The Person with Sickle Cell Disease

Dr Felix I.D Konotey-Ahulu (photo left) continues his medical theme.

People suffer from hereditary rheumatism when two ‘Ache’ genes are inherited together. ‘S’ from mother and ‘S’ from father gives "SS" disease (also called "Sickle Cell Anaemia" - Ache/Ache).

There is no such thing as 'SC Trait'. When one uses the term 'Trait', a Haemoglobin-A gene must be present in the phenotype. Sickle Cell Trait is 'AS Trait', with the proportion of 'A' greater than 'S'. Indeed, the proportion of the abnormal Haemoglobin-S in Sickle Cell Trait is always less than 40%, and can be as little as 20%. 

Signs and symptoms

The "signs and symptoms" described below apply to persons with the disease, not trait:

Hand-foot syndrome

"Doctor, my child's tummy is getting bigger, and he complains of pains". Sickled red cells sometimes collect in the spleen (left side of the abdomen) or in the liver (right side of the abdomen), or both. When this happens suddenly, it constitutes a form of sickle cell crisis which makes the child very pale (judged by the nails, palms, and within the lower eyelids). This so-called Sequestration Syndrome with much of the child's blood collecting in the spleen and liver, can be very dangerous. Again, an infection in the body, with the child not eating or drinking properly can set this up. Parents are taught to feel the child's tummy gently to see if these organs are getting suddenly bigger, and to seek medical attention.

When this happens in the tiny vessels of the hands, it results in the 'hand-foot syndrome'. What parents need to know is that there is always a precipitating cause of this "sickle cell crisis" (red cells sickling within the body). Dehydration, for example from lack of drinking due to severe sore throat, and high body temperature from any infection (including malaria) can cause 'hand-foot syndrome' in a child with sickle cell disease.

There is urgent need to replace lost fluid and treat the infection.

When a child with sickle cell disease has no problems, and is quite happy running about and playing, that means the red cells have not altered their shape to sickle-shape, and are moving freely round the body. The child is said to be "in the steady state".

But if circumstances like teething or immunisation or infections, produce fever and cause a significant proportion of the red cells in the body to change from round to sickle shape and clog up blood vessels and start sudden rheumatic and bone pains, then he/she is said to be "in sickle cell crisis".

To give only pain-killers in severe sickle cell crisis and to forget the cause of the crisis and fluid replacement is to court disaster.

Sequestration Syndrome

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It is amazing how many parents have been taught to educate their doctors and nurses: "I think my child may be getting the sudden big spleen syndrome. She has been off colour recently."

As these circumstances are almost invariably the cause of a sickle cell crisis, parents should be watchful and anticipate problems. Daily oral Penicillin has been known to prevent not only frequent sore throats and many sickle crises, but also the sequestration syndrome.

Folic acid tablets (5 milligrams daily) helps to replenish the "blood factory". Those who live in the tropics or visit there on holidays should...
remember that, contrary to what they hear from some scientists, malaria is the commonest cause of sickle cell crisis in Africa! You had better believe me, and not those scientists!

Priapism
"Doctor, my son sometimes gets erection of the penis and it takes hours and hours before it settles". Wherever red cells change in the body from round to sickle shape, expect to have some problems. If this happens in the vessels of the penis, then persistent erection could occur. Priapism is the medical term for it, and it can be most distressing, especially in the older person where the tendency is to hide the problem and not tell the nurse or even the doctor about it.

Again, what are the circumstances for this problem? It starts most often after midnight, waking the person up. Sometimes a urinary tract infection needs to be treated before it disappears. On other occasions, dehydration requires to be corrected.

In the older person who arrives in hospital several days after priapism had begun, successful treatment occurs with rehydration and partial exchange blood transfusion.

A drop in the oxygen content of the blood during the night, I suspect, could be another factor, hence priapism occurs most often after midnight.

Snoring reduces arterial oxygen content, so patients need examination by an ear/nose/throat specialist to make sure the tonsils (which are often huge in sickle cell disease patients) are not obstructing the airways.

I have known some young men cut short a priapism attack by getting out of bed and walking about to drive sluggish blood round the body, or even taking a bath.

Sickle Cell Anaemia
"Doctor, I get easily tired when I play football or swim". Well, the 'SS' phenotype is known as 'Sickle Cell Anaemia'. Because sickled red cells are often removed from the blood by the spleen, the number of red cells in the body is constantly reduced, producing anaemia.

The haemoglobin level in the 'steady state' in Sickle Cell Anaemia is seldom above 9 grams per deci-Litre of blood (Normal value is between 13 and 15 g/dL). The 'SS' person's body tissues adjust beautifully to cope with a steady-state Hb of 9 g/dL, but this is not enough to make Kwesi or Yomi exercise his football skills without getting out of breath. They should stick to swimming and running.

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The 'hand-foot syndrome': Swollen, painful hands are usually the first indication of Sickle Cell Disease

impediment. She was rushed to Nairobi Hospital. Fortunately she lived, but things could have been much worse.

As 'Genius' genes were inherited from her parents alongside their 'S' genes, Ruth managed to educate her Left hand to be used in computer work in which she presently excels. Her speech has recovered, though she walks with a paralytic gait.

If the vessel blockage had occurred on the Right side of the brain, Ruth would have been paralysed on the Left side of the body, and her speech would not have been affected, being a right-handed person.

Enough teaching for this month, but test yourself:

1. Can an 'AS Trait' suffer from hand-foot syndrome?
2. Does Sickle Cell Trait mean 'SC' phenotype?
3. Is there such a person as 'SC' Trait?
4. Can malaria kill a Sickle Cell Disease child suddenly?
5. What circumstances can cause a stroke in a Sickle Cell Disease person?
6. Why are pain-killers not necessarily the most important requirement for sickle cell crisis when the patient first arrives in hospital?
7. What is priapism?
8. How can snoring affect the wellbeing of the sickle cell disease person?
9. Can a Sickle Cell Trait person have only 20% of Haemoglobin-S?
10. The Sickle Cell Test on Dr Konotey-Abulu's mother was POSITIVE, but on his father's blood the test was NEGATIVE. How was it possible for 3 of their 11 children to have suffered from hand-foot syndrome?

NB: One in 3 West Africans reading this article have the 'AS' or 'AC' haemoglobin phenotype and are probably not aware of this.

The KAGE Foundation, and the University of Cape Coast, Ghana, have undertaken to help Africans and their Diaspora relatives to identify their haemoglobin phenotype, so that they can ascertain the chances of Sickle Cell Disease in their offspring. This will help us all in taking informed decisions about procreation and family size limitation.

If you yourself are among the 2 in 3 West Africans who are Norm/Norm (AA), but would nevertheless like to help others identify their phenotype, please contact the "Kweyer Aggrey Distinguished Professor of Human Genetics, Faculty of Science, University of Cape Coast, Ghana", mentioning how many people you could sponsor for laboratory tests and genetic counselling.

Those in the UK may find that their Family Practitioner may not agree to do the Sickle Cell Test plus Haemoglobin Electrophoresis. Most doctors outside Nigeria and Ghana have never heard of Haemoglobin-C, so they will not ask for Haemoglobin Electrophoresis to identify the 'AC' Trait person.

If the GP is not prepared to do this test, then West Africans in the UK should tell their doctor they are willing to pay for the tests themselves. Had they better not spend some money to find out their phenotype, than stand amazed when their first, second, or third child begins to suffer hand-foot syndrome at 18 months of age?

(Readers could write to me, care of New African, or direct to me at konotey-abulu@sicklecell.md or at my website: www.sicklecell.md)