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Sickle cell disease is spreading through the UK

It's one of the most under researched diseases prevalent in the UK and it is spreading throughout the country



Judy Hobson

Pamela Gyebi-Ababio, 18, first realised she had sickle cell disease when she started primary school. "I was always cold and had to wear socks and tights and I remember the head teacher picking on me for this in front of the whole school. It was horrid and I cried. As a result my parents moved me to another school."

When she was eight, dancers from the Royal Ballet School came to the school to encourage more children to dance. "I loved dancing and got into the final of a competition for a place at the Royal Ballet School but had to pull out," says Pamela.

"The pain starts at my waist, goes down my legs and back up again. It feels as though my thighs are being stabbed with a knife and that someone is chiselling away inside my shins while someone else is hammering them from the outside. I think my legs are about to snap in two."

Pamela is one of thousands of children and adults in the UK living with sickle cell disease, a condition that puts sufferers at increased risk of stroke and damage to their vital organs.

There are estimated to be between 12,500 and 15,000 people in the UK with the disease and more than one in nine of the population are carriers. In the main, sickle cell is found in people whose families come from the Caribbean, Africa, the eastern Mediterranean, the Middle East and Asia, but carriers are also being found in the white population as a result of intermixing many generations ago.

Although numbers are very low, they indicate that sickle cell disease can no longer be regarded as confined to specific sections of the population and highlight the importance of antenatal and newborn screening. The NHS's newborn screening programme, which has covered the whole of England since 2006, shows that sickle cell is increasingly present anywhere in the country and not just confined to areas of London and the West Midlands, where Afro-Caribbean families have traditionally settled. There are, however, still high concentrations in certain London boroughs such as Lambeth, Lewisham and Southwark where there are an estimated 3,000 cases, 600 of them children.

Disturbingly, the National Confidential Enquiry into Patient Outcome and Death (NCEPOD), published in May this year, found that of 19 sickle cell patients who had complained of pain and subsequently died in hospital, nine had been given excessive doses of painkillers and five had died because of complications owing to overdose.

The Archbishop of York Dr John Sentamu, who is calling for an all-party parliamentary group to look into sickle cell disease, says: "Although sickle cell is the most common genetic disorder in the country, recognition and treatment varies widely and is woefully inadequate in some areas. The NCEPOD survey found that many cases were not fully supported by experienced medical staff, and heartbreakingly this has led to deaths that could have been avoided. This is unacceptable. I will shout from the rooftops to ensure that everyone with sickle cell gets better care." It tears apart Pamela's mother Hannah, 45, when she sees her teenage daughters racked with pain, in hospital on morphine drips during a crisis. In addition to monthly five-hour-long blood transfusions, Pamela now has to struggle with what is known as iron chelation therapy to rid her body of iron overload, the cumulative effect of the transfusions. Left untreated, the excess iron could damage her heart, liver and endocrinal glands. It means that Pamela has to wear a cumbersome portable infusion pump for at least eight hours a day, five days a week, to deliver desferrioxamine, a drug that helps to prevent a toxic build-up of iron.

Hannah, a care worker from Coulsdon, Surrey, says: "I feel so guilty for putting my child in this situation. I hate to see her suffering. I shed a lot of

tears watching them connect the device to her belly to remove the excess iron. She finds it very uncomfortable and has to hold on to it whenever she moves because it's so heavy, she is frightened she might pull it out."

There is, however, a new drug for iron overload that could make Pamela's life easier - a daily tablet taken in a glass of water. Called Exjade, it costs about £20,000 a year, twice the cost of her desferrioxamine treatment. Pamela's haematologist, Dr Mary Clarke, is so convinced that the new drug would improve the teenager's quality of life that she is making a strong case for her to get it: "I hope that within two months Pamela will get Exjade, but like all drugs, it has side-effects and so her liver and renal functions will have to be closely monitored."

Sickle cell disease has already cost Pamela a place at the Royal Ballet School and put her a year behind at Nonsuch High School for Girls, Cheam, Surrey. Yet she is determined to do her maths, chemistry and biology A levels to become a pathologist researching Aids. She says: "It would be wonderful to learn more about sickle cell but I wouldn't be happy doing that because I live with it every day and would forever be checking myself. I hate being ill, so when I can I try to think about something else."

In spite of her condition, the teenager has put her name down to work during the holidays as a volunteer at St Helier's University Hospital, Carshalton, the hospital she attends as a patient. However, she can only work on "good days".

The day I spoke to her she should have been at an open day at Imperial College, London, but was unable to attend because of her illness. She says: "I couldn't go because last night I felt feverish and my eyes were yellow, a telltale sign that I was getting ill again. I tried to pretend I was OK but my mum knows just by looking at me when I am about to have another crisis."

Hannah, who also has three younger daughters, had no idea that she was a sickle cell carrier until she was screened at Newham Hospital when she was 15 weeks pregnant with Pamela.

She says: "I spent the rest of the pregnancy praying my baby would not be a sickler. When she was born she looked healthy but at three months her hands and legs began to swell and she cried the whole time.

"Growing up in Ghana I didn't know anything about sickle cell. My mother died when I was 10 and I have a gut feeling she had it. More information should be given during antenatal screening. Although I was told I was a

carrier, I had no idea what sickle cell was. If I had known more about the disease, it would not have come as such a shock when I had to start dealing with it.

“Pamela and I have discussed relationships and I have told her before she falls in love to make sure the boy is not a carrier. If he is and she can't stop her feelings, then instead of having children of their own, they should adopt.”

Jacqueline Cox, 39, is another mother whose daughter suffers from sickle cell disease. Her eldest daughter, Alicia, 15, walks with a limp, the result of a stroke when she was 9. Children with sickle cell are at increased risk of stroke.

Every day Alicia, from East Dulwich, southeast London, is tired all the time, struggles to get out of bed and has pain somewhere in her body. At least three times a year she is admitted to King's College Hospital, southeast London, for up to two weeks at a time when she has a crisis and the pain becomes unbearable.

It means that her mother, who has two sons and three other daughters, lives “on a knife's edge” wondering what each day will bring. The strain, she says, is never-ending.

Alicia's crises started when she was 3 and have increased as she has grown older.

Jacqueline, who is of Caribbean descent, says: “She doubles over, is unable to walk and cries out in pain. She is back and forth to hospital three or four times a year, put on a morphine drip and can be up to two weeks in hospital.”

Although Jacqueline's brother died from sickle cell at 3 when she was 10, she did not understand the implications. She has since found out that there was a lot of sickle cell on her father's side of the family. She and Alicia's father, who now lives in Jamaica, were not screened before she became pregnant with Alicia. Jacqueline says: “In those days you weren't screened during pregnancy. Even if I had been I wouldn't have fully understood what sickle cell meant and would still have gone ahead with the pregnancy. “What you need is a sickler to go in and talk to girls in secondary school before they get pregnant, telling them how serious and debilitating this disease can be. They should be made aware of what can happen if both they and their boyfriends have the trait.”

One problem facing Alicia is that when she is not experiencing a crisis, she

looks like a normal, healthy person. As a result her teachers, schoolfriends and sometimes her sisters forget how overwhelmingly tired the condition makes her feel.

Jacqueline says: "People see a normal person in front of them and think she is being lazy because they don't understand the nature of the illness. At primary school she sometimes fell asleep during lessons and the teacher would get angry. I tried to explain that it was because her body was so tired as a result of the anaemia."

Alicia "is up and down all night", finding it impossible to get to sleep during a crisis. Jacqueline does her best to wake her and get her up for school, even though this frequently means that she and her sisters are late. Her attendance is only 50 per cent.

Jacqueline says: "She is highly intelligent but cannot attend lessons often enough. I have tried to get work sent home but the education people don't seem to understand why she can't go to school. In an ideal world there would be mediators to help families like ours. They could explain to everyone what having sickle cell means."

In 2004, the NHS started rolling out its antenatal sickle cell screening programme - due to cover the whole of England by the autumn - to see if expectant mothers carry the gene. If they do, their partners are called for screening, and if both are found to have the gene, they are offered counselling and the option of a termination.

But as Iyamide Thomas, the Sickle Cell Society's regional care adviser for South London, says: "There is still something of a stigma attached to sickle cell and some men are reluctant to be screened."

Ideally, she would like to see preconception screening introduced, because then couples would know whether they risk having a child with sickle cell before the woman becomes pregnant and can make an informed decision about what to do.

Lack of public awareness of this disease means that the Sickle Cell Society managed to raise only £300,000 last year and is struggling to survive. This sum is a drop in the ocean compared with that received by other niche charities such as the Multiple Sclerosis Society, which received £31 million; the Cystic Fibrosis Trust (£11.5 million) and the British Society for Rheumatology (£2.68 million).

At the Sickle Cell Society's invitation, a multidisciplinary group of professionals and patients has drawn up the UK's first standards for the

clinical care of adults with sickle cell disease with the aim of addressing inequalities in care and treatment.

These recommend that preconceptual counselling, including genetic screening and partner testing, should be offered to all men and women with sickle cell disease.

Norman Lamb, the Liberal Democrats' spokesman on health, and Dawn Butler, Labour MP for Brent South, pledged to set up an all-party parliamentary group for sickle cell disease.

Launching the new standards at the House of Commons last month, the Archbishop said: "They have the potential to improve the lives of the thousands of people in the UK and are the first step in raising our game."

Sickle cell disease: the facts

What is sickle cell disease? It is an inherited blood disorder where the red blood cells that transport oxygen from the lungs to the rest of the body contain an abnormal haemoglobin - haemoglobin S. This causes the cells to change in texture and shape, becoming sickle-like in appearance, making it difficult for them to pass through small blood vessels. The cells die prematurely, leaving sufferers with chronic anaemia.

What is a crisis? This occurs when these sickle-shaped cells get stuck in blood vessels, causing excruciating pain in the arms, legs, chest and abdomen that can last from a few hours to weeks. This can lead to stroke, blindness, and, over time, damage to the liver, kidneys, lungs, heart and spleen.

Who gets it? If both the mother and father have the sickle gene, there is a one in four chance that their child will have the disease. If only one of them has the gene, then the child will not have sickle cell disease but can be a carrier, and may pass the gene on to his or her children.

For more info log on to:

www.sicklecellsociety.org;
www.sct.screening.nhs.uk