers which are long stiff rods and those deform the red blood cells, so they make the crescent sickle shape. That makes the red blood cells fragile, so they tend to break down. And they can also block the tiny capillaries in the body, and that can cause tissue damage and a variety of health complications.

Patients with sickle cell disease can benefit from a blood transfusion, both when they're acutely unwell and in the long term to prevent the complications. Part of my job is ensuring we get the best blood for them.



Dr Rachel Carling – Consultant Biochemist

Hello my name is Rachel Carling and I'm a consultant biochemist at Thomas' Hospital. I'm the clinical lead for biochemical sciences and I'm also director at the southeast Thames regional newborn screening laboratory.

Newborn screening is offered to every baby born in the UK. The screening test is done on the blood spot sample which is collected from a heel prick on day five of life. Every baby is tested for nine different disorders and one of these disorders are sickle cell disease. The reason we screen babies with sickle cell disease is because affected babies are at risk of life-threatening infections but if we can identify the babies with sickle-cell disease in the first few weeks of life we can offer them prophylactic antibiotics and refer them into specialist clinical care.

So, my lab screens around 16,000 babies each year and our job is to test blood spots samples for the presence of haemoglobin and its variants. We measure haemoglobin A, F, S, C, D, E and O and we do this by a technique called tandem mass spectrometry.

If we find a baby with haemoglobin S present or one of the other variant haemoglobins, we call this a condition suspected result and we test the sample again using a different method. We test it with high performance liquid chromatography to confirm the screening result.

Last year we identified 52 babies with sickle cell disease and 1321 carriers.

I'm now going to pass you over to my colleague Yvonne Daniel who will you a little bit more about the screening program.

Dr Yvonne Daniel – Chemical Pathologist

Hello my name is Yvonne Daniel and I'm the lead scientist in the special haematology laboratory here at Guys' and St Thomas's Hospital. And I'm also a scientific adviser to the Public Health England Sickle Cell and Thalassaemia Screening Program.

Public Health England provides standards, guidance and oversight for the linked antenatal and newborn screening programs in England.

In addition to newborn screening, antenatal screening in early pregnancy is offered to all woman and depending on the results obtained to the biological father of the baby.

As Rachel has said, screening babies saves lives by ensuring that they have the right treatment at the right time. Last year this laboratory identified 52 babies with sickle cell disease but globally it has been estimated that every year between 150 to 300 thousand babies are born with sickle cell disease. Those estimates show a huge variation and in reality, we do not know the true number.

This is because there is little or no screening in much of the world and in particular in sub-Saharan Africa and India where the numbers are highest.

There are now projects which involve experts like me hoping to change this and the aim is that in the future, babies will be identified and get the right treatment just as they do here in England no matter where they are born.



Dr Sara Trompeter – Consultant Haematologist

Hi, morning! My name is Sarah Trumpeter. I'm a haematologist and I'm off to work today. This is the Cancer Centre and this is where I'll be doing a haematology clinic this morning.

So, this is the inside the Cancer Centre. It's pretty quiet today because of coronavirus keeping people at home unless they absolutely need to be here. So, I'm going to go up the lift and see you up there.

Here's the waiting area where people will wait to see us once they've checked in. I'm now going to take you further on to see some other clinical areas before I start clinic.

So, this is the apheresis unit and this is where we do red cell exchanges or apheresis procedures for other patients. There's the lovely nursing staff getting all the machines ready. Aren't you, Clarissa?

So, this is the clinic room. We've got a bed so I can examine patients and there's emergency kit on the wall which you can see. Suction, oxygen, that's a sharps box that yellow thing. You can wash your hands which is always a good thing. And a bin. That is a bench which actually has my scarf on it but usually has medical students on it. And see that? Lots of coffee!

And a computer. We are very lucky at my hospital. We've got, if you see that thing left of the computer, that is a voice recognition software. Anyhow, so that's my clinic room. So soon, I'm going to look at the clinic list and invite the first patient in.

Here on the ward round and Ashley who's one of our junior team, is going to tell me a bit about how our patient's doing.

So, our patient had her red cell exchange overnight. She had 12 units. The procedure went successfully. We've sent off her blood tests this morning. Some of them are back. Some are not, so we're just waiting for those. I've spoken to the lab to make sure they're all in process. We're now going to see our patient in the cubicle.

Just finished ward rounds. All the patients have plans. The junior team go off and enact those plans. Some will be going home. Some need to be staying in the hospital. Some have had changes in their medications. But they've all got plans. Some of them will get reviewed later on today.

End of a long day. I have driven home which is unusual for me because I usually take public transport but I haven't been doing during the pandemic. So, I'm now home and I'm looking forward to seeing my family after a long day at work. It's, what time is it? Quarter past 7. Okay hopefully my husband and sons have made me dinner.



June Okochi – Sickle Cell Society Representative

Hi, my name is June. I live in London and I work for the National Health Service. I was diagnosed with sickle cell at the age of one. Actually, just before I turned one. I was an infant.

The way sickle cell affects me is predominantly, like most people, I get pain. Sometimes quite intense pain that requires me to go into hospital for treatment. I also have some complications as a result of sickle cell. So, retinopathy of the eyes and avascular necrosis in my hips which will require me to have a hip replacement. I'm also prone to infections and have suffered a number of pneumonias actually.

So, there are some stigmas around sickle cell. Some of which are, you know, sickle cell is a black disease, people who live with sickle cell are drug-seeking people and obviously that if you live with sickle cell you cannot have a good quality of life. And even some around early mortality.

As debilitating as the disease can be, with the right support around people living with the disease, whether it's the best care, proper treatment, regular monitoring, holistic therapies, I can assure you that people living with sickle cell do have a better quality of life and are not always in hospital.

If they are actually being supported in the right way. I currently receive absolutely amazing support from my haematology team and I get blood exchange transfusion every six weeks as a form of treatment, which has really supported me to stay well and do all the amazing things I like to do.

The way I am involved in the sickle cell community is through volunteering for the Sickle Cell Society, fundraising and also mentoring children and young people living with sickle cell in East London.

I hope you can learn a thing or two and good luck. Thank you.

Courteney Mclune-Calvin – Sickle Cell Society Representative

Hi, my name is Courteney. I'm 22 years old. I'm a Sickle Cell Society Representative. I volunteered with them and I am a mentee within their mentor program, and have been for about a year and a half.

So, as a sickle cell sufferer, what I'd like to talk to you about today is one of the common misconceptions to do with the sickle cell disease. And that is the pain levels that we experience. People tend to think that the pain we go through is always high intensity like, not being able to walk or you're incapacitated for a number of hours.

And although that does happen, for me, that happens a bit more rarely. And I tend to go through more minor pains. So that can be like hand cramps or a series of shooting pains throughout the day. But the only difference is this can happen every single day.